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COMPROMISED AFFECT AND LEARNING ASSOCIATED WITH CROUZON SYNDROME – A CLINICAL CASE STUDY

by

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This dissertation is dedicated to all those who made this research project possible.

‘We trust in God’

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DECLARATIONS

I, Björn Opper (99099332), hereby declare that **COMPROMISED AFFECT AND LEARNING ASSOCIATED WITH CROUZON SYNDROME – A CLINICAL CASE STUDY** is my own original work and that all the resources that were consulted are included in the reference list.

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Björn Opper

September 2006

I, Rika Opper, hereby declare that I undertook the editing of the grammatical and language aspects of this dissertation.

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SUMMARY

COMPROMISED AFFECT AND LEARNING ASSOCIATED WITH CROUZON SYNDROMME – A CLINICAL CASE STUDY

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DEGREE: MAGISTER EDUCATIONIS
(Specializing in Educational Psychology)
DEPARTMENT: EDUCATIONAL PSYCHOLOGY
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Several reported case studies suggest that *Crouzon Syndrome* is characterized by a specific pattern of lesioned brain areas, which led to the exploration of how these structural brain lesions relate to the learner's affective, social, and cognitive behaviour. However, these case studies were reported from highly specialized neurological and genetic perspectives, with no attempt at merging the triad, i.e., specific brain circuitry, learning and affect. This research project thus aims to describe compromised affect and learning associated with *Crouzon Syndrome* on the basis of a clinical case study, and of three critical inferences based upon a review of relevant literature. The first inference is that a craniofacial condition such as *Crouzon Syndrome* might be classified as a pervasive developmental disorder, since the brain is not fully developed at the time of diagnosis, while the concept *pervasive* suggests that these impairments significantly affect individuals throughout their lives. The second inference implicates a neural substrate to compromised learning associated with *Crouzon Syndrome*, therefore the expectation is that individuals diagnosed with *Crouzon Syndrome* might show a particular profile of compromised learning. As a result of specific lesioned brain areas, the third inference is that compromised affect associated with *Crouzon Syndrome* is delicately entwined with compromised learning. Human cognitive behaviour and emotions involve specific and delicately intertwined brain-operating systems, and it can be expected that the same brain-operating systems also underlie compromised affect associated with *Crouzon Syndrome*. Therefore, this project aims to scrutinize the neural makeup of *Crouzon Syndrome* based on a clinical case study, in order to compile a detailed explanatory profile of

compromised affect and learning associated with *Crouzon Syndrome*. This is done using a mixed-method approach which involves both quantitative and qualitative research.

Key words: Crouzon syndrome, compromized learning, compromized affect, neural condition, genetic disorder, fresh mutation, atypical brain development.

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CHAPTER 1

AWARENESS OF THE PROBLEM, RATIONALE FOR THE STUDY AND ANALYSIS OF THE RESEARCH PROBLEM, PROBLEM STATEMENT, PURPOSE AND AIM OF THE STUDY, THEORETICAL FRAMEWORK AND PARADIGMATIC PERSPECTIVE, RESEARCH DESIGN AND METHODOLOGY

1.1 INTRODUCTION

This research project aims to describe compromised affect and learning associated with *Crouzon Syndrome* on the basis of a clinical case study, and of three critical inferences based upon a review of relevant literature. The first inference is that a craniofacial condition such as *Crouzon Syndrome* might be classified as a pervasive developmental disorder, since the brain is not fully developed at the time of diagnosis, while the concept *pervasive* suggests that these impairments significantly affect individuals throughout their lives (Barlow & Durand, 2002:464). The second inference implicates a neural substrate to compromised learning associated with *Crouzon Syndrome,* therefore the expectation is that individuals diagnosed with *Crouzon Syndrome* might show a particular profile of compromised learning. As a result of specific lesioned brain areas, the third inference is that compromised affect associated with *Crouzon Syndrome* is delicately entwined with compromised learning. Human cognitive behaviour and emotions involve specific and delicately intertwined brain-operating systems, and it can be expected that the same brain-operating systems also underlie compromised affect associated with *Crouzon Syndrome*. Therefore, this project aims to scrutinize the neural makeup of *Crouzon Syndrome* based on a clinical case study, in order to compile a detailed explanatory profile of compromised affect and learning associated with *Crouzon Syndrome*.

1.2 AWARENESS OF THE PROBLEM

During my internship training this year I am stationed at a school for neurally impaired learners in Pretoria. The outcome of various consultations with learners, their parents and

teachers directed me towards a neuropsychological explanation for compromised learning, behaviour and affect that are particularly prevalent within this specialized school setting. One of the learners referred to me was diagnosed with *Crouzon Syndrome* at the age of two years, due to early fusion of the cranial sutures, causing a misshapen (egg shaped) head.

She is currently nine years old and is experiencing significant delays in learning and affect. I proceeded with in-depth reading in order to support the learner's parents and teachers with accurate information on *Crouzon Syndrome* and related learning and affective difficulties. Several reported case studies suggested that *Crouzon Syndrome* is characterized by a specific pattern of lesioned brain areas, which led me to further explore how these structural brain lesions relate to the learner's affective, social, and cognitive behaviour. However, case studies were reported from highly specialized neurological and genetic perspectives, with no attempt at merging the triad, i.e., specific brain circuitry, learning and affect. I discussed these limitations with my supervisor, and we concluded that a highly focussed research project based on a clinical case study might significantly expand the existing knowledge base. I then consulted with both the parents and the learner and explained my particular interest in *Crouzon Syndrome* in order to obtain their informed consent to proceed with such a research project. Keeping in mind that the parents or the learner might later reconsider involvement, I discussed the advantages and disadvantages of such an endeavour with the parents and the learner, upon which I was granted informed consent for this research project.

1.3 ACTUALITY AND ANALYSIS OF THE RESEARCH PROBLEM

Crouzon syndrome was first described by a French neurosurgeon, O. Crouzon, in 1912. *Crouzon syndrome*, with a reported incidence of 1:25000 births, is the most common of more than 70 conditions in which premature fusion of the cranial sutures may be a feature (Singer, Walpole, Brogan & Goldblatt, 1997:11), suggesting a neural substrate to compromised affect and learning. Premature synostosis may be evident at birth or develop during the first year of life and is completed by the second or third year (Cohen, 2000:362). The sutures affected include the coronal, the sagittal, and occasionally the lambdoidal sutures. According to Kummer (2001:92), the major feature of Crouzon syndrome is craniosynostosis, usually involving the coronal sutures. Premature fusion usually results in an abnormal head shape and, in severe cases, this condition could also cause increased

pressure on the developing brain. Compensatory growth occurs at the remaining open sutures to allow continued postnatal brain growth, causing midfacial hypoplasia, shallow orbits, a foreshortened nasal dorsum, maxillary hypoplasia, occasional upper airway obstruction and brachycephaly, which is characterized by a short skull (Chen, 2006:2). If the sagittal suture is involved, the lateral growth of the skull will be prevented, resulting in frontal bossing and scaphacephaly, where the skull appears oblong from front to back. However, when multiple sutures are involved, this may result in asymmetry of the skull, which is referred to as plagiocephaly (Kummer, 2001:91). From the preceding it follows that the prenatal brain might have developed in a typical fashion, but that atypical postnatal brain development, due to intracranial pressure, might result in compromised affect and learning, consequently prenatal brain development will not feature strongly within this research project, and the main focus will be on atypical postnatal brain development and plasticity.

Cerebral cortex development involves an intricate progression of precisely timed events, resulting in finely tuned neural circuitry. This circuitry mediates the integration of sensory information that underpins cognitive functions of diverse nature (Berger-Sweeney & Hohmann, 1997:121). Typically, developing cortical circuitry is dependent on the synchronized generation, migration and differentiation of neurons and glia (Kolb & Whishaw, 2003:610). In the presence of *Crouzon syndrom*e this typical cortical development is disrupted, which might have profound and long-lasting influences on the individual's affect and learning (Humphreys, Kaufmann & Galaburda, 1990: 727-738).

> 'Synchronizing these developmental events is essential so that nerve cells from different parts of the brain assemble at the appropriate times and places to form functional circuits. Considering the complexity of events in the ontogeny of cortical circuitry and the complexity of the functions that will later be mediated by this circuitry, it is not surprising that these events are exquisitely vulnerable to disruptions during the developmental period' (Berger-Sweeney & Hohmann, 1997:121-122).

Having read the research conducted on autism, Claassen (2006:86) suggests that human brain development follows a programmed continuum, during which programmed development might be disrupted at any stage. This statement is also applicable to Crouzon

syndrome as a developmental disorder, despite diagnostic differences between autism and Crouzon syndrome. According to Kolb and Whishaw (2003:609) impairments resulting from neural insults can be studied in three ways. The first approach is to examine nervous system maturation and to correlate maturation with specific impairments. The basic premise of this approach is that, as the brain structures develop and mature, their associated functions emerge and manifest as observable behaviours. Some structures develop quickly, and associated behaviours become evident quickly, as opposed to some structures and associated manifest behaviour that develop at a slower rate, e.g. the frontal lobes, which continue to develop well into adolescence. Inferences about impairments can thus be drawn in terms of typical neural development.

The second approach, the converse of the first, is to consider a developing child's impairments and then make inferences about neural maturation, in other words, because we observe new abilities emerging at a later developmental stage, we infer that these abilities must be controlled by later-maturing neural structures (Kolb & Whishaw 2003:610).

The third approach to studying the relation between neural development and impairments is to identify and study factors that influence both (Kolb & Whishaw 2003:610). This approach relies on the study of brain structures, as well as on experiences, where the concept 'experience' refers to pre- and postnatal incidents such as neurochemical disruptions, cortical lesioning and/or the developmental effects of atypical genetic coding. For purposes of this research project, the latter approach will apply. Kolb and Whishaw (2003:610) explains this approach as follows:

> 'Logically, if behaviour is influenced by one of these experiences, then structures in the brain that are changed by that experience are responsible for the behavioural outcomes. For example, we might study how an abnormal[1] secretion of a hormone affects both a certain brain structure and a certain behaviour. We can then infer that, because the observed behavioural abnormality results from the abnormal functioning of the brain structure, that structure must normally play some role in controlling the behaviour.'

[1] In this report the concepts 'abnormal' and 'normal' are being replaced by the concepts 'typical' and 'atypical', except when excerpts are quoted directly. In this sense the concept 'typical' refers to typical programmed neural development, while 'atypical' refers to atypical or disrupted programmed neural development.

Crouzon syndrome might be viewed as a closed-head injury, which causes intracranial pressure that affects brain functioning, by disrupting blood supply and producing scarring of brain tissue (Camfield, Camfield & Cohen, 2000:177; Kummer, 2001:133; Kabbani & Raghuveer, 2004:2868; Shprintzen, 2000:200). Two kinds of behavioural effects result from closed-head injuries, namely discrete impairment of the specific functions mediated by the cortex at the site of the lesion, and more generalized impairments from widespread trauma throughout the brain. Behavioural effects include loss of complex cognitive functions, including reductions in mental speed, concentration and overall cognitive efficiency (Kolb & Whishaw 2003:703). Closed-head injury might also significantly impact on personality and social behaviour (Levin, Benton & Grossman 1982:12), and this implicates compromised affect. Levin *et al*., (1982) suggested that recovery of social skills and personality, areas which often change significantly, is less favourable with closed-head injuries, suggesting the pervasive nature of compromised affect with Crouzon syndrome. To further illustrate this pervasive nature, Levin *et al*., (1982) also demonstrated that these individuals' quality of life in respect of social interactions, perceived stress levels and enjoyment of leisure activities is significantly reduced after closed-head injury, and that this reduction is chronic.

In keeping with the aim of this research project, which is to investigate the precise relationship between developmental brain insults associated with *Crouzon syndrome* and compromised affect and learning, my first research hypothesis is that compromised affect and learning observed in the clinical case study might be related to atypical cortical architecture and connectivity resulting from closed-head injury. My second research hypothesis is that an individual diagnosed with *Crouzon syndrome* might meet a specific profile of compromised affect and learning. To best examine this phenomenon, one could consider to first explore research findings on closed-head injury, as well as models and experimental manipulations which simulate lesioned cortical architecture and circuitry, and which interfere with programmed postnatal cortical development, resulting in compromised affect and learning. The reason why programmed postnatal cortical development is considered here, is because Berger-Sweeney and Hohmann (1997:122) demonstrated that postnatal manipulations affect a nervous system that is not fully developed and might consequently alter its developmental course, which includes cortical

neurogenesis, gliogenesis, cell migration and differentiation, dendrite and axon growth, formation of synapses, pruning and formation of myelin.

In keeping with the three critical inferences offered in the introduction of this proposal, analysis of the research problem in terms of the link between atypical cortical development and associated compromised learning will be dealt with first, followed by analysis of the research problem in terms of associated compromised affect. By first appreciating some of the features of normal cortical development, one could better understand the effects of closed-head injury associated with *Crouzon syndrome* that might alter postnatal cortical development.

Cortical development takes place through a series of overlapping phases (Teicher, Andersen, Polcari, Anderson, & Navalta, 2002:397), and these phases can simplistically be divided into seven discrete developmental time windows, namely cell birth (neurogenesis; gliogenesis), cell migration, cell differentiation, cell maturation (dendrite and axon growth); synaptogenesis (formation of synapses), cell death and synaptic pruning, followed by myelogenesis (formation of myelin) (Kolb & Whishaw, 2003:611). For purposes of this research project cortical development during gestation will be reviewed only briefly, with the main focus on those developmental time windows that stretch beyond gestation into peri- and postnatal phases, because the effects of *Crouzon syndrome* are likely to be most prominent during these developmental phases. The following general principles emerged from available studies, which provide insights into functional abnormalities following early postnatal brain insult.

1.3.1 Cell birth (neurogenesis and gliogenesis)

In humans, most cortical neurons are generated in the proliferative zone near the cerebral ventricles during early and middle parts of gestation (Rakic & Caviness, 1995:1101-14). The laminar structure of the cerebral cortex is formed by the migration of successively generated neurons along radial glial scaffolding (Berger-Sweeney and Hohmann, 1997:123). An inside-out layered pattern is formed, whereby neurons that are generated first, settle, whereas neurons generated subsequently push past the older neurons. This process is called neurulation (Bayer, Altman, Russo & Zhang, 1993: 83). After neurulation subsequent processes such as cell migration and differentiation, dendrite and

axon growth, synaptogenesis, programmed apoptosis and myelogenesis start after gestational day 28, while glial and synapse formation continue until approximately three years of age (Schmid & Rotenberg 2005:4). While neurogenesis (production of new neurons) ceases in most brain regions at birth, stem cells continue throughout life to generate neurons and glial, even in an aging brain, at least within the olfactory bulb and the hippocampal dentate gyrus (Eriksson, Perfilieva & Bjork-Eriksson 1998:1313; Teicher *et al.*, 2002:398; Kolb & Whishaw 2003:612). This cell division is programmed, but in *Crouzon syndrome* cell division might be suppressed due to intracranial pressure following premature fusion of the cranial sutures, resulting in abnormal brain size (Clark, 2002:5) or a misshapen head (Abou-Sleiman, Apessos, Harper, Serhal, & Delhanty, 2002:307-308; Kummer, 2001:91). From birth to five years of age, the brain triples in mass, and this gain in brain size is mostly ascribed to the vigorous myelination of fiber tracts (Teicher *et al*., 2002:398). In humans myelination peaks during the third trimester of gestation and continues into the young adult years. After birth the brain increases in mass coinciding with growth spurts (Kolb & Whishaw 2003:620). Elbert, Heim & Rockstroh (2001: 191-204) report on consistent spurts in brain growth from 3 to 10 months (accounting for an increase of 30% in brain weight by the age of 1½ years), as well as between ages 2 to 4 years, 6 to 8 years, 10 to 12 years, and 14 to 16+ years. Brain weight increases by about 5% to 10% in each of these 2-year periods, most likely due to the growth of glial and synapses. "Although synapses themselves would be unlikely to add much weight to the brain, the growth of synapses is accompanied by increased metabolic demands, which causes neurons to become larger, new blood vessels to form, and new astrocytes to be produced" (Kolb & Whishaw 2003:620).

Prolonged synapse formation, continued generation of neurons, glia and astrocytes, as well as programmed postnatal myelination of fiber tracts, account for the developing brain's longer period of vulnerability associated with *Crouzon syndrome* (Coleman 1994:107). Although compensatory growth at the remaining open sutures allows some continued synapse formation, generation of neurons, glia and astrocytes, as well as myelination of fiber tracts and consequent brain growth (Chen, 2006:2), these processes are typically inhibited and slowed down in *Crouzon syndrome*. Research suspects that disrupted glial, astrocyte and synapse formation, inhibited cell migration, and poorly myelinated fibre tracts, with a specific focus on the olfactory bulb and the hippocampal dentate gyrus, might contribute significantly to compromised affect and learning associated with *Crouzon*

syndrome. In addition, with *Crouzon syndrome* the initial two growth spurts, i.e. 3 to 10 months, and 2 to 4 years, are interrupted, and less glia, synapses and astrocytes might be produced, each playing a significant role in postnatal learning.

1.3.2 Cell migration

At about 4½ months of gestation the neuroblasts that eventually form the cerebral cortex have reached completion, but cell migration continues and extends up until about eight months after birth (Kolb & Whishaw 2003:613). Migration takes place along radial glial in a process known as *radial migration* or by *somal translocation* (Clark 2002:7). During migration the deepest layer of the cortical plate migrates and deposits prior to the formation of subsequent layers, suggesting that the first migrating neurons are predestined to form layer VI, followed by neurons of layers V, IV, III, II, and I respectively. The cortex therefore is formed in an inside-out fashion, as described above (Clark 2002:7). When programmed migration is interrupted prematurely, a group of cells that belong in an outer cortical layer will remain scattered among inner cortical layers. Because cell migration extends into the postnatal period, *Crouzon syndrome* might interrupt this process, causing the formation of cortical layers to be faulty. These disorders of migration can be identified by means of MR-imaging, and faulty migration in humans is usually associated with dyslexia (Kolb & Whishaw 2003:614), in keeping with Kummer's (2001:133) observation of reading impairment among individuals diagnosed with *Crouzon syndrome*.

1.3.3 Cell differentiation and cell maturation (dendrite and axon growth)

When migrating neurons have reached their final cortical regions and differentiated into specific neuron types, these neurons start to grow dendrites to extend synaptic surface areas and to expand neural circuitry (Kolb & Whishaw 2003:614). This process involves dendritic arborization (or branching) and dendritic spine growth, on which most dendritic synapses take place (Kolb & Whishaw 2003:614). Cell differentiation and cell maturation commence during gestation, but during the postnatal phase and during childhood marked expansion of axonal and dendritic arborization take place, with rapid increase in synaptic contacts (Teicher *et al.*, 2002:398). Axons grow at the rate of a millimeter per day, whereas dendrites grow at a daily rate of only one micrometer. This dissimilar growth

allows the axon to influence dendritic differentiation (Kolb & Whishaw 2003:615). In the case of Crouzon syndrome, the formation of these neural circuitry might be disrupted, resulting in the failure of axons to reach programmed targets (Courchesne 2004:106; Schmid & Rotenberg 2005:5). Cerebellar granule cells and Purkinje cells might be deficient (Tatter, Galpern & Isacson 1995:286-297; Panksepp 1998:114; Bailey, Luthert, Harding *et al*., 1998:880-905), accompanied by other intracranial anomalies such as agenesis of the corpus callosum and hindbrain herniation (Kabbani & Raghuveer, 2004:2868; Kummer, 2001:92), which relate to compromised learning of specific nature.

1.3.4 Neuronal modification by selective depletion

The process of synaptogenesis, cell death and synaptic pruning is known as *neuronal modification by selective depletion* (Teicher *et al*., 2002:399), which refers to the genetically programmed overproduction of synapses, followed by elimination of synapses based on programmed cell death and synaptic pruning, also known as apoptosis. The early embryonic phase is characterized by low-density synaptogenesis, but synapse formation continues until approximately two years of age. During the postnatal phase synaptogenesis is accelerated, i.e. synapses are generated at about 40 000 synapses per second, after which a plateau is reached, followed by the elimination of synapses through synaptic pruning (apoptosis) (Kolb & Whishaw 2003:616; Teicher *et al*., 2002:398). It follows that within the enclosed cranial chamber, brought about by premature fusion of cranial sutures, accelerated synaptogenesis and synaptic pruning might proceed in an atypical fashion.

1.3.5 Myelogenesis (formation of myelin)

Myelogenesis is a prolonged process that starts during gestation and continues in the CNS through childhood and into adulthood. Different regions are myelinated differentially, for example, myelination of the vestibular system takes place prenatally, while tertiary association areas (tertiary PTO cortices) are myelinated during middle childhood and adolescence (Schmid & Rotenberg, 2005:1). These tertiary association areas thus show prolonged vulnerability, and are likely to be affected by *Crouzon syndrome*. Tertiary association areas mediate complex activities such as language, planning, memory, and attention (Kolb & Whishaw, 2003:65), which closely relate to compromised learning.

Considering the enclosed cranial chamber that denotes *Crouzon syndrome*, maturation of the frontal regions might also be at risk, especially the prefrontal cortex, since motor systems myelinate at an early age, whereas the prefrontal cortex only myelinate during adolescence (Teicher *et al*., 2002:398). The frontal gray matter volumes increase 20% between early childhood and the end of childhood (Carper, Moses, Tigue & Courchesne 2002:1038), while cerebral white matter volumes increase 59% between the second or third postnatal year until adolescence is reached (Courchesne, Karns, Davis, *et al*., 2001:245). Maximum brain volumes are not reached until about 10 to 12 years of age (Courchesne, 2004:109). Cerebral white matter volumes continue to increase through middle childhood and adolescence (Courchesne *et al*., 2000:672), and the corpus callosum shows continued development throughout childhood into adulthood (Pujol, Vendrell, Junquae *et al*., 1993:71). Since myelination significantly increases the tempo of information exchange (Teicher *et al*., 2002:398), research suggests that individuals diagnosed with *Crouzon syndrome* might reveal reduced tempo of information exchange, coupled with impaired associative learning, language, planning, memory, and attention.

The preceding review of cortical development and associated postnatal behaviour emphasizes the importance of each of these developmental phases (Berger-Sweeney & Hohmann 1997:123), making these phases most relevant to the study of compromised affect and learning associated with *Crouzon syndrome*.

1.4 LITERATURE REVIEW AND RATIONALE FOR THE STUDY

Based upon the preceding typical brain development, it is essential to synthesize biological, psychological, and neurological perspectives on compromised affect and learning associated with Crouzon syndrome. Various disciplines have contributed towards a synthesised conceptualization of learning and affect. The various *cognitive sciences* eloquently explain human learning (Eysenck, 2003:5), but the cognitive sciences do not adequately address the subservient affect to human learning, which is also neurally based. For example, the *cognitive neuropsychological approach* involves studying patterns of cognitive impairment shown by brain-damaged patients in order to understand normal human cognition (Eysenck, 2003:5), whereas the *cognitive neuroscientific approach* involves using several techniques for studying brain functioning (e.g., brain scans) in order to identify the processes and structures used in cognition (Eysenck 2003:5). For purposes

of this research project, elements characteristic of both of these two approaches will be employed, in other words, principles explaining brain damage, and relevant techniques to explore processes and structures used in cognition. In addition, the *neuropsychological approach* aims to diagnose and explain the presence of cortical lesioning and to localize it where possible at hand of specific assessment media (Kolb & Whishaw, 2003:756), which also closely reflects the aim of this research project. By also employing the *affective neuroscientific* approach of Panksepp (1998), this research project seeks to combine several elements of more than one approach to learning and affect respectively, in order to adequately explain compromised affect and learning associated with Crouzon syndrome, as it will become clear from the following paragraphs. The contribution of some of these approaches will be discussed under the heading that deals with the embedded paradigm; yet these approaches are mentioned here in order to illustrate the blurred distinctions and the complexities inherent to a suitable rationale.

Considering the sites of potential lesioning included in the foregoing discussion of the research problem, these sites of potential lesioning form part of the justification of and motivation for this research project.

A study of saccadic eye movements demonstrated the link with frontal systems circuitry (Reichle, Pallatsek, Fisher & Rayner, 1998). Therefore it follows that if Crouzon syndrome negatively impacts on frontal lobe development (Kummer 2001:91), saccadic eye movements will also be affected. Reichle *et al*., (1998:125-157) illustrated the important role of saccadic eye movements in reading and learning. These researchers argue that the next eye movement in reading is programmed after only *part* of the processing of the currently fixated word has occurred. Completion of lexical access to the currently fixated word produces a shift of covert attention to the next word. This greatly reduces the time between completion of processing on the current word and movement of the eyes to the next word. Any spare time is used to start processing the next word. According to Eysenck (2003:259) this model specifies the major factors determining eye movements in reading, and illustrates that reading occurs on a word-by-word basis, and that parafoveal processing increases the efficiency of the reading process. In addition, stem cells' prolonged generation of neurons and glia within the olfactory bulb might be affected by Crouzon syndrome, implicating that vision might also be compromised (Eriksson, Perfilieva & Bjork-Eriksson, 1998:1313; Teicher *et al*., 2002:398; Kolb &

Whishaw 2003:612). Faulty migration in humans is also associated with dyslexia (Kolb & Whishaw, 2003:614), in keeping with Kummer's (2001:133) observation of reading disability among individuals diagnosed with *Crouzon syndrome*. Considering the important role that vision plays in learning, these findings provide justification for research into compromised learning associated with Crouzon syndrome.

Research conducted by Kabbani and Raghuveer (2004:2868) and Kummer (2001:92) demonstrated agenesis of the corpus callosum and hindbrain herniation, which relate to compromised learning of specific nature. Bauman and Kemper (1995:1-26) provided evidence that explains the link between disconnection syndrome and learning, especially between cerebellar and limbic zones with other higher brain areas. Due to the role of the limbic system in affect, these findings also justify research into compromised affect associated with Crouson syndrome. In keeping with a disconnection syndrome, neural systems that should be working in close unison appear not to have developed normal synaptic interchange in various brain areas that control socialization, communication, and imagination (Panksepp, 1998:113).

The reduction in the total cross-sectional area of the corpus callosum observed relative to total brain volume may indicate a decrease in interhemispheric connectivity (Kolb & Whishaw, 2003:67). Higher-level cognitive functions, such as language and linguistic processes, depend upon hemispheric specialization, as the callosal pathways are involved in integrating processes embedded in affect and learning, particularly when one considers the importance of cerebral asymmetry in functions such as language.

There have also been reports of deficits in executive function and spatial working memory (Bauman & Kemper, 1994), all relating to compromised affect and learning. Furthermore, temporal lobe abnormalities are implicated in compromised explicit memories (i.e. memories for daily events), whereas cerebellar abnormalities are implicated in implicit memory (i.e. skills and conditioned responses) (Kolb & Whishaw, 2003:658). In the limbic system, the hippocampal complex, subiculum, entorhinal cortex, amygdala, mammillary body, anterior cingulate gyrus, and septum are connected by neuronal circuits, and it is suspected that the brains of individuals diagnosed with Crouzon syndrome might show reduced neuronal cell size and increased cell-packing density in these areas due to intracranial pressure.

The prefrontal cortex, the amygdala, the superior temporal sulcus and the insular cortex form part of the neural network underlying affect and social cognition (Panksepp, 1998:272). Panksepp (1998:272) theorizes that specific regions such as the cingulate gyrus, septal area, bed nucleus of the stria terminalis, preoptic area, dorsomedial thalamus and the periaqueductal gray (PAG) all play an important role in social cognition and social bonding. In addition, Eriksson *et al*. (1998:1313) and Teicher *et al*. (2002:398) provided evidence towards a compromised hippocampal dentate gyrus and limbic system in Crouzon sydrome, which findings justify exploration of compromised affect. The limbic system, particularly the amygdala, plays a crucial role in behavioral responses to emotional stimuli and in emotional learning (Du Preez, Naudé & Pretorius, 2004:27; Naudé, Pretorius, Van Schoor & Becker, 2005:47; Pretorius, Naudé & Pretorius, 2005:310). Amygdalar damage impairs recognition of emotional faces (Adolphs, Tranel, Damasio *et al*., 1994:669) and has been implicated in an impaired ability to link visual perception of emotionally relevant stimuli (Adolphs, Sears & Piven, 2001:232).

From an African perspective one can argue that Crouzon syndrome and the resulting craniosynostosis can easily be overlooked due to a lack of resources and technology. Therefore a profile of resulting compromsied affect and learning can be valauble in supporting affected individuals.

From the preceding it is clear that recent research findings are supporting and justifying investigation into the compromised affect and learning associated with Crouzon syndrome.

1.5 DEFINITION OF KEY CONCEPTS

1.5.1 Compromised learning

For purposes of this research project the concept ‘compromised learning’ encompasses two distinct concepts, namely ‘learning disability’ and ‘learning impairment’. Not all learning problems are learning disabilities or impairments. According to Lerner (2003:7-9), learning disabilities can be defined as a group of heterogeneous disorders manifested by significant difficulties in the acquisition and use of listening, speaking, reading, writing, reasoning, or mathematical abilities. These disorders are intrinsic to the individual and are presumed to be due to central nervous system dysfunction resulting in conditions such as

perceptual handicaps, brain injury, minimal brain dysfunction, dyslexia, and developmental aphasia. Problems in self-regulatory behaviours, social perception, and social interactions may exist with learning disabilities, but on their own do not constitute a learning disability. Furthermore, the term excludes individuals who experience learning problems as a result of hearing, visual, motor, intellectual impairments, emotional disturbances and extrinsic influences such as cultural differences, insufficient instruction or economic disadvantage (Dednam, 2005:364).

The neuroscience perspective on the concept *learning impairment* or *learning disability* is that all learning originates within the brain, and therefore learning disabilities are presumed to be related to a central nervous system dysfunction. Due to developments in neuroscience and medical research, there is growing evidence that learning disabilities have a neurological basis and are caused by impairment in brain function (Lerner, 2003:11).

The concept *learning impairment* suggests a lower level of functioning than before, due to some cortical lesioning of specific nature. Considering that Crouzon syndrome only manifests as the brain continues to develop, there is a postnatal time window characterised by an absence of atypical neural functioning, before scarring sets in, and during which time window the child shows typical development in the absence of any disabilities. Thus, during this time window the mastery of basic developmental skills is not compromised; yet, when Crouzon syndrome starts to manifest, the condition contributes to sensory, motor, affective and cognitive impairment or deterioration, and mastery of basic developmental skills become compromised. In order to conceptualize the decline from typical development and neural functioning to atypical neural functioning due to scarring, the concept *impairment* best describe this decline, while the relative permanence of the condition is implicated by the concept *disability*. Thus, by merging the embedded meanings of *learning disabilities* and *learning impairments* into the concept *compromised learning*, one comes to understand that there is a decline to lower levels of neural functioning, and that this decline is of relative permanent nature.

1.5.2 Compromised affect

For purposes of this research project the concept *compromised affect* is preferred to 'emotional difficulty', since the concept *compromised affect* signifies that the child's current functioning is on a lower level than premorbid functioning due to neurological lesioning associated with Crouzon syndrome. The concept *compromised affect* implicates a continuum of emotional functioning, reflecting the fact that emotional functioning might have been on par before neurological scarring due to Crouzon syndrome took it's course, resulting in deterioration of affect.

Compromised affect can be defined as difficulties related to emotions and relationships, and how these affect behaviour. A wide range of possible emotions underlie compromised affect. Examples include feeling threatened, inadequate, lonely, afraid, insecure, guilty, frustrated, conflicted and angry (Donald, Lazarus, & Lolwana, 2002:287).

From an affective neuroscience perspective, Salovey (2004:32) defines 'emotion' as a person's genetic and acquired motivational predisposition resulting in certain behaviour. Affective neuroscience however, seeks to link our understanding of basic neural circuitry for emotions with our straightforward cognitive and psychological views of the human mind. Thus affective neuroscience is deeply rooted within physiological psychology, behavioural biology, and behavioural neuroscience in an attempt to understand human emotions (Panksepp, 1998:9).

In conclusion, the concept *compromised affect* denotes a decline in emotional functioning from a previous, premorbid higher level of functioning to a lower level of functioning due to neurological lesioning associated with Crouzon syndrome.

1.5.3 Crouzon syndrome

Crouzon syndrome belongs to a group of conditions marked by abnormalities in the shape of the skull. These conditions occur when the bones of the skull, which normally are separated by a narrow space at birth, fuse together too early. This is known as craniosynostosis, which is not lethal, but treatment requires surgical intervention from a

young age to alleviate increased pressure on the growing brain. (Abou-Sleiman *et al*., 2002:307-308; Kummer, 2001:91).

Crouzon syndrome is distinguished from most other craniosynostois syndromes by the lack of obvious hand or foot abnormalities. The manifestations of this condition are generally limited to the skull and face, and its chief components are craniosynostosis, maxillary hypoplasia, shallow orbits and ocular proptosis (Vajo, Francomano, & Wilkin, 2000:23).

1.5.4 Developmental disorder

A developmental disorder manifests itself during the early developmental years, and often persists as the person grows older. The concept *early developmental disorder* or *childhood disorder* therefore seems to be a misnomer, because these conditions are relatively permanent in nature and the duration thereof persists through adulthood (Barlow & Durand 2002:455). According to Claassen (2006:21) Educational Psychologists take special interest in these developmental disorders, since significant focus is placed on the social, affective and cognitive development of children diagnosed with these disorders. According to this author these disorders are of clinical significance within the field of Educational Psychology, since the child's normal development is affected by the condition, implicating compromised mastery of developmental milestone. She states that when the full range of diagnostic criteria only manifests during adulthood, the mastery of basic developmental skills is not compromised; yet, adult psychopathology might contribute to sensory, motor and cognitive impairment or deterioration. In addition to non-mastery of certain developmental milestones, developmental disorders usually first diagnosed in infancy or childhood also affect family life, educational needs, education planning and provision.

1.6 PROBLEM STATEMENT

Based upon the rationale of this research project and the conceptualisation of the topic of interest, the research problem can be formulated as follows:

What is the relationship between developmental brain insults associated with Crouzon syndrome and compromised affect and learning?

1.6.1 Sub questions

- What atypical brain development associated with *Crouzon Syndrome* can be identified through MR imaging and radiographic techniques?
- Which brain areas are typically lesioned with *Crouzon Syndrome*?
- How does atypical brain architecture and circuitry associated with *Crouzon syndrome* relate to compromised affect and learning?

1.6.2 Research hypothesis

The following research hypothesis is formulated for this research project:

There is a neural substrate to compromised affect and learning associated with *Crouzon Syndrome,* resulting in a particular affective and learning profile.

1.7 PURPOSE OF THE STUDY

The purpose of my research study is to scrutinize the neural makeup of *Crouzon Syndrome* on the basis of a clinical case study, in order to compile a detailed explanatory profile of compromised affect and learning associated with *Crouzon Syndrome*.

1.8 THEORETICAL FRAMEWORK AND PARADIGMATIC PERSPECTIVE

This research design is firmly embedded in a positivist paradigm. Positivism proposes that an objective truth exists and is based upon the utilization of research methods and practices derived from the natural sciences and application of these to the social sciences (Human-Vogel, 2004:17). Research findings are based on objective, observable facts and are interpreted in terms of quantifiable units. Data is viewed to be linear and objective in nature, value free and theory independent, as well as relatively free from researcher contamination or bias (Jansen, 2004:380). Reality is thus perceived as external, and knowledge about human behaviour is attained through methods of scientific inquiry using sensory experience only. Positivism assumes that reality can be best understood by breaking it up into its smallest parts. There are many examples of this approach from various fields of study such as physics, biology and medicine. This type of design is

experimental in nature and is seen as either irrefutable or refutable (Donald *et al.*, 2002:99).

In addition, this project is firmly rooted in the theoretical framework of cognitive neuroscience. Cognitive neuroscience attempts to investigate the neural underpinnings of human cognitive, affective and volitional capacities (Bennett & Hacker, 2003:1). Cognitive neuroscience thus incorporates both neurophysiology and psychology and revolves around higher mental functions such as perception, attention, language, memory, thinking, and spatial awareness. More recently, behavioural and cognitive neuroscience has benefited from new approaches, such as non-invasive neuroimaging techniques that allow for images of the brain in action (Zigmond, Bloom, Roberts, Landis & Squire, 1999:4). Research to date has identified many brain features unique to individuals with learning impairments. What remains now, however, is to map such features to distinct developmental profiles of neuropsychological functioning (Collins & Rourke, 2003:1029).

Despite the recent developments in research, cognitive neuroscience does, to a great extent, lack interpretation of the neurobiological systems that underlie affective human behaviour. Thus, affective neuroscience has developed from cognitive science and seeks to provide conceptual bridges that can link our understanding of basic neural circuits of emotions with straightforward *cognitive* and p*sychological* views of the human mind leading us to understand the neurobiological underpinnings of our human emotions (Panksepp 1998:304). Thus, our introspective access to emotions supplements 'hard' scientific data and therefore results in an in-depth psychological understanding of developmental disorders. Panksepp's (1998) summary of the major premises of affective neuroscience is captured in table 1.2 below.

Table 1.2 The major premises of affective neuroscience (Panksepp, 1998:14-15)

'Emotional processes … play a key role in the causal chain of events that control the actions of humans. They provide various types of natural internal values upon which many complex behavioural choices in humans are based. However, such internal feelings are not simply mental events; rather, they arise from neurobiological events. In other words, emotional states arise from material events at the neural level." "Emotional feelings not only sustain certain unconditioned behavioural tendencies but also help guide new behaviours by providing simple value-coding mechanisms that provide self-referential salience, thereby allowing humans to categorize world events efficiently so as to control future behaviours.'
'A series of basic emotional processes arises from distinct neurobiological systems and everyday emotional

concepts such as anger, fear, joy, and loneliness are not merely the arbitrary taxonomic inventions of noncritical thinkers. These brain systems have several common characteristics. The core function of emotional systems is to coordinate many types of behavioural and physiological processes in the brain and body.'
'When such neural activities continue at low levels for extended periods of time, they generate moods and, ultimately, such personality dimensions as the differential tendency to be happy, irritable, fearful, or melancholy. These systems help create a substantial portion of what is traditionally considered universal "human nature"'.
'A complete study of emotional systems is also essential for understanding the many psychiatric disturbances that assail humans – schizophrenia, autism, mania, depression, anxiety, panic, obsessive-compulsive disorders, post-traumatic stress disorders, neuroses, and other vexations of the human spirit.'
'We will not understand the underlying neurodynamics of emotional systems without a great deal of concurrent brain research.'

1.9 RESEARCH DESIGN AND METHODOLOGY

This study will take the form of an individual case study which by definition involves a detailed account of one person and the examination of multiple variables (Babbie & Mouton, 2001:281). In terms of sampling, a single case will thus be investigated to gain information and various perspectives will be taken into account when attempting to understand the influence of Crouzon syndrome on affect and learning. Multiple sources of data will also be used to increase the reliability of the findings.

This research project represents a mixed-method approach which involves both quantitative and qualitative research. This study however is predominantly quantitative in nature. The aim of quantitative research is to explain and predict human behaviour, whereas the focus of qualitative research is on describing and understanding (Babbie & Mouton, 2001:53).

Quantitative research is deductive in nature and the research hypothesis, which flows from the research problem, directs the scientific inquiry (Garbers, 1996:287), leading to hypothesis-testing. This approach deals with relations, correlations and covariance of variables. Pertaining to this research project, the mode of inquiry is non-experimental at hand of a clinical case study which, in nature, is *ex post facto*, i.e. 'after the fact'. Experimental control, which heightens validity of the results, is recognized by making use

of the norm scales of the applicable measuring instruments and psychological assessment media. This will allow replication at hand of future studies. Replicating findings usually convince researchers that the findings cannot merely be ascribed to coincidence (Cohen, Manion & Morrison, 2000:181). 'The strength of a research program is in its ability to replicate findings in different ways to build confidence in the results' (Barlow & Durand 2002:104). Therefore, this research study involves establishing a hypothesis that is then tested. In abnormal psychology, research focuses on hypotheses meant to explain the nature and causes of disorders (Barlow & Durand, 2002:110).

Denzin and Lincoln, in De Vos (2000:240), explains qualitative research as a multiperspective approach to social interaction, aimed at describing, making sense of, interpreting or reconstructing this interaction in terms of the meanings that the subjects attach to it. Often unanticipated information can be identified through qualitative research, since the discussion is not limited by predetermined closed questions. Thus, by conducting a historicity interview and by including projective and graphic media in my study, information about how participants think, feel and act, as well as what they believe, can be collected.

1.9.1 Data generating strategies

The following data-generating strategies will be employed: MR-imaging, cranial X-rays prior to surgery, optometric assessment data, cognitive assessment, affective assessment, scholastic assessment, and a review of retrieved clinical reports from neurologists, occupational, speech and physiotherapists.

1.9.1.1 MR-imaging

MR-imaging is a procedure using radio signals generated in a strong magnetic field and passed through body tissue to produce detailed, even layered, images of brain structures, which are useful in detecting very small brain lesions (Eysenck, 2003:9). In brain research, MR-imaging is commonly used to locate tumours and lesions, or to identify other abnormalities. This is done by generating images of multiple sections of the brain, indicating the shape and location of various brain structures. An example of its validity and reliability is illustrated by research conducted with MRI scans that show that the frontal regions of the brains of individuals with dyslexia and learning disabilities are symmetrical

and smaller when compared with those of individuals without these learning disabilities (Wolfe, 2001:8 & Lerner, 2003:329).

1.9.1.2 Cortical X-Rays

The journey to where we are today with regard to imaging techniques began with the development of the X-ray, discovered in 1895. According to Wolfe (2001:4) X-rays are of little use in depicting the brain, but may be useful in determining the order and rate of suture fusion with regard to craniosynostosis. This is not a lethal condition, but treatment requires surgical intervention from a young age to alleviate pressure build-up on the cranial vault.

1.9.1.3 Additional media and techniques

Optometric, cognitive, neuropsychological and personality assessment will complement brain imaging studies. In order to meet these demands the following instruments will be administered: the Senior South African Individual Scale – Revised (SSAIS-R), the Quick Neurological Screening Test, the Wisconsin Card Sorting Test, the Children's Personality Questionnaire, the Children's Apperception Test (CAT), graphic media, as well as a scholastic assessment battery. The majority of these assessment tools are known to the critical reader and therefore only the Wisconsin Card Sorting Test will be discussed at this stage.

Traditionally, the focus of neuropsychology has been on the assessment of cognitive processes in order to anatomically localize structural brain injuries, as it is believed that lesions in any one structural component of the brain may cause a measurable disturbance in functional activity. Neuropsychological testing thus involves the application of statistical methods to define quantitative abnormalities in cognitive functioning (McCrory, Makdissi, Davis, & Collie, 2005:58).

> 'With regards to children and adolescents neuropsychological testing addresses the diagnosis of disturbances in speech, and/or language, reading, writing, spelling and computation. These disorders frequently are associated with other behaviours such

as hyperactivity, lack of attention span for visualizing and auditorizing and perceptual handicaps' (Fryburg, 1997:38).

Therefore, by using neuropsychological testing as part of our research study, it will allow for the exploration and understanding of the influence of closed-brain injury on the development of affect and learning from a neuroscience perspective. The Wisconsin Card Sorting Test (WCST) has increasingly been employed as a clinical neurological instrument and much of this test's current popularity among clinicians stems from its reported specific sensitivity to brain dysfunction affecting the frontal lobes. Interest in the cognitive and developmental effects of early frontal-lobe injury among children has also sparked considerable interest in the use of the WCST as a potential measure of executive function among school-age children (Heaton, Chelune, Talley, Kay & Curtiss, 1993:1).
Heaton *et al*. (1993:1) states that, owing to its apparent sensitivity to the effects of fontal-lobe lesions, the WCST is often referred to as a measure of 'frontal' or 'prefrontal' functioning. However, this labelling represents an oversimplification and research thus strongly recommends that the clinical interpretation of WCST performance should be conducted within the context of a comprehensive neuropsychological evaluation that integrates neuropsychological data with medical, psychosocial and historical information.

1.9.2 Process of data generation and collection

After my subject was referred to me by her teacher, I conducted a telephonic interview with her mother to gain further background information. The initial referral problem was of an affective nature and later learning difficulties were also reported. Since there is limited information on how affect and learning are compromised by Crouzon syndrome I discussed the possibility of conducting my research on this topic with her and her parents, after which I was granted informed consent. I will conduct a complete cognitive, scholastic and affective assessment and compare it to brain imaging results as well as make use of existing clinical reports. An optometric assessment will also be incorporated in the data generation process.

1.9.3 Data analysis and interpretation

The data will be quantified and interpreted along linear lines of thought, i.e. according to specific conceptual schemes and parameters of pre- and postnatal endocrinological and central nervous system development. Where applicable, significance of deviations will be calculated at the 0.001 and 0.005 levels of reliability.

Additional external expertise will be provided by Dr De Schmed (optometrist), Dr Van Rensburg (radiologist at MR-centre, Willows Hospital), and Prof. Marius Bosman (neuro-anatomist, University of Pretoria). An independent statistical analyst employed by Deloitte & Touche will assist towards the statistical calculations.

1.10 ROLE OF THE RESEARCHER

In this research project I have a dual relationship in dealing with my subject, as I am conducting research as well as providing emotional support in the form of psychotherapy. Thus my research has a strong focus on possible intervention strategies. Commonalities between these two roles however include confidentiality and doing what is in the best interest of the child.

1.11 ETHICAL CONSIDERATIONS

Planning a research project involves much more than just selecting the appropriate design – it also includes ethical considerations. We are working with human beings and great care should be taken to ensure that participants will not be placed at risk or harm of any kind. Participants should be thoroughly informed about the potential impact of the research and it is crucial that informed consent be obtained prior to commencement of the research project as demonstrated with **Annexure A**. The basic components of informed consent are competence, voluntarism, full information and comprehension on the part of the research participant. In other words, research participants must be capable of consenting to participation in the research, they must volunteer or not be coerced into participating, they must have all the information they need to make the decision, and they must understand that they will be at liberty to withdraw from the project at any time. Furthermore, confidentiality should also be ensured, refering to the handling of information in a

confidential manner which limits other's access to private information (De Vos, 2000:25-26).

In addition to the principles of informed consent, protection against potential harm, and the right to confidentiality, the Society for Research in Child Development (1990) has endorsed ethical guidelines for research that addresses some of the issues unique to research with children. Not only do these guidelines call for confidentiality, protection from harm, and debriefing, but they also require informed consent from children's caregivers and from the children themselves if they are age seven and older. These guidelines specify that the research must be explained to children in language they can understand so they can decide whether they wish to participate.

Many other ethical issues extend beyond the protection of participants. These concerns will be adhered to, as explained in the Ethics and Research Statement of the Faculty of Education, University of Pretoria.

1.12 CHAPTER PLANNING

Chapter one consists of the orientational introduction and actualisation of the research problem, the problem statement, the research hypothesis, as well as the research methodology and research design. Ethical considerations are also included.

Chapter two describes the pathogenesis and manifestation of Crouzon syndrome from various relevant perspectives, i.e. neurobiology, affective neuroscience and neuropsychology.

Chapter three explains the process of data collection and describes the empirical research and related findings. Based upon the findings, the research hypothesis will be accepted or rejected.

Chapter four represents an in-depth literature review linking those areas of the brain that are affected by Crouzon Syndrome with compromised affect and learning that might flow from these lesioned areas.

Chapter five gives an overview of the research findings, as well as deductions and conclusions derived at. Relevant recommendations will be made, and shortcomings inherent to the design will be pointed out.

1.13 KEY WORDS

- Crouzon syndrome
- Compromized learning
- Compromized affect
- Neural condition
- Genetic disorder
- Fresh mutation
- Atypical brain development

1.14 LIMITATIONS AND CONTRIBUTIONS

The existing body of knowledge covering compromised affect and learning with Crouzon syndrome is relatively restricted. The majority of sources that do describe this phenomenon represent a rather clinical, neuroscientific perspective, which does not adequately provide the answers to the research problem.

With this study we are hoping to contribute to a better understanding of how affect and learning are compromised as a result of the onset of Crouzon syndrome. We are eager to interpret this information in order to provide teachers, parents and psychologists with guidelines and understanding in terms of possible interventions and support strategies.

1.15 LIST OF REFERENCES

Abou-Sleiman, P.M., Apessos, A., Harper, J.C., Serhal, P. & Delhanty, J.D.A. 2002. Pregnancy following preimplantation genetic diagnosis for Crouzon syndrome. *Molecular Human Reproduction*, 8(3): 304-309.

Adolphs, R., Sears, L. & Piven, J. 2001. Abnormal processing of social information from faces in autism. *Journal of Cognitive Neuroscience*, 13:232-240.

Adolphs, R., Tranel, D., Damasio, H. & Damasio, A. 1994. Impaired recognition of emotion in facial expressions following bilateral damage to the human amygdala. *Nature*, 372:669-672.

Babbie, E. & Mouton, J. 2001. *The Practice of Social Research*. New York: Oxford University Press.

Bailey, A., Le Couteur, A., Gottesman, O., Bolton, P., Simonoff, E., Yuzda, E. & Rutter, M. 1995. Autism as a strongly genetic disorder: evidence from a British twin study. *Psychological Medicine*, 25: 63-77.

Barlow, D.H. & Durand, V.M. 2002. *Abnormal Psychology*. 3rd edition. Belmont: Wadsworth.

Bauman, M.L. & Kemper, T.L. 1995. Neuroanatomical observations of the brain in autism. In J. Panksepp, *Advances in biological psychiatry*, 1:1-26. Greenwich, Conn.: JAI Press.

Bauman, M.L. & Kemper, T.L. (Eds.). 1994. *The neurobiology of autism*. Baltimore: Johns Hopkins University Press.

Bayer, S.A., Altman, J., Russo, R.J. & Zhang, X. 1993. Timetables of neurogenesis in the human brain based on experimentally determined patterns in the rat. *Neurotoxicology*, 14: 83 - 144.

Bennett, M.R. & Hacker, P.M.S. 2003. *Philosophical Foundations of Neuroscience*. Oxford: Blackwell Publishing.

Berger-Sweeney, J. & Hohmann, C.F. 1997. Behavioral consequences of abnormal cortical development: insights into developmental disabilities. *Behavioural Brain Research*, 86: 121-142.

Camfield, P.R., Camfield, C.S. & Cohen, M.M. 2000. Neurologic Aspects of Craniosynostosis. In Cohen, M.M. & MacLean, R.E. (Eds.). *Craniosynostosis: Diagnosis, Evaluation, and Management.* 2nd edition. Oxford: Oxford University Press.

Carper, R.A., Moses, P., Tigue, Z.D. & Courchesne, E. 2002. Cerebral lobes in autism: Early hyperplasia and abnormal age effects. *Neuroimage*, 16:1038-1051.

Chen, H. 2006. *Crouzon Syndrome*. eMedicine. Retrieved from the World Wide Web on 17 March 2006 from: http://www.emedicine.com/ped/topic511.htm

Claassen, M. 2006. *Exploring the contribution of prenatal stress to the pathogenesis of autism as a neurobiological developmental disorder: a dizygotic twin study.* Unpublished M.Ed.-dissertation, University of Pretoria.

Clark, G. D. 2002. Brain development and the genetics of brain development. *Neurologic Clinics*, 20(4):1-16. http://minedump.redi.co.za:2198/das/articles/body/46623206-2/jorg=journal&source=MI. Date of MD website access 2006/05/22.

Clark, P.M. 2002. Programming of the hypothalamo–pituitary–adrenal axis and the fetal origins of adult disease hypothesis. *European Journal of Pediatrics*, 157:7–10.

Cohen, M.M, 2000. Crouzon Syndrome. In Cohen, M.M. & MacLean, R.E. (Eds.). *Craniosynostosis: Diagnosis, Evaluation, and Management.* 2nd edition. Oxford: Oxford University Press.

Cohen, L., Manion, L. & Morrison, K. 2000. *Research Methods in Education*, 5th edition. New York: RoutledgeFalmer.

Coleman, M. 1994. Second Trimester of Gestation: A Time of Risk for Classical Autism? *Developmental Brain Dysfunction*, 7:104-109.

Collins, D.W. & Rourke, B.P. 2003. Learning-disabled Brains: A Review of the Literature. *Journal of Clinical and Experimental Neuropsychology*, 25(7):1011-1034.

Courchesne, E., Chisum, H.J., Townsend, J., Cowles, A., Covington, J., Egaas, B. 2000. Normal brain development and aging: Quantative analysis at in vivo MR imaging in healthy volunteers. *Radiology*, 216:672-682.

Courchesne, E., Karns, C.M., Davis, H.R., Ziccardie, R., Carper, R.A., Tigue, Z.D., Chisum, H.J., Moses, P., Pierce, K., Lord,C., Lincoln, A.J., Pizzo, S., Schreiman, L., Haas, R.H., Akshoomoff, N.A. & Courchesne, R.Y. 2001. Unusual brain growth patterns in early life in patients with autistic disorder: an MRI study. *Neurology*, 57:245-254.

Courchesne, E. 2004. Brain Development in Autism: Early overgrowth followed by premature arrest of growth. *Mental Retardation and Developmental Disabilities Research Reviews*, 10:106-111.

Dednam, A. 2005. Learning Impairment. In Landsberg, E., Krüger, D. & Nel, N. (Eds.). *Addressing Barriers to Learning: A South African Perspective*. Pretoria: Van Schaik.

De Vos, A.S. 2000. *Research at Grass Roots: A primer for the caring professions*. Pretoria: Van Schaik.

Donald, D., Lazarus, S. & Lolwana, P. 2002. *Educational Psychology in Social Context*. 2nd edition. Cape Town: Oxford University Press.

Du Preez, C.S., Naudé, H. & Pretorius, E. 2004. Executive emotional system disruption as causal agent in frontal lobishness among abused children. *Early Child Development and Care*, 174(5):437-460.

Elbert, T., Heim, S., & Rockstroh, B. 2001. Neural plasticity and development. In C.A. Nelson & M. Luciana, (Eds.). *Handbook of Developmental Cognitive Neuroscience*, pp. 191-204. Cambridge, MA: MIT Press.

Eriksson, P.S., Perfilieva, E. & Bjork-Eriksson, T. 1998. Neurogenesis in the adult human hippocampus. *Nature & Medicine*, 4:1313-1317.

Eysenck, M.W. 2003. *Principles of Cognitive Psychology*. 2nd edition. New York: Psychology Press.

Fryburg, E.L. 1997. *Reading and Learning Disability: A Neurological Approach to Evaluation and Instruction*. New York: Charles C Thomas Publishers.

Garbers, J.G. 1996. *Doeltreffende geesteswetenskaplike navorsing*. Pretoria: Van Schaik.

Heaton, R.K., Chelune, G.J., Talley, J.L., Kay, G.G. & Curtiss, G. 1993. *Wisconsin Card Sorting Test Manual: Revised and Expanded*. Florida: Psychological Assessment Resources, Inc.

Human-Vogel, S. 2004. Cognition and Learning. In Eloff, I. & Ebersöhn, L. (Eds.). *Keys to Educational Psychology*. Cape Town: UCT Press.

Humphreys, P., Kaufmann, W.E. & Galaburda, A.M. 1990. Developmental dyslexia in women: neuropathological findings in three patients. *Annals of Neurology*, 28: 727-738.

Jansen, J. 2004. An Introduction to Education Theory. In Eloff, I. & Ebersöhn, L. (Eds.). *Keys to Educational Psychology*. Cape Town: UCT Press.

Kabbani, H. & Raghuveer, T. 2004. Craniosynostosis. *American Family Physician*, 69(12): 2863-2870.

Kolb, B. & Whishaw, I.Q. 2003. *Fundamentals of Human Neuropsychology*, 5th edition. New York: Worth Publishers.

Kummer, A.W. 2001. *Cleft Palate and Craniofacial Anomalies: Effects on Speech and Resonance*. San Diego: Singular Publishing Group, Inc.

Lerner, J.W. 2003. *Learning disabilities.* 7th edition. New York: Houghton Mifflin Company.

Levin, H.S., Benton, A.L. & Grossman, R.G. 1982. *Neurobehavioral Consequences of Closed Head Injury*. New York: Oxford University Press.

McCrory, P., Makdissi, M., Davis, G. & Collie, A. 2005. Value of neuropsychological testing after head injuries in football. *Brittish Journal of Sports Medicine*, 39(1):58-68.

Naudé, H., Pretorius, E., Van Schoor, A.H. & Becker, J. 2005. Cognitive and learning strategies for longstanding temporal lobe lesions in a child who suffered from Herpes Simplex virus encephalitis: a case study over 10 years. *Early Child Development and Care*, 174(5):487-500.

Panksepp, J. 1998. *Affective Neuroscience: The foundations of human and animal emotions.* New York: Oxford University Press.

Pretorius, E., Naudé, H. & Pretorius, U. 2005. Training the hippocampus and amygdala of preschool children by means of priming tasks: should parents rather focus on learning of facts than reading fairytales? *Early Child Development and Care*,175(4): 303 – 312.

Pujol, J., Vendrell, P., Junquae, C., Martai-Vilalta, J.L. & Capdevila, A. 1993. When does human brain development end? Evidence of corpus callosum growth up to adulthood. *Annuals of Neurology*, 34:71-75.

Rakic, P. & Caviness, V.S. 1995. Cortical development: view from neurological mutant two decades later. *Neuron*, 14:1101-1114.

Reichle, E.D., Pollatsek, A., Fisher, D.L. & Rayner, K. 1998. Toward a model of eye movement control in reading. *Psychological Review*, 105:125-157.

Salovey, P. 2004. Emotions and Emotional Intelligence for Educators. In Eloff, I. & Ebersöhn, L. (Eds.). *Keys to Educational Psychology*. Cape Town: UCT Press.

Schmid, C. & Rotenberg, J.S. 2005. Neurodevelopmental Toxicology. *Neurologic Clinics*, 23(2):1-13. http://minedump.redi.co.za:2198/das/articles/body/46471010-2/jorg=journal&source=MI. Date of MD website access 2006/05/25.

Singer, S.L., Walpole, I., Brogan, W.F. & Goldblatt, J. 1997. Dentofacial features of a family with Crouzon syndrome. Case reports. *Australian Dental Journal*, 42(1): 11-17.

Shprintzen, R.J. 2000. Speech and Language Disorders in Syndromes of Craniosynostosis. In Cohen, M.M. & MacLean, R.E. (Eds.). *Craniosynostosis: Diagnosis, Evaluation, and Management.* 2nd edition. Oxford: Oxford University Press.

Society for Research in Child Development, Committee for Ethical Conduct in Child Development Research. 1990. SRCD ethical standards for research with children. *SRCD Newletter*. Chicago: SRCD.

Tatter, S.B., Galpern, W.R., & Isacson, O. 1995. Neurotrophic factor protection against excitotoxic neuronal death. *The Neuroscientist*, 1:286-297.

Teicher, M.H., Andersen, S.L., Polcari, A., Anderson, C.M., & Navalta, C.P. 2002. Developmental neurobiology of childhood stress and trauma. *Psychiatric Clinics of North America*, 25(2):397-426. http://minedump.redi.co.za:2198/das/article/body/46518972-2/jorg-journal&source=… MD Consult website accessed on 2006/05/23.

Vajo, Z., Francomano, C.A. & Wilkin, D.J. 2000. The Molecular and Genetic Basis of fibroblast Growth Factor Receptor 3 Disorders: The Achondroplasia Family of Skeletal Dysplasias, Muenke Craniosyntosis, and Crouzon Syndrome with Acanthosis Nigricans. *Endocrine Reviews*, 21(1): 23-39.

Wolfe, P. 2001. *Brain Matters: Translating Research into Classroom Practice*. Alexandria: Association for Supervision and Curriculum development.

Zigmond, M.J., Bloom, F.E., Roberts, J.L., Landis, S.C. & Squire, L.R. 1999. *Fundamental Neuroscience*. London: Academic Press.

CHAPTER 2

THE MANIFESTATION AND PATHOGENESIS OF CROUZON SYNDROME

2.1 INTRODUCTION

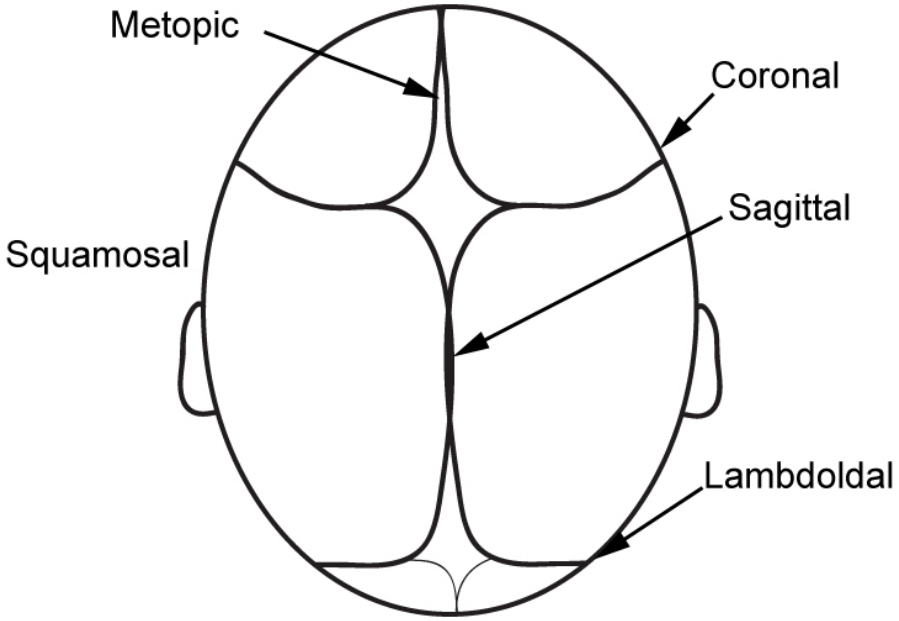
This chapter describes the pathogenesis and manifestation of Crouzon syndrome from various relevant perspectives, i.e. neurobiology, affective neuroscience and neuropsychology. A clarification of the term Crouzon syndrome is followed by a discussion of the manifestations of this condition and a description of the causes thereof.

2.2 DEFINING CROUZON SYNDROME

Crouzon syndrome was first described by a French neurosurgeon, O. Crouzon, in 1912. He reported a mother and her son who had a widened skull with protrusion in the region of the anterior fontanel, exophthalmoses, a beaked nose, and hypoplasia of the maxilla. More than 100 additional patients had been reported by 1966. Currently Crouzon syndrome, with a reported incidence of 1:25000 births, is the most common of more than 70 conditions in which premature fusion of the cranial sutures may be a feature (Singer *et al.*, 1997:11).

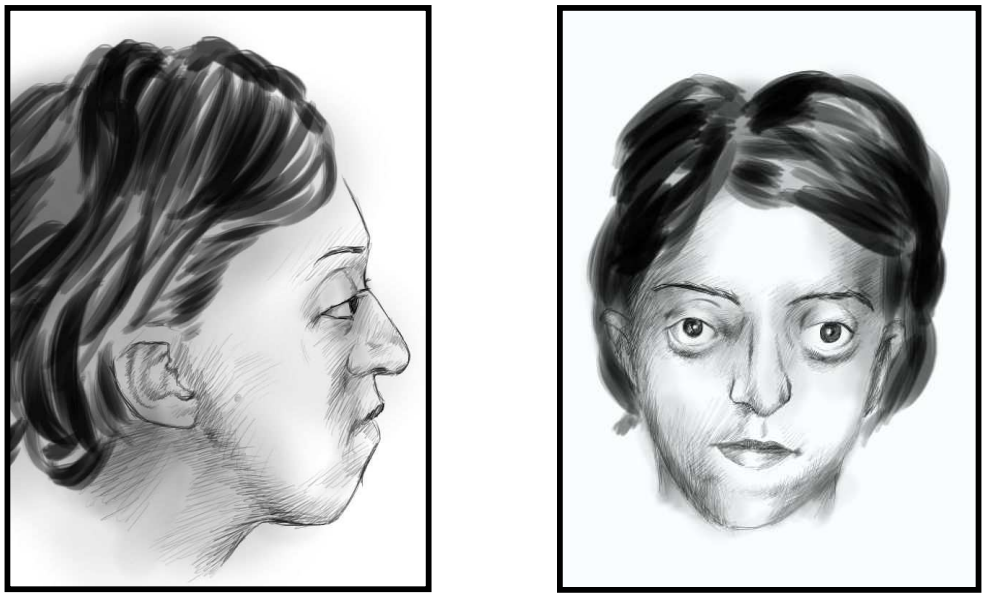
Crouzon syndrome belongs to a group of conditions marked by abnormalities in the shape of the skull. These conditions occur when the bones of the skull, which normally are separated by a narrow space at birth, fuse together too early. This is known as craniosynostosis, which is not lethal, but treatment requires surgical intervention from a young age to alleviate pressure build-up on the cranial vault. Craniotomy and skull reshaping procedures are often required (Abou-Sleiman *et al.*, 2002:307-308; Kummer, 2001:91). The order and rate of suture fusion determine the degree of deformity and disability. Premature synostosis may be evident at birth, or may develop during the first year of life and is completed by the second or third year (Cohen, 2000:362). The sutures affected include the coronal, the sagittal, and occasionally the lambdoidal sutures, as illustrated in Figure 2.1 below.

Figure 2.1 A diagrammatic representation of the different sutures involved in Crouzon syndrome



According to Kummer (2001:92), the major feature of Crouzon syndrome is craniosynostosis, usually involving the coronal sutures. As illustrated in Figure 2.1, abnormal fusion of the coronal sutures can result in an abnormal head shape, and in severe cases this condition can also cause increased pressure on the growing brain. Compensatory growth occurs at the remaining open sutures to allow continued postnatal brain growth, causing midfacial hypoplasia, shallow orbits, a foreshortened nasal dorsum, maxillary hypoplasia, occasional upper airway obstruction and brachycephaly, which is a short skull (Chen, 2006:2). If the sagittal suture is involved, the lateral growth of the skull will be compromised, resulting in frontal bossing and scaphacephaly, where the skull appears oblong from front to back. However, when multiple sutures are involved, this may result in asymmetry of the skull, which is referred to as plagiocephaly (Kummer, 2001:91).

The appearance of Crouzon syndrome can, however, vary in severity from a mild presentation with subtle midfacial characteristics to severe forms with multiple fused cranial sutures and marked midfacial features, as illustrated in Figures 2.2 to 2.3 below.



In order to fully appreciate the manifestations of Crouzon syndrome, a literature review was undertaken to learn more about human developmental neuroscience and its intersection with Crouzon syndrome. Because Crouzon syndrome involves postnatal brain development, this review focuses on the postnatal neuroanatomic changes that arise during the first years of human life. Human neurodevelopment is characterized by two major organizational periods. The first period starts at conception and includes events such as neurulation, proliferation, migration, and differentiation (Webb, Monk & Nelson, 2001:147), which events might be controlled genetically and epigenetically. The second period, which is a time of reorganization in the human cortex, occurs during gestation and continues postnatal, possibly through the second decade of life. This second period is characterized by dendritic and axonal growth, synapse production, neuronal and synaptic pruning, and changes in neurotransmitter sensitivity. Although the initiation of these events is influenced by endogenous signals, further neural maturation is primarily influenced by exogenous signals (Webb *et al.*, 2001:147) and it is most likely that Crouzon syndrome would impact on this second period of neurodevelopment; therefore the focus of this discussion will be on this period, starting with dendritic and axonal growth.

2.2.1 Dendritic and axonal growth

Once neurons have migrated to their correct location, they begin the process of extending axons and dendrites to provide the surface area for synapses with other cells (Webb *et al.*, 2001:148). As axons are the primary mechanism by which neurons signal other neurons in

the cortex, these axons extend to appropriate targets to initiate the formation of other synapses. These processes are part of neural maturation (Kolb & Whishaw, 2003:614), and it is thought that these processes are extremely vulnerable to the impact of Crouzon syndrome, for example around Broca's area. The neurons first display simple dendritic fields, and these fields become progressively more complex until a child reaches the age of about two years (Kolb & Whishaw, 2003:614). At least four different mechanisms serve to guide axon path-finding: contact attraction, contact repulsion, chemo attraction, and chemo repulsion (Tessier-Lavigne & Goodman, 1996:1123), and according to Webb and co-workers (2001:149), these mechanisms work in combination to balance the attraction and repulsion cues. The complexity of growth cues is due to the fact that different populations of cortical axons react differently to membrane-associated molecules from a given cortical layer, and it is most likely that different cortical layers express different sets of receptors and have different signalling pathways (Bolz, Castellani, Mann, & Henke-Fahle, 1996:41). Korsching (1993:2739) and Thoenen (1995:593) found layer-specific differences in growth factors, neurotrophins, and the expression of their receptors, which regulate both axonal and dendritic growth. It is possible that several such mechanisms operate simultaneously or sequentially (Kolb & Whishaw, 2003:615). Dendritic sprouting begins as soon as neurons reach the cortical plate (at approximately 15 weeks), with spines typically appearing on both pyramidal and nonpyramidal neurons between the 25th and the 27th week of gestation and increasing in some cortical regions through the 24th postnatal month (Mrzljak, Uylings, Van Eden & Judas, 1990:185), i.e., at the time that Crouzon syndrome has fully manifested.

Initially there is an overproduction of dendrites, dendritic spines, and axons, which are later pruned to the final number of dendrites, dendritic spines and axons through the process of competitive elimination during the postnatal period (Webb *et al.*, 2001:150). LaMantia and Rakic (1990:2156) suggest an overproduction of axons in infragranular Layers IV and V, which are later eliminated, while axons in later projection areas such as in supergranular Layers II and III seem to survive. This process of initial overproduction and elimination seems to be closely associated with the excessive synaptogenesis during the early postnatal period (Webb *et al.*, 2001:151), and it is most likely that intracranial pressure owing to Crouzon syndrome might inhibit this initial process of overproduction and elimination. For example, within the visual cortex the number of axon spines peaks at about five postnatal months, while synaptogenesis peaks around the eighth postnatal

month. Consequently the visual cortex shows rapid development between the second and fourth postnatal month, with maximum dendritic arborization occurring at approximately five months and regression to an adult level by two years (Michel & Garey, 1984:223), therefore the visual cortex is most vulnerable to the impact of Crouzon syndrome. Webb and co-workers (2001:151) report that during the first postnatal year, dendritic sprouting and branching can be seen in all six layers, although these spines are still immature, suggesting that the process of dendritic sprouting and branching in all six layers coincide with the progression of Crouzon syndrome, i.e., increased intracranial pressure brought about by a decrease of intracranial space for expansion due to premature closure of the relevant sutures. Furthermore, the length of prefrontal dendrites increases 5 to 10 times during the first six months of postnatal life, thereby demanding an expansion of cranial size. In general, pyramidal neuron branching increases rapidly at 28 weeks of gestation, with slower increases in branching continuing up to the age of seven years (Becker, Armstrong, Chand, & Wood, 1984:117). Dendritic arborization in the frontal cortex at birth is delayed compared with the visual cortex and the hippocampus, where the full number is present by approximately six months post term (Webb *et al.*, 2001:151), while production of pyramidal neuron dendrites peaks during the second year of life (congruent with the peak of synaptogenesis between the ages of two and three years) (Mrzljak *et al.*, 1990). In addition, in the prefrontal cortex, Layer IIIc basilar dendrites of pyramidal neurons undergo a rapid dendritic growth phase postnatal to around one year of age, with increases in branching until early adulthood (Koenderink, Uylings, & Mrzljak, 1994:173). According to these researchers, by seven and a half months of age, the number of basal dendrites per neuron is at a constant level; however, the length of the basal dendritic field shows a marked increase between seven and a half months and one year. This increase is due to larger segments and additional branches. Similarly, in Layer V, the number of basal dendrites per pyramidal neuron of the prefrontal cortex (efferent neurons that provide callosal, association, and subcortical projections) stabilizes at the postnatal age of one year, although there is progressive elongation of the basal dendrite field through five years of age when they are morphologically mature (Webb *et al.*, 2001:150).

The preceding findings thus demonstrate that Crouzon syndrome might significantly inhibit cortical growth and maturation due to premature fusion of relevant sutures, and it is most likely that the visual cortex and the prefrontal cortex are the areas that are most vulnerable to the impact of decreased intracranial space and concurrent increased

intracranial pressure. The formation of appropriate neural pathways can be disrupted in a number of ways, for example, in a case study described by Lyon and co-workers (Lyon, Arita, Le Galloudec, Valee, Misson & Ferriere, 1990:193), three infants with congenital encephalopathy, profound weakness and hypotonia presented marked deficiency in cerebral axons. Due to intracranial pressure, axons might fail to reach target areas in the early months of postnatal life (Kolb & Whishaw, 2003:615), for example, in the visual and prefrontal cortex, as well as in the corpus callosum and the hippocampal system. In a similar way optic pathways might be reduced in size and area of distribution, or axonal development might be disrupted if the axonal system's target is damaged, in which case the system might degenerate or might connect with an inappropriate target (Kolb & Whishaw, 2003:615). If the axon's target neuron has been damaged, either aberrant pathways may form to other cells, or the axon will not receive the trophic support it needs, and thus both the axon and its cell body may die (Webb *et al.*, 2001:151). Should the axonal system connect with an inappropriate target, the behaviour supported by the invaded area might be affected as well. The abnormalities of posture and movement seen in children with certain kinds of athetosis (slow involuntary movement) and dystonia (imbalances in muscle tone) associated with Crouzon syndrome might be ascribed to fibre systems that were meant to support posture and movement, but that have connected to the wrong target (Kolb & Whishaw, 2003:615). However, there are many ways that a developing brain can adjust its growth to achieve functional connections if its normal development is hindered, especially if the prematurely closed sutures are surgically corrected.

Crouzon syndrome may cause abnormalities of dendritic development and mental retardation associated with defects in the number, length, and spatial arrangement of dendritic branching and dendritic spines, dendritic spines might be thinner, there might be a decrease in the number of dendritic spines, and dendritic spines might have shorter stalks. Purpura and colleagues (Purpura, Bodick, Suzuki, Rapin, & Wurzelmann, 1982:287) suggested that the decreased dendritic number results from a failure of cytoskeletal structure, where the microtubules do not align properly such that the cell shape is abnormal and cannot support the outgrowth of dendritic branches. In addition, because the rate at which the brain matures is disturbed, especially the medulla oblongata might show decreased numbers of dendritic spines, abnormally thin dendrites, and long, thin dendritic spines (Tasashima & Mito, 1985:76).

2.2.2 Synapse production

Neurons transmit information to other neurons at their synaptic contacts (Webb *et al.*, 2001:152). There are several kinds of synapses. In one kind, called an axodendritic synapse, the axon terminal of a neuron meets a dendrite or dendrite spine of another neuron. Another kind of synapse is an axomuscular synapse in which an axon synapses with a muscle. The many other types of synapses include axosomatic synapses, in which an axon terminal ends on a cell body; axoaxonic synapses, in which an axon terminal ends on another axon, and axosynaptic synapses, in which an axon terminal ends at another synapse (Kolb & Whishaw, 2003:105). Axon terminals that have no specific target, but instead secrete their transmitter chemicals nonspecifically into the extra cellular fluid, are called axoextra-cellular synapses. There are axosecretory synapses as well, in which an axon terminal synapses with a capillary (a tiny blood vessel) and secretes its transmitter directly into the bloodstream. Synapses need not include even a single axon terminal – dendrites may send messages to other dendrites through dendrodendritic synapses (Kolb & Whishaw, 2003:105). All these different types of synapses contribute to form an extremely versatile chemical delivery system. A distinction is made between electrical synapses (e.g. gap junctions) and chemical synapses, although very little is known about electrical synapses (Webb *et al.*, 2001:152). At a chemical synapse, an electrical signal from the presynaptic cell is converted into a chemical signal that can be transferred through extra cellular space to the postsynaptic cell. In synaptic transmission, an electrical signal is transferred from the soma (cell body) down the axon and signals the release of chemical messengers into extra cellular space. The chemical messengers (typically neurotransmitters and neuropeptides), when combined with a receptor protein, can open or close ion channels on dendritic spines, changing the electrical current in the postsynaptic cell. This process allows for intercellular communication, with most synapses occurring between axons and dendrites (but also from axon to soma, dendrite to dendrite, and axon to axon). Similar to axon and dendritic growth, both spontaneous and environmentally induced neuronal activity lead to the formation and stabilization of synapses (Webb *et al.*, 2001:152).

How does an understanding of synapse production benefit our understanding of Crouzon syndrome? These early synapses are labile, and the formation of mature patterns of connection involves the elimination of a limited number of immature labile connections

with the elaboration and addition of appropriate connections (Webb *et al.*, 2001:152). Those synapses that make functional connections receive a larger amount of coordinated activity and are stabilised, but those that do not may be eliminated or reabsorbed, suggesting that synaptic stability might be delayed in Crouzon syndrome. Synapse stabilisation may take place through the local release of neurotrophins (nerve-growth factor [NGF], neurotrophin-3 [NT-3], and brain-derived neurotrophic factor [BDNF]). It has been hypothesized that postsynaptic cells release neurotrophins, and that axons whose parent cells have recently been activated are able to respond to these signals (Katz & Shatz, 1996:1133; Thoenen, 1995:593). Thus, in Crouzon syndrome, the number of synapses that are contributing to information transfer might be inhibited, resulting in a lesser amount of coordinated activity. Molliver *et al.* (1973:403) identified the first synaptic junctions in the cortical plate at about 23 weeks gestation. However, peaks in the quantity of synapses typically occur during the first year of life, once the clinical features of Crouzon syndrome have already manifested. Although the timing of synaptogenesis is varied, adult values and peak levels of synaptic density in the auditory cortex, visual cortex, and medial frontal gyrus show similar aggregate values, which suggests that peak densities and synaptic elimination occur to a similar degree throughout the cortex (Huttenlocher & Dabholkar, 1997:167). This in turn suggests that auditory and visual functioning might be particularly compromised with Crouzon syndrome.

In addition, Huttenlocher and colleagues (Huttenlocher, 1979; 1984; Huttenlocher & Dabholkar, 1997; Huttenlocher & de Courten, 1987) documented the postnatal period of synaptogenesis in the visual cortex and prefrontal cortex. In the visual cortex (Huttenlocher & de Courten, 1987:1-9) the greatest increases in synaptogenesis occur between the ages of two and a half and eight postnatal months, with the most rapid increases between two and four months. Physically, the shape and structure of the eye and the retina also undergo massive transformations (Webb *et al.*, 2001:154). It is thought that improvements in vision (from 20/400 to 20/40) are caused by the physical maturation of the eyes, and not by the synaptic levels, and that this maturation is delayed in children with Crouzon syndrome. However, it is not known whether further maturation of the visual cortex is correlated with improvements in depth perception and other associated visual abilities, considering that maturation might be inhibited by properties such as defects in the number, length, and spatial arrangement of dendritic branching and dendritic spines,

dendritic spines might be thinner, there might be a decrease in the number of dendritic spines, and dendritic spines might have shorter stalks.

Similar to other cortical areas, the visual cortex matures at different rates (Webb *et al.*, 2001:154). In a postmortem analysis of the visual cortex, Huttenlocher and de Courten (1987:488) found synaptic profiles in Brodmann's area 17 of the human striate cortex as early as at 28 weeks gestation. However, the synaptic density at this time is only 2% of adult value and at birth it is only 17% of adult value, suggesting that the bulk of synapse formation in the visual cortex takes place postnatal. Maximum synaptic density is attained in the upper cortical Layers I to IVb by four months (projects to other cortex areas), by eight months in Layer IVc (projects to Layer IVb), 11 months in Layer V (projects to the superior colliculus), and 19 months in Layer VI (projects to the lateral geniculate). Later developing synapses may reflect top-down modulation of the early visual areas (superior colliculus and lateral geniculate). These findings suggest that the visual cortex matures at different rates and that the visual cortex might be particularly at risk with Crouzon syndrome.

In the frontal cortex, synapse formation begins at 27 weeks gestation and does not reach its maximum density until after 15 postnatal months (Webb *et al.*, 2001:154), suggesting that synapse formation in the frontal cortex is equally at risk. In the middle frontal gyrus, which is implicated in abstract thinking and reasoning, synaptic density reaches its maximum number of synapses at three and a half years (Huttenlocher & Dabholkar, 1997:167-178), and synaptic density might be compromised due to intracranial pressure associated with Crouzon syndrome. At three months the middle frontal gyrus is at 50% of peak; however, compared to the adult morphology, the synapses are immature (Webb *et al.*, 2001:154). According to Huttenlocher's findings (1979:195-205), synapses only gradually become adult-like at six to 24 postnatal months, which is usually the time when Crouzon syndrome also peaks. Synaptic density further increases significantly between childhood (six months to seven years) and adulthood (16 to 72 years) (Webb *et al.*, 2001:154). In addition, neuronal density shows significant layer-related variations that also vary with age. Layer IV, which is the primary afferent layer, and Layers V and VI, which give rise to efferent fibres, develop more rapidly than Layers II and III, which are concerned with information processing (Huttenlocher & Dabholkar, 1997:167). These

findings suggest that Layers IV, V and VI might be more likely to be lesioned due to Crouzon syndrome.

Data on the development of postnatal synaptogenesis in the hippocampus is relatively sparse compared with the data available on the visual and frontal cortex. In general, the onset of synaptogenesis appears long before interaction with the environment and the onset of cognitive function (Webb *et al.*, 2001:154). In the hippocampus, synapses (asymmetric, axodendritic) are present in the marginal zone, cortical plate, and sub-plate zone as early as at 15 weeks gestational age (Kostovic, Seress, Mrzljak, & Judas, 1989:105-116), which suggests that the hippocampus might also be morphologically and functionally compromised due to Crouzon syndrome.

This overproduction of synapses might be a mechanism by which the brain is made ready to receive specific input from the environment (Webb *et al.*, 2001:155). Studies of synaptogenesis demonstrate important developmental increases in the postnatal period and Goldman-Rakic (1987:601-622) proposed that the period of early overgrowth is important for the onset of cognitive function in human infants. For example, in the human infant, the frontal cortex begins to dramatically increase its synaptic density at eight months and peaks at two years (Huttenlocher, 1979:195), which coincides with the period of language acquisition and increased perceptual and cognitive awareness. The implication of these findings is that children diagnosed with Crouzon syndrome might show functional impairment of visual, somatosensory, motor, limbic and associative functions.

2.2.3 Neuronal and synaptic pruning

Pruning, or loss of synapses in the absence of cell death, refers to environmentally regulated changes in the density of synapse per unit of dendritic length, not the loss of the whole neuron (Webb *et al.*, 2001:156). During infancy there is a period of rapid synaptogenesis, followed by a plateau in childhood (although synaptic density still exceeds adult synaptic density), followed by synapse elimination during late childhood and adolescence, during which the number of synapses seen at peak during childhood is reduced by approximately 40% to reach adult density (Huttenlocher & Dabholkar, 1997:167). It thus follows that if synaptogenesis is compromised during infancy due to the presence of Crouzon syndrome, synaptic density might later be reduced further through

programmed synapse elimination during late childhood and adolescence, despite reconstructive surgery.

Furthermore, it is hypothesized that the cerebral metabolic rate for glucose might also be compromised due to inhibited synapse formation associated with intracranial pressure. In general, the cerebral metabolic rate for glucose rises rapidly during infancy when synaptic density peaks, remains high throughout childhood, and decreases during adolescence when there is a fall in synaptic density due to programmed synapse elimination (Chugani, Phelps, & Mazziotta, 1987:487). In particular, the prefrontal cortex lags by about five to eight months in amount of cerebral energy metabolism (Chugani *et al.*, 1987:490) as compared to the rest of the cortex, similar to the lag in synaptic density values seen in the prefrontal cortex (Webb *et al.*, 2001:157).

2.2.4 Myelination

Myelin is a fatty sheath that insulates axons in both the peripheral and central nervous systems (Webb *et al.*, 2001:160). Schwann cells in the peripheral nervous system and oligodendroglia in the central nervous system wrap around each axon, insulating it, except for the small region between each glial cell. This insulation, referred to as myelin or a myelin sheath (Kolb & Whishaw, 2003:96), provides for more rapid impulse conduction and energy efficiency, for example on the largest myelinated mammalian axons the nerve impulse can travel at a rate as high as 120 metres per second, compared with only about 30 metres per second on smaller, less-myelinated axons (Kolb & Whishaw, 2003:96). Webb and co-workers (2001:160) report that myelination occurs in several stages, i.e. 'first, after the extension of an axon, glial cell hyperplasia occurs in the vicinity of the axon. The glial cells accumulate myelin lipid components cytoplasmically before the actual appearance of myelin. Second, myelin becomes visible to the microscope (Stage 1), then to the naked eye (Stage 2), and in Stage 3 reaches adult density levels. These stages do not occur linearly; the shift from Stage 1 to 2 may occur rapidly, but the shift from Stage 2 to 3 may take many months or years'. Kolb and Whishaw (2003:97) report that myelination occurs in a caudal to rostral direction, behavioral systems such as the primary sensory and motor projection areas of the cortex myelinate in advance of the association areas, and the layers sub-serving communication with the brain stem and spinal cord (i.e. Layers I, IV, V, VI) myelinate prior to layers sub-serving communication with the cortex (i.e. Layers II, III).

Stimulation tracts carrying postural, orientation, and vestibular impulses are fully myelinated prenatally and myelination of these areas is unlikely to be affected by Crouzon syndrome. However, major tracts of the visual system (superior colliculus, optic tract and optic nerve) begin to myelinate prior to birth and reach maturity by nine months of age (Brody, Kinney, Kloman, & Gilles, 1987:283-301), suggesting that the visual system might be compromised. Some systems, such as the autonomic nervous system, do not myelinate, while some neurological functioning starts prior to the appearance of myelin (Webb *et al.*, 2001:160).

'Disruptions in myelination are likely to contribute to decreased conduction velocity, increased refractory periods after synaptic firing, more frequent conduction failures, temporal dispersion of impulses, and increased susceptibility to extraneous influences' (Webb *et al.*, 2001:161). As a general link between structure and function, Harbord *et al.* (1990:295-303) examined a heterogeneous group of children with developmental delay (in addition to associated neurological abnormalities) using MR imaging techniques and concluded that almost two thirds showed delayed or absent myelination as the only visible abnormality. Glia cells undergo a constant cycle of proliferation and cell death and may contribute to brain-volume measures. Changes in total brain volume reflect changes in the ratio of cerebral grey matter, white matter, and cerebrospinal fluid volumes (Webb *et al.*, 2001:161), and brain volumetric differences have been implicated in attention deficit hyperactivity disorder (ADHD). Kolb and Whishaw (2003:618) report delayed myelination and slightly smaller (by 4%) total brain volume (both white and grey matter), abnormalities of the basal ganglia, and a striking (15%) decrease in the volume of a restricted region of the posterior cerebellum among children diagnosed with ADHD. It therefore follows that children diagnosed with Crouzon syndrome might suffer from attention deficits with or without hyperactivity.

2.2.5 Whole brain growth

Dramatic changes in the volume of the human brain occur after birth with brain weight increasing fourfold from birth to the age of 10 years (Webb *et al.*, 2001:161). The number, size, and density of neurons and glia, as well as dendritic and axonal number and density determine brain size. Apart from the association between brain size and ADHD, reduction in the size of the mid-sagittal *corpus callosum* and the *planum temporale* have been

implicated in dyslexia and other learning disorders (Giedd *et al.*, 1996:551-560). Total brain volume also differs in the normal population, but Reiss, Abrams, Singer, Ross and Denckla (1996:1763-1774) concluded that total brain volume predicts 20% of the variance in IQ in a bell-shaped function, whereas cerebral volume outside of the normal range has been associated with sub-optimal functioning in children (Webb *et al.*, 2001:161). From birth to five years of age, the brain triples in mass, and this gain in brain size is mostly ascribed to the vigorous myelination of fibre tracts (Teicher *et al.*, 2002:398), yet a reduction in intracranial volume often characterizes Crouzon syndrome. Grey matter decreases with age, whereas white matter increases with age (Reiss *et al.*, 1996:1767). Grey matter changes are likely to reflect cell growth, arborisation, synaptogenesis, and cell proliferation, while decreases in grey matter are thought to reflect pruning and normal neuronal elimination during childhood. At 30 weeks of gestation, 35% of the total brain volume is from grey matter, increasing to 50% at birth (Huppi *et al.*, 1998:224-235). These changes are primarily due to increase in cortical grey matter, not sub-cortical structures. However, white matter seems to be increasing during development. From 29 to 41 weeks of gestation, white matter increases fivefold (Huppi *et al.*, 1998:229), and it continues to increase through childhood (Reiss *et al.*, 1996:1767), with increases in the dorsal, frontal, and parietal regions continuing through adolescence (Sowell *et al.*, 1999:587-597). Maximum hippocampal growth takes place at more or less 1.5 months (Webb *et al.*, 2001:162), while intracranial pressure due to Crouzon syndrome increases at the same time, resulting in the malformation and/or size reduction of various brain structures. A reduction in intracranial volume thus suggests lowered intellectual functioning, attention deficits, as well as a possible diagnosis of dyslexia and other learning disorders.

2.2.6 Cortical circuits

Because Crouzon syndrome manifests during infancy, which is a critical period of development, the rapidly growing structures are more sensitive to damage, e.g. Crouzon syndrome manifests during a period of rapid synaptogenesis (Taylor & Alden, 1997:555-567). In addition, various children with early brain lesions, including those who sustained traumatic brain injury, are susceptible to both immediate and long-term neurobehavioural deficits (Vargha-Khadem *et al.*, 1997:376-380). However, the infant brain also demonstrates greater ability to recover from some types of injury than is the case during later stages of development. For example, age is an important determinant of the effects of

early lesions, and three critical age divisions have been identified (Kolb & Whishaw, 2003:626). Lesions incurred before the age of one year tend to produce disproportionately greater impairments than do those incurred later. Lesions incurred between the ages of one and five years are followed by some reorganisation of brain function, including rescuing of language functions. Lesions incurred after the age of five years permit little or no sparing of function (Kolb & Whishaw, 2003:626). However, Bates (1999:214-253) reviewed two studies of recovery of function after developmental lesion and found that worse cognitive outcomes were associated with children who suffered their injuries between the ages of one and five years, compared with those with congenital injuries or those who sustained injuries between the ages of five and 12 years. These results suggest that we are not likely to find a general linear relation between age and recovery of function (Webb *et al.*, 2001:164), and therefore the manifestations of Crouzon syndrome are now reviewed in subsequent paragraphs.

2.3 MANIFESTATIONS OF CROUZON SYNDROME

Crouzon syndrome is distinguished from most other craniosynostosis syndromes by the lack of obvious hand or foot abnormalities. The manifestations of this condition are generally limited to the skull and face, and its chief components are craniosynostosis, maxillary hypoplasia, shallow orbits and ocular proptosis (Vajo *et al.*, 2000:23). However, cervical spine abnormalities are fairly common, and a smaller percentage of patients may have subtle elbow, hand, musculoskeletal, or internal anomalies (Singer *et al.*, 1997:11; Cohen, 2000:364).

2.3.1 Neurological aspects of Crouzon syndrome

According to Camfield *et al.* (2000:177), neurological abnormalities resulting from Crouzon Syndrome may be caused by premature synostosis resulting in distorted brain shape secondary to abnormal skull shape, or by primary abnormalities of the brain, such as hydrocephalus and agenesis of the corpus callosum, or by a combination of these. However, the degree of cranial malformation in Crouzon syndrome depends on the order and rate of progression of sutural synostosis.

2.3.2 Increased intracranial pressure and mental functioning

In Crouzon syndrome, hydrocephalus with increased intracranial pressure may occur due to craniosynostosis. Furthermore, increased intracranial pressure is found most commonly in patients with multiple sutural synostoses. Research shows that decreased mental functioning is more likely when more than one suture is involved. For example, oxycephaly involving the coronal and sagittal sutures has been associated with between 35% and 50% of the cases where decreased mental function has been observed. If left untreated, this can have a permanent effect on intelligence and cognitive functioning (Kummer, 2001:133). However, when mental handicap accompanies single-suture synostosis, it is most likely caused by an underlying brain malformation rather than by brain distortion resulting from synostosis (Camfield *et al.*, 2000:177). According to Chen (2006:2), decreased mental function is present in approximately 12% of all patients with Crouzon syndrome. Further intracranial anomalies include malformation, agenesis of the corpus callosum, and hindbrain herniation (Kabbani, 2004:2868; Kummer, 2001:92). According to research conducted, children who had intracranial pressure and were more than three years old, had higher rates of intellectual handicap than children whose increased intracranial pressure was discovered before age three (Camfield *et al.*, 2000:178). This supports the idea that the longer the intracranial pressure is elevated, the greater the effect on intellect. A further warning sign of increasing intracranial pressure includes the presence of progressive speech impairment (Shprintzen, 2000:200).

2.3.3 Otologic manifestations

Patients with Crouzon syndrome can exhibit various pathological features of the ear. While external malformations are unusual, middle-ear disease and hearing loss are more common (Orvidas *et al.*, 1999:1372).

According to Sulicia and Grundfast (2000:204), an examination of 30 patients revealed evidence of chronic middle-ear disease in more than half the participants. The involvement of the cranial nerve VIII has also been identified as resulting in deafness in patients with Crouzon syndrome. The cause of the deafness may be the cranial nerve compression in the auditory canal, or changes in the middle ear, either in the cochlea or in the ossicular chain. According to Kummer (2001:180), abnormal formation of the ossicles

may affect the transmission of sound to the inner ear, causing conductive hearing loss. Furthermore, Orvidas *et al.* (1999:1372) also note a higher incidence of senorineural hearing loss than was previously reported. The incidence of hearing loss is not well established, but may be as high as 10%. Hearing should therefore be carefully checked, especially in young children with craniostenosis, due to its critical role in language development (Camfield *et al.*, 2000:181).

2.3.4 Ocular features

Shallow orbits with ocular proptosis are an important diagnostic feature for Crouzon syndrome. Headaches and failing vision are also attributable to elevated intracranial pressure and further visual disturbances can result from corneal injury due to exposed conjunctivitis or keratitis (Chen, 2006:3). The orbits are shallow, causing exophthalmoses, or protrusion of the eyeballs (Kummer, 2001:92). According to Cohen (2000:363), low-frequency findings include nystagmus, iris coloboma, aniridia, anisocoria, corectopia, micro-cornea, megalocornea, keratoconus, cataract, ectopia lentis, blue sclera, and glaucoma.

2.3.5 Nasal and oral manifestations

Many children with Crouzon syndrome have hypoplasia of the mid face with diminished nasal and nasopharyngeal spaces. This is evident when the nose and upper jaw appears sunken in because of poor bone growth in the face. This increases nasal airway resistance and forces the infant to breath primarily through the mouth. This type of breathing may result in inadequate breathing (Posnick, 2000:272).

Cleft palates and sub-mucous cleft palates are occasionally seen in patients with Crouzon syndrome, but overall this developmental anomaly is an uncommon finding (Kummer, 2001:92). Furthermore, due to maxillary hypoplasia in Crouzon syndrome, the anteroposterior dimension of the maxillary dental arch is shortened, which gives the appearance of a highly arched palate (Cohen, 2000:364).

2.3.6 Speech and language disorders

It is accepted that speech refers to the ability to communicate verbally and is made up of several separate but interdependent components. These include language, voice, resonance, rate and articulation. The incidence of Crouzon syndrome may, however, have a strong influence on some, if not all of the components of communication. This is substantiated by the fact that effective speech and communication requires an intact hearing mechanism, complete skeleton and normal soft tissues (Shprintzen, 2000:197).

2.3.6.1 Resonance

Resonance, in relation to speech, refers to the supraglottic modulation of the noise produced by the vocal cords. In syndromes of craniosyntosis, hyponasal resonance during speech is a common finding, which is due to the stenotic suture lines between the facial bones and cranial base (Shprintzen, 2000:197; Kummer, 2001:451). Resonance is said to be hyponasal if no nasal resonance is present where there should be some (as in /m/, /n/, and /ng/).

2.3.6.2 Articulation

Articulation is probably the component of speech that is most consistently impaired in individuals with Crouzon syndrome, due to the malocclusion that is so common in syndromes of craniosynostosis (Shprintzen, 2000:200). Articulation errors can be grouped into four basic categories on the basis of congenital malformations: developmental, obligatory, compensatory, and neurogenic articulation errors.

Developmental articulation errors, which are errors not caused by structural anomalies, also occur in many individuals in the general population. Obligatory errors are influenced by abnormal anatomy that makes the correct acoustic pronunciation of a sound almost impossible. It is thus strongly recommended that the structural anomaly be resolved before speech therapy is introduced. Compensatory substitutions are alternative strategies used by many patients with Crouzon syndrome to replicate the correct acoustic production by altering the place or manner of articulation. For example, the individual with Crouzon syndrome may have difficulty putting his/her lips together to make a /p/ or /b/ sound, and

thus, in an attempt to create an acceptable-sounding substitute for /p/ or /b/, may curl the tongue towards the upper lip to produce the sound. Lastly, neurogenic errors are those associated with disorders in which the central nervous system is impaired, therefore neurogenetic disorders of speech are uncommon in Crouzon syndrome. Thus sluggishness, slurring of speech, dysarthria, poor planning of speech, or dyspraxia are not common articulation errors found in Crouzon syndrome (Shprintzen, 2000:200).

2.3.6.3 Voice

Another aspect of speech that is often affected in syndromes of craniosynostosis is the volume of voice. This stands to reason, as conductive hearing loss is a common feature in Crouzon syndrome. Individuals with conductive hearing loss have reduced voice volume because they hear their own voices more loudly, in comparison to other sounds in their environment (Shprintzen, 2000:202).

2.3.7 Language and learning disabilities

According to Kummer (2001:133), learning disabilities are also commonly found in individuals with craniofacial syndromes and these learning problems can also affect language. Language may be defined as the ability to use a system of symbols, sounds or signs to convey meaning to another person. Therefore, normal language development is heavily dependent on the central nervous system, more specifically the brain, and thus any impairment or anomaly of the brain may result in language impairment (Shprintzen, 2000:202).

In syndromes of craniosynostosis, there are a number of possible contributors to language impairment, some being primary and others secondary. While most individuals with Crouzon syndrome have normal intellect, the secondary effects of increased intracranial pressure resulting in brain damage or hydrocephalus could potentially result in language impairment, discoordination of the speech mechanism and cognitive impairment. However, with the availability of advanced forms of cranial surgery currently available, it is now unusual to see children with Crouzon syndrome who have abnormal language or intellectual function (Shprintzen, 2000:202).

According to Shprintzen (2000:202), language disorders are common in children with hydrocephalus who have otherwise normal brain and craniofacial development. Therefore, when hydrocephalus occurs in a child with Crouzon syndrome, it may be inferred that the effect is additive.

Another major concern with regard to language development is the presence of persistent conductive hearing loss. It is not known whether the language impairment observed in patients with Crouzon syndrome is compounded to any extent by the presence of conductive hearing loss, but it is possible that the effect is additive. It is important to realise that abnormal development of the brain as a cause of Crouzon syndrome cannot be corrected, but early intervention for language stimulation and development should be scheduled as soon as possible (Shprintzen, 2000:202). These language delays can be addressed through the use of speech therapy (Chen, 2006:8).

Kummer (2001:133) states that although intellectual ability may test within normal limits in the early years, the intelligence scores tend to deteriorate in later years due to the individual's significant difficulty with abstract thinking. Mathematics and reading comprehension are therefore the most severely affected learning skills due to this difficulty with abstraction.

2.4 CAUSES OF CROUZON SYNDROME

Crouzon syndrome is inherited through an autosomal dominant pattern that can be caused by mutations in the fibroblast growth factor receptor 2 (FGFR2) gene, which is highly localized to two axons termed IIIa and IIIc and accounts for the majority of classical cases (Glaser *et al.*, 2000:768; de Ravel *et al.*, 2005:505).

In autosomal dominant disorders, heterozygous individuals have a recognisable phenotype, which is a group of characteristics associated with the genetic condition. Autosomal dominant pedigrees will frequently show that a parent is affected and many cases are associated with paternal age older than 35 years. However, if neither parent is affected, the affected individual is presumed to carry a new mutation that is causing the condition (Kummer, 2001:44). According to Kabbani and Raghuveer (2004:2868), nearly 60 percent of cases are not inherited and are the result of new mutations. Crouzon syndrome is

therefore thought to arise due to a mutation in both sporadic and inherited cases (Singer *et al.*, 1997:11).

Recent research identified a new cluster of relatively rare FGFR2 mutations within the intracellular tyrosine kinase domain (de Ravel *et al.*, 2005:503). According to Tsai *et al.* (2001:265) it is very likely that Crouzon syndrome is a heterogeneous phenotype and that other mutations in the FGFR2 gene or other genes not yet identified may be responsible for the Crouzon phenotype. However, a specific point mutation in the FGFR3 gene has also been shown to result in Crouzon syndrome associated with acanthosis nigricans (Abou-Sleiman *et al.*, 2002:304). Approximately 5% of patients with Crouzon syndrome have acanthosis nigricans, which can be identified by darkened thickened skin with accentuated markings and a velvety feel (Chen, 2006:4).

2.5 SYNOPSIS

In this chapter the manifestations and pathogenesis of Crouzon syndrome were described. According to research findings, several learning areas might be compromised due to compromised postnatal cortical development related to premature fusion of different sutures. In the following chapter the impact on these different learning areas are further investigated.

2.6 LIST OF REFERENCES

Abou-Sleiman, P.M., Apessos, A., Harper, J.C., Serhal, P. & Delhanty, J.D.A. (2002). Pregnancy following pre-implantation genetic diagnosis for Crouzon syndrome. *Molecular Human Reproduction*, **8**(3): 304-309.

Bates, E. (1999). Plasticity, location and language development. In S. Broman & J. Fletcher (Eds.), *The changing nervous system* (pp. 214-253). New York: Oxford University Press.

Becker, L., Armstrong, D., Chand, F., & Wood, M. (1984). Dendritic development in the human occipital cortical neurons. *Developmental Brain Research*, 13: 117-124.

Bolz, J., Castellani, V., Mann, F. & Henke-Fahle, S. (1996). Specification of layer-specific connections in the developing cortex. *Progress in Brain Research*, 108: 41-54.

Brody, B., Kinney, H., Kloman, A., & Gilles, F. (1987). Sequence of central nervous system myelination in human infancy: An autopsy study of myelination. *Journal of Neuropathology & Experimental Neurology*, 46: 283-301.

Broman, S. & Fletcher, J. (Eds.). 1999. *The changing nervous system*. New York: Oxford University Press.

Camfield, P.R., Camfield, C.S. & Cohen, M.M. (2000). Neurologic Aspects of Craniosynostosis. In Cohen, M.M. & MacLean, R.E. (eds). *Craniosynostosis: Diagnosis, Evaluation, and Management* (2nd ed.). Oxford: Oxford University Press.

Chen, H. (2006). *Crouzon Syndrome*. eMedicine. Retrieved from the World Wide Web on 17 March 2006 from: http://www.emedicine.com/ped/topic511.htm

Cohen, M.M, (2000). Crouzon Syndrome. In Cohen, M.M. & MacLean, R.E. (eds). *Craniosynostosis: Diagnosis, Evaluation, and Management* (2nd ed.). Oxford: Oxford University Press.

Chugani, H., Phelps, M., & Mazziotta, J. (1987). Positron emission tomography study of human brain functional development. *Annals of Neurology*, 22: 487-497.

De Ravel, T.J.L., Taylor, I.B., Van Oostveldt, A.J.T., Fryns, J.P. & Wilkie, A.O.M. (2005). A further mutation of the FGFR2 tyrosine kinase domain in mild Crouzon syndrome. *European Journal of Human Genetics*, **13**: 503-505.

Giedd, J., Snell, J., Lange, N., Rajapakse, J., Casey, B., Kozuch, P., Vaituzis, A., Vauss, Y., Hamburger, S., Kaysen, D., & Rapoport, J. (1996). Quantitative magnetic resonance imaging of human brain development: Ages 4 to 18. *Cerebral Cortex*, 6: 551-560.

Glaser, R.L., Jiang, W., Boyadjiev, S.A., Tran, A.K., Zachary, A.A., Van Maldergem., Johnson, D., Walsh, S., Oldridge, M., Wall, S.A., Wilkie, A.O.M. & Jabs, E.W. (2000).

Paternal Origin of FGFR2 Mutations in Sporadic Cases of Crouzon Syndrome and Pfeiffer Syndrome. *American Journal of Human Genetics*, **66**: 768-777.

Goldman-Rakic, P. (1987). Development of cortical circuitry and cognitive function. *Child Development*, 58: 601-622.

Harbord, M., Finn, J., Hall-Cragg, M., Robb, S., Kendall,B., & Boyd, S. (1990). Myelination patterns on magnetic resonance of children with developmental delay. *Developmental Medicine & Child Neurology*, 32: 295-303.

Huppi, P., Warfield, S., Kikinis, R., Barnes, P., Zientara, G., Jolesz, F., Tsuji, M., & Volpe, J. (1998). Quantitative magnetic resonance imaging of brain development in premature and mature newborns. *Annals of Neurology*, 43: 224-235.

Huttenlocher, P. (1979). Synaptic density in human frontal cortex: Developmental changes and effects of aging. *Brain Research*, 163: 195-205.

Huttenlocher, P. (1984). Synapse elimination and plasticity in developing human cerebral cortex. *American Journal of Mental Deficiency*, 88: 488-496.

Huttenlocher, P. & de Courten, C. (1987). The development of synapses in striate cortex of man. *Human Neurobiology*, 6: 1-9.

Huttenlocher, P., & Dabholkar, A. (1997). Regional differences in synaptogenesis in human cerebral cortex. *The Journal of Comparative Neurology*, 387: 167-178.

Kabbani, H. & Raghuveer, T. (2004). Craniosynostosis. *American Family Physician*, **69**(12): 2863-2870.

Katz, L. & Shatz, C. (1996). Synaptic activity and the construction of cortical circuits. *Science*, 274: 1133.

Khan, A. (2000). *Craniofacial Anomalies: A Beginners Guide for Speech-Language Pathologists*. California: Singular Publishing Group, Inc.

Koenderink, M., Uylings, H., & Mrzljak, L. (1994). Postnatal maturation of the layer III pyramidal neurons in the human prefrontal cortex: A quantitative Golgi analysis. *Brain Research*, 653: 173-182.

Kolb, B. & Whishaw, I.Q. (2003). *Fundamentals of Human Neuropsychology*, fifth edition. New York: Worth Publishers.

Korsching, S. (1993). The neurotrophic factor concept: A reexamination. *Journal of Neuroscience*, 13: 2739-2748.

Kostovic, I., Seress, L., Mrzljak, L., & Judas, M. (1989). Early onset of synapse formation in the human hippocampus: A correlation with Nissl Golgi architectonics in 15- and 16.5-week old fetuses. *Neuroscience*, 30: 105-116.

Kummer, A.W. (2001). *Cleft Palate and Craniofacial Anomalies: Effects on Speech and Resonance*. San Diego: Singular Publishing Group, Inc.

LaMantia, A., & Rakic, P. (1990). Axon overproduction and elimination in the corpus callosum of the developing rhesus monkey. *Journal of Neuroscience*, 10: 2156-2175.

Lyon, G., Arita, F., Le Galloudec, E., Valee, L., Misson, J., & Ferriere, G. (1990). A disorder of axonal development, necrotizing, myopathy, cardiomyopathy, and cataracts: A new familiar disease. *Annals of Neurology*, 27: 193-199.

Michel, A., & Garey, L. (1984). The development of dendritic spines in the human visual cortex. *Human Neurobiology*, 3: 223-227.

Molliver, M., Kostovic, I., & Van der Loos, H. (1973). The development of synapses in the human fetus. *Brain Research*, 50: 403-407.

Mrzljak, L., Uylings, H., Van Eden, C., & Judas, M. (1990). Neuronal development in human prefrontal cortex in prenatal and postnatal stages. *Progress in Brain Research*, 85: 185-222.

Orvidas, L.J., Fabry, L,B., Diacava, S. & McDonald, T,J. (1999). Hearing and Otopathology in Crouzon Syndrome. *Laryngoscope*, **109**(9): 1372-1375.

Posnick, J.C. (2000). *Craniofacial and Maxillofacial Surgery in Children and Young Adults*, Volume One. Philadelphia: W.B. Saunders Company.

Purpura, D., Bodick, N., Suzuki, K., Rapin, I., & Wurzelmann, S. (1982). Microtubules disarray in cortical dendrites and neurobehavioral failure. Golgi and electron microscopic studies. *Brain Research*, 281: 287-297.

Reiss, A., Abrams, M., Singer, H., Ross, L., & Denckla, M. (1996). Brain development, gender and IQ in children. A volumetric imaging study. *Brain*, 119: 1763-1774.

Shprintzen, R.J. (2000). Speech and Language Disorders in Syndromes of Craniosynostosis. In Cohen, M.M. & MacLean, R.E. (eds). *Craniosynostosis: Diagnosis, Evaluation, and Management* (2nd ed.). Oxford: Oxford University Press.

Singer, S.L., Walpole, I., Brogan, W.F. & Goldblatt, J. (1997). Dentofacial features of a family with Crouzon syndrome. Case reports. *Australian Dental Journal*, **42**(1): 11-17.

Sowell, E., Thompson, P., Holmes, C., Batth, R., Jernigan, T., & Toga, A. (1999). Localizing age-related changes in brain structure between childhood and adolescence using statistical parametric mapping. *Neuroimage*, 9: 587-597.

Sulica, R.L. & Grundfast, K.M. (2000). Otologic Manifestaions of Craniosyntosis Syndromes. In Cohen, M.M. & MacLean, R.E. (eds). *Craniosynostosis: Diagnosis, Evaluation, and Management* (2nd ed.). Oxford: Oxford University Press.

Tasashima, S., & Mito, T. (1985). Neuronal development in the medullary reticular formation in sudden infant death syndrome and premature infants. *Neuropediatrics*, 16: 76.

Taylor, H., & Alden, J. (1997). Age-related differences in outcomes following childhood brain insults: An introduction and overview. *Journal of the International Neuropsychological Society*, 3: 555-567.

Teicher, M.H., Andersen, S.L., Polcari, A., Anderson, C.M., & Navalta, C.P. 2002. Developmental neurobiology of childhood stress and trauma. *Psychiatric Clinics of North America*, 25(2):397-426. http://minedump.redi.co.za:2198/das/article/body/46518972-2/jorg-journal&source=… MD Consult website accessed on 2006/05/23.

Tessier-Lavigne, M. & Goodman, C. (1996). The molecular biology of axon guidance. *Science*, 274: 1123.

Thoenen, H. (1995). Neurotrophins and neuronal plasticity. *Science*, 279: 593-598.

Tsai, F.J., Yang, C.F., Wu, J.Y., Tsai, C.H. & Lee, C.C. (2001). Mutation analysis of Crouzon syndrome and identification of one novel mutation in Taiwanese patients. *Paediatrics International*, **43**: 263-266.

Vajo, Z., Francomano, C.A. & Wilkin, D.J. (2000). The Molecular and Genetic Basis of fibroblast Growth Factor Receptor 3 Disorders: The Achondroplasia Family of Skeletal Dysplasias, Muenke Craniosyntosis, and Crouzon Syndrome with Acanthosis Nigricans. *Endocrine Reviews*, **21**(1): 23-39.

Vargha-Khadem, F., Gadian, D., Watkins, K., Connelly, A., Van Paesschen, W., & Mishkin, M. (1997). Differential effects of early hippocampal pathology on episodic and semantic memory. *Science*, 277(5324): 376-380.

Webb S.J., Monk, C.S. & Nelson, C.A. (2001). Mechanisms of Postnatal Neurobiological Development: Implications for Human Development. *Developmental Neuropsychology*, **19**(2): 147-171.

CHAPTER 3

THE EMPIRICAL STUDY

3.1 INTRODUCTION

The preceding chapters described the nature of Crouzon syndrome. A critical review of the relevant literature suggested that programmed neural development might be compromised due to intracranial pressure caused by premature fusion of the sutures, which might result in compromised affect and learning. This chapter describes the empirical research and related findings. The purpose of this research project is to scrutinize the neural makeup of Crouzon syndrome on the basis of a clinical case study, in order to compile a detailed explanatory profile of compromised affect and learning associated with Crouzon syndrome. Based upon the findings, the research hypothesis will be accepted or rejected.

3.2 PROBLEM STATEMENT

Once the nature of Crouzon syndrome had been conceptualized, the research problem was formulated as follows:

What is the relationship between developmental brain insults associated with Crouzon syndrome and compromised affect and learning?

3.2.1 Sub-questions

The following sub-questions support the main research question:

- What atypical brain development associated with Crouzon syndrome can be identified through MR imaging and radiographic techniques?
- Which brain areas are typically lesioned where Crouzon syndrome is present?
- How does atypical brain architecture and circuitry associated with Crouzon syndrome relate to compromised affect and learning?

3.2.2 Research hypothesis

The following research hypothesis was formulated for this research project:

There is a neural substrate to compromised affect and learning associated with Crouzon syndrome, resulting in a particular affective and learning profile.

3.3 PURPOSE OF THE STUDY

The purpose of this research project is to scrutinize the neural makeup of Crouzon syndrome on the basis of a clinical case study, in order to compile a detailed explanatory profile of compromised affect and learning associated with Crouzon syndrome.

3.4 RESEARCH DESIGN

Mixed-method research designs are usually employed in order to collect, analyse and combine both quantitative and qualitative data in a single study in order to better understand a research problem. In this study, an exploratory mixed-method design was implemented, as the initial emphasis had been on gathering qualitative data to explore the phenomenon of Crouzon syndrome, followed by quantitative data collection in order to explain relationships found in the qualitative data (Creswell, 2005:516).

3.5 METHODS, MATERIALS AND PROCEDURES

Before the process of data collection begins, it is imperative to identify boundaries or parameters for the data collection and the method of data analysis (De Vos, 2000:42). Thus, in this chapter, the emphasis is on providing a detailed description of the specific measuring instruments that were employed, as well as a description of the related test results.

This research project comprises of a single case study of a girl diagnosed with Crouzon syndrome who, at the time of test taking, was 9 years and 10 months of age and in Grade 3 (hereafter referred to as *the research participant*). Information was obtained from case history data, which included a detailed interview with the parents concerning prenatal,

perinatal and postnatal developmental histories, clinical, neurological and psychological assessment data, paramedical reports, scholastic progress reports, and social adjustment data. The following data generating strategies were employed:

3.5.1 Interviews

Qualitative interviews were conducted with the research respondent's parents and teacher. These interviews might be viewed as 'guided approaches', where the topics for discussion were listed on an interview schedule, allowing for topics to be rephrased during the interview, if necessary, and to be covered by the interviewer in any sequence fitting to the nature of the interview (Johnson & Turner, 2003:305-306).

3.5.2 Standardized tests and questionnaires

Standardized tests and questionnaires are tools for measuring, observing, or documenting quantitative data. They are used to measure achievement, intelligence, aptitude, personality and self-perceptions (Johnson & Turner, 2003:155).

The term standardized refers to those tests and questionnaires that have been standardized for a certain norm population and the test results are usually expressed in terms of a norm score. A norm score could be a stanine, a sten score or a percentile ranking. The test results are thus quantified in order to obtain a valid comparison (Naudé, 1998:14).

3.6 DATA-GENERATING STRATEGIES

Optometric, cognitive, neuropsychological and personality assessment were employed to complement brain-imaging studies. In order to meet these demands, the following instruments were administered: the Senior South African Individual Scale – Revised (SSAIS-R) (Van Eeden, 1992), the Quick Neurological Screening Test (Mutti, Sterling & Spalding, 1998), the Wisconsin Card Sorting Test (Heaton, Chelune, Talley, Kay & Curtiss, 1993), the Children's Personality Questionnaire (Du Toit & Madge, 1988), the Children's Apperception Test (CAT) (Bellak & Bellak, 1965), graphic media (DAP and K-F-D), and the *Southern California Sensory Integration Tests* (Ayers, 1972). A brief

description of the nature and applicability of each of these instruments is provided in **Annexture B** to this dissertation.

3.7 RESULTS OF THE CASE STUDY

At the time of assessment the participant was 9 years and 10 months old and in Grade 3. She is the younger of two children and her sister is in Grade 8. The clinical background, as reported by the parents, as well as the obstetric and developmental data relating to the research participant is presented in the following paragraphs:

3.7.1 Obstetric and developmental data

The pregnancy was normal and the participant was born by caesarean section without any complications. She was born two weeks prematurely and weighed 2,98 kg. She reached her physical milestones within the normal limits, started walking at the age of 12 months, and concurrently started using single-syllable words such as ‘mom and ‘dad’. At the age of two years craniosynostosis, caused by the early fusion of the bones of the skull that resulted in a misshapen head, was *first observed*. However, the research participant was *only diagnosed* with Crouzon syndrome at the age of three years, and at the age of three years and one month this diagnosis was followed by surgery during which the fused cranial bones were loosened to prevent any further intracranial pressure. The procedure lasted five and a half hours. Following surgery, the research participant suffered partial loss of speech and her language development was generally delayed. There was limited sparing of vocabulary and the research participant had to relearn known words, e.g. the names of colours and shapes, which progressed slowly. Following this surgery at the age of three years, she received speech and occupational therapy, followed by physiotherapy from the age of five years.

3.7.2 Scholastic data

The research participant started school at a mainstream primary school, but was referred to a school for neurally impaired children where she repeated Grade 1. She is still attending this school and, according to her parents, she struggles with creative writing. Other problems experienced include the reversal of letters and words. At times she is reluctant to

attend school. It is suspected that she presents with a low academic self-concept. Regarding her schoolwork, she becomes aggressive and frustrated. She unfavourably compares herself to other children and feels that she is different. The research participant reportedly also realizes that other children can master things more quickly than she does. Furthermore, her parents have noticed that it takes her some time to grasp new ideas. Until April 2006, the drug *Concerta*™ was administered to assist with concentration in class.

3.7.3 Social adjustment data

The research participant reportedly experiences social interaction with her peers as problematic and struggles to read social queues. Whereas she reportedly experiences feelings of alienation among her peers, she enjoys playing with younger children and during these interactions she prefers to take the lead. However, when she spends time with older children and adults she likes to be babied. According to the parents' self-reports, she often misinterprets many things within her social environment and she does not always understand what people are saying to her. She might be described as stubborn and she likes things to be done her way. At times she might become aggressive and frustrated. She does not respond well to change and prefers a fixed routine. The research participant dislikes loud noises, but she likes music, colouring in and helping with household chores. She also likes making up her own games that require the involvement of other people. She reportedly talks to herself regularly, and she enjoys talking to the dogs and pretends to teach them things.

3.8 DATA ANALYSIS AND INTERPRETATION

The empirical data are dealt with under the following headings: cognitive functioning, neurological screening, screening for frontal lobishness, personality structure, visual perceptual ability, auditory perceptual ability, and samples of handwriting.

3.8.1 Cognitive functioning

The Senior South African Individual Scale (SSAIS-R) was administered and the results are presented in Table 3.1 below. Following the administration of the SSAIS-R, the research participant was referred for optometric assessment and far-sightedness was diagnosed. After a period of six months those subtests of the SSAIS-R that heavily rely on visual acuity were re-administered after optometric correction by means of prescription spectacles had been done. The results of the second administration are presented in Table 3.2 below to allow for a comparison of the two sets of data.

Table 3.1 Scatter analysis of the SSAIS-R <u>before</u> optometric correction

Subtests	Scaled score	Deviation	Significant deviation	
			5% level	1 % level
Verbal Scale: 86 (low average)				
Vocabulary	8	+0.2	1.93	2.54
Comprehension	9	+1.2	2.40	3.16
Similarities	7	- 0.8	2.33	3.07
Number Problems	4	-3.8	2.11*	2.78**
Story Memory	11	+3.2	2.18*	2.86**
Sum of scaled scores	39			
Average	7.8			
Memory for Digits	8	+0.2	2.79	3.76
Non-verbal Scale: 61 (cognitively handicapped)				
Pattern Completion	4	-0.5	2.56	3.36
Block Designs	6	+1.5	2.05	2.69
Missing Parts	3	-1.5	2.87	3.77
Form Board	5	+0.5	2.72	3.57
Sum of scaled scores	18			
Average	4.5			
Coding	3	-1.5	3.27	4.30
Full Scale: 72 (borderline intellectual functioning)				
Vocabulary	8	+1.7	2.04	2.68
Comprehension	9	+2.7	2.61*	3.43
Similarities	7	+0.7	2.53	3.33
Number Problems	4	-2.3	2.26*	2.97
Story Memory	11	+4.7	2.34*	3.07**
Pattern Completion	4	-2.3	2.74	3.60
Block Designs	6	-0.3	1.96	2.58
Missing Parts	3	-3.3	3.18*	4.19
Form Board	5	-1.3	2.97	3.90
Sum of scaled scores	57			
Average	6.3			
Memory for Digits	8	+1.7	2.73	3.59
Coding	3	-3.3	3.05*	4.01

Tables of norms used: Non-Environmentally Disadvantaged Norm Group
SED index: High-average to advantaged Socio Economic Status (SES)
* = significant deviation at 5% level
** = significant deviation at 1% level

Table 3.2 Scatter analysis of the SSAIS-R <u>after</u> optometric correction

Subtests	Scaled score	Deviation	Significant deviation	
			5% level	1 % level
Verbal Scale: 86 (low average)				
Vocabulary (re-do)	8	+0.2	1.88	2.48
Comprehension	9	+1.2	2.49	3.28
Similarities	7	- 0.8	2.40	3.15
Number Problems	4	-3.8	2.13*	2.81**
Story Memory	11	+3.2	2.16*	2.84**
Sum of scaled scores	39			
Average	7.8			
Memory for Digits	8	+0.2	2.79	3.76
Non-verbal Scale: 76 (borderline intellectual functioning)				
Pattern Completion (re-do)	8	+1.5	2.42	3.18
Block Designs (re-do)	6	-0.5	2.04	2.69
Missing Parts (re-do)	7	+0.5	3.03	3.99
Form Board	5	-1.5	2.66	3.51
Sum of scaled scores	26			
Average	6.5			
Coding (re-do)	2	-4.5	3.02*	3.98**
Full Scale: 79 (borderline intellectual functioning)				
Vocabulary (re-do)	8	+0.8	1.95	2.57
Comprehension	9	+1.8	2.69	3.54
Similarities	7	-0.2	2.58	3.39
Number Problems	4	-3.2	2.26*	2.97**
Story Memory	11	+3.8	2.29*	3.02**
Pattern Completion (re-do)	8	+0.8	2.55	3.35
Block Designs (re-do)	6	-1.2	1.96	2.59
Missing Parts (re-do)	7	-0.2	3.41	4.49
Form Board	5	-2.2	2.90	3.82
Sum of scaled scores	65			
Average	7.2			
Memory for Digits	8	+0.8	2.86	3.77
Coding (re-do)	2	-5.2	2.79*	3.67**

Tables of norms used: Non-Environmentally Disadvantaged Norm Group
SED index: High-average to advantaged Socio Economic Status (SES)
* = significant deviation at 5% level
** = significant deviation at 1% level

3.8.1.1 Analysis and interpretation of results

The research participant's general intellectual functioning before and after optometric correction is at borderline level (Full Scale 72 and 79 respectively) (Van Eeden, 1992:48). The verbal-auditory processing before and after optometric correction is at low average level of intellectual functioning (Verbal Scale 86) (Van Eeden, 1992:48), while the visual-motor processing before optometric correction is at cognitively handicapped level (Non-verbal Scale 61), but rises to borderline level after optometric correction (Non-verbal Scale

76) (Van Eeden, 1992:48), indicating a gain of 15 non-verbal IQ points after optometric correction.

The scaled score difference between the Verbal and the Non-verbal Scales before optometric correction is statistically significant at the 1% level of reliability (Van Eeden, 1992:57), but after optometric correction no statistically significant difference between these two scales is noted. Before optometric correction there was a twenty-six point superiority of Verbal over Non-verbal IQ, suggesting an advantage of auditory-verbal abilities (e.g. subtests Vocabulary, Comprehension, Similarities, Story Memory) over constructional praxis tasks involving visual-motor abilities (e.g. subtest Pattern Completion, Block Designs, Form Board). After optometric correction the auditory-verbal superiority over visual-motor abilities prevailed, yet was no longer statistically significant. Before optometric correction only 7% of subjects from the norm population showed a similar difference between the Verbal and the Non-verbal scales (Van Eeden, 1992:58); after optometric correction this difference decreased to equal the performance of 52% of subjects from the norm population (Van Eeden, 1992:58). This latter verbal/non-verbal difference after optometric correction is therefore ascribed to better developed crystallized abilities that were acquired through previous training, education and acculturation, as opposed to fluid abilities that involve immediate problem solving and reasoning (without prior experience). Immediate problem solving and reasoning require cognitive adaptability and flexibility when faced with an unfamiliar situation, and the inference is that the initial verbal/non-verbal difference before optometric correction could be ascribed to peripheral visual deficits (far-sightedness), while the latter difference after optometric correction might be ascribed to differences in central processing of information, as expressed by *inter alia*:

- a relative sparing and/or recovery of those cortical areas that process auditory-verbal information, while the cortical areas involved in visual-motor tasks, constructional praxis, planning, and problem solving are more significantly lesioned;
- typical expressions of and preferences in respect of cognitive style;
- sensory and/or visual perceptual deficiencies (e.g. due to suspected lesioning of the optic nerve due to intracranial pressure);

- weaknesses in the processing of visual-motor information, e.g. due to suspected lesioning of the parietal, temporal and frontal lobes; and
- gains in the processing of auditory-verbal information due to ongoing speech and language therapy following surgery.

A higher verbal than non-verbal performance can thus be ascribed to:

- language abilities that are better developed than non-verbal abilities;
- auditory processing modalities that were spared and/or that have recovered to a greater extent than the visual non-verbal modalities;
- difficulties with comprehension and the execution of practical tasks associated with lesioning of specific cortical areas involved in cortical processing of this nature;
- possible frontal lobishness associated with limited skill in respect of planning and the execution of motor tasks;
- lack of willingness to venture in the execution of motor tasks, for example due to certain personality traits associated with frontal lobishness and/or lesioning of other cortical areas that are linked to affect and personality, as well as parental overprotection that might have set in following the initial diagnosis, surgery and attempts at recovery through ongoing therapy;
- subtle cortical lesioning due to intracranial pressure, because not all lesions are visible by means of magnetic resonance imaging (MR-imaging), e.g. in closed-head injury.

These preceding interpretations are further refined by means of analyses of the subtest scatter. Statistically significant subtest scatter was found on the verbal and full scales at the 1% and 5% levels of reliability before and after optometric correction. Relative to the research participant's borderline level of general intellectual functioning (Full scale performance) before optometric correction, she performed significantly higher on subtest **Story Memory** (+4.7 at both 5% and 1% levels of reliability) and on subtest **Comprehension** (+2.7 at 5% level of reliability). However, relative to the research participant's borderline level of general intellectual functioning (Full scale performance) before optometric correction, she performed significantly lower on subtest **Number Problems** (-2.3 at 5% level of reliability), on subtest **Missing Parts** (-3.3 at 5% level of reliability) and on subtest **Coding** (-3.3 at 5% level of reliability). Before optometric

correction almost similar deviations were found on the Verbal Scale, i.e. a significantly higher performance on subtest **Story Memory** (+3.2 at both the 5% and 1% levels of reliability), and a significantly lower performance on subtest **Number Problems** (-3.8 at both the 5% and 1% levels of reliability).

However, some of the preceding deviations from the respective average scaled scores, i.e. average Verbal, Non-verbal and Full Scale scores, are ascribed to the research participant's far-sightedness, since only some of these deviations were preserved on the scatter profile after optometric correction, which suggests that the improved performance on certain non-verbal, vision-dependent subtests could be ascribed to improved functional vision after optometric correction, while deviations on subtests that are only partially vision dependent or non-vision-dependent could be ascribed to central processing deficits, which would explain why these deviations still persist after optometric correction. Although it is acknowledged that far-sightedness may result from defiant eye muscle tone, as well as from swelling of the optic disk (papilledema) due to intracranial pressure (Kolb & Whishaw, 2003:709), far-sightedness is expressed as a peripheral 'hardware' deficit, while persisting deviations on subtests that are only partially vision dependent or non-vision-dependent are ascribed to impaired central processing due to lesioning of certain cortical areas brought about by Crouzon syndrome.

Persisting deviations on the scatter profile after optometric correction were found on all three scales, i.e. on the Verbal, the Non-verbal and the Full Scales. On the Verbal Scale a statistically significant positive deviation at both the 5% and 1% levels of reliability was found on subtest **Story Memory** (+3.2), indicating higher performance within this specific modality. In addition, a statistically significant negative deviation at both the 5% and 1% levels of reliability was found on subtest **Number Problems** (-3.8), indicating lower performance within this specific modality. Statistically significant deviations pertaining these two subtests were also noted on the Full Scale after optometric correction, i.e. subtest **Story Memory** (+3.8) and subtest **Number Problems** (-3.2). On the Non-verbal Scale a statistically significant negative deviation at both the 5% and 1% levels of reliability was found on subtest **Coding** (-4.5), indicating lower performance within this specific modality. This negative deviation also repeated on the Full Scale, i.e. subtest **Coding** (-5.2). The following interpretations are offered pertaining to the research participant's statistically significant higher and lower performances on these respective subtests:

A relatively higher performance on subtest **Story Memory** implies that the research participant shows adequate short-term verbal memory and recall, yet this performance is merely average in terms of the norm population. 'Short-term and long-term memory are systems in which material is processed separately and simultaneously (Kolb & Whishaw, 2003:475). Lesions at the junction of the parietal, temporal and occipital cortexes usually produce short-term-memory deficits; yet the research participant's average perfomance on verbal memory tasks suggests a relative sparing of these areas, contrary to her low performance on the Non-verbal items of the SSAIS-R and the QNST-II, which suggests impaired temporal lobe functioning. This contrast is explained by the nature of the verbal content of subtest Story Memory. Since one can relate to this story by relying on past experiences, it seems that the research participant relied on her long-term memory system, allowing recognition and retrieval of several of the stimulus words inherent to subtest Story Memory from her long-term memory. The results might therefore be biased towards a false positive strength.

Kolb and Whishaw (2003:475) point out that short-term memory can be doubly dissociated from long-term memory with respect to the kinds of impairments seen in the different systems and with respect to the different kinds of structural damage from which those deficits arise. For example, one patient's left posterior temporal lesion resulted in an almost total inability to repeat verbal stimuli such as digits, letters, words, and sentences. In contrast, this patient's long-term recall of paired-associated words and short stories was nearly normal. These researchers further report that some patients with temporal lesions apparently have defects in short-term recall of visually presented digits or letters, but have normal short-term recall of the same stimuli presented aurally, while other patients present with just the opposite difficulty: specific deficits for aurally presented items, but not visually presented verbal items, because short-term-memory deficits can also result from damage to the polymodal sensory areas of the posterior parietal cortex and posterior temporal cortex, or from lesioning of the pathways linking these areas to the frontal lobe, or from lesioning of the two systems for short-term memory in the frontal cortex itself, i.e. Brodmann's area 8 and areas 9 and 46 respectively (Kolb & Whishaw, 2003:477). Therefore, in this case of prolonged intracranial pressure it might be almost impossible to pinpoint specific areas of sparing and/or lesioning, as we will learn from the research participant's MR images, which will be presented in the next chapter. Lesioning might rather be diffuse, following a mosaic pattern across the entire cortex because, due to their

immaturity, some areas are more vulnerable than others, resulting in a wide spectrum of diffuse impairments, coupled with an overall decrease in general intellectual capacity, as observed in this research participant's intellectual profile (SSAIS-R).

The research participant could master only the first three concrete items on subtest **Number Problems.** This suggests dyscalculia, which refers to extreme difficulty in performing arithmetical operations (Kolb & Whishaw, 2003:G-8). The research participant's ability to do mental arithmetic is so poor that she could not solve even simple additions and subtractions, which suggests lesioning of the parietal, temporal and frontal areas, with a specific focus on the left parietal area.

A lower performance on the subtest **Coding** suggests poor visual-associative learning. Poor figure recognition and production is associated with inadequate handwriting performance, but also with poor visual-motor integrative skills development, because it measures non-verbal concept formation, including perceptual organisation, spatial visualisation and orientation. Mainly three cortical lobes are involved in processing the information required in order to perform in the subtest Coding, i.e. the parietal, the temporal and the frontal lobes, with a strong focus on frontal-lobe functioning. Although visual memory does play a role, the problem more likely lies in learning to select, from a set of competing stimuli, the appropriate ones for the execution of this task (Kolb & Whishaw, 2003:410), which suggests frontal-lobe lesioning. Damage to either the left or the right frontal lobe results in an impaired ability to learn arbitrary associations between one kind of stimulus and another (Kolb & Whishaw, 2003:410).

3.8.2 Neurological screening

The Quick Neurological Screening Test – Revised Edition (QNST-II) was administered and the research participant obtained a suspicious total score on the QNST-II, which is indicative of developmental delays in certain areas. Mixed dominance was established, i.e. left eye, right hand and left foot dominance. The research participant avoids cursive writing and still prefers print. She experienced moderate to suspicious difficulty on the following subtests: Palm form recognition, eye tracking, sound patterns, finger to nose, thumb and finger circle (only regarding sequential memory), rapidly reversing repetitive hand movements, arm and leg extension, tandem walk (only regarding dynamic balance

with eyes closed), standing on one leg (only regarding dynamic balance with eyes closed) and skipping. The following behavioural irregularities were noted: perseveration, distractibility and impulsivity.

3.8.2.1 Analysis and interpretation of results

Print:

The preference given to print as opposed to cursive writing suggests deviations of muscle tone, which intuitively directs the research participant to printing, rather than doing cursive writing. Poor muscle tone could be associated with lesioning of the basal nuclei. The basal nuclei modulate the activation of upper motor neurons that are required for the normal initiation of voluntary movements, either during the anticipation of a movement or during the movement itself. Basal nuclei dysfunction results in the loss of a person's ability to switch smoothly between the cerebral commands that initiate a movement and those that terminate a movement. Lesions to the secondary somatosensory area of the parietal lobe also give rise to poorly controlled handwriting movements. Minor lesions to the motor and premotor cortices may disrupt the rhythm and automatism of handwriting (Luria, 1973). Apart from adequate muscle tone and fine motor skills development, numerous functional systems subserving language formulation, visio-spatial construction, praxic organization and so forth are involved in handwriting. Most often the cerebral lesion disturbs several systems so that the mechanism of poor hand skill and subsequent poor handwriting are complex in the majority of children with learning difficulties.

Palm form recognition:

'Palm Form Recognition' seems to tell us more about developmental readiness with regard to numbers and arithmetic than about neurological deficits. 'If number concepts are intact and arithmetic skills are adequate, then a deficit in Palm Form recognition is of broader neuro-educational concern. Sense of touch may be awry. A tactile deficit correlates with learning deficits' (Mutti *et al.*, 1998). In order to demonstrate fluent handwriting, the child has to have a clear image of the letters or numbers, and of various related shapes. Thus revisualization by means of haptic-tactile recognition and perception underlies handwriting proficiency at levels of automatism. A typical demonstration of such ability is when a child does not know how to spell a word, but when writing the word, he seemingly instinctively remembers how the word should be written. This ability is mediated by the

parietal and parts of the occipital lobes. The primary somatosensory area or zone of the parietal lobe is responsible for the interpretation of tactile and kinaesthetic sensation. This lobe mediates haptic processing, i.e. recognition and processing of sensual impressions from the skin, muscles, joints and vestibular organ. Lesions to the primary somatosensory area usually result in a loss of sensation with regards to touch, temperature, and position of the body. The secondary area mediates analysis, synthesis and interpretation of haptic information, i.e. the recognition of objects, numbers, letters, shapes, and so forth by means of touch and movement. Damage to the primary and secondary somatosensory zones of the parietal lobe might result in an inability to analyse and synthesize tactile-kinaesthetic information to such an extent that the person cannot recognize stimuli by means of touch, known as *astereognosis* (Kolb & Whishaw, 2003). Writing problems are thus to some extent the result of an inability to recall the visual image and symbols or the sequence of symbols by means of tactile processing of information.

Inability to detect touch might be due to impaired proprioception, indicating an inability to perceive objects using fine touch and pressure, disturbed perception of body awareness, and also disturbances in perceiving pain and temperature. The research participant might overreact very negatively to stimuli, and she might have been labelled tactile defensive. This condition might lead to behavioural difficulties among the peers, because she might tend to push around, bump into, and invade the personal space of others. She might be equally sensitive to certain sounds, flavours and textures, e.g. certain clothing, food textures and certain brands of toothpaste, which could lead to neglect of personal hygiene.

Eye tracking:

This task is given to determine whether both eyes are able to simultaneously track a visual stimulus, i.e. identification of horizontal or vertical jerkiness, incoordination, visual distractability, inability to cross the visual midline (associated with below par laterality), as well as poorly developed optic muscles. Simultaneous inputs from both eyes play an important role in merging the images from each eye. If the visual images cannot be merged, this might result in faulty saccades, reading problems and learning difficulties. If the muscles of either one of the eyes are poorly developed, this condition might result in inhibition of the functionality of one eye (the ‘lazy eye’). Consequently, the inhibited eye will be further inhibited by the remaining strong eye, and so the defect is reinforced. Even so, removal of the inhibition can permit some degree of recovery. In addition, if the

environment is so arranged that the visual system is exposed to stimuli of one type, the cells in the system develop a preference for those stimuli, and visual neglect might result (Kolb & Whishaw, 2003). Faulty eye movements might result in poor scanning ability, which plays a major role in reading, writing and learning.

Sound patterns:

This subtest determines the individual's ability to process auditory information in the form of sound patterns, and to reproduce patterns both orally and in terms of motor activity. It also measures auditory memory, because the individual must receive, visualize, retain and express the sound patterns either orally, or in terms of a motor response. Sound patterns are mainly processed in the right temporal lobes and the superior temporal gyrus. Inability to detect sound patterns might adversely impact on expressive language, spelling and reading. Owing to the possible inadequate development of the auditory pathways, she might relate visually to the world around her, with the result that ideas and directions that are stated verbally might never become part of the research participant's world, as her world can be decoded visually only. She may come across as being concretely oriented and nonverbal, therefore planning, decision-making and judging are not skills that would have been automatically developed by school age. Auditory ability is one of the most critical of the sensory-motor skills required for academic achievement, particularly reading. She may thus have had trouble with phonics and may have learnt reading and spelling visually or kinaesthetically. Owing to the difficulty she appears to have with processing auditory information, she may struggle when given more than one direction at a time and may come across as not paying attention. If bombarded with auditory information, the research participant may come across as being distractible, very verbal and often hyperactive, and she may act impulsively. She will show definite difficulties regarding sequencing and organising due to poor auditory processing ability.

Finger to nose:

Inability to perform this task points towards disturbed orientation of concrete space, associated with parietal lobe functioning. Results derived from this item suggest poor direction within personal space, i.e. the ability to discriminate between left and right, in front and behind, above and beneath, and the research participant is likely to experience difficulties regarding motor planning and control. Frontal cortex area 8 participates in short-term memory for the spatial location of objects, i.e. searching for an object, and it

receives projections from the parietal cortex. Thus, frontal cortex 8 (together with the parietal areas) is involved in spatial vision and search when a stimulus is presented, while frontal cortex areas 9 and 46 receive information from the inferior temporal cortex, and are involved in visual memory for objects that are identified sequentially (Kolb & Whishaw, 2003:477).

Significant scores on subtest 'Finger to Nose' also suggest poor sense of body placement. A poor sense of body placement carries over to objects outside the self, such as words on the written page or placement of the pencil in a specific place on the page. This may adversely impact on all learning areas, such as reading, writing and numerical reasoning where letters, words, and numbers are reversed (i.e. 'on' instead of 'no'), rotated ('b' instead of 'd'), or inverted ('p' instead of 'd'). Disorientation of concrete space is also associated with localization of sounds, and the research participant might find it hard to determine the origin of sounds from her environment. This may hamper her visual-auditory integration and might result in emotional insecurity.

Rapidly reversing repetitive hand movements:

This task is associated with rhythm and synchronization and also, to some degree, muscle tone. 'Many researchers find a positive relationship between a child's ability at rapidly reversing repetitive hand movements and school performance. … An unusual rate of execution, particularly if extremely slow, is evidence of poor motor-planning ability. Asymmetry says that messages from the central nervous system (CNS) are not translating into motor output in a balanced fashion. Because one side is receiving messages differently from the other, there will be problems in reading, hitting a baseball, running, or doing any task where the right and left sides need to work together' (Mutti *et al.*, 1998). The performance of rapidly alternating movements with the hands and/or fingers is a function of the frontal lobes, and inability to perform is associated specifically with lesioning of the supplementary motor cortex (Kolb & Whishaw, 2003:400). Some clinicians refer to this inability to incorporate certain voluntary motor actions into a single action as *apraxia* (Du Preez & Steenkamp, 1986). The term *apraxia* is applied to testees with *no* visible weakness, ataxia (incoordination), or other extra-pyramidal disruptions, and *no defect* of the primary modes of sensation which consequently results in one loosing *the ability to execute complex and previously learned skills and* gestures (Halsband *et al.*, 2001; Binkofski, 2001; Adams & Victor, 1997). The term pyramidal disruption refers to a

lesioned pathway from the pyramidal cells of the fifth and sixth layers of the neocortex to the spinal cord (Kolb & Whishaw, 2003). *Apraxia* is therefore used where a lesion erases the memory of the pattern of movements necessary for an intended action. In children with learning difficulties, impaired motor planning is evident, suggesting frontal lobe involvement.

Arm and leg extension:

This task measures muscle tone and muscle strength. Inadequate muscle tone and strength suggest that the research participant might tire easily, that she finds it hard to remain seated for extended periods of time, and that she might not be able to concentrate (because of exhaustion). 'Investigators report more than a 90 percent probability of a reading problem when there is an abnormality noted in execution of arm and leg extension. … The implication is that, in children with learning disorders, … the brain may be having trouble integrating and processing information' (Mutti *et al*., 1998). This item links closely with the previous item, especially with regard to motor planning ability. Motor planning is the CNS function of knowing where body parts will be in any motion, such as writing, drawing, or running. 'The subject who tires or whose performance seems to deteriorate on this and other QNST motor tasks may have poor muscle tone or other CNS difficulties' (Mutti *et al*., 1998).

Skip:

This task requires the research participant to move bilaterally while in forward motion in a movement between a walk and a run. This is a large-motor skill and relates to balance, sequencing and motor planning, and a sense of place in space. Furthermore, the poor balance can be linked to the research participant's difficulties regarding auditory-perceptual skills (Mutti, *et al.*, 1998:26-30). The research participant also demonstrated below par sequencial memory involving several tasks, e.g. copying sound patterns and repetitive thumb and finger movements. The performance of rapidly alternating movements with the hands and/or fingers is a function of the frontal lobes, specifically the supplementary motor cortex (Kolb & Whishaw, 2003:400), while frontal cortex areas 8, 9 and 46, as well as the parietal lobes and the inferior temporal cortex, also play a significant role in performance in this task (Kolb & Whishaw, 2003:477). In addition, the research participant demonstrated poor dynamic balance on items 'tandem walk' and 'skip', while the item 'stand on one leg' produced poor static balance.

'Poor balance and tasks notably more difficult on one side than the other relate to body symmetry, sequencing, motor planning, and sense of place in space. Sequencing of words and letters, handwriting, and reading skills are more proficient when these neuromotor skills develop at an age-appropriate pace' (Mutti *et al*., 1998). The item 'Skip' measures the integration of rhythm, synchronization and dynamic balance. Rhythm and synchronization play an important role in syllabification. 'Balance is often considered to be closely related to auditory-perceptual skills' (Mutti *et al*., 1998), and it is thought that the research participant's below par auditory-perceptual development impacts on all related subskills.

3.8.3 Screening for frontal lobishness

Lesions to the frontal lobes usually result in impaired response inhibition and inflexible behaviour, and the research participant finds it difficult to use information from environmental cues (feedback) to regulate or change her behaviour. This difficulty manifests in perseveration on responses in a variety of test situations, particularly those in which there are changing demands. This difficulty also manifests in behavioural irregularities such as distractibility and impulsivity noted on the QNST-II. The ***Wisconsin Card-Sorting Test***, which is one of the standard clinical tests for frontal lobe injury, was therefore administered to the research participant.

3.8.3.1 Analysis and interpretation of results

The research participant demonstrated below par ability to shift response strategies and perseveration was noted. 'Such perseveration is common on any task in which a frontal-lobe patient is required to shift response strategies, demonstrating that the frontal lobe is necessary for flexibility in behaviour' (Kolb & Whishaw, 2003:408). Based on various researchers' findings (Milner, Corsi & Leonard, 1991:601; Milner & Petrides, 1984:403; Owen, Downes, Sahakian, Polkey & Robbins, 1990:1021), the principal locus of the Wisconsin Card-Sorting effect appears to be roughly around Brodmann's area 9 in the left hemisphere. Lesions elsewhere in the left frontal lobe, and often in the right, will also produce a deficit on this task, although an attenuated one (Kolb & Whishaw, 2003:408).

3.8.4 Personality structure

Table 3.3 Children's Personality Questionnaire (CPQ)

	STEN SCORE	LOW SCORE DESCRIPTION	1	2	3	4	5	6	7	8	9	10	HIGH SCORE DESCRIPTION
A	2	Critical, reserved, cool		■									Warm, soft-hearted, participating
B	3	Dull, less intelligent			■								More intelligent, bright
C	5	Emotionally immature & unstable					■						Emotionally mature, stable, realistic
D	7	Deliberate, stodgy, placid							■				Unrestrained, nervous, excitability
E	7	Obedient, mild, dependent							■				Assertive, aggressive, rebellious
F	2	Sober, silent, serious		■									Happy-go-lucky, enthusiastic
G	5	Casual, quitting, undependable					■						Conscientious, persevering
H	2	Timid, threat-sensitive, shy		■									Venturesome, thick-skinned
I	4	Practical, tough-minded				■							Tender-minded, sensitive, protected
J	7	Vigorous, goes readily with group							■				Individualistic, obstructive, reflective
N	7	Socially un-alert, naïve							■				Socially percipient & skilful
O	6	Secure, resilient, confident						■					Discouraged, worrying, self-reproaching
Q_3	4	Careless, ignores standards, lax				■							Self-controlled, self-respecting
Q_4	9	Relaxed, composed									■		Tense, driven, irritable

Frontal lobe lesions usually also result in impaired social behaviour and pseudodepression, as demonstrated by the results of the CPQ in paragraphs below. The wide range of symptoms of frontal-lobe lesions can conceptually be grouped into several categories, i.e. disturbances of motor functions, loss of divergent thinking, impaired response inhibition and inflexible behaviour, poor temporal memory and impaired social behaviour. There is a complementary effect of left and right frontal lesions, in that left frontal lesions are more likely to affect language or movement-related behaviours, whereas right frontal lesions are more likely to alter non-language functions such as emotions (Kolb & Whishaw, 2003:422).

3.8.4.1 Analysis and interpretation of results

Factor A: Sizothymia versus affectothymia: The warm-cool social orientation

The research participant's exceptionally low Factor A sten score, indicates that she does not find social activities rewarding, mostly because she does not seek or enjoy social interactions. She does not easily warm up to people, and she prefers to spend her time in solitude or with a selected few friends (or relatives) (Cattell, 1989:19). The research participant's choices about how to spend her time are strongly influenced by her talents and interests, and therefore she might invest much time and effort in these hobbies and personal activities – as substitutes for social involvements (Cattell, 1989:20). The research participant strongly prefers to work alone, yet she might invest too much energy in negative emotions; however, her time and energy is usually not consumed by social needs. The preceding traits have certain implications for the classroom, for example that she might not be willing to collaborate towards the completion of a group project, might avoid group projects, group activities and team work, and she might dislike school because she basically wants to avoid large-scale social meetings. The research participant's *personal strengths* are likely to include the following: She tends to be trustworthy and willing to stick to her commitments (Du Toit & Madge, 1988:3). However, she is likely to channel most of her energy into non-social activities, therefore she might have only a few close friends and confidantes. This observation is confirmed by the background information, referring to her feelings of alienation and her preference to play with her dogs. Although the research participant is able to recount objective facts by mere observation, she might also overlook the importance of the social and emotional contexts of observed occurrences (Cattell, 1989:21); she might thus benefit from developing a sense for social cues and the ability to interpret the body language of other people to better understand the social and emotional contexts of occurrences. The research participant's *leadership style* is likely to include the following: The research participant tends to be a 'loner', therefore she is not easily persuaded (by praise or other people's disapproval or dislike) to compromise standards, views, and values (Cattell, 1989:22). Consequently, in certain contexts, she might be viewed as stubborn, because she reveals some kind of inflexibility that has obvious advantages and disadvantages, i.e. she might be reluctant to accept a new train of thought or view on a specific topic, and she is unlikely to willingly subject to others' opinions (Du Toit & Madge, 1988:3). *Interpersonal difficulties* are likely to include the

following: The research participant might be extremely withdrawn, to such extent that she neither desires nor enjoys close relationships and social gatherings. Her relationships are usually ritualistic with little affective responses, except towards her first-degree relatives (Cattell, 1989:25). Although the research participant might feel only indifference to others, the effect of her cool detachment is apt to evoke not just indifference, but negative responses from those with whom she interacts, based on the combination of factors -A/-I sten scores (Cattell, 1989:26). In addition, other people may find conversations (even small talk) with her demanding/difficult/tiring, due to her lack of sociability and her disinterest in human affairs (Du Toit & Madge, 1988:3). She might benefit from developing some conversational skills.

Factor B: The ability to discern relationships (Intelligence)

The research participant's low sten score on Factor B suggests a weakened capacity to discern relationships in terms of how things stand, relative to one another (Cattell, 1989:30). Her thinking style is more concrete, she finds it hard to handle abstract problems (Du Toit & Madge, 1988:3) and she seems to be less well organized (Cattell, 1989:32). This sten score is in keeping with the research participant's performance on the SSAIS-R.

Factor C: Adaptation to the environment

In humans, because of their decision-making capabilities, adaptation as a process occurs in two ways: Humans may act (a) alloplastically, i.e. by changing their environments to meet their needs, or they may act (b) autoplastically, i.e. by changing themselves in order to survive or live more harmoniously with exogenous conditions. Essentially good adaptation requires the use of these two actions and proper judgment about which is more appropriate for a given situation. Problem solving requires anticipation, judgment, planning, reality testing, memory and self-regulation. It also requires the ability to integrate novel experiences and new knowledge in such a way as to provide the best probability of incurring healthy survival and maximizing overall satisfaction (Cattell, 1989:38).

The research participant's average Factor C sten score indicates average emotional control and adaptation to her social environment (Du Toit & Madge, 1988:4). The research

participant might sometimes be accepting of conditions she cannot change; yet at other times she might become upset. However, she more or less applies 'common sense' to solve problems, considering her cognitive abilities and age (Cattell, 1989:40).

Factor D: Phlegmatic temperament versus irritability

The relatively high sten score on Factor D suggests that the research participant might often be irritable and demanding, and when frustrated she might show uncontrolled emotional behaviour (Cattell, 1989:298). She tends to be impatient, thereby demanding that her needs be attended to. In addition, she tends to view her social environment from an egocentric perspective, coupled with subjective feelings of jealousy, based upon an unfavourable comparison of self to others (Du Toit & Madge, 1988:4).

The research participant's interpersonal relations are likely to be dominated by 'defensive projection', leading to interpersonal difficulties of diverse nature. Thus, the 'green-eyed monster' characteristically colours her perceptions (Cattell, 1989:174). The essential component of her jealousy is a subjective experience of deprivation, a sense that something that would have made a meaningful difference to her happiness has been denied, e.g. material possessions or certain privileges. She unfavourably compares herself to others, which results in subjective feelings of deprivation and alienation (Du Toit & Madge, 1988:4). This subjective experience of deprivation is often accompanied by concomitant feelings of sadness, emptiness, and even anger (relatively high factor E sten score). These dynamics inevitably complicate parent-child relations, peer-group relations, as well as the prevailing atmosphere at home and within the classroom. The second component of jealously is expressed as a self-judgment of inadequacy (Cattell, 1989:173). Others are perceived, either in actuality or fantasy, as owning or having whatever the research participant thinks she has been deprived of, e.g. she feels that her classmates are able to master certain tasks with relative ease, or that others are being favoured. A strong feeling to right this disparity typically accompanies this subjective sense of disparity, resulting in feelings of resentment and animosity. These perceptions always create emotional tension, which is often relieved by finding a substitute gratification, e.g. demanding attention, or preferring to play with animals instead. If the research participant cannot right this subjectively felt disparity, hostility towards these individuals prevails, and she then enters into blaming. The combination of factors +D/+E suggests that she insists on getting her

point across, and that she suspects classmates to be gossiping about her, to which she responds by being oppositional and quick to take offence (Du Toit & Madge, 1988:4).

Factor E: Control versus submissiveness in human relations

The research participant's relatively high Factor E sten score suggests that she prefers to play a dominant (controlling) role in interpersonal relationships, and this dominance might be mostly expressed in a somewhat aggressive style (Cattell, 1989:72). She is likely to often steer other people in a specific direction, and when they show resistance in subtle ways, she tends to experience confusion (Du Toit & Madge, 1988:5). The research participant's *personal strengths* are likely to be some of the following: She usually maintains a commanding presence, she shows a strong self-will, and she holds strong opinions, despite the fact that others might differ from her (Cattell, 1989:73). The research participant might experience the following *interpersonal difficulties*: It is likely that people who are at the receiving end of the research participant's controlling behaviour are those who suffer most. Although she prefers to behave dominantly, she has to exert unusually large quantities of effort to stretch her personal limits, to overcome obstacles, and to respond to frustrations head-on (Cattell, 1989:76). For this reason, though the research participant might not get depressed, she is likely to experience stress-related symptoms. The research participant might also not cope well with loss and grief situations. The following *areas for personal growth* are suggested: The research participant should learn that she cannot ALWAYS control ALL situations that she meets. She should assume more democratic processes and decision-making by learning to appreciate the opinions of others (although opinions might differ).

Factor F: Exuberant versus sombre (serious) orientations

Based upon the research participant's low F sten score, she is likely to show inhibited thinking, cautious speech and behaviour, consistency and regularity (predictability) (Cattell, 1989:98). It seems that she is preserving a cautious life orientation, i.e. that she anticipates difficulties, avoids making mistakes, and hesitates to take risks (Cattell, 1989:100). She dislikes change (e.g. changing responsibilities, changing teachers, changing desks in the classroom, changing the furniture in her room, and changing

neighbourhoods), and she prefers to experience security and predictability in all aspects of her life. The research participant's *personal strengths* might include careful speech and behaviour, and deep thinking. She might move from thought to thought ponderously, critically, checking and rechecking for possible mistakes, thereby revealing some degree of doubt (Du Toit & Madge, 1988:5). She experiences deep feelings, which she does not usually discuss with others. She usually takes her work seriously, although others may view her as an introverted daydreamer (Cattell, 1989:101). However, she often feels that she has to cope with realities that are too advanced for her experience and/or capabilities (Du Toit & Madge, 1988:5).

The research participant's low F sten score indicates that she might experience the following *interpersonal difficulties*: She might often feel that she 'can't loosen up and have fun', or that she 'takes everything too seriously', which might result in interpersonal conflict with her peers, especially because she lacks humour and experiences even an innocent joke as ego-threatening. Although the research participant might have deep needs and thoughts, she often experiences difficulty expressing these feelings, thoughts and needs (Du Toit & Madge, 1988:5). The following areas for *personal growth* are suggested: She reveals a restricted range of interests, but she could become absorbed in one specific activity. Because she has a restricted range of interests, she might find little to talk about in situations that call for general conversation, e.g. within the peer group, and, for precisely this reason, she often feels alienated and ill at ease during social gatherings. Hence she might benefit from developing a few more interests in order to ensure that she has something to talk about at social gatherings, will be invited more often, and will be able to respond more spontaneously to social invitations and gatherings in general. Furthermore, the research participant should learn to entrust others with her deep thoughts and feelings, and to ask for what she wants. Although desurgency (F-) does not in itself represent depression, the research participant's quietness and seriousness may easily be misinterpreted as a sign of depression. If, in addition to her quietness and seriousness, she also experiences a sense of helplessness and hopelessness, this may indicate depression and professional consultation should become an option (Cattell, 1989:102; Du Toit & Madge, 1988:5). In conclusion, the research participant might have a narrow concentration span (Cattell, 1989:101).

The combination of F- with all five other factors, i.e. H-, Q3+, A-, E-, and N+, suggests that the research participant is quiet and serious, socially aloof and self-reliant, with limited involvement with others, but that she takes care not to be socially offensive (Cattell, 1989:102).

Factor G: The content and action of moral values

The research participant's average G sten score suggests that the content and action of her moral values are in line with mainstream values (Cattell, 1989:113). She is moderately rule-bound, behaves accordingly, and conforms to prevailing values, customs and rules (Du Toit & Madge, 1988:6).

Factor H: Courage versus timidity in human temperament

The research participant's low H sten score suggests that she does not attach undue social importance to herself, and that she is finely attuned to and considerate of others' feelings and emotions (Cattell, 1989:144). She prefers certainty and predictability, therefore she also prefers to preserve sameness and avoid radical changes (Cattell, 1989:145). Sometimes she might appear to be 'living in a shell' – to protect herself from the harsh and painful outside world (Du Toit & Madge, 1988:6). She is likely to camouflage the self in order to become almost barely visible (not to be noticed) in order to feel safe (Cattell, 1989:144). The research participant tends to minimize stress by avoiding risks, competition and new experiences, which suggests an unwillingness to venture. She prefers not to be singled out for attention, especially when such attention is negative. She usually prefers non-competitive activities and one or two close friends to groups of people (Du Toit & Madge, 1988:6). She needs to receive precise and accurate instructions in order to preserve her sense of personal security and confidence. The following might reflect the research participant's *personal strengths*: The research participant is likely to be well behaved, reliable and considerate of the feelings and emotions of others. She is a diligent and conscientious worker, mostly because she wants to avoid stressful social interaction, such as negative criticism, therefore she also tends to be rule-bound and she readily conforms to conventional norms and standards of behaviour (Cattell, 1989:148). She is emotionally guarded and controlled, and during group activities she is apt not to interact socially (Du Toit & Madge, 1988:6). The following are potential areas for *personal*

growth: The research participant has such a low tolerance for fear and arousal that she needs to continuously protect herself from situations and stimuli that might threaten her delicate internal homeostasis. By nature she tends to avoid all experiences that might increase her fear, nervousness, and uncertainty, thereby concurrently she closes off opportunities for personal gain, friendships and adventure. Because she tends to hide away so much, she neglects to enjoy life, and due to her shyness and anxiety, she fails to participate and compete, despite her strong desire to also enjoy acknowledgement for achieved success. The research participant sometimes becomes so absorbed in her own internal processes, i.e. her own thoughts and feelings, that she neglects to attend sufficiently to what is going on around her. She usually contributes very little to conversations, and classmates might misinterpret her silence as rejection or silent criticism. She also easily becomes too dependent within relationships. Because she prefers to cling to what is known, she shows a narrow band of interests; consequently, she easily feels alienated and disillusioned, because she does not share activities and interests with peers (Cattell, 1989:146). In addition, the combination -H/+N suggests that she is able to keep up a fine social façade (Cattell, 1989:147). Based on the combination -H/-F she not only tends to be shy and timid, she is also sober, reserved and serious (Cattell, 1989:147). The combination -H/-A suggests that she is reserved, detached, critical and distant, but determined (Cattell, 1989:147). The combination -H/+Q4 suggests that she is also tense, frustrated, driven and overwrought (Cattell, 1989:149).

Factor I: Feeling versus Thinking – contrasting modes of evaluating experience

The research participant's middle range sten score on Factor I suggests that she moves with versatility between thinking and feeling responses during assessment and decision-making (Du Toit & Madge, 1988:7). This versatility allows for the incorporation of both subjective and objective reality, which can lead to a more complete understanding than when either only feeling, or only thinking, is overemphasized to the detriment of the other. Thus, the research participant uses both her feelings and thoughts to make evaluations, instead of relying heavily on one modality only, which trait might be beneficial when having to consider the interests of others (Cattell, 1989:152).

Factor J: Adventurous and sociable versus emotionally cautious orientation

The research participant's relatively high sten score on Factor J suggests that she prefers to do things on her own, that she thinks about her mistakes and about how to avoid them, that she tends not to forget if she was treated unfairly, and that she has strong private views differing from those of the group, but that she prefers to keep in the background and avoid arguments (Du Toit & Madge, 1988:7).

Factor N: Self-presentation in social situations

A relatively high sten score on Factor N suggests that the research participant is successful at wearing a social mask, e.g. telling a white lie or act in ways not congruent with her true feelings or motives, in order to avoid embarrassment, avoid hurting feelings, or win positive responses (Cattell, 1989:210). She is likely to keep her social mask firmly in place with most people and in most situations (Du Toit & Madge, 1988:8).

Factor O: Guilt proneness versus self-confidence and resilience

The research participant's average sten score on Factor O suggests that she might be relatively free from unrealistic feelings of guilt (Du Toit & Madge, 1988:8). She reveals an adequate ability to generate empathetic and appeasement responses (Cattell, 1989:222).

Factor Q3: Investment in maintaining a socially approved self-image

Factor Q3 reflects the self-concept and sense of identity. There might be a difference between the research participant's wished-for self-concept and her subjectively perceived self-concept, conversely her observation that she is failing to live up to her personal ideals often causes significant psychological discomfort, e.g. she engages in self-degrading self-talk and experiences feelings of shame and anxiety (Cattell, 1989:288). In addition, the degree of discomfort that she experiences when she cannot meet certain standards may obscure her observation of reality, thereby either partially or fully blocking recognition of the incongruence between the wished-for self-concept and the perceived self-concept from awareness (Du Toit & Madge, 1988:9). This blocked observation of reality then becomes

a mechanism of defense, and she is likely to use rationalisation, denial, repression, or other forms of self-deception (self-delusion) to get relief from inner discomfort and cognitive dissonance (Cattell, 1989:290). The clinical history supports this observation, e.g. she likes to play with younger children and in such situations she takes the lead, yet among adults she prefers to be 'babied'. Consequently, within interpersonal relationships she might not fare as well, e.g. she might be chronically seeking guidance and support.

Factor Q4: Tense versus relaxed temperaments

Factor Q4 measures the unpleasant sensations that accompany autonomic arousal and nervous tension, also known as *free-floating anxiety*, meaning pervasive, generalized fears that are not attached to any particular idea, object, or event (Cattell, 1989:296). A high Q4 sten score on a profile usually indicates that nervous tension is a personality trait. In this case the research participant is characteristically a tense, volatile and easily upset individual (Du Toit & Madge, 1988:9). She might habitually experience trouble relaxing or even just sitting still for extended periods, because she 'always needs to be doing something' (Cattell, 1989:298). It is likely that she is experiencing enduring stress, because she feels unable to cope with her academic and social environment. In this case the research participant's 'nervous' quality of energy does not come from a surplus of healthy vitality, but from her need to relieve her overtaxed autonomic nervous system (Du Toit & Madge, 1988:9). A high Q4 sten score suggests that she might be impatient, accident prone and irritable. Being anxious and tense, she is likely to do some spur-of-the-moment thing, simply to release tension. She also has a narrow concentration span (Cattell, 1989:300). These observations are in keeping with her clinical history, showing that she has been on *Concerta*™ for quite some time to address her attention deficits.

Draw-a-Person (D-A-P) and Kinetic Family Drawings (K-F-D)

The research participant's D-A-P and K-F-D drawings are inserted as Figure 3.1 to Figure 3.4 below.

3.8.4.2 Analysis and interpretation of results

The participant's drawings indicate that she may experience tension, aggression, anxiety and feelings of inadequacy. She may also come across as infantile at times and shows a reluctance to explore. Generally the drawings indicate a need to envelope and control the entire environment, which may reflect her subjective feelings of not being in control. She may come across as rebellious and suspicious by nature, and may also present as being introverted and self-conscious, and as experiencing basic life uncertainty. These interpretations are in keeping with the CPQ protocols.

Figure 3.1

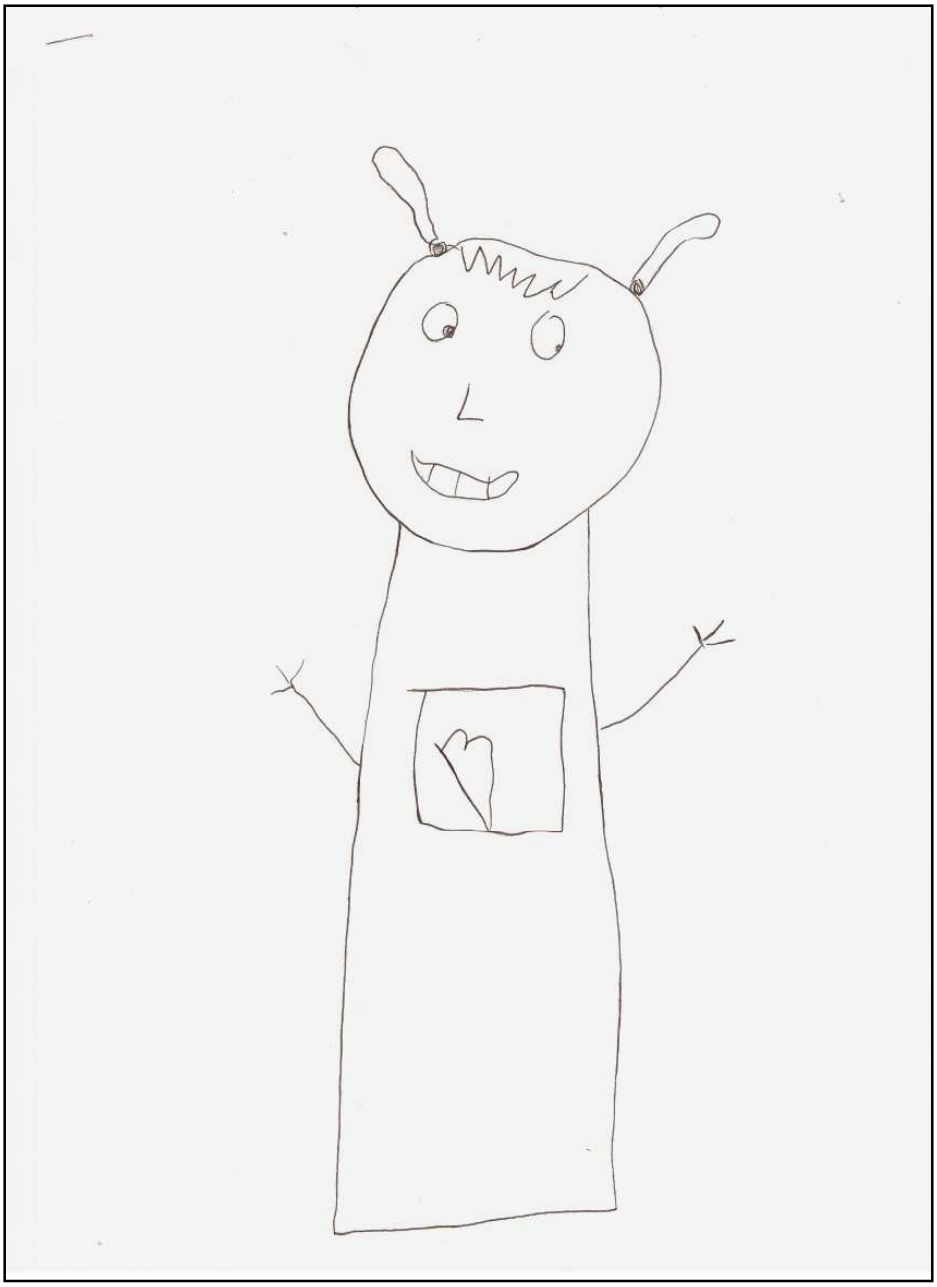


Figure 3.2

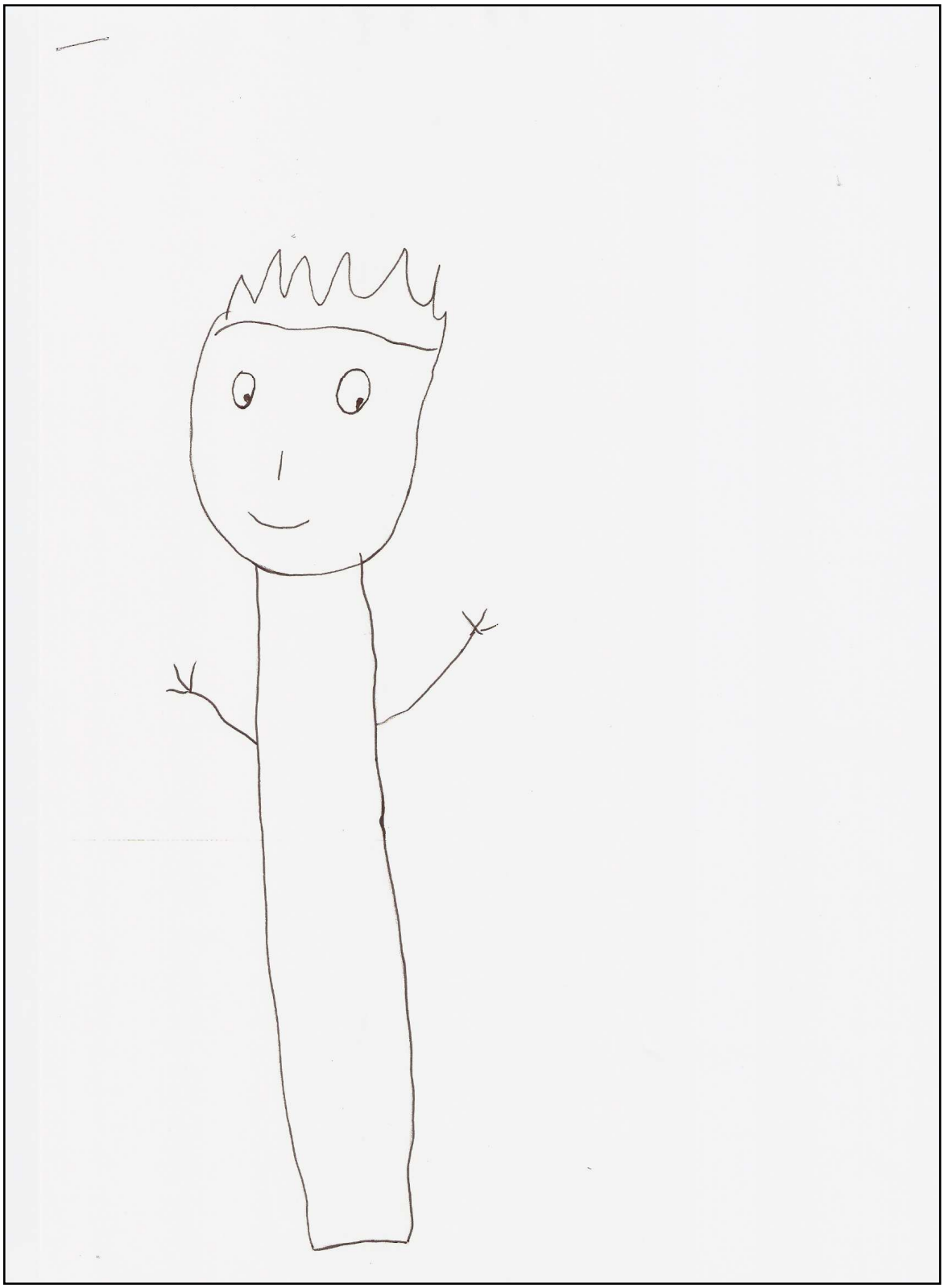


Figure 3.3

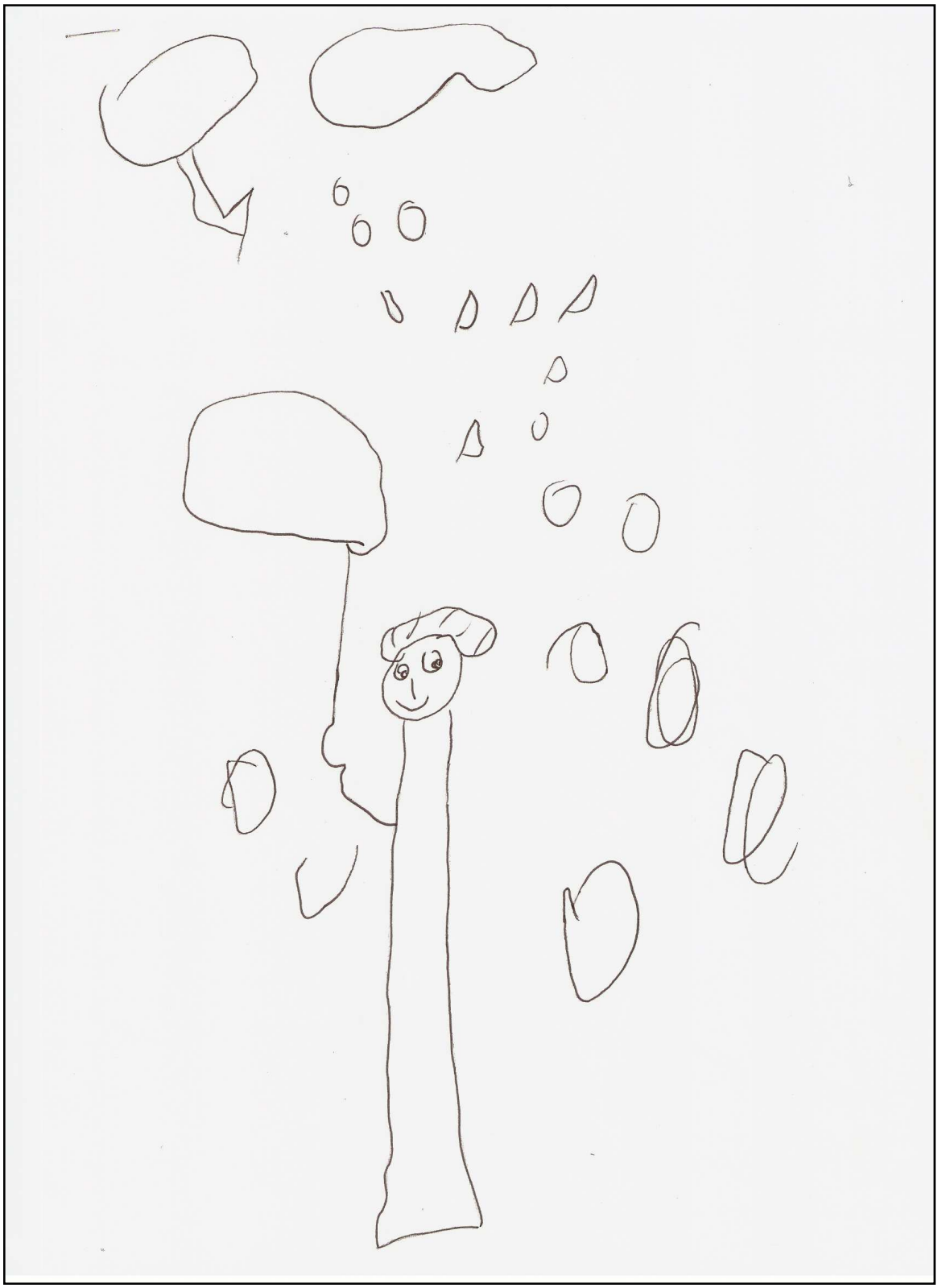


Figure 3.4

Children's Apperception Test (CAT)

A transcription of the verbatim responses to the CAT cards is provided in **Annexure C**.

3.8.4.3 Analysis and interpretation of results

In order to enhance objectivity, the preceding verbatim protocols were analysed and interpreted in terms of the s*chedule of adaptive mechanisms in CAT responses*, developed by Haworth (1963:181-184; reprinted with CAT manual in 1990).

SCHEDULE OF ADAPTIVE MECHANISMS IN CAT RESPONSES

DEFENSE MECHANISMS

TOTALS

A. *Reaction-formation* (only one check per story). [A + B = minimum of 5 confirmatory checks regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Exaggerated goodness, or cleanliness on **Card 6** (washing to do);
2. Oppositional attitudes, rebellion, stubbornness on **Card 2**; **Card 7**; **Card 8**; **Card 10**;
3. Story tone opposed to picture content on **Card 4**;

B. *Undoing and Ambivalence* (only one check per story). [A + B = minimum of 5 confirmatory checks regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Undoing on **Card 2**; **Card 3**; **Card 4**; **Card 5**; **Card 6**; **Card 7**; **Card 8**; **Card 9**; **Card 10**.
2. Gives alternatives; balanced phrases (asleep – awake; hot – cold, etc.) on **Card 3**; **Card 5**; **Card 6**; **Card 7**; **Card 10**.
3. Indecision by *S* or story character on **Card 4**; **Card 5**; **Card 6**;
4. Restates (e.g. 'that …, no this …,' 'he was going to, but …') on **Card 4**; **Card 6**; **Card 7**; **Card 8**.

C. *Isolation.* [Total of 6 confirmatory checks regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Detached attitude ('It couldn't happen.'; It's a cartoon.')
2. Literal ('It doesn't show, so I can't tell.') on **Card 10** ('I have a cousin …').
3. Comments on story or picture ('That is hard.'; 'I told a good one.') on **Card 4**; **Card 7**; **Card 10**.
4. Laughs at card, exclamations

5. Use of fairy-tale, comic-book, or 'olden-days' themes of characters on **Card 2**; **Card 6** (reminds of Hansel and Gretel);
6. Describes in detail, logical; 'the end'; gives title to story on **Card 3**; **Card 4**; **Card 5**; **Card 6**; **Card 7**; **Card 8**; **Card 9** (that's all); **Card 10**.
7. Specific details, names or quotes ('four hours'; she said, ' … ') on **Card 2**; **Card 3**; **Card 4**; **Card 5**; **Card 6**; **Card 7**; **Card 8**; **Card 9**; **Card 10**.
8. Character gets lots on **Card 6**; **Card 8** (lots of bananas); **Card 10** (bones).
9. Character runs away due to anger **Card 4**; **Card 6**; **Card 7**.
10. *S* aligns with parent against 'naughty' child character; disapproves of child's actions on **Card 8**; **Card 10**.

D. *Repression and Denial.* [A total of 5 confirmatory checks is regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Child character waits, controls self, conforms, is good, learned lesson.
2. Accepts fate, did not want it anyway.
3. Prolonged or remote punishment on **Card 10**.
4. 'It was just a dream.'
5. Forgets, or loses something on **Card 9** (forgot to close the door).
6. Omits figures or objects from story (on #10 must omit mention of toilet *and* tub or washing) on **Card 10** (omits toilet, tub and washing).
7. Omits usual story content on **Card 4**; **Card 6**; **Card 10**.
8. No fantasy or story (describes card blandly) on **Card 1**; **Card 9**.
9. Refuses card.

E. *Deception.* [A total of 3 confirmatory checks is regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Child superior to adult, laughs at adult, is smarter, tricks adult, sneaks, pretends, hides from, steals from, peeks at or spies on adult (only one check per story) on **Card 7**; **Card 8**; **Card 10**.
2. Adult tricks child, is not what he/she appears to be (only one check per story).

F. *Symbolization.* [A total of 4 confirmatory checks is regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Children play in bed
2. See parents in bed (#5)
3. Open window (#5, #9); dig, or fall into a hole on **Card 7** (slipped on banana peel); **Card 9** (open door).
4. Babies born
5. Rope breaks (#2); chair or cane breaks (#3); balloon breaks (#4); tail pulled or bitten (#4, 7); crib broken (#9)
6. Rain, river, water, storms, cold, dark on **Card 9** (dark)
7. Fire, explosions, destruction
8. Sticks, knives, guns on **Card 4**
9. Cuts, stings, injuries, actual killings (other than by eating) on **Card 2**; **Card 4**
10. Oral deprivation on **Card 6** (hunger).

G. *Projection and Introjection.* [A total of 4 confirmatory checks is regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. The attacker is attacked, 'eat and be eaten' on **Card 6**; **Card 7.**
2. The innocent one is eaten or attacked on **Card 4**; **Card 7**.
3. The child is the active aggressor (bites, hits, throws; do not include verbal or teasing attacks) on **Card 8**; **Card 10**.
4. Characters blame others on **Card 6** (What did you do to my baby?); **Card 8**.
5. Others have secrets or make fun of somebody on **Card 3**.
6. *S* adds details, objects, characters, or oral themes on **Card 4**; **Card 6**; **Card 7** (banana); **Card 8** (fridge); **Card 10** (draws on themes from cards 7 and 8).
7. Magic or magical powers.

PHOBIC, IMMATURE OR DISORGANIZED

H. *Fear and Anxiety.* [A total of 3 confirmatory checks is regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Child hides from danger, runs away because of fear on **Card 4**; **Card 7**.
2. Fears outside forces (wind, ghosts, hunters, wild animals, monsters) on **Card 4**; **Card 6**; **Card 9**.
3. Dreams of danger.
4. Parent dead, goes away, or does not want child on **Card 4**; **Card 5**; **Card 6**.
5. Slips of tongue by *S* on **Card 7** (tiger/lion); **Card 8** (auntie/mom).

I. *Regression.* [A total of 2 confirmatory checks is regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Much affect in telling story on **Card 1**; **Card 2**; **Card 4**; **Card 6**; **Card 10**.
2. Personal references on **Card 4** (I don't like sad stories); **Card 8** (She always shouts at me when I do that stuff.); **Card 10**.
3. Food spilled on **Card 7**.
4. Bed or pants wet, water splashed.
5. Dirty, messing, smelly; person or object falls into toilet on **Card 7** (banana had worms).
6. Ghosts, witches, haunted house on **Card 6**.

J. *Controls weak or absent.* [A total of only <u>one</u> confirmatory check is regarded as a 'critical score' for this category (Haworth, 1990:8)].

1. Bones, blood
2. Poison
3. Slang or nonsensical words
4. Perseveration of unusual content from a previous story on **Card 5**; **Card 8** (bananas re-introduced); **Card 10**.
5. Bizarre content on **Card 7** (still like worms).

IDENTIFICATION

K. *Adequate, same-sex.* For the identification measure, the equivalent of a critical score is secured by comparing the relative number of checks for categories K and

L. If the sum of checks for L is equal to, or exceeds the sum for K, identification is considered to be 'confused' and contributes one unit to the total of critical scores (Haworth, 1990:8).

1. *S* identifies with same-sex parent or child character.
2. Child jealous of, or scolded or punished by same-sex parent on **Card 8**; **Card 10**.
3. Child loves, or is helped by parent of opposite sex on **Card 3**; **Card 6**.

L. *Confused, or opposite sex*

1. *S* identifies with opposite-sex parent or child character.
2. Child fears, or is scolded or punished by opposite-sex parent on **Card 8.**
3. Misrecognition by *S* of sex or species on **Card 2**; **Card 5**; **Card 7**.
4. Slips of tongue with respect to sex of figures.

The final quantitative measure consists of the number of categories receiving critical scores, and not of the total number of checks for all categories. On the basis of research findings by Haworth (1963:181-184), five or more critical scores would indicate enough disturbance to warrant clinical intervention.

Based on the data from the preceding *Schedule of adaptive mechanisms in CAT responses*, developed by Haworth (1963:181-184; reprinted with CAT manual in 1990) the research participant obtained critical scores in a total number of nine (out of a possible 10) categories, which indicates social adjustment and affective difficulties.

It has already been established, on the basis of interpretations of the test results, that the research participant is likely to have suffered diffused lesioning across all cortical areas, with a specific focus on the parietal, temporal and frontal areas. Lesioning of the temporal and frontal areas is particularly associated with impaired socialization, affect and exacerbation of certain personality traits. Based on the results of the CAT, it is suggested that the research participant is employing a large number of unhealthy defence mechanisms, i.e. reaction-formation, undoing and feelings of ambivalence, isolation and concurrent feelings of alienation, repression and denial, symbolization, projection and introjection, excessive fear and anxiety, regression (likes to be babied), and confused identification, while her behavioural controls seem to be poorly developed, in keeping with the results of the CPQ.

3.8.5 Sensory-motor integration

Kolb and Whishaw (2003:684) report that on average partial postoperative recovery from brain surgery and closed-head injury is noticeable within two years. In the light of this, the two-year postoperative assessment data of the research participant will now be scrutinized to determine the nature of enduring after effects of Crouzon syndrome and corrective surgery, based on the results of the *Southern California Sensory Integration Tests* (Ayres, 1972).

3.8.5.1 Analysis and interpretation of results

Speech abilities:

At the time of the two-year postoperative assessment, the research participant did not use the 'th' sound in speech. She now pronounces it correctly.

Oral functioning:

Face

At the time of the two-year postoperative assessment, the muscle tone in the facial area appeared to be low. Sucking and blowing exercises were prescribed and she now has functional muscle tone.

Jaws and teeth

At the time of the two-year postoperative assessment, jaw movements appeared to be within the normal range. At this stage there appears to be a slight asymmetry, but this does not affect oral functioning. Her teeth are healthy and the development of dentures is age appropriate.

Lips

At the time of the two-year postoperative assessment, the lips showed low muscle tone. Attention was paid to active lip closure and the building of muscle tone in the oral area. Her lips are now functional.

Tongue

At the time of the two-year postoperative assessment, the tongue had low muscle tone. She can now move her tongue in all directions outside her mouth, but needs her jaw to assist in movements.

Throat

At the time of the two-year postoperative assessment, the throat movements appeared to be normal.

Speed of movements and voicing

At the time of the two-year postoperative assessment, she had a slow speech production rate. This has remained unchanged, but does not affect speech comprehensibility.

Hearing:

At the time of the two-year postoperative assessment, informal assessment of hearing showed functional hearing.

Language development:

Language comprehension

At the time of the two-year postoperative assessment, the level of the research participant's vocabulary was 4 years and 9 months, while her verbal comprehension of language structure in sentence form was at a 4 years and 5 months level (her chronological age at the time was 5 years and 5 months). She has shown vast improvement following intense speech therapy, and currently her performance on the Verbal scale of the SSAIS-R is at the low average level.

Listening skills (auditory processing of speech and environmental sounds):

Auditory sequential memory

At the time of the two-year postoperative assessment, the research participant could remember only three or four facts (test items). Currently she is able to recall six digits forward, but only two digits backward (SSAIS-R subtest Memory for Digits). Because

recall of digits backward relies on mental manipulation associated with frontal lobe functioning, this still indicates significant impairment.

Auditory discrimination of sound

At the time of the two-year postoperative assessment, auditory discrimination was significantly impaired, and currently speech sound discrimination is still sometimes a problem, especially when she has to listen to words with which she is not familiar.

Somato-sensory system:

Sensory Modulation

At the time of the two-year postoperative assessment, sensory modulation was very low in respect of all modalities, as depicted in Table 3.4 below. Currently she still functions at below average levels. Sensory modulation is a function that protects the body against potential danger. It enables one to automatically appraise a response according to incoming stimuli in order not to under-react or overreact to it. Insufficient sensory modulation causes an over(hyper)-sensitivity or an under(hypo)-sensitivity to sensory stimulation, which is generally associated with attention deficits, impulsivity and fidgety behaviour.

Table 3.4 Level of two-year postoperative sensory modulation

	Below average				**Average**					**Above average**		
	-1.8	-1.5	-1.2	-0.9	-0.6	-0.3	0.0	+0.3	+0.6	+0.9	+1.2	+1.5
Visual sensitivity						**x**						
Auditory sensitivity				**x**								
Olfactory sensitivity							**x**					
Tactile sensitivity					**x**							
Postural insecurity						**x**						

(A score of 0.0 is normal.)

Sensory discrimination

At the time of the two-year postoperative assessment, sensory discrimination was very low with regards to kinaesthesia, form perception and praphaestesia, as depicted in Table 3.5 below. Sensory discrimination is the ability to interpret information that relate to objects in the environment. It enables a person to plan movement on a subconscious level, to accurately perceive the direction and speed of movement, and to accurately react on a motor level. These abilities are necessary for daily functioning, especially when tasks are executed without being able to see, e.g. when fastening buttons or searching for something in the dark. It is also extremely important when doing written tasks.

Table 3.5 Level of two-year postoperative sensory discrimination

	Below average				**Average**					**Above average**		
	-1.8	-1.5	-1.2	-0.9	-0.6	-0.3	0.0	+0.3	+0.6	+0.9	+1.2	+1.5
Kinaesthesia					**x**							
Form perception				**x**								
Finger identification								**x**				
Graphaestesia	Below -1.8											
Localisation of tactile stimuli										**x**		

(A score of 0.0 is normal.)

As depicted in Table 4.5, the research participant performed as follows:

Kinaesthesia: Left: -0.4; Right: +0.2; Total -0.5
The ability to recognize the position of the limbs, that cannot be seen is below par on left, but within normal limits on the right side.

Form perception: Left: -1.3; Right: -0.7; Total -0.9
The ability to identify shapes by touch without vision is below par on both sides.

Finger identification: Left: +0.4; Right: +0.1; Total +0.3
The ability to identify fingers without vision is within normal limits, suggesting absence of finger agnosia.

Graphestesia: Left: -1.2; Right: -2.9; Total -2.3
The identification and recognition of shapes that are drawn on the palm of the hand is below par on both sides.

Localisation of tactile stimuli: Left: +0.1; Right: +0.9; Total +0.8
The ability to identify the body part that was touched (tactile stimulation) is on par on both sides.

Considering the results of the QNST-II, the sensory discrimination profile has not hanged since the two-year postoperative assessment.

Postural motor functioning:

At the time of the two-year postoperative assessment, the research participant functioned at below average level with regards to gross motor coordination, as depicted in Table 3.6 below. According to Ayres (1972), the vestibular system plays an important role in the regulation of behaviour. The research participant performed as follows:

Nystagmus (Normal range is between -0.5 and +0.5)

Left eye: -0.8; Right eye: -1.0; Total -0.9

This score is obtained by turning the research participant in a specific direction in order to evaluate the eye movement that is associated with the rotation. Performance was below par on both sides, and she lost her balance to both sides. Considering the results of the QNST-II, the eye movement profile has not changed since the two-year postoperative assessment.

Muscle tone (normal range is between –0.9 and +0.9)
At the time of the two-year postoperative assessment, the research participant's muscle tone was at below average level, as manifested by the following:

- she struggled to maintain the anti gravity posture for the required time;
- she would at times use her hand to support her head while sitting at the desk; and

- she complained of getting tired.

Postural adjustment mechanisms

At the time of the two-year postoperative assessment, it seemed as if the research participant's primitive postural reflexes have not been fully integrated, which has resulted in inadequate and uncomfortable seating and posture while working at her desk.

Balance -1.0 with eyes open
-1.0 with eyes closed

The research participant could execute simple tasks, such as hopping on one leg only. She was unable to perform tasks that required more advanced motor planning and bilateral integration, e.g. skipping. Considering the results of the QNST-II, the muscle tone and postural adjustment profile has not changed since the two-year postoperative assessment.

Table 3.6 Level of two-year postoperative postural motor functioning

	Below average				**Average**					**Above average**		
	-1.8	-1.5	-1.2	-0.9	-0.6	-0.3	0.0	+0.3	+0.6	+0.9	+1.2	+1.5
Vestibular proprioception												
Nystagmus				**x**								
Balance (with eyes open)				**x**								
Balance (with eyes closed)				**x**								
Total extension				**x**								
Total flexion					**x**							
Postural behaviour												
Muscle tone					**x**							
Shoulder stability							**x**					
Equilibrium						**x**						
Protective extension							**x**					
Dynamic balance					**x**							
Postural background movements				**x**								

Postural reflexes				**x**								

(A score of 0.0 is normal.)

Eye movements and tracking:

There is a distinct connection between eye movement and the vestibular system. The vestibular system exerts a strong influence on the extra-ocular muscles of the eye, for when the head is moved, the half-circular canals of the inner ear are stimulated and this affects the extra-ocular muscles to adjust tracking and eye movements accordingly. There is a strong link between reading and writing, and eye convergence, focusing and tracking to allow for adequate scanning while reading. At the time of the two-year postoperative assessment, eye movements and tracking were below par, as illustrated in Table 4.7 below. The results of the QNST-II indicate that the eye movements and tracking profile has remained unchanged since the two-year postoperative assessment.

Table 3.7 Level of two-year postoperative eye movement and tracking

	Below average				**Average**					**Above average**		
	-1.8	-1.5	-1.2	-0.9	-0.6	-0.3	0.0	+0.3	+0.6	+0.9	+1.2	+1.5
Tracking							**x**					
Convergence			**x**									
Rapid localisation					**x**							

(A score of 0.0 is normal.)

Bilateral motor coordination: (BMC)

Bilateral integration refers to the ability of the research participant to coordinate movements of one side of the body, as well as simultaneous movements of both sides. As illustrated in Table 3.8 below, the research participant performed below par with regards to midline crossing (CMLX -1.2). Bilateral gross motor activities, e.g. jumping jacks and hopping, as well as fine motor task, such as diadokineses and repetitive thumb-finger circles, also revealed poorly integrated bilateral integration, as illustrated by the below par average score (BMC -0.9) on the standardized test. Left/right discrimination was on par (RLD -0.2). Considering the results of the QNST-II, the bilateral motor coordination profile has remained unchanged since the two-year postoperative assessment.

Table 3.8 Level of two-year postoperative bilateral motor coordination

	Below average				Average					Above average		
	-1.8	-1.5	-1.2	-0.9	-0.6	-0.3	0.0	+0.3	+0.6	+0.9	+1.2	+1.5
Bilateral motor coordination				**x**								
Gross motor coordination					**x**							
Fine motor coordination			**x**									
Midline crossing			**x**									
Right/left discrimination						**x**						
Functional dominance	Hand: Right; Foot: Right; Eye: Left; Ear: Left											

(A score of 0.0 is normal.)

Eye-hand coordination: (EH)

At the time of the two-year postoperative assessment the research participant's eye-hand coordination was below par, as illustrated in Table 3.9 below. According to the results of the QNST-II, the eye-hand coordination profile has not changed since the two-year postoperative assessment. The results are as follows:

Developmental Test of Visual Perception:

Eye-hand coordination	-1.3
Copying	-1.0
Visual Motor Speed	-1.6
Beery	-1.0

Table 3.9 Level of two-year postoperative eye-hand coordination

	Below average				Average					Above average		
	-1.8	-1.5	-1.2	-0.9	-0.6	-0.3	0.0	+0.3	+0.6	+0.9	+1.2	+1.5
Ball sense and skill						**x**						
Pencil grip						**x**						
Eye-hand coordination			**x**									
Visual motor speed		**X**										
Copying				**x**								
Colouring					**x**							
Cutting (scissors)					**x**							
Beery				**x**								

(A score of 0.0 is normal.)

Praxis

Developmental dyspraxia is regarded as a '… disorder of sensory integration interfering with the ability to plan and execute skilled or non-habitual motor tasks' (Ayres, 1972:12). The postoperative assessment revealed an overall below par praxis performance (IP – 1.7), as depicted in Table 3.10 below. The results of the QNST-II indicate that the praxis profile has not changed since the two-year postoperative assessment. The results are as follows:

Table 3.10 Level of two-year postoperative praxis

	Below average				Average					Above average		
	-1.8	-1.5	-1.2	-0.9	-0.6	-0.3	0.0	+0.3	+0.6	+0.9	+1.2	+1.5
Imitation of postures	**x**											
Gross motor planning			**x**									
Fine motor planning			**x**									

(A score of 0.0 is normal.)

Perceptual functioning:

At the time of the two-year postoperative assessment, the research participant functioned at below average level with regards to all modalities of perception, as depicted in Table 3.11 below. However, memory for digits was not impaired, in keeping with the current status

reflected by the SSAIS-R results. The results of the QNST-II indicate that the praxis profile has remained unchanged since the two-year postoperative assessment. The results are as follows:

Table 3.11 Level of two-year postoperative perceptual functioning

	Below average				**Average**					**Above average**		
	-1.8	-1.5	-1.2	-0.9	-0.6	-0.3	0.0	+0.3	+0.6	+0.9	+1.2	+1.5
Form constancy			**x**									
Figure-ground		**x**										
Spatial relations				**x**								
Position in space			**x**									
Visual motor integration				**x**								
Discrimination and matching						**x**						
Visual analysis and synthesis					**x**							
Visual memory		**x**										
Visual closure	**x**											
Memory for digits							**x**					
Sequential memory			**x**									

(A score of 0.0 is normal.)

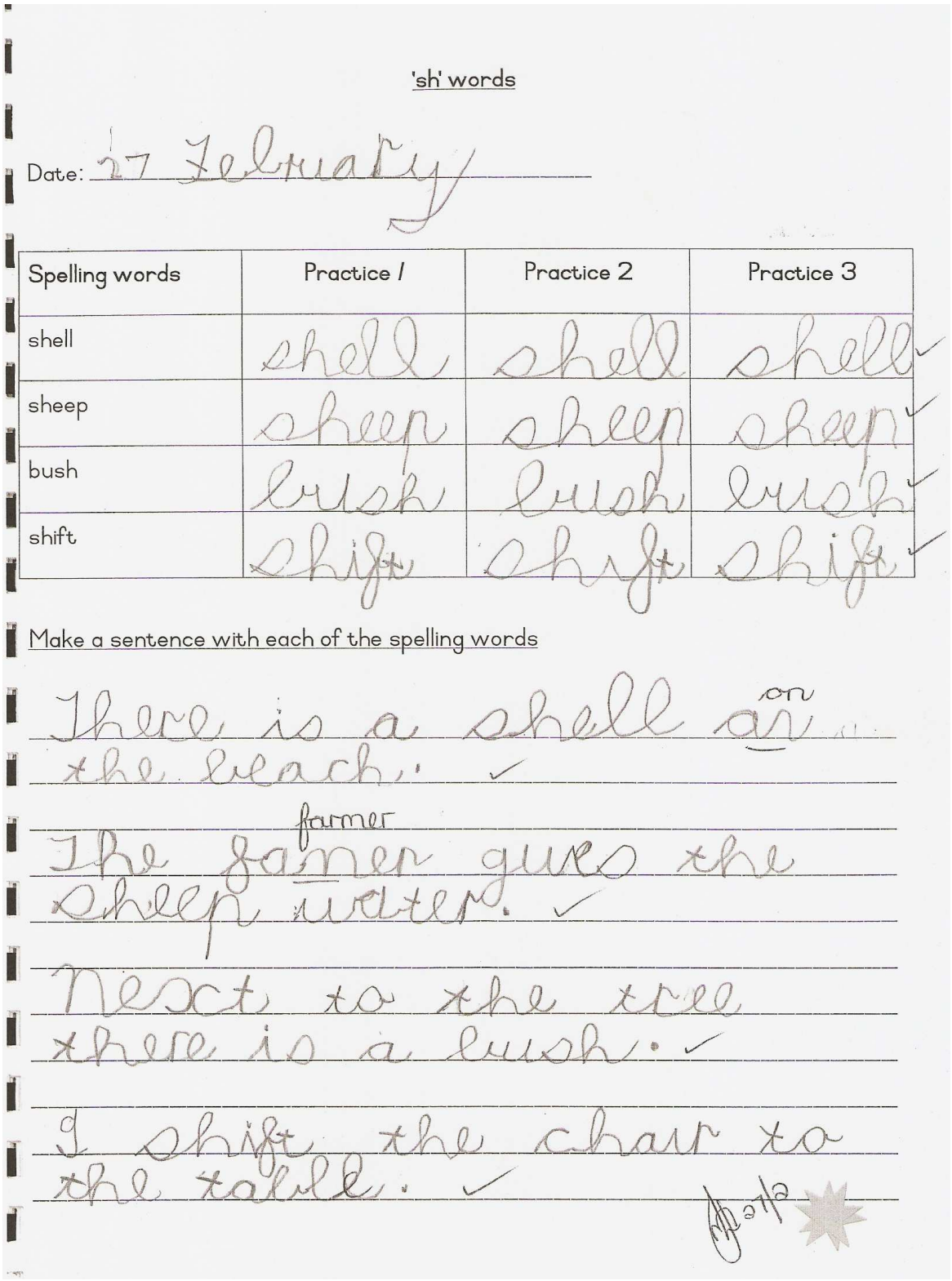
3.8.6 Samples of handwriting

Samples of the research participant's handwriting are inserted as Figures 3.5 to 3.7.

3.8.6.1 Analysis and interpretation of results

As can be noted from the samples of handwriting, the quality of the research participant's handwriting varies. There is a subtle link between differentiation at motor levels and the learning process. Considering maturation at other relevant levels, differentiation might not be equally on par across all writing and drawing tasks.

Figure 3.5



'sh' words

Date: 27 February

Spelling words	Practice 1	Practice 2	Practice 3
shell	shell	shell	shell ✓
sheep	sheep	sheep	sheep ✓
bush	bush	bush	bush ✓
shift	shift	shift	shift ✓

Make a sentence with each of the spelling words

There is a shell on the beach. ✓

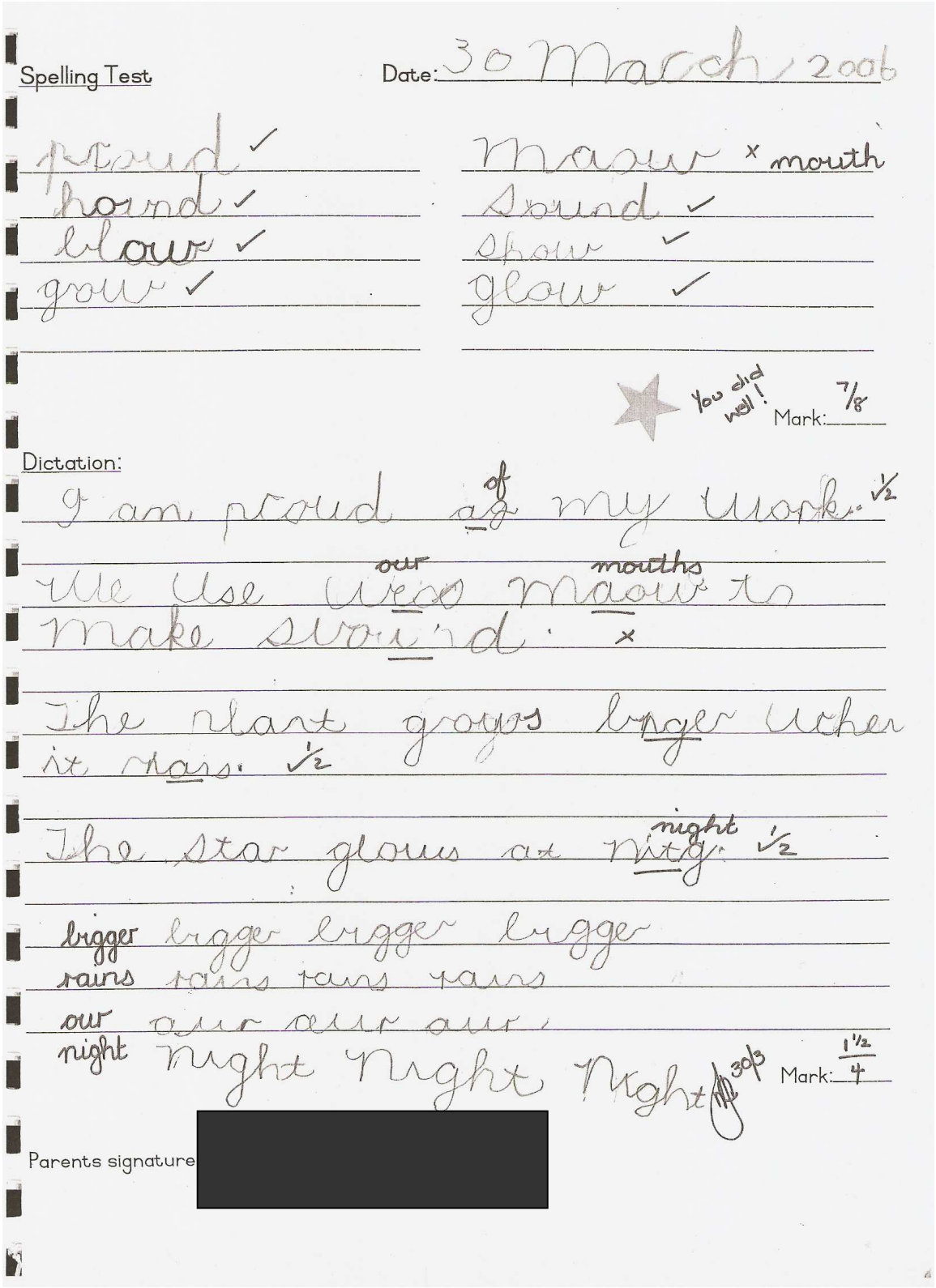
The farmer gives the sheep water. ✓

Next to the tree there is a bush. ✓

I shift the chair to the table. ✓

2/2

Figure 3.6



Spelling Test Date: 30 March / 2006

proud ✓	maour × mouth
hound ✓	Sround ✓
blaur ✓	shaur ✓
graur ✓	glaur ✓

You did well! Mark: 7/8

Dictation:

I am proud ~~a~~ of my work. ½

We use our wero mouths maour to make sround. ×

The plant graggs bnger ucher it nars. ½

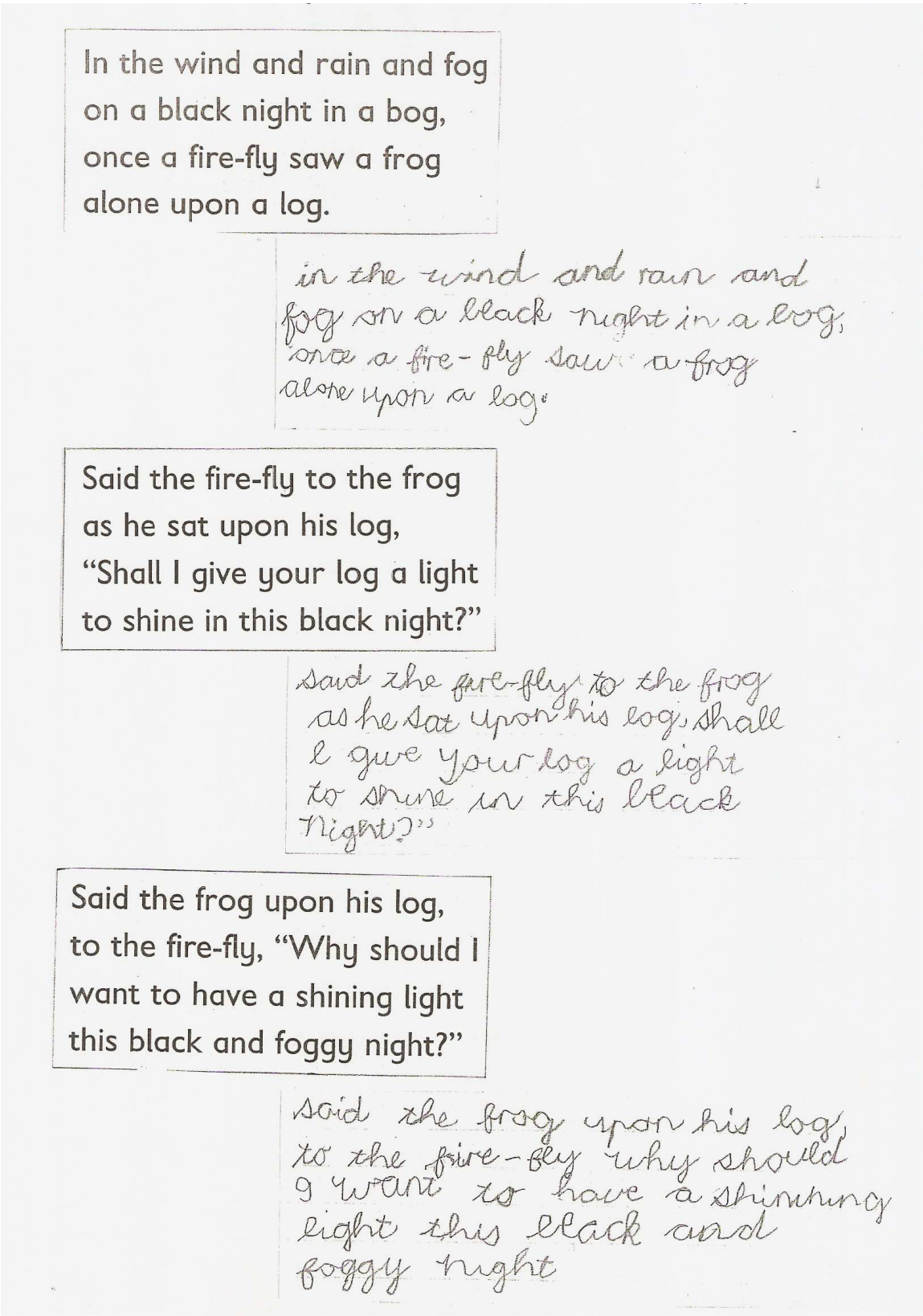
The star glaurs at nitg night ½

bigger bngge bngger bngge
rains rains rains rains
our aur aur aur
night Night Night Night 3of3

Mark: 1½/4

Parents signature [redacted]

Figure 3.7


In the wind and rain and fog
on a black night in a bog,
once a fire-fly saw a frog
alone upon a log.
in the wind and rain and
fog on a black night in a bog,
once a fire-fly saw a frog
alone upon a log.
Said the fire-fly to the frog
as he sat upon his log,
"Shall I give your log a light
to shine in this black night?"
said the fire-fly to the frog
as he sat upon his log, shall
I give your log a light
to shine in this black
night?"
Said the frog upon his log,
to the fire-fly, "Why should I
want to have a shining light
this black and foggy night?"
said the frog upon his log,
to the fire-fly why should
I want to have a shinning
light this black and
foggy night

The mastery of a specific writing movement often requires an isolated splinter skill, which is not embedded into the general developmental pattern of the learner. Slow maturation of these splinter skills might hamper the completion of differentiation at motor levels, or might cause development to stagnate to such an extent that flexible integration of motor movements is hampered (Du Preez & Steenkamp, 1986). Handwriting problems can thus be ascribed to delayed or slow maturation of these splinter skills. When muscle patterns are well integrated, the writing task requires less attention and the learner can focus her attention on the aim of the movement, rather than on its execution. A developmental delay with regard to splinter skills forces the learner to memorize each and every writing movement in detail. This hampers not only the tempo, but also the quality of written expression. Delayed motor differentiation also accounts for overflow of movement to the opposite hand.

3.9 INFERENCES AND DISCUSSION

The preceding results of the empirical study demonstrated that intracranial pressure due to Crouzon syndrome manifest in a variety of learning and effective impairments, in keeping with the problem statement of this research project. The research participant presents with an overall reduction in intellectual capacity, with the visual-motor modalities being more seriously impaired. Based upon the nature of identified impairments, the inference is that lesions might not be narrowly localized, but that lesioning is of a diffuse nature, impacting on almost all levels of cognitive functioning. However, analyses of the symptoms suggest a stronger focus of lesioning on the temporal, parietal and frontal lobes. This is in keeping with observations made by Kolb and Whishaw (2003:680):

> Although we may be able to point to a specific immediate cause of brain injury, the damage that is then wrought on the brain is not the result of a single causative event. Rather, the initial event is followed by a cascade of cellular events that can seriously compromise not only the injured part of the brain but other regions as well.

Based on a comparison between the results of the two-year postoperative assessment and the results of the current assessment, it seems that the research participant either compensated for, or recovered some of her deficits, e.g. short-term memory. Kolb and

Whishaw (2003:679) suggest that both might take place – recovery facilitated by ongoing therapy, and compensation achieved by developing new ways of functioning to compensate for lost abilities. Is all post-brain-injury improvement compensation, or do some improvements actually constitute functional restitution? Kolb and Whishaw (2003:680) state that some functional recovery, especially the return of language functions, is clearly possible if the individual is still young, but that even this recovery should be seen as a complete recovery of all functions. A goal for rehabilitation, therefore, is to find ways of stimulating plastic responses in the brain to provide the best possible compensation.

Considering the sub-questions that directed this research project, the preceding results thus provide evidence towards the following inferences:

- Which brain areas are typically lesioned with Crouzon syndrome? Depending on the nature of premature fusion, the age of the patient and the specific sutures involved in premature fusion, lesioning might be widespread across the entire cortex, or it might be localized to only those areas that were mostly subjected to intracranial pressure. However, the results of this empirical study suggest lesioning of the parietal, the temporal and the frontal lobes.

- How does atypical brain architecture and circuitry associated with Crouzon syndrome relate to compromised affect and learning? Because the neural pathways between different regions are intricately linked to form functional units, the results suggest that compromised affect and learning cannot possibly be narrowly localized, yet there might be sparing of some functions and capabilities because specific regions and their neural pathways are not affected. However, a discussion of the precise relation between atypical brain architecture and circuitry and compromised affect and learning will follow in Chapter Four.

The main research question, as well as the research hypothesis, will be addressed at the end of Chapter Four.

3.10 SYNOPSIS

This chapter presented the results of the empirical study. On the basis of these results, it was inferred that lesioning of certain regions of the cortex takes place as a result of Crouzon syndrome, but that such lesioning can be so subtle that it may impossible to identify specific lesioned sites. Nevertheless even such subtle lesioning does impact on learning and affect, as demonstrated on the basis of the empirical study reported in this chapter.

The next chapter, which deals with the research participant's pre- and postoperative MR-images, also presents an in-depth discussion of the relation between atypical brain architecture and circuitry due to Crouzon syndrome and compromised affect and learning.

3.11 LIST OF REFERENCES

Adams, R. D. & Victor, M. (1997). *Principles of Neurology,* 6th edition. New York: McGraw-Hill.

Ayres, A.J. (1972). *Southern California Sensory Integration Tests*, Manual, 4th printing 1975. Los Angeles: Western Psychological Services.

Bellak, L. & Bellak S.S. (1965). *Children's Apperception Test*, revised 1980, 10th printing 1990. New York: C.P.S. Inc.

Bellak, L. & Hurvich, M.S. (1990). *Manual for the Children's Apperception Test (Human Figures)*. New York: C.P.S. Inc.

Binkofski, F., Kunesch E., Classen, J., Seitz, R. J. & Freund, H. J. (2001). Unimodal apractic disorder of tactile object exploration associated with parietal lobe lesions. *Brain,* 124(1): 132-144.

Burns, R.C. & Kaufman, S.H. (1970). *Kinetic family drawings (K-F-D): An introduction to understanding children through kinetic drawings*. New York: Brunner/Mazel.

Burns, R.C. & Kaufman, S.H. (1972). *Actions, styles and symbols in kinetic family drawings (K-F-D)*. New York: Brunner/Mazel.

Cattell, H.B. (1989). *The 16-PF: Personality in Depth*. Illinois: Institute for Personality and Ability Testing, Inc.

Creswell, J. W. (2005). *Educational Research: Planning, Conducting, and Evaluating Quantitative and Qualitative Research.* New Jersey: Pearson Education Ltd.

Crumbaugh, J.C. (Ed.). (1990). *A Primer of Projective Techniques of Psychological Assessment*. San Diego: Libra Publishers.

Crumbaugh, J.C. (1980). Graphoanalytic cues to personality assessment. In R.H. Woody (Ed.), *Encyclopedia of clinical assessment:* Volume II (pp. 919-929). San Francisco: Jossey-Bass.

De Vos, A.S. (2000). *Research at Grass Roots: A primer for the caring professions*. Pretoria: Van Schaik.

Du Preez, J. J., Steenkamp, W. L. (1986). *Spesifieke leergestremdhede – 'n neuropsigologiese perspektief*, 2de uitgawe. Durban: Butterworths.

Du Toit, L. & Madge, E.M. (1988). *Manual for the Children's Personality Questionnaire (CPQ)*. Pretoria: Human Science Research Council (HSRC). [Original publisher Institute for Personality and Ability Testing (IPAT)]

Goodenough, F. (1926; revised 1986). *Measurement of intelligence by drawing*. New York: World Book Co.

Hammer, E.F. (1958; fourth printing 1975). *The clinical application of projective drawings*. Springfield, IL.: Charles C Thomas.

Halsband, U., Ito, N., Tanji, J. & Freund, H.J. (2001). The role of premotor cortex and the supplementary motor area in the temporal control of movement in man. *Brain*, 116 (1): 243-266.

Haworth, M.R. (1963). A Schedule for the Analysis of CAT Responses. *Journal of Projective Techniques & Personality Assessment*, 27(2): 181-184).

Heaton, R.K., Chelune, G.J., Talley, J.L., Kay, G.G. & Curtiss, G. (1993). *Wisconsin Card Sorting Test Manual: Revised and Expanded.* Florida: Psychological Assessment Resources, Inc.

Holzberg, J.D. & Wexler, M. (1950). The validity of human figure drawings as a measure of personality deviation. *Journal of Projective Techniques*, 14, 343-361.

Johnson, B. & Turner, L.A. (2003). Data Collection Strategies in Mixed Methods Research. In Tashakkori, A. & Teddue, C. (Eds.). *Handbook of Mixed Methods in Social & Behavioural Research.* London: Sage Publications.

Kolb, B. & Whishaw, I.Q. (2003). *Fundamentals of Human Neuropsychology*, 5th edition. New York: Worth Publishers.

Luria, A.R. (1973). *The Working Brain*. New York: Penquin.

Machover, K. (1949). *Personality projection in the drawing of the human figure.* Springfield, IL.: Charles C Thomas.

Milner, B., Corsi, P. & Leonard, G. (1991). Frontal cortex contribution to recency judgements. *Neuropsychologia*, 29:601-618.

Milner, B. & Petrides, M. (1984). Behavioural effects of frontal-lobe lesions in man. *Trends in Neuroscience*, 7:403-407.

Murstein, B.I. (1965). *Handbook of projective techniques*. New York: Basic Books.

Mutti, M., Sterling, H.M. & Spalding, N.V. (1998). *QNST: Quick Neurological Screening Test – Revised Edition.* Novato: Academic Therapy Publications.

Naudé, H. (1998). *Practical Psychometrics 401: Study Guide.* Braamfontein: College Publications.

Neale, M.D. (1997). *Neale Analysis of Reading Ability – Revised: Manual For Schools.* Oxford: NFER-NELSON Publishing Company Ltd.

Owen, A.M., Downes, J.J., Sahakian, B.J., Polkey, C.E. & Robbins, T.W. (1990). Planning and spatial working memory following frontal lobe lesions in man. *Neuropsychologia*, 28:1021-1034.

Purves, D., Augustine, G.J., Firtzoatrick, D., Katz, L.C., LaMantia, A-S., McNamara, J.O., & Wiliams, S.M. (Eds.). (2001). *Neuroscience*, 2nd edition. Sunderland: Sinauer Associates.

Van Eeden, R. (1992). *Manual for the Senior South African Individual Scale – Revised (SSAIS-R): Background and standardization.* Pretoria: Human Sciences Research Council.

Woody, R.H. (Ed.). (1980). *Encyclopedia of clinical assessment:* Volume II. San Francisco: Jossey-Bass.

CHAPTER 4

THE NEURAL SUBSTRATE TO COMPROMISED LEARNING AND AFFECT ASSOCIATED WITH CROUZON SYNDROME

4.1 INTRODUCTION

This chapter aims to describe compromised learning and affect associated with Crouzon Syndrome on the basis of the results of the empirical study presented in Chapter Three, and on the basis of MR-imaging protocols. The expectation is that the research participant who was diagnosed with Crouzon syndrome might show a particular profile of compromised learning and affect stemming from specific lesioned brain areas, therefore the aim is to describe the link between specific lesioned cortical regions and specific learning and affective outcomes in order to compile a tailor-made profile.

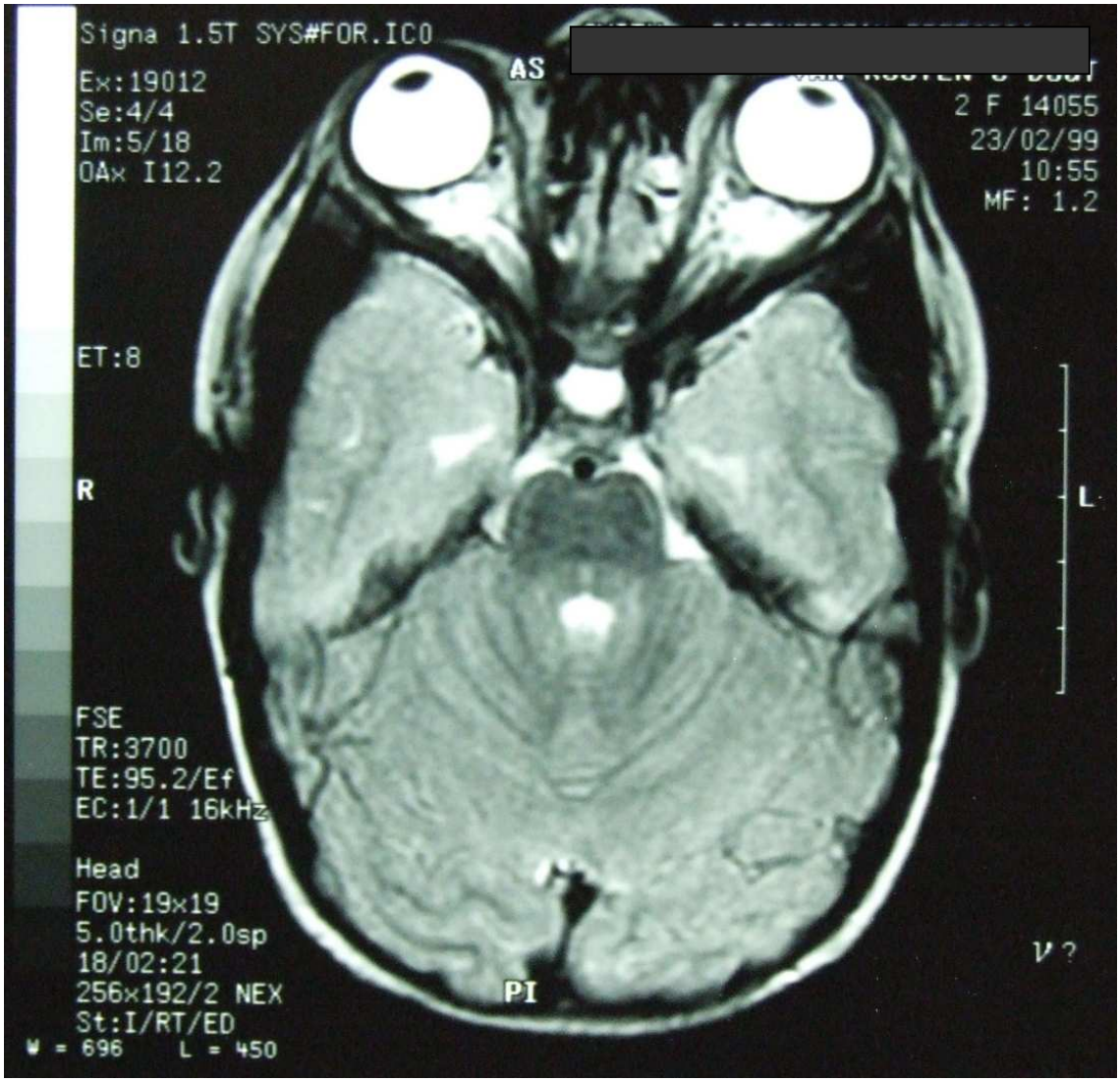
4.2 MR-IMAGING STUDIES

As already reported in Chapter Two, craniosynostosis is a congenital developmental disorder involving premature fusion of cranial sutures, often associated with multiple neurological manifestations (Flores-Sarnat, 2002:274-91; Carinci *et al.*, 2005:361-8). These neurological manifestations were demonstrated by means of the empirical study described in Chapter Three. The more than 100 syndromes described as craniosynostosis (Flores-Sarnat, 2002:274-91) are usually associated with molecular defects, particularly fibroblast growth factor receptor (FGFR) mutations, with and without limb and/or dermatological anomalies (Okajima, *et al.*, 1999:160-70). In this case, the research participant presents with no limb or dermatological anomalies.

This research project deals only with Crouzon syndrome, which is one well-known form of craniostenosis, which represents approximately 5% of craniosynostoses (Cohen & Kreiborg, 1992:12-15) and occurs equally in males and females, with an incidence of between 1/25 000 and 1/65 000 births (Cohen, 1986:453-461; Cohen & Kreiborg, 1992:12-15; Orvidas, Fabry, Diacova & McDonald, *et al.*, 1999:1372-5). The clinical features shown by the research participant are cranial synostosis, namely hypertelorism,

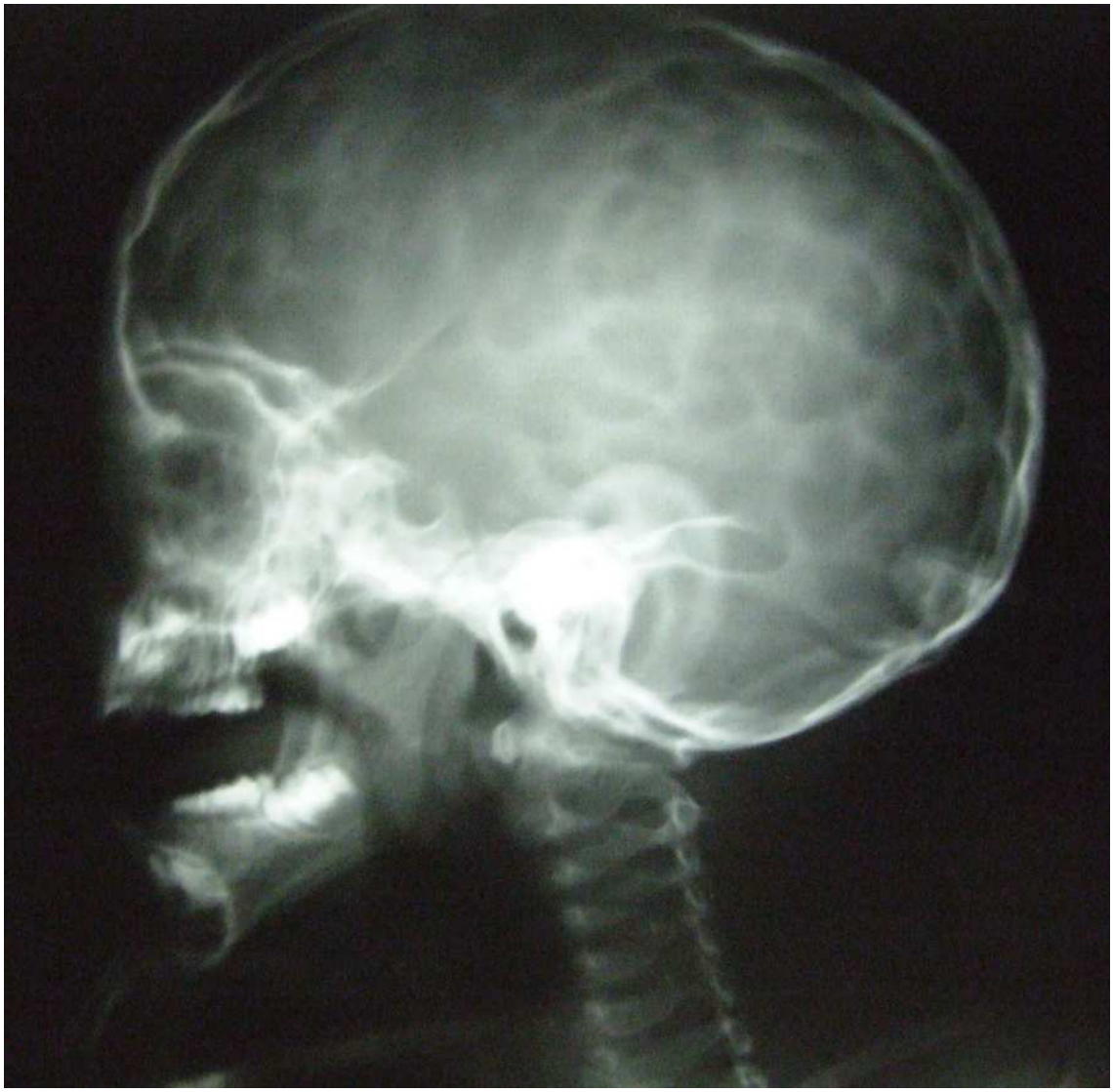
exophthalmos, parrot-beaked nose, short upper lip, hypoplastic maxilla, and a relative mandibular prognathism, in keeping with features first published by Crouzon in 1912. The research participant also has shallow orbits and ocular proptosis is distinctive, in keeping with features published by Orividas and co-workers in 1999, and Lowe and co-workers in 2000. These features are illustrated by MR-images presented in Figures 4.1 and 4.2.

Figure 4.1 MR-image demonstrating shallow orbits and ocular proptosis



The syndrome may present with calcification of the stylohyoid ligament, cervical spine abnormalities, elbow malformations, minor hand deformities, visceral anomalies, various musculoskeletal deformities, skin lesions, and cervical spine fusion anomalies affecting C2 to C5 (Prowdman, *et al.*, 1994:218-222). None of these anomalies are present in the research participant, but she doe present a pertinently misshapen cranium due to intracranial pressure, as can be noted in Figure 4.3 below.

Figure 4.2 MR-image demonstrating facial anomalies



Hydrocephalus is also associated with Crouzon syndrome (Tokumaru, 1996: 619-630; Lowe, *et al.*, 2000:907-22). Lowe and colleagues (2000) noted steep clivus, a vertically oriented brainstem and a Chiari I malformation in a 14-year-old Crouzon syndrome patient, as well as signal voids in the calvaria, probably caused by sutures from previous cranioplasty. These researchers also reported markedly abnormal venous drainage, including absence of flow in the region of the jugular bulbs, with multiple collateral veins and hypoplastic transverse sinuses. The research participant presented with hydrocephalus and a valve was inserted to relieve intracranial pressure. No other anomalies could be identified by means of MR-imaging, and no anomalies of the sinuses could be identified by means of MR-imaging, as illustrated in Figure 4.4 below.

Figure 4.3 MR-image demonstrating misshapen cranium

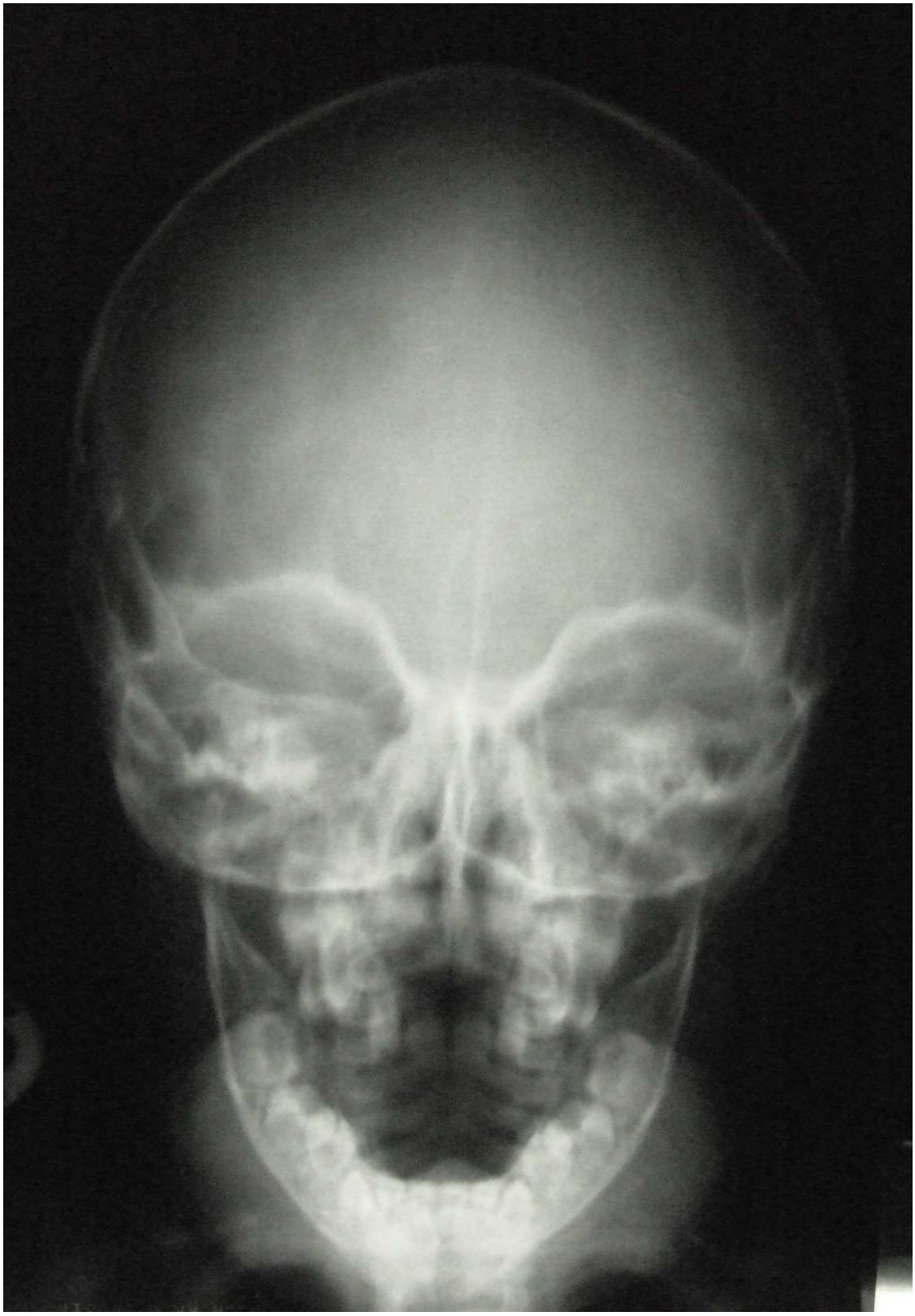
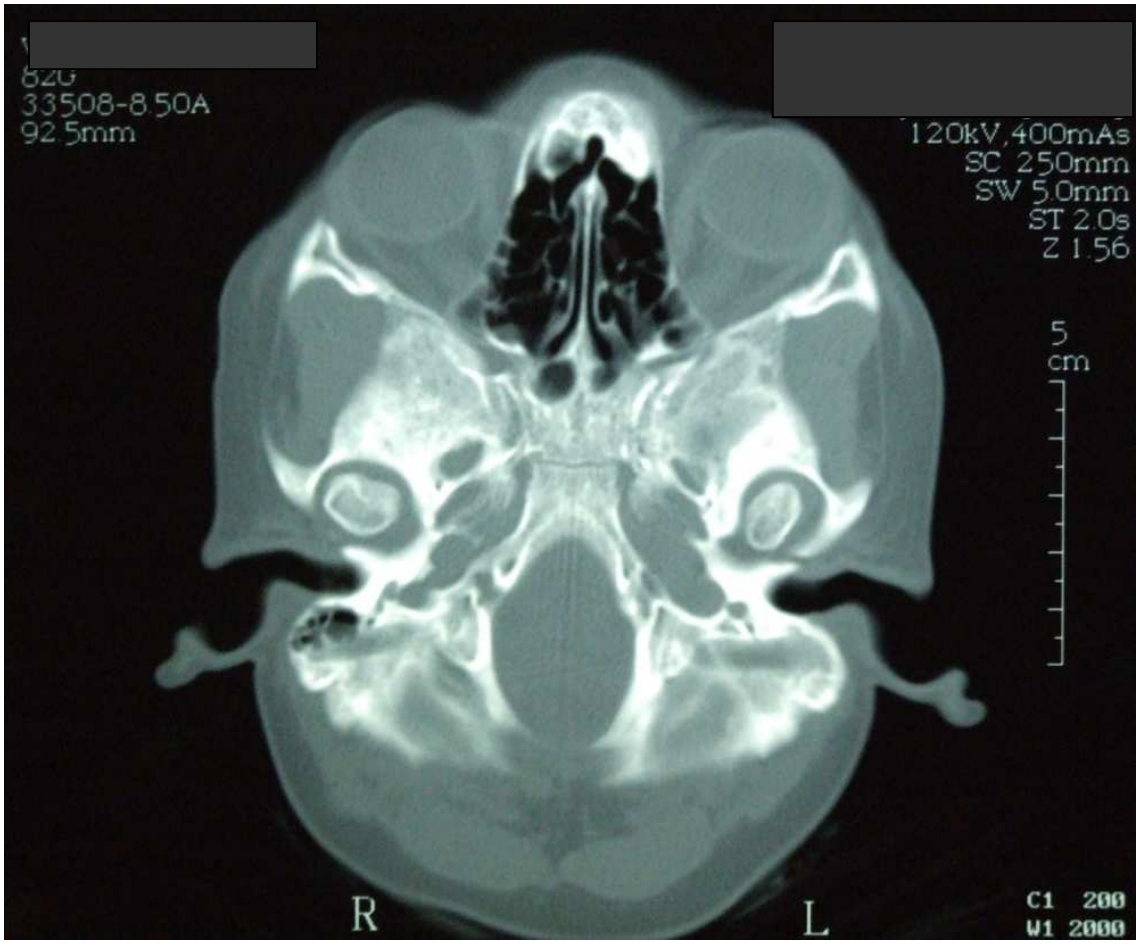


Figure 4.4 MR-image (postoperative) of sinuses



Orvidas and co-workers (1999:1372-5) reported various pathological features of the ear associated with Crouzon syndrome. Although external malformations are unusual, middle-ear anomalies and hearing loss are commonly associated with Crouzon syndrome. The research participant's inner ear structures were not postoperatively assessed, yet the results of the *Southern California Sensory Integration Tests* (SCSIT) revealed mild auditory deficits such as impaired auditory analysis, synthesis and discrimination, which suggests either malformation of the inner-ear structures, or nerve damage associated with intracranial pressure that might have resulted in conductive hearing loss.

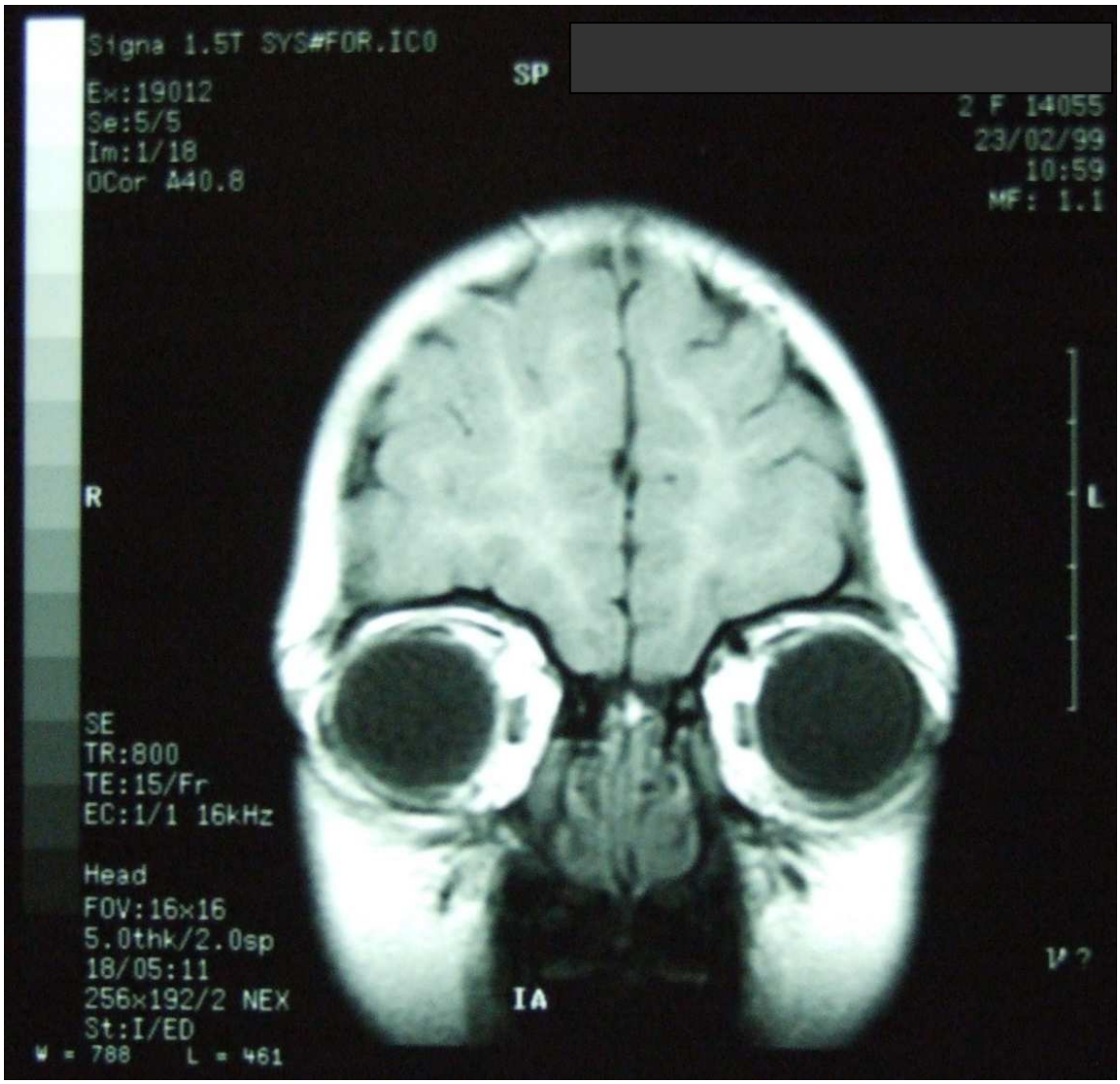
Crouzon syndrome is an autosomal dominant genetic disorder, but a large number of cases are sporadic and represent new mutations. The syndrome is associated with a mutation in the gene encoding for the fibroblast growth factor receptor 2 (FGFR2) and it is specifically a point mutation in the extra cellular or transmembrane domains of FGFR2 (Carinci, *et al.*, 2005:361-8). Approximately 44% of Crouzon syndrome cases are familial cases, while 56% are sporadic or new mutations (Kreiborg, 1981:21). Carinci and colleagues (2005)

suggested that 63% of cases represented new or sporadic mutations, while increased paternal age may be linked to sporadic mutations (Carinci, *et al.*, 2005:361-8). Since the research participant's family history does not reveal any incidents of Crouzon syndrome and no paternal age risk, this case might be ascribed to a spontaneous new mutation.

Ocular problems particularly associated with Crouzon syndrome have been associated with FGFR2. Clinical presentation disorders associated with FGFR2 include shallow orbits, proptosis, strabismus, and hypertelorism, but no ocular anterior chamber structural abnormalities have yet been reported (Okajima, *et al.*, 1999:160-70). The research participant presents with similar features, therefore FGRF2 involvement is suggested.

Fibroblasts are one of the main connective tissue cells and are responsible for the formation of collagen fibres. Bodo and colleagues noted from various studies (1999; 2000; 2002) that fibroblasts found in Crouzon syndrome have an accelerated proliferation rate and that type I and III collagen were raised in Crouzon fibroblast medium, while the concentration of fibronectin was lower than in normal cells. This phenomenon was also reported by Baroni, Lilli and Marinucci (2002:94), who suggested that this might precipitate premature fusion and overgrowth of the cranial sutures. MR-imaging shows multisuture synostosis in the research participant, as illustrated in Figure 4.5 below, with a noticeable copper beaten effect on the cranial radiographs presented in Figures 4.6-a and 4.6-b.

Figure 4.5 MR-image of multisuture synostosis



A diffuse beaten copper pattern, erosion of the dorsum sella, and suture diastases were seen more commonly in patients with craniosynostosis than in controls ($P < 0.05$), but the presence of the beaten copper pattern was no more common in children with craniosynostosis. The likelihood of intracranial pressure is greater when a diffuse beaten copper pattern, dorsum sellar erosion, suture diastases, or narrowing of basal cisterns is present ($P < 0.05$) (Tuite, Evanson, Chong, *et al.*, 1996:691-699), confirming the presence of preoperative intracranial pressure in the research participant. Although some cranial radiographic and computed tomographic findings do correlate with elevated intracranial pressure, the sensitivity of radiological methods for detecting elevated intracranial pressure is universally low and they are not recommended to screen for elevated intracranial pressure in children with craniosynostosis, nor can these methods be employed to reliably screen for optic nerve damage.

Figure 4.6-a Cranial radiograph (X-Ray) demonstrating the copper beaten effect

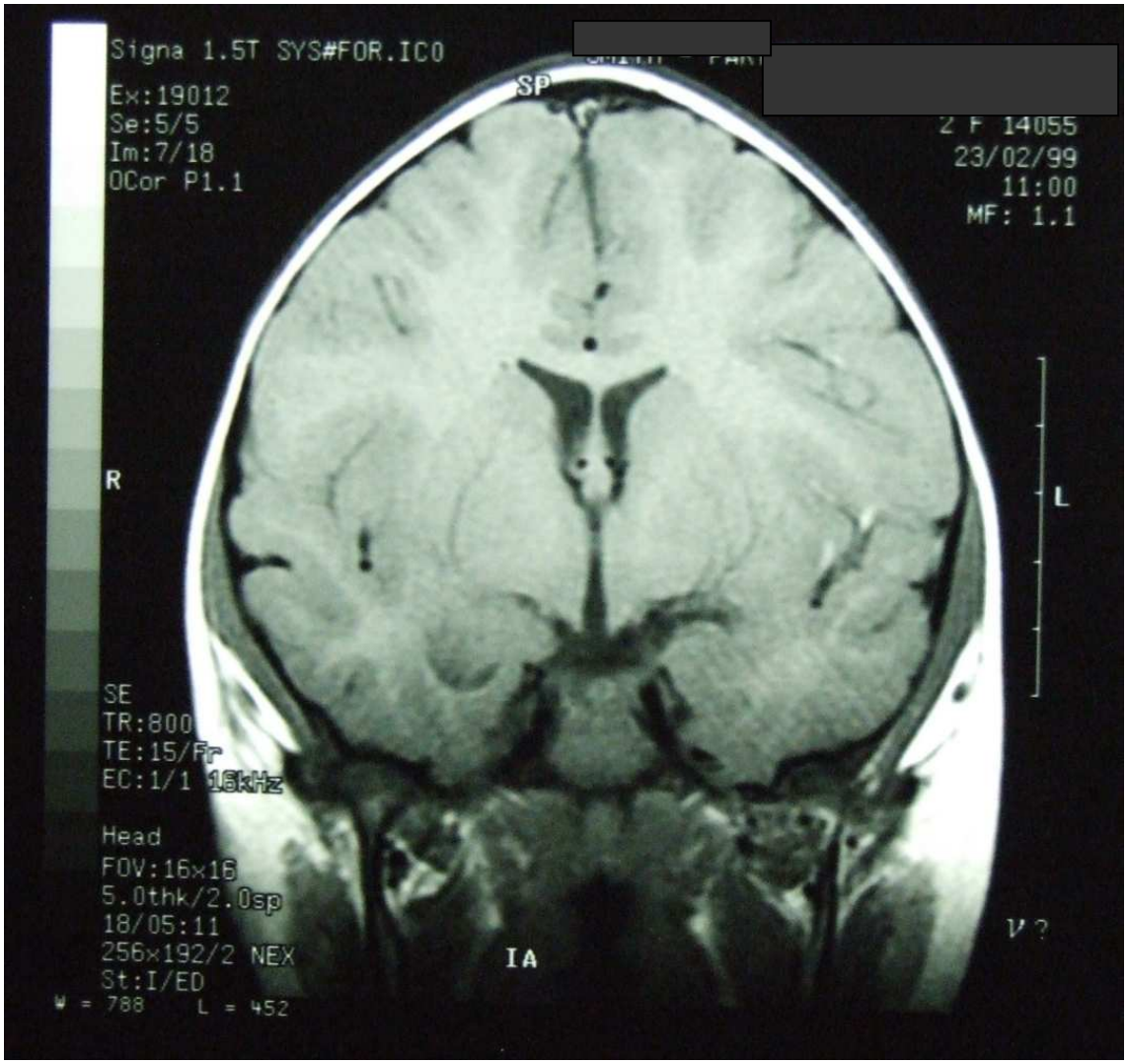
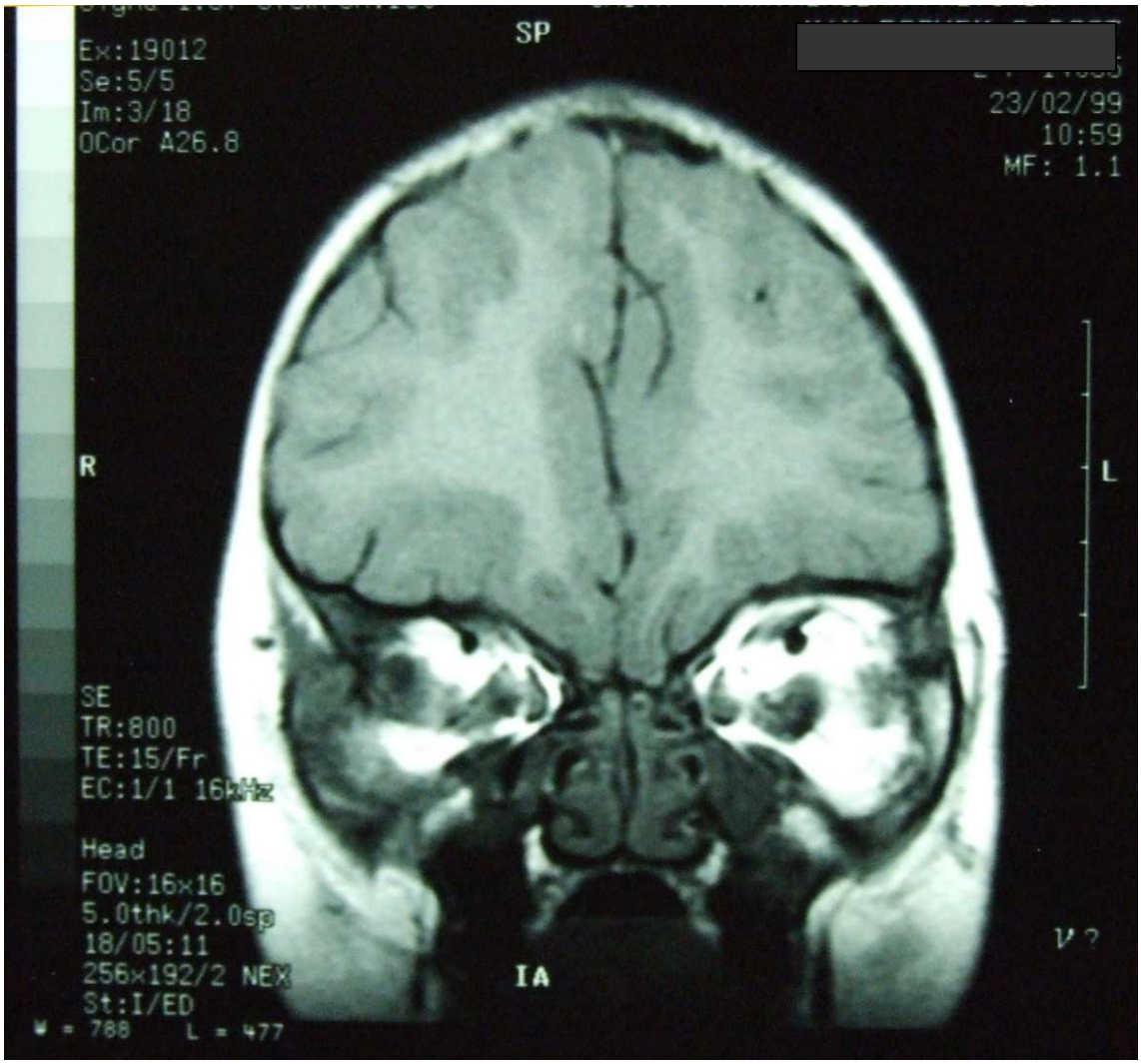


Figure 4.6-b Cranial radiograph (X-Ray) demonstrating the beaten copper effect



The preceding MR-imaging results confirm brain injury in the research participant, yet the research question relates to how injury is linked to compromised learning and affect, considering the research participant's age at the time of lesioning, as well as the plasticity of the brain that might have resulted in the sparing and recovery of certain functions. The generalization that sparing of function follows infant lesions became known as the *Kennard principle* (Kolb & Whishaw, 2003:626). For a time the idea was widely accepted, but neuroscientists began to realize that earlier may not always be better and might sometimes be worse. Kolb and Whishaw (2003:626) report that children who incur prefrontal injuries in infancy or early childhood have very poor outcomes. The ultimate effect of a brain injury depends on the behaviour affected, the extent and location of the damage, and the precise age at which the injury occurs. With respect to cognitive function in humans, it is clear that speech survives early brain damage, but some elements of syntax

and some non-language functions may not survive, and general intellectual ability may decline. Based on the results of the empirical study, the research participant shows a similar profile, reflecting reasonable recovery of language functions, but impairment of non-language functions, coupled with inhibition of general intelligence.

Riva and Cazzaniga (1986:423-428) identified three critical age divisions with regards to brain damage: before one year of age, between one and five years, and older than five years. Lesions incurred before the age of one tend to produce disproportionately greater impairments than do those incurred later. Lesions incurred between one and five years of age are followed by some reorganization of brain function, including rescuing of language functions. Lesions incurred later than five years permit little or no sparing of function. As early as in 1965, Alajouanine and Lhermitte noted writing and reading deficits in 32 brain-damaged children. Similarly, Hécaen (1976:114) followed the post-injury recovery of 15 children and noted disorders of writing and calculation to be most prominent, followed by less prominent disorders of speech. Of these children, five showed complete recovery within two years. Most of the remaining children showed considerable improvement, and in many cases the only remaining deficit was difficulty in writing. Woods (1987:519-525) studied about 50 individuals with prenatal or early postnatal brain damage to either the left or the right hemisphere. Using normal siblings as controls, they arrived at the following conclusion:

- Language survives after early left-hemisphere injury;
- Much of this survival seems attributable to appropriation of a potential language zone in the right hemisphere;
- This shift of language location has a price: specifically, some kinds of visuospatial orientation are impaired;
- Early lesions of the right hemisphere produce deficits similar to those produced by such lesions in adulthood, i.e. these functional impairments tend to be persistent.

Kolb and Whishaw (2003:629) report that brain injury to the left hemisphere during infancy result in depressed IQ scores on both the verbal and the non-verbal scales, whereas only non-verbal scores are depressed by right-hemisphere lesions, with an average IQ score of about 100. The results suggest that, if language moves to the right hemisphere, its

usual functions are sacrificed to accommodate the shift. The results also suggest that right-hemisphere functions do not shift sufficiently to interfere with language.

Kolb and Whishaw (2003:628) report the following symptoms caused by left-hemisphere lesions in childhood, and which might result in compromised learning: mutism, disorders of articulation, auditory verbal comprehension disorders, naming disorders, paraphasia, reading disorders, agraphia, facial apraxia and acalculia.

The results of the empirical study reported in Chapter Three demonstrated that the research participant endures similar difficulties as noted in the preceding paragraphs. She also revealed depressed general intellectual capacity, suggesting bilateral injury, with relative post-injury recovery of language functions and with the visuospatial functions being significantly depressed. The research participant's recovery of auditory-verbal memory is noteworthy, relative to her memory for digits, suggesting some moving of language functions to the right hemisphere, consequently functions mediated by the right hemisphere might have been sacrificed to accommodate this shift.

In the following paragraphs the research participant's injury will be linked to specific regions and impaired post-injury functioning will be discussed.

4.3 LEARNING AND AFFECT LINKED TO THE TEMPORAL LOBES

The temporal lobe comprises all the tissue that lies below the Sylvian sulcus and anterior to the occipital cortex. Subcortical temporal-lobe structures include the limbic cortex, the amygdala, and the hippocampal formation (Kolb & Whishaw, 2003:371). Because neural pathways to and from the temporal lobe extend throughout the brain, typical symptoms of temporal-lobe lesions include radical changes in affect and personality, memory disturbance, and at least a transient disturbance of language. Table 4.1 below presents a summary of the research participant's major symptoms associated with temporal-lobe lesions, which is used as conceptual base for understanding the links with compromised learning and affect.

Table 4.1 A summary of the major symptoms of temporal-lobe lesions

Symptoms	Most probable lesions site	Examples of research participant's deficits as demonstrated in chapter three
Disturbance of auditory sensation.	Areas 41, 42, 22	Overly sensitive to sounds;
Disturbance of selection of visual and auditory input	Areas TE, superior temporal sulcus	Visual and auditory distractibility noted; low performance on Sound Patterns (QNST-II);
Disorders of visual perception	Areas TE, superior temporal sulcus, amygdala	Poor performance on Non-verbal scale (SSAIS-R);
Disorders of auditory perception	Areas 41, 42, 22	Poor auditory analysis and closure on SCSIT; Poor performance on Sound Patterns (QNST-II);
Disorders of music perception	Superior temporal gyrus	Observed poor rhythm and music perception;
Impaired organization and categorization of material	Areas TE, superior temporal sulcus	Poor performance on subtest Similarities (SSAIS-R);
Poor contextual use	Area TE	Cannot follow social contextual cues (observed and CPQ);
Disturbance of language comprehension	Area 22 left	Average performance, but reportedly cannot follow conversations and instructions;
Poor long-term memory	Areas TE, TF, TH, 28	Below-average performance on subtests Comprehension and Similarities (SSAIS-R); difficulty mastering subject content;
Changes in personality and affect	Areas TE, plus amygdala	Anxiety; depression; suspiciousness; extremely withdrawn;

Adapted from: Kolb & Whishaw, 2003:377

Considering the empirical data presented in Chapter Three, the research participant's identified difficulties are mapped and linked to probable lesion sites indicated in Table 4.1. These links are now used as conceptual basis to discuss compromised learning and affect that might stem from temporal lobe functioning. Based upon the probable sites of temporal-lobe lesioning, the following learning difficulties might emerge:

4.3.1 Learning linked to temporal lobe functioning

The process of matching visual and auditory information is called cross-modal matching, consequently the research participant might find it difficult to apply visually presented material, such as in dictation, to reading, writing, arithmetic and oral class work. She might also find it difficult to follow speech on television, and similarly she might get lost if instructions that are given orally have to be applied to written work in the classroom. She might not benefit adequately from a teaching style that relies heavily on audio-visual presentations, especially since her auditory perception proves to be somewhat below average, i.e. auditory analysis, synthesis and sequencing. She might therefore be unable to grasp the meaning of words and instructions, as will be particularly noticeable in reading and/or written work, and also in oral work. In addition, she might have difficulty in differentiating between very similar sounds, such as m/n; b/d; t/d, or between words such as bell/dell; mane/name and tear/dear. She might often spell incorrectly, e.g. dean instead of bean, and she might mispronounce words. She experiences sensory 'noise' due to an over-taxed sensory system, therefore she might find it difficult to ignore irrelevant auditory information and to keep it in the background, which might narrow her attention span and impair her ability to focus on the teacher's voice, because she cannot differentiate between relevant and irrelevant sensory stimuli and cross-modal matching is impaired. In addition, the temporal lobe also plays an important role in the selection of visual and auditory input, consequently selective attention to visual and auditory input might be impaired, and the research participant might experience concomitant attention deficits.

Because of impaired auditory long-term memory, she might not be able to tell what happened in the classroom on the previous day, might find reproduction of facts difficult during tests and examination, might struggle to memorise verses, poems and rhymes, and also to remember addition, subtraction, division and multiplication combinations, and important daes in history, telephone numbers, addresses or birthdays (Naudé, 1998:53).

Furthermore, the ability to organize material is especially important for language and memory. For example, categorization makes it possible to comprehend complex, extended sentences, including both the meanings of individual clauses and the information inferred from them (Kolb & Whishaw, 2003:385). Sometimes the broader categories can be recognized, but she might experience difficulty with recognising the more specific ones.

The research participant's inability to use contextual information might hamper her ability to master comprehension tests, and she might find it difficult to recall information on the basis of a single contextual clue, e.g. providing the correct answer to a question, or responding appropriately to instructions, because the meanings of identical stimuli can vary, depending on their contexts.

4.3.2 Affect linked to temporal lobe functioning

Lesions to the temporal lobe may result in an inability to use contextual cues in social and interpersonal relations. A more complex example of extracting meaning from context can be found in social situations; therefore the research participant's social interaction might be compromised. The interpretation of events, and her role in events, depend on the social context, therefore stimuli may be interpreted in one way when she is with her parents, and in a different way when she is with her peers, which suggests impaired social cognition (Kolb & Whishaw, 2003:386).

In addition, temporal-lobe lesions have been associated with high levels of fear, anxiety and depression, due to involvement of the amygdala, as well as the anterior and medial temporal regions. Kolb and Whishaw (2003:387) note that temporal-lobe lesions are associated with specific personality traits, i.e. '… personality characteristics that overemphasize trivia and the petty details of daily life'. Pincus and Tucker (1974:57) associate temporal-lobe lesions with egocentricity and perseveration in discussions of personal significance, as well as proneness to aggressive outbursts.

4.4 LEARNING AND AFFECT LINKED TO THE PARIETAL CORTEX

The parietal lobe is the region of the cerebral cortex between the frontal and occipital lobes, underlying the parietal bone at the roof of the skull. This area is roughly demarcated anteriorly by the central fissure, ventrally by the Sylvian fissure, dorsally by die cingulate gyrus, and posteriorly by the parieto-occipital sulcus. The principal regions of the parietal lobe include post-central gyrus (Brodmann's Areas 1, 2, and 3), the superior parietal lobule (Areas 5 and 7), the parietal operculum (Area 43), the supramarginal gyrus (Area 40), and the angular gyrus (Area 39). Together, the supramarginal gyrus and angular gyrus are

often referred to as the inferior parietal lobe. The parietal lobe can be divided into two functional zones: an anterior zone including areas 1, 2, 3, and 43; and a posterior zone, which includes the remaining areas. The anterior zone is the somatosensory cortex, and the posterior zone is the posterior parietal cortex (Kolb & Whishaw, 2003:346).

The parietal cortex processes and integrates somatosensory and visual information, especially with regard to the control of movement (Kolb & Whishaw, 2003:345). Based upon an understanding of these subdivisions of the parietal cortex, this section presents a theoretical model of the links between the parietal cortex and compromised learning and affect. Damage to the anterior parietal cortex, including area PE, produces deficits in various somatosensory functions, while damage to the posterior parietal regions produces most of the other disorders ascribed to parietal-lobe lesions. Table 4.2 below presents a summary of the research participant's major symptoms associated with parietal-lobe lesions, which is used as conceptual base for understanding the links with compromised learning and affect.

Table 4.2 A summary of the major symptoms of parietal-lobe lesions

Symptom	Most probable lesion site	Examples of research participant's deficits as demonstrated in chapter three
Disorders of tactile perception; Changes in somatosensory thresholds and impaired position sense	Areas 1, 2, 3	Inability to detect a light touch by one or two sharp points; inability to localize points of touch on skin contralateral to lesion; inability to tell whether fingers of contralateral hand were passively moved;
Astereognosis	Areas 1, 2, 3	Inability to recognize the nature of an object by touch (blindfolded);
Simultaneous extinction	PE (right), PF (right)	Inability to recognize individual sensory stimulus if many sensory stimuli impinge simultaneously to the same of different body parts
Afferent paresis	Postcentral gyrus	Movement of fingers are clumsy because necessary feedback about their exact position is lost, e.g. handwriting and repetitive finger movements on QNST-II;
Tactile agnosia	Area PE	Impaired tactile sense on QNST-II;
Defects in eye movement	Areas PE, PF	Impaired eye movements and tracking;
Poor manipulation of objects	Areas PF, PG	Poor performance on subtest

		Block Designs and Form Board (SSAIS-R);
Apraxia	Areas PF. PG, left	Dyspraxia on SCSIT[2]
Constructional apraxia	Area PG	Poor performance on subtest Block Designs and Form Board (SSAIS-R);
Acalculia	Areas PG, Superior temporal sulcus (STS)	Poor performance on subtest Number Problems (SSAIS-R);
Impaired cross-modal matching	Areas PG, Superior temporal sulcus (STS)	Poor performance on subtest Coding (SSAIS-R) and WCST[3];
Impaired object recognition	Area PG right	Poor object recognition on SCSIT
Disorders of body image	Area PE	Poor body image on D-A-P and K-F-D;
Right-left confusion	Areas PF, PG	Handwriting and reversal or digits
Disorders of spatial ability	Areas PE, PG	Finger to nose on QNST-II
Disorders of drawing	Area PG	Drawings

Considering the empirical data presented in Chapter Three, the research participant's identified difficulties are mapped and linked to probable lesion sites, as presented in Table 4.2. These links are now used as conceptual basis to discuss compromised learning and affect that might stem from parietal lobe functioning respectively. Based upon the probable sites of parietal-lobe lesioning, the following learning difficulties might emerge:

4.4.1 Learning linked to parietal lobe functioning

Lesioning of the parietal lobe will inevitably affect almost all areas of learning due to this region's neural pathways that are linked to all other regions of the brain, even to the superior temporal sulcus and hippocampal formation. The posterior cortex has a role in the viewer-centred system, and contributes to many types of viewer-centred movements, e.g. eyes, head, limbs, body, and combinations thereof. The research participant's ability to apply spatial information might also be compromised (Kolb & Whishaw, 2003:351). Because mathematics and arithmetic have a quasi-spatial nature analogous to the mental manipulation of concrete shapes, but entailing abstract symbols, lesioning of the parietal lobe most significantly result in acalculia, and in the worst-case scenario, in Gertsmann's syndrome (Hynd & Obrzut, 1981). Gerstmann's syndrome, a disorder of cognition, is characterized by finger agnosia, dysgraphia, dyscalculia, and right-left disorientation. Acquired Gerstmann's syndrome is caused by lesions to the dominant (usually left) parietal

[2] Southern California Sensory Integration Tests
[3] Wisconson Card Sorting Test

lobe. The angular gyrus or subjacent white matter (Adams, *et al.*, 1998: 457), the left angular gyrus in particular, is usually involved (Mazzoni, Pardossi, Cantini, Giorgetti & Arena, 1990:459).

Dehaene and Cohen (1997:219) reported on two subjects diagnosed with acalculia/dyscalculia, one with a left subcortical lesion and the other with a right inferior parietal lesion. The subject with the subcortical lesion revealed impaired selective rote verbal knowledge, including but not limited to arithmetic tables, while her semantic knowledge of numerical quantities was intact. The subject with the right inferior parietal lesion revealed a category-specific impairment of quantitative numerical knowledge, particularly salient in subtraction and number bisection tasks, with preserved knowledge of rote arithmetic facts, suggesting that there are two distinct cerebral pathways in processing numerical knowledge. Dehaene and Cohen (1997:219) propose that a left subcortical network contributes to the storage and retrieval of rote verbal arithmetic facts, while a bilateral inferior parietal network is dedicated to the mental manipulation of numerical quantities.

Learning in all its modalities might be further compromised due to impaired tactile form recognition, impaired perception and processing of visual stimuli, compromised spatial relations, and language application. Language has many of the same demands as arithmetics, e.g. the phrases ‘my son’s wife’ and ‘my wife’s son’ contain the same words, but the spatial organization is different, therefore almost identical words and phrases have different meanings. Language could thus be seen as quasispatial, and is dependent on the polysensory region at the temporoparietal junction (Kolb & Whishaw, 2003:354). Similarly, drawing and handwriting might be compromised. A selective writing, reading, or calculation anomaly in the presence of normal oral communication triggers several interesting possibilities for the brain mechanisms behind normal language processing (Suresh & Sebastian, 2000:267). Similarly, the association of acalculia with finger anomia and agraphia with right-left disorientation may have specific implications in the neuropsychological processing of calculation and writing.

4.4.2 Affect linked to parietal lobe functioning

Very little has been published about the affective links with parietal lobe injury, however, due to tactile imperception, the research participant might violate personal space, she might accidentally bump into other children, and her affective bonding might be compromised, basically due to an aversion to touch, or due to violation of personal boundaries. Her misreaching for objects and overall lethargy and clumsiness might also contribute to social embarrassment. Furthermore, impaired sense of spatial relations coupled with impaired visual processing usually contributes to anxiety, feelings of insecurity and a fear of the dark (Naudé, 1998:45). Compromised body image might result in negative self-esteem and self-doubt, coupled with little courage to venture.

4.5 LEARNING AND AFFECT LINKED TO THE FRONTAL LOBES

In a real sense, all neural pathways eventually lead to the frontal lobes, as these lobes have a central executive function. The frontal lobes comprise all the tissue in front of the central sulcus, this region consists of 20% of the neocortex, and is made up of three functionally distinct regions, i.e. the motor, premotor, and prefrontal areas (Kolb & Whishaw, 2003:392).

The motor cortex is Brodmann's Area 4, while the premotor cortex includes Brodmann's Areas 6 and 8. The premotor cortex can be subdivided into four regions, i.e. the lateral Area 6 (lateral premotor cortex), the medial Area 6 (supplementary motor cortex), Area 8 (frontal eye field), and Area 8A (supplementary eye field). The lateral premotor cortex is expanded as Broca's Area (Brodmann's Area 44) developed in humans (Kolb & Whishaw, 2003:392).

The prefrontal cortex can also be divided intro three distinct regions, namely the dorsolateral prefrontal cortex (Brodmann's Areas 9 and 46), the inferior (ventral) prefrontal cortex (Brodmann's Areas 11, 12, 13, and 14), and the medial frontal cortex (Brodmann's Areas 25 and 32). The inferior (ventral) prefrontal cortex is sometimes referred to as the orbital frontal cortex, because of its relation to the eye socket, while the medial frontal area is sometimes considered part of the anterior cingulate gyrus rather than part of the prefrontal cortex, even though it may receive dorsomedial projections. This is

because many areas in the frontal lobe are multimodal, as in the temporal lobes, e.g. the lateral premotor cortex (Brodmann's Area 6) and the dorsolateral prefrontal cortex (Brodmann's Area 46) are responsive to visual, auditory, and somatic stimuli (Kolb & Whishaw, 2003:393). Table 4.3 below presents a summary of the research participant's major symptoms associated with frontal-lobe lesions, which is used as conceptual base for understanding the links with compromised learning and affect.

Table 4.3 A summary of the major symptoms of frontal-lobe lesions

Symptoms	Most probable lesion site	Examples of research participant's deficits as demonstrated in chapter three
Disturbances of Motor Function		
Loss of fine movements	Area 4	Handwriting; repetitive finger movements on QNST-II
Loss of strength	Areas 4 and 6; dorsolateral	Poor muscle tone on QNST-II and SCSIT
Poor movement programming	Premotor; Dorsolateral	Alternate hand movements on QNST-II; poor performance on subtests Block Designs and Form Board (SSAIS-R); poor handwriting
Poor voluntary eye gaze	Frontal eye fields	Poor performance on subtest Coding (SSAIS-R); Poor performance on WCST; Poor visual search and saccade task on subtest Missing Parts (SSAIS-R)
Poor corollary discharge	Dorsolateral; premotor	Looses balance to both sides with rotated vision on SCSIT
Broca's aphasia	Area 44	Adequate verbal comprehension but impaired expressive language
Loss of Divergent Thinking		
Poor strategy formation	Dorsolateral	Impaired Non-verbal scale (SSAIS-R); poor performance on WCST
Poor frequency estimate	Dorsolateral	Poor frequency estimate on QNST-II and SCSIT
Environmental Control of Behaviour		
Poor response inhibition	Prefrontal	Perseveration, impulsivity and distractibility
Impaired associative learning	Dorsolateral	Poor performance on subtest Coding (SSAIS-R) and on WCST
Risk taking and rule breaking	Prefrontal	Social behaviour; Results of CAT analyses
Self-regulatory disorder	Orbital	Perseveration, impulsivity and distractibility
Poor Temporal Memory		
Poor working memory	Dorsolateral	Only significant with regards to subtest Number Problems (SSAIS-R)
Poor delayed response	Dorsolateral	Impulsivity noted

Other Symptoms		
Impaired social behaviour	Orbital; dorsolateral	Clinical history; observations; analyses of CPQ and CAT
Disorders associated with damage to the facial area	Face	Low facial muscle tone on SCSIT

In the light of the empirical data presented in Chapter Three, the research participant's identified difficulties are mapped and linked to probable lesion sites in Table 4.3. These links are now used as conceptual basis to discuss compromised learning and affect that might stem from frontal lobe functioning respectively. Based upon the probable sites of frontal lobe lesioning, the following learning difficulties might emerge:

4.5.1 Learning linked to frontal lobe functioning

Because the frontal lobes assume an executive function, almost all areas of learning will be affected. Frontal lesions might result in tactlessness, disinhibition, moria (silliness), coarseness, perseveration, social imperception and decreased attentiveness (Benson, 1994:20), which might impact on overall classroom behaviour and learning. In addition, frontal lobe injury is also associated with depressed explicit-declarative memory (especially semantic memory), as well as poor error detection and restoration, consequently the research participant might not be able to recognize and correct her own mistakes, e.g., written work as well as oral work. Stirling (2002:135) considers both episodic and semantic memory to be part of the explicit-declarative memory, which might be compromised. Therefore, all tasks requiring recall might be performed at below par level.

This ability to work independently in class, to recognize and correct own mistakes, and to apply metacognitive strategies to learning might be significantly impaired. Some researchers refer to this inability as frontal lobishness (Mazzoni & Nelson, 1998; Benson, 1994), while others call it an executive dysfunction (Mezzacappa, Kindlon & Earls, 2001; Stirling, 2002), and yet another group of researchers refer to this phenomenon as a self-monitoring disturbance (Hacker, Dunlosky & Graesser, 1998; Pressley, 1994; Mazzoni & Nelson, 1998). However, all of these phenomena stemming from frontal lobe injury might result in an inability to plan, initiate and terminate actions, to think in conceptual terms, to adapt to changing circumstances and to respond in socially appropriate ways. The research

participant's classroom behaviour might therefore be characterized by marked dependent behaviour, impulsivity, poor decision-making, and sometimes even perseveration on the same task.

The frontal lobes receive information from, and send information to most other cortical and subcortical regions such as the basal ganglia, the limbic system and the cerebellum (Stirling, 2002:208). Due to lesioned frontal lobes, relay of information to these structures might be compromised (Panksepp 1998:34), resulting in affective over- or underreactivity to the learning matter and learning task. Some researchers regard the anterior cingulate gyrus as part of the frontal cortex, some as part of the sub-cortical structures (Kolb & Whishaw, 2003). However, the anterior cingulate gyrus plays an important role in the selection of appropriate responses and the inhibition of inappropriate ones within this domain of behaviour. Therefore, selection and inhibition can be impaired because memory that subserve error detection and restoration is either poorly retrieved, or inaccessible. Panksepp (1998:34) describes these mechanisms as hierarchical representations throughout the EES that course between higher and lower levels of the brain *concurrently*. It is therefore important to consider how basic emotional systems might modify the higher cerebral processes needed for adequate classroom performance.

4.5.2 Affect linked to frontal lobe functioning

It is thought that metacognitive knowledge develops from children's interactions with peers and adults (Anderson *et al.*, 2001:45; Hacker *et al.*, 1998:71), therefore a child learns to recognise and restore mistakes through social interaction with other individuals, and if social interaction is compromised, metacognitive knowledge might not be acquired (Hacker, *et al.*, 1998:368). Research examining the relationship between metacognitive knowledge and error detection and restoration indicates that children who are aware of *why*, *when* and *how* strategies should be used, are more likely to be able to use monitoring strategies successfully (Pressley, 1994:259), implicating that frontal lobe injury might result in a lack of specific *strategy knowledge*, coupled with deficient monitoring skills (Hacker, *et al.*, 1998:72). Based upon the cognitive psychological perspective, frontal lobe injury might result in a self-monitoring disturbance (Mazzoni & Nelson, 1998) or in an executive dysfunction (Stirling, 2002), coupled with poor memory access and retrieval, which might result in limited error detection and restoration. One needs metacognitive

strategies in order to sufficiently address behaviour difficulties, poor planning, impulsiveness, and perseveration (on feelings or manual tasks). The frontal lobes are also involved in conceptual thinking and one's ability to adapt to changing circumstances and contexts, allowing one to respond to these changes in socially appropriate ways. It was demonstrated at hand of the CPQ that the research participant finds it difficult to monitor her own behaviour, that she reveals social imperception, coupled with an inability to asimmilate change.

Balance between the prefrontal cortex and amygdala is an important aspect of mental health (Restak, 2000:130). Despite adequate understanding and sufficient social knowledge, the research participant might not be able to evaluate social situations, apply information to other situations in a socially acceptable way, detect and restore mistakes, and regulate emotional behaviour (Newport & Nemeroff, 2000:216). In addition, functional impairment of the orbitofrontal cortex will hamper explanation of social rules and knowledge, coupled by an inability to regulate own behaviour. The research participant might also be unable to apply her social knowledge to relevant social situations (Carlson, 1994:344). Because various neural pathways link the temporal, parietal and frontal regions, injury to the hippocampus might produce poor regulation of behaviour, as stored social information cannot be sufficiently retrieved from the hippocampus. Generally, frontal lobe injury is associated with an inability to regulate own behaviour, to make choices, to detect and restore mistakes, and to regulate emotional behaviour (Restak, 2000:77), therefore the research participant's emotions might be erratic and unpredictable. Due to inhibited anterior cingulate gyrus and orbitofrontal cortex functioning, attention and problem solving might also be impaired (Newport & Nemeroff, 2000:216), exacerbating her feelings of insecurity and dependency.

4.6 REVISITING THE RESEARCH STATEMENT

The research statement that directed this research project was formulated as follows:

What is the relationship between developmental brain insults associated with Crouzon syndrome and compromised affect and learning?

Without repeating all the results, the answer to this research statement could be summarized as follows:

As a general rule, one can say that brain injury represents a major change in programmed neural development, with implications for future social and affective adjustment, as well as for learning. However, as demonstrated in the preceding chapters, these changes also imply a certain degree of neural change and recovery that should be considered when attempting to compile a profile based on brain injury stemming from Crouzon syndrome.

This research project demonstrated that Crouzon syndrome might result in depressed general intellectual capacity, but because this syndrome manifests during infancy or early childhood, it is likely that the language functions will be fully or partially recovered. The possibility also exists that some language functions might be reorganized or relocated to the right hemisphere, causing a discrepancy between the verbal and the performance (non-verbal) scores attained on an IQ test, because the right hemisphere cannot accommodate these functional changes without further sacrifice of some right hemisphere functions. Brain injury associated with Crouzon syndrome can usually be ascribed to heightened intracranial pressure, and lesioning might be widespread across the entire cortex. However, MR-imaging usually does not identify these subtle insults, and impaired functioning has to be inferred by employing applicable psychological testing.

4.7 REVISITING THE RESEARCH HYPOTHESIS

The following research hypothesis was formulated for this research project:

There is a neural substrate to compromised affect and learning associated with Crouzon syndrome, resulting in a particular affective and learning profile.

By way of the empirical study it was demonstrated that there is a neural substrate to compromised affect and learning associated with Crouzon syndrome, which allows one to compile a particular affective and learning profile upon which further affective and learning support could be based. The research hypothesis is therefore accepted.

4.8 SYNOPSIS

This chapter dealt with the link between specific lesioned cortical regions and specific learning and affective outcomes, and an individualized affective and learning profile of the research participant was compiled. Findings from the empirical study and MR-imaging studies were used to describe the affective and learning difficulties that the research participant might be experiencing as a result of early brain injury caused by Crouzon syndrome.

The following chapter will present the findings, conclusion and recommendations for further research.

4.9 LIST OF REFERENCES

Adams, R.D., Victor, M. & Ropper, A.H. 1998. *Principles of Neurology*. McGraw-Hill.

Anderson, V., Northam, E., Hendy, J. & Wrennall, J. 2001. *Developmental neuropsychology: a clinical approach.* Philadelphia: Taylor & Francis Inc.

Alajouanine, T. & Lhermitte, F. 1965. Acquired aphasia in children. *Brain*, 88:653-662.

Benson, D.F. 1994. *The neurology of thinking*. New York: Oxford University Press.

Bodo, M., Baroni, T. & Carinci, F. 1999. A regulatory role of fibroblast growth factor in the expression of decorin, biglycan, betaglycan and syndecan in osteoblasts from patients with Crouzon syndrome. *European Journal of Cellular Biology*, 78:323-330.

Bodo, M., Baroni, T. & Carinci, F. 2000. Interleukin secretion, proteoglycan and procollagen alpha (I) gene expression in Crouzon fibroblasts treated with basic fibroblast growth factor. *Cytokine*, 12:1280-1283.

Bodo, M., Lilli, C. & Aisa, M.C. 2002. Basic fibroblast growth factor: effects on matrix remodeling, receptor expression, and transduction pathway in human periosteal fibroblasts with FGFR2 gene mutation. *Journal of Interferon Cytokine Research*, 22:621-630.

Baroni, T., Lilli, C. & Marinucci, L. 2002. Crouzon syndrome: differential in vitro secretion of bFGF, TGFbeta isoforms and extracellular matrix macromolecules in patients with FGFR2 gene mutation. *Cytokine*, 19:94-98.

Carinci, F., Pezzetti, F., Locci, P., Becchetti, E., Carls, F., Avantaggiato, A., Becchetti, A., Carinci, P., Baroni, T. & Bodo, M. 2005. Apert and Crouzon syndromes: clinical findings, genes and extracellular matrix. *Journal of Craniofacial Surgery*, May, 16(3):361-8.

Carlson, N.L. 1994. *Physiology of behaviour*. 5th edition. London: Allyn & Bacon.

Cohen, M.M. 1986. *Craniosynostosis: Diagnosis, Evaluation and Management*. New York: Raven Press.

Cohen, M.M. & Kreiborg, S. 1992. Birth prevalence studies of the Crouzon syndrome: comparison of direct and indirect methods. *Clinical Genetics*, 41:12-15.

Dehaene, S. & Cohen, L. 1997. Cerebral pathways for calculation: double dissociation between rote verbal and quantitative knowledge of arithmetic. *Cortex*, 33(2):219-50.

Flores-Sarnat, L. 2002. New insights into craniosynostosis. *Seminars in Pediatric Neurology*, December, 9(4):274-91.

Hacker, D.J., Dunlosky, J. & Graesser, A.C. (Eds.). 1998. *Metacognition in Educational Theory and Practice*. London: Lawrence Erlbaum Associates.

Hécaen, H. 1976. Acquired aphasia in children and the ontogenesis of hemipspheric functional specialization. *Brain and Language*, 3:114-134.

Hynd, G.W. & Obrzut, J.E. 1981. *Neuropsychological Assessment and the School-Age Child. Issues and Procedures.* New York: Grune & Stratton.

Kolb, B. & Whishaw, I.Q. 2003. *Fundamentals of Human Neuropsychology*, 5th edition. New York: Worth Publishers.

Kreiborg, S. 1981. Crouzon syndrome. A clinical and roentgencephalometric study. *Scandinawian Journal of Plastic Reconstructive Surgery*, 19(Suppl): 1-198.

Lowe, L.H., Booth, T.N., Joglar, J.M. & Rollins, N.K. 2000. Midface anomalies in children. *Radiographics*, Jul-Aug, 20(4):907-22.

Mazzoni, M., Pardossi, L., Cantini, R., Giorgetti, V. & Arena, R. 1990. Gerstmann syndrome: a case report. *Cortex.* 26(3):459-67.

Mazzoni, G. & Nelson, T.O. 1998. *Metacognition and cognitive neuropsychology – monitoring and control processes*. London: Lawrence Erlbaum Associates.

Mezzacappa, E., Kindlon, D. & Earls, F. 2001. Child abuse and performance task assessment of executive functions in boys. *Journal of Child Psychological Psychiatry*, 42 (8):1041-1048.

Naudé, H. 1998. *Practical Psychometrics*. Braamfontein: College publications.

Newport, D.J. & Nemeroff, C.B. 2000. Neurobiology of posttraumatic stress disorder. *Current Opinion in Neurobiology*, 10 (2):211-218.

Okajima, K., Robinson, L.K., Hart, M.A., Abuelo, D.N., Cowan, L.S., Hasegawa, T., Maumenee, I.H. & Jabs, E.W. 1999. Ocular anterior chamber dysgenesis in craniosynostosis syndromes with a fibroblast growth factor receptor 2 mutation. *American Journal of Medical Genetics*, July, 16,85(2):160-70.

Orvidas, L.J., Fabry, L.B., Diacova, S. & McDonald, T.J. 1999. Hearing and otopathology in Crouzon syndrome. *Laryngoscope*. September, 109(9):1372-5.

Panksepp, J. (Ed.). 1998. *Affective Neuroscience*. New York: Oxford University Press.

Prowdman, T.W., Moore, M.H., Abbott, A.H. & David, D.J. 1994. Noncraniofacial manifestations of Crouzon disease. *Journal of Craniofacial Surgery*, 5:218-222.

Pincus, J.H. & Tucker, G.J. 1974. *Behavioural Neurology*. New York: Oxford University Press.

Pressley, M. 1994. Embracing the complexity of individual differences in cognition: Studying good information processing and how it might develop. *Learning and Individual Differences,* 6:259-284.

Restak, R. 2000. *Mysteries of the Mind*. Washington DC: National Geographic Publishers.

Riva, D. & Cazzaniga, L. 1986. Late effects of unilateral brain lesions sustained before and after age one. *Neuropsychologia*, 24:423-428.

Stirling, J. 2002. *Introducing Neuropsychology*. New York: Taylor & Francis Inc.

Suresh, P.A. & Sebastian, S. 2000. Developmental Gerstmann's syndrome: a distinct clinical entity of learning disabilities. *Pediatric Neurology*, 22(4):267-78.

Tokumaru, A.M., Barkovich, A.J., Ciricillo, S.F. & Edwards, M.S. 1996. Skull base and calvarial deformities: association with intracranial changes in craniofacial syndromes. *American Journal of Neuroradiololgy*, 17:619-630.

Tuite, G. F. , Evanson, J., Chong, W. K., Thompson, D., Harkness, W., Jones, B. M. & Hayward, R.D. 1996. The Beaten Copper Cranium: A Correlation between Intracranial Pressure, Cranial Radiographs, and Computed Tomographic Scans in Children with Craniosynostosis. *Neurosurgery*, October 1996, 39(4):691-699.

Woods, B.T. 1987. Impaired speech shadowing after early lesions of either hemisphere. *Neuropsychologia*, 25:519-525.

CHAPTER 5

FINDINGS, CONCLUSIONS AND RECOMMENDATIONS

5.1 INTRODUCTION

This research project, which included a clinical case study, made it possible to describe the compromised affect and learning associated wit Crouzon syndrome. Three critical inferences were based upon a review of relevant literature: The first inference was that a craniofacial condition such as Crouzon Syndrome might be classified as a pervasive developmental disorder, since the brain is not fully developed at the time of diagnosis, while the concept pervasive suggests that these impairments significantly affect individuals throughout their lives (Barlow & Durand, 2002:464). The second inference implicated a neural substrate to compromised learning associated with Crouzon syndrome, therefore the expectation was that individuals diagnosed with Crouzon Syndrome might show a particular profile of compromised learning. As a result of specific lesioned brain areas, the third inference was that compromised affect associated with Crouzon Syndrome might be delicately intertwined with compromised learning.

In wrapping up this research project the most significant findings, conclusions and recommendations are summarized in this chapter.

5.2 OVERVIEW

The research statement that directed this project was formulated as follows:

What is the relationship between developmental brain insults associated with Crouzon syndrome and compromised affect and learning?

This research question was answered by means of an in-depth literature review, and at hand of psychological assessment data and MR-imaging studies.

The research hypothesis was defined as follows:

There is a neural substrate to compromised affect and learning associated with Crouzon syndrome, resulting in a particular affective and learning profile.

Based on the results of the empirical study it was concluded that there is a neural substrate to compromised affect and learning associated with Crouzon syndrome, which results in a particular affective and learning profile. The research hypothesis was thus accepted.

5.3 FINDINGS

In response to the research statement and sub-questions, the findings are summarized as follows:

5.3.1 Significant findings related to the manifestation and pathogenesis of Crouzon syndrome

- Crouzon syndrome is characterized by marked by abnormalities in the shape of the skull due to premature fusion of the sutures.
- Craniotomy and skull reshaping procedures are often required to relieve intracranial pressure, and to prevent facial deformity.
- Premature synostosis may be evident at birth or develop during the first year of life and is completed by the second or third year.
- The sutures affected include the coronal, the sagittal, and occasionally the lambdoidal sutures, but usually involving the coronal sutures.
- Compensatory growth occurs at the remaining open sutures to allow continued postnatal brain growth, causing midfacial hypoplasia, shallow orbits, a foreshortened nasal dorsum, maxillary hypoplasia, occasional upper airway obstruction and brachycephaly, which is a short skull.
- The degree of cranial malformation in Crouzon syndrome depends on the order and rate of progression of sutural synostosis.

- It is most likely that Crouzon syndrome would impact on the second period of neurodevelopment, i.e., dendritic and axonal growth, synapse production, neuronal and synaptic pruning, and changes in neurotransmitter sensitivity.
- Hydrocephalus with increased intracranial pressure may occur due to craniosynostosis.
- Crouzon syndrome can cause malformation of the structures of the middle ear with accompanying hearing loss.
- Shallow orbits with ocular proptosis often result from Crouzon Syndrome.
- Headaches and failing vision are also attributable to elevated intracranial pressure and further visual disturbances can result from corneal injury due to exposed conjunctivitis or keratitis.
- Hypoplasia of the mid face with diminished nasal and nasopharyngeal spaces may occur.
- Cleft palate and sub mucous cleft palates are occasionally seen in Crouzon syndrome, but overall, this developmental anomaly is an uncommon finding.
- In syndromes of craniosyntosis, hyponasal resonance during speech is a common finding, which is due to the stenotic suture lines between the facial bones and cranial base.
- Articulation is probably the component of speech that is most consistently impaired in individuals with Crouzon syndrome, due to the malocclusion that is so common in syndromes of craniosynostosis.
- Articulation errors can be grouped into four basic categories based on congenital malformations: developmental, obligatory, compensatory, and neurogenic articulation errors.
- Learning disabilities are also commonly found in individuals with craniofacial syndromes.
- The secondary effects of increased intracranial pressure might include language impairment, discoordination of the speech mechanism, as well as cognitive impairment.
- Conductive hearing loss might further impair language development.
- The intelligence scores tend to deteriorate in later years due to the individual's significant difficulty with abstract thinking. Mathematics and reading comprehension might be the most severely affected learning skills due to this difficulty with abstraction.

5.3.2 Significant findings related to the empirical investigation

Psychological assessment media

- Optometric, cognitive, neuropsychological and personality assessment were employed to complement brain imaging studies and these proved to be a valid and reliable test battery.

Cognitive functioning

- The Senior South African Individual Scale (SSAIS-R) was administered before and after optometric correction, with a lapse of six months in between.
- The research participant's general intellectual functioning before and after optometric correction was found to be at borderline level (Van Eeden, 1992:48).
- The verbal-auditory processing before and after optometric correction was at low average level of intellectual functioning.
- The visual-motor processing before optometric correction was at cognitively handicapped level, but rose to borderline level after optometric correction, indicating a gain of 15 Non-verbal IQ points after optometric correction.
- The scaled score difference between the Verbal and the Non-verbal Scales before optometric correction was statistically significant at the 1% level of reliability, but after optometric correction no statistically significant difference between these two scales was noted.
- Before optometric correction there was a twenty-six-point superiority of Verbal over Non-verbal IQ, suggesting an advantage of auditory-verbal abilities over constructional praxis tasks involving visual-motor abilities.
- After optometric correction the auditory-verbal superiority over visual-motor abilities could still be noted, yet it was no longer statistically significant.
- This verbal/non-verbal difference after optometric correction was ascribed to better develop crystallized abilities that were acquired through previous training, education and acculturation, as opposed to fluid abilities that involve immediate problem solving and reasoning (without prior experience).

- Immediate problem solving and reasoning required for cognitive adaptability and flexibility was found to be below par, suggesting frontal lobe injury.
- A relative sparing and/or recovery was found of those cortical areas that process auditory-verbal information, while those cortical areas involved in visual-motor tasks, constructional praxis, planning, and problem solving were significantly lesioned, without any indications of recovery.
- Weaknesses in the processing of visual-motor information were found, e.g., due to lesioning of the parietal, temporal and frontal lobes.
- Persistent deviations on the scatter profile after optometric were found on all three scales, i.e., on the Verbal, the Non-verbal and the Full Scales of the SSAIS-R.
- Average performance with regards to short-term verbal memory and recall was found in comparison to the norm population.
- An average performance on verbal memory tasks suggested a relative sparing of the regions that mediate these functions; alternatively these functions might have been reorganized in the left hemisphere; alternatively the long-term memory system could have supported recognition and retrieval of various of the stimulus words, resulting in a false positive strength.
- Extreme concrete thinking in performing arithmetical operations was noted, which manifested as dyscalculia.
- Poor visual-associative learning was found.

Neurological screening

- Mixed dominance is established, i.e., left eye, right hand and left foot dominance.
- The following developmental delays were identified: low muscle tone with inhibition of cursive writing and voluntary finger movements, difficulty in switching smoothly between cerebral commands that initiate a movement and those that terminate a movement, poorly controlled handwriting movements, disrupted rhythm and automated handwriting.
- Impaired revisualization by means of haptic-tactile recognition and perception was noted, which hampered handwriting proficiency.

- It was found that handwriting deficits resulted from an inability to recall the visual image and symbols, or the sequence of symbols by means of tactile processing of information.
- Impaired proprioception was found.
- Impaired eye movements and tracking manifested as horizontal and vertical jerkiness, incoordination, visual distractibility, inability to cross the visual midline (associated with below par laterality), as well as poorly developed optic muscles.
- In ability to process auditory information in the form of sound patterns, and to reproduce patterns both orally and in terms of motor activity were found.
- Disturbed orientation of concrete space was found, suggesting poor direction within personal space, i.e. to discriminate between left and right, in front and behind, above and beneath, and difficulties regarding motor planning and control, as well as poor sense of body placement.
- A poor sense of body placement carried over to objects outside the self, such as words on the written page or placement of the pencil in a specific place on the page.
- Disorientation of concrete space was associated with impaired localization of sounds and compromised visual-auditory integration, which was found to have resulted in emotional insecurity.
- Rhythm and synchronization were found to be below par, coupled with poor motor-planning ability.
- Dyspraxia was noted, based on an inability to incorporate certain voluntary motor actions into a single action.
- Below par gross-motor skill related to poor dynamic balance, sequencing, motor planning and sense of place in space was found.

Screening for frontal lobe injury

- The *Wisconsin Card-Sorting Test* proved to be a reliable and valid clinical test of frontal lobe injury.
- Impaired response inhibition and inflexible behaviour, coupled with difficulty to use environmental cues (feedback) to regulate or change behaviour were identified. This difficulty manifested in perseveration on responses in a variety of situations during test administration, particularly those that demanded a change of strategy. This difficulty

also manifested in behavioural irregularities such as distractibility, impulsivity, and below par ability to shift response strategies (perseveration was noted).

Personality structure

- The *Children's Personality Questionnaire* was found to render valid and reliable data, even in the presence of low-average cognitive ability.
- Social inhibition and withdrawn behaviour were noted. Choices about how to spend free time are strongly influenced by personal interests, which are employed as substitutes for social involvement.
- A tendency to channel the bulk of psychological energy into non-social activities was noted, resulting in feelings of alienation and a preference to play with animals and inanimate objects.
- Inability to notice the importance of the social and emotional contexts was observed, coupled with impaired sense for social cues.
- An inflexible and concrete mind resulted in stubbornness and reluctance to accept a new trail of thought.
- A weakened capacity to discern relationships was identified.
- Capacity for emotional control was found to be weakened.
- Due to an inability to make sense of the social context, various defence mechanisms are employed.
- The content and action of moral values are in line with mainstream values.
- A preference for certainty and predictability was found, coupled with a tendency to preserve sameness and avoid radical changes.
- Impaired willingness to venture was found, which is closely associated with a low tolerance for fear and arousal, and a need to protect the delicate internal homeostasis.
- Dependency, coupled with a low self-concept and confused sense of identity was found. There seems to be a vast difference between the wished-for self-concept and the subjectively perceived self-concept, conversely self-degrading self-talk and feelings of shame and anxiety prevail.
- In the case of below-average non-verbal cognitive ability, and due to concurrent compromised drawing ability, the D-A-P and the K-F-D were found to render less valid

and reliable data in terms of personality structure, although all interpretations were in keeping with the CPQ protocols.

- It was found that the *Schedule of Adaptive Mechanisms in CAT responses*, developed by Haworth (1963:181-184; reprinted with CAT manual in 1990), allowed objective, quantifiable data, which compared favourably with the CPQ protocols.

MR-imaging studies

- The following clinical features of cranial synostosis were identified: hypertelorism, exophthalmos, parrot-beaked nose, short upper lip, hypoplastic maxilla, and a relative mandibular prognathism, in keeping with features first published by Crouzon in 1912.
- Shallow orbits, proptosis, a pertinent misshapen cranium and ocular proptosis are distinctive features.
- Intracranial pressure may or may not manifest as hydrocephalus.
- No family history of Crouzon syndrome was revealed and there was no paternal age risk; therefore this case was ascribed to a spontaneous new mutation.
- A diffuse beaten copper pattern, erosion of the dorsum sella, and suture diastases were noted, indicative of preoperative intracranial pressure.
- Although some cranial radiographic and computed tomographic findings do correlate with elevated intracranial pressure, the sensitivity of radiological methods for detecting elevated intracranial pressure is universally low and they are not recommended to screen for elevated intracranial pressure in children with craniosynostosis, nor can these methods be employed to reliably screen for optic nerve damage.
- The MR-imaging results confirmed brain injury.
- It was established that the ultimate effect of a brain injury depends on the behaviour affected, the extent and location of the damage, and the precise age at which the injury occurs.
- With respect to cognitive function in humans, speech usually survives early brain damage, but some elements of syntax and some non-language functions may not survive, and general intellectual ability also tend to decline.
- It was found that reasonable recovery of language functions can be expected with brain injury between the ages of one and five years, but non-language functions were found to be impaired, coupled with inhibition of general intelligence.

- Lesions incurred before the age of one year tend to produce disproportionately greater impairments than do those incurred later.
- Lesions incurred between the ages of one and five years are followed by some reorganization of brain function, including rescuing of language functions.
- Lesions incurred after the age of five years permit little or no sparing of function.
- The following post-injury impairments were found: disorders of writing and calculation tend to be most prominent, followed by less prominent disorders of speech. Language usually survives after early left-hemisphere injury.
- Much of this survival seems attributable to appropriation of a potential language zone in the right hemisphere.
- This shift of language location has a price: specifically, some kinds of visuospatial orientation are impaired.
- Early lesions of the right hemisphere produce deficits similar to those produced by such lesions in adulthood, i.e. these functional impairments tend to be persistent.
- If language functions are reorganized to the right hemisphere, the right hemisphere's usual functions are sacrificed to accommodate the shift. Right-hemisphere functions do not shift sufficiently to interfere with language.

Areas of compromised affect and learning stemming from temporal lobe lesions

The data demonstrated the following:

- Disturbance of auditory sensation: probable lesioned sites are Brodmann's areas 41, 42, 22.
- Disturbance of selection of visual and auditory input: probable lesioned sites are Brodmann's areas TE and the superior temporal sulcus.
- Disorders of visual perception: probable lesioned sites are Brodmann's areas TE, superior temporal sulcus, amygdala.
- Disorders of auditory perception: probable lesioned sites are Brodmann's areas 41, 42, 22.
- Disorders of music perception: probable lesioned site is the superior temporal gyrus.
- Impaired organization and categorization of material: probable lesioned sites are Brodmann's areas TE and the superior temporal sulcus.
- Poor contextual use: probable lesioned site is Brodmann's area TE.

- Disturbance of language comprehension: probable lesioned site is Brodmann's area 22 (left).
- Poor long-term memory: probable lesioned sites are Brodmann's areas TE, TF, TH and 28.
- Changes in personality and affect: probable lesioned site is Brodmann's area TE plus the amygdala.

Areas of compromised affect and learning stemming from parietal lobe lesions

The data demonstrated the following:

- Disorders of tactile perception and changes in somatosensory thresholds, with impaired position sense: probable lesioned sites are Brodmann's areas 1, 2, and 3.
- Astereognosis: probable lesioned sites are Brodmann's areas 1, 2, 3.
- Simultaneous extinction: probable lesioned sites are Brodmann's areas PE (right) and PF (right).
- Afferent paresis: probable lesioned site is Brodmann's Postcentral gyrus.
- Tactile agnosia: probable lesioned site is Brodmann's area PE.
- Defects in eye movement: probable lesioned sites are Brodmann's areas PE and PF.
- Poor manipulation of objects: probable lesioned sites are Brodmann's areas PF and PG.
- Apraxia: probable lesioned sites are Brodmann's areas PF and PG (left).
- Constructional apraxia: probable lesioned site is Brodmann's area PG.
- Acalculia: probable lesioned sites are Brodmann's areas PG and the superior temporal sulcus (STS).
- Impaired cross-modal matching: probable lesioned sites are Brodmann's areas PG and the superior temporal sulcus (STS).
- Impaired object recognition: probable lesioned site is Brodmann's area PG (right).
- Disorders of body image: probable lesioned site is Brodmann's area PE.
- Right-left confusion: probable lesioned sites are Brodmann's areas PF and PG.
- Disorders of spatial ability: probable lesioned sites are Brodmann's areas PE and PG.
- Disorders of drawing: probable lesioned site is Brodmann's area PG.

Areas of compromised affect and learning stemming from frontal lobe lesions

The data demonstrated the following regarding disturbances of motor function:

- Loss of fine movements: probable lesioned site is Brodmann's area 4.
- Loss of strength: probable lesioned sites are Brodmann's areas 4 and 6, as well as the dorsolateral region.
- Poor movement programming: probable lesioned sites are Brodmann's premotor and dorsolateral regions.
- Poor voluntary eye gaze: probable lesioned sites are Brodmann's frontal eye fields.
- Poor corollary discharge: probable lesioned sites are Brodmann's dorsolateral and premotor regions.

The data demonstrated the following regarding disturbances of divergent thinking:

- Poor strategy formation: probable lesioned site is Brodmann's dorsolateral region.
- Poor frequency estimate: probable lesioned site is Brodmann's dorsolateral region.

The data demonstrated the following regarding disturbances of environmental control of behaviour:

- Poor response inhibition: probable lesioned site is Brodmann's prefrontal region.
- Impaired associative learning: probable lesioned site is Brodmann's dorsolateral region.
- Risk taking and rule breaking: probable lesioned site is Brodmann's prefrontal region.
- Self-regulatory disorder: probable lesioned site is Brodmann's orbital region.
- Impaired social behaviour: probable lesioned sites are Brodmann's orbital and dorsolateral regions.

The data demonstrated the following regarding disturbances of temporal memory:

- Poor working memory: probable lesioned site is Brodmann's dorsolateral region.
- Poor delayed response: probable lesioned site is Brodmann's dorsolateral region.

5.4 CONCLUSIONS

5.4.1 Conclusions pertaining to compromised learning linked to temporal lobe functioning

- Impaired cross-modal matching will result in difficulty to apply visually presented material, such as in dictation, to reading, writing, arithmetic and oral class work. There might be difficulty to follow speech on television, to follow oral instructions that have to be applied to written work in the classroom.
- Teaching styles that rely heavily on audio-visual presentations might not benefit a learner with cross-modal impairments.
- Inability to organize material result in compromised language applications (written and oral work) and memory.
- Inability to use contextual information will hamper mastering of comprehension tests, recall of information on the basis of a single contextual clue, and responding appropriately to instructions.

5.4.2 Conclusions pertaining to compromised affect linked to temporal lobe functioning

- There is an inability to use contextual cues in social and interpersonal relations.
- Social interaction might be compromised.
- Temporal-lobe lesions are associated with high levels of fear, anxiety, depression, egocentricity and perseveration in discussions of personal significance, as well as with proneness to aggressive outbursts.

5.4.3 Conclusions pertaining to compromised learning linked to parietal lobe functioning

- Parietal-lobe lesioning will affect almost all areas of classroom learning.
- The learner’s ability to apply spatial information will be compromised.

- Because mathematics and arithmetic have a quasi-spatial nature involving abstract symbols, lesioning of the parietal lobe will result in acalculia, dyscalculia, or in Gertsmann's syndrome.
- Learning in all its modalities might be further compromised due to impaired tactile form recognition, impaired perception and processing of visual stimuli, compromised spatial relations, and language application.
- Language has many of the same quasi-spatial demands as arithmetic; therefore language application might be significantly compromised.

5.4.4 Conclusions pertaining to compromised affect linked to parietal lobe functioning

- Due to tactile imperception the learner might violate others' personal space in a variety of ways and context.
- Due to misreaching for objects, overall lethargy and clumsiness the learner might often experience social embarrassment.
- Anxiety, feelings of insecurity and fear of the dark might be significant.
- Compromised body image might result in negative self-esteem and self-doubt, coupled with little courage to venture.

5.4.5 Conclusions pertaining to compromised learning linked to frontal lobe functioning

- Due to the frontal lobe's executive function, almost all areas of learning will be affected.
- Tactlessness, disinhibition, moria (silliness), coarseness, perseveration, social imperception and decreased attentiveness might surface in the classroom and might impact on overall classroom behaviour and learning.
- Explicit-declarative memory (especially semantic memory) might be compromised.
- The learner might show poor error detection and restoration, consequently the learner might not recognize and correct own mistakes, e.g., written work as well as oral work.
- The ability to work independently in class, to recognize and correct own mistakes, and to apply metacognitive strategies to learning might be significantly impaired.

- The learner might present with an inability to plan, initiate and terminate actions, to think in conceptual terms, to adapt to changing circumstances and to respond in socially appropriate ways. Classroom behaviour might therefore be characterized by marked dependent behaviour, impulsivity, poor decision-making, and sometimes even perseveration on the same task.
- Affective over- or under reactivity to the learning matter and the learning task might be prevalent.

5.4.6 Conclusions pertaining to compromised affect linked to frontal lobe functioning

- Social interaction might be compromised; consequently the learner might not develop metacognitive knowledge from interactions with peers and adults.
- The learner might not know why, when, and how self-monitoring strategies should be used.
- Behaviour difficulties such as poor planning, impulsiveness, and perseveration (on feelings or manual tasks) might be prevalent.
- Conceptual thinking might be restricted.
- The learner will experience an inability to adapt to changing circumstances and contexts, not being able to respond to these changes in socially appropriate ways.
- Difficulty to monitor own behaviour, social imperception, and an inability to assimilate change might inhibit social adjustment.
- The learner might not be able to evaluate social situations, apply information to other situations in a socially acceptable way, detect and restore mistakes, and regulate emotional behaviour.
- The learner's emotions might be erratic and unpredictable.
- Due to inhibited anterior cingulate gyrus and orbitofrontal cortex functioning, attention and problem solving will be impaired.
- Feelings of insecurity and dependency might prevail and extend into adolescence and young adulthood.

5.5 RECOMMENDATIONS

5.5.1 Recommendations regarding teaching and classroom practice

- Teaching styles should be adjusted to also meet the needs of this highly specific group of learners in terms of their compromised learning and affect. These learners may benefit from closer teacher supervision, more frequent positive reinforcement, shorter assignments and seating near the teacher, etc.
- Parent-teacher meetings should focus on empowering parents towards adequate learning support at home – academically, socially and affectively.
- Academic programmes should be adjusted to focus on skills training in order to prepare these individuals to be able to sustain themselves within the adult world of work and everyday living.
- Because these children's affective responses are erratic, disciplinary practices should be aimed at channelling behaviour into more favourable directions, instead of on punishment per se.
- Group projects should be carefully considered so that these children could also feel invited, appreciated and willing to contribute towards a group effort.
- As these children may find it difficult to adapt to change, an attempt should be made to provide a structured classroom environment. Having a routine at school will assist in providing a feeling of security.

5.5.2 Recommendations regarding parent guidance

- A Crouzon syndrome interest group needs to be established. Considering the limited number of diagnosed cases, such an interest group could be headed by an Educational Psychologist, and administered by means of a website and e-mails.
- Parents need to understand the existence of compromised affect and learning associated with Crouzon syndrome and have realistic expectations.
- Focus group discussions at schools for neurally impaired learners should be implemented in order to support the parents towards discipline, social support, etc.

- Structure and routine plays a large part in providing these children with boundaries. They function on a concrete level of thinking and thus one should work with them using behaviour modification strategies, such as penalties and positive reinforcement.

5.5.3 Recommendations regarding the Educational Psychological Practice

- A widespread acceptance and acknowledgement of the value of neuropsychological assessment should be encouraged in schools.
- The basic training of Educational Psychologists should include neuropsychological assessment, diagnosis and treatment.
- The findings and conclusions of this research project once again accentuated the need for multiprofessional collaboration. Students in Educational Psychology should be trained to collaborate meaningfully towards such projects.
- Our role in providing effective learning support and early intervention is crucial. By having a working knowledge of neuropsychology, one can make certain inferences that will lead to early diagnosis and appropriate intervention.

5.5.4 Recommendations regarding further research

- The development of a clinical neuropsychological test battery tailored for Educational Psychological use should be considered. Although there are various tests available, the majority of these tests are not standardized for South African use.
- The findings and conclusions are based on a single case study. It is therefore recommended that a similar research project be conducted with a larger sample population.

5.6 CLOSING

Because this research project was of limited scope, and based on a single case study, findings and conclusions should not be extrapolated to all learners diagnosed with a neural impairment.

However, doing this research has been an enriching experience, and I could only hope that the knowledge I have gained may contribute to a better understanding of how affect and learning are compromised as a result of the onset of Crouzon syndrome. Working with the research participant and her parents made me realize that research is about searching for meaning and understanding. It is about giving people possible answers to their questions.

In conclusion, I believe I have succeeded in compiling an exploratory profile of compromised affect and learning associated with Crouzon Syndrome by linking specific brain circuitry to learning and affect. This was done in an attempt to provide teachers, parents and psychologists with guidelines and understanding in terms of possible interventions and support strategies. I feel that this research study definitely contributes to the existing body of research and I hope that it encourages further research in this field.

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ANNEXURE A

Request for informed consent

04/04/2006

Dear [redacted]

I am a Masters student at the University of Pretoria and doing my internship at [redacted], where your daughter is currently in Grade 3. As part of my training, I'm also expected to conduct a research project. The focus of my research project is a case study into Crouzon Syndrome. For this purpose I am kindly requesting your consent to include your daughter as a participant in the research project. The working title of my research is, ***"Exploring learning and emotional difficulties associated with Crouzon Syndrome - a case study"***.

Data collection will comprise of a psychometric test battery, which includes administration of an IQ scale, reading and mathematics assessment, assessment of visual and auditory processing, assessment of neurological integrative functioning, and assessment of the child's emotional status. These assessment media will be discussed with you during an interview, and prior to commencement of the research project. Relevant clinical reports, i.e., previous neurological, occupational and speech therapy reports are thought to contribute to the value of the research project. Subject to your consent, these reports, if available, might be retrieved and included in my research report. My request for informed consent thus includes (a) your child's participation in the project, (b) assessment of your child, (c) the inclusion of information from related clinical reports, and (d) a parent interview on the child's prenatal and postnatal history, early development, and current functioning. Should the need for additional assessments arise, these will be treated on an ad hoc basis, subject to your prior consent.

The assessment will take place during the April school holidays, and the facilities of the University of Pretoria at Groenkloof Campus will be used for this purpose. Assessment might stretch over two days, pending your personal schedule.

The following ethical principles will apply:

- Participation is voluntary.
- You and your daughter are free to withdraw from the project at any stage if any party wishes to do so.
- No participating party will be placed at risk or harm of any kind.
- All information provided by you and your daughter will be treated confidentially and anonymously.
- No monetary compensation is involved in participation.
- As soon as the project is concluded, the research findings will be communicated to you and your daughter during an interview.
- The research findings might be used to plan an individualized treatment plan for your daughter.
- The research findings might be published in an accredited research journal, but confidentiality and anonymity will be honoured.

If you are willing to assist me, kindly complete the attached letter of consent and return it to me. If you have any further questions before consenting to this project, please don't hesitate to contact me.

Kind regards,

Björn Opper
Intern Psychologist
New Hope School

Phone number: 072 6239556
E-mail address: bjorn_opper@yahoo.com

Informed consent

Having read the attached request for informed consent, I declare that I am fully aware of the nature and purpose of the study conducted by Björn Opper. I understand that all information will be treated anonymously and as strictly confidential. I further understand that all ethical considerations as outlined in the request for consent will be adhered to.

I hereby consent to participation in the following: (a) my daughter's participation to the project, (b) assessment of my daughter, (c) the retrieval and inclusion of information from related clinical reports, and (d) a parent interview on my daughter's prenatal and postnatal history, early development, and current functioning. I also consent to the publication of the research findings, subject to anonymity and confidentiality.

Signature: ..... Date: .....

ANNEXURE B

DESCRIPTION OF STANDARDIZED AND NON-STANDARDIZED TESTS

1.1 The Senior South African Individual Scale – Revised (SSAIS-R)

Aim

The SSAIS-R is used to obtain a differential image of certain cognitive abilities. Firstly, the level of general intelligence is determined, for instance to predict scholastic achievement. Secondly, relative strengths and weaknesses in certain important facets of intelligence are measured to obtain diagnostic and prognostic information (Van Eden, 1992:3).

Rationale

The point of departure in constructing the SSAIS-R was that intelligence is a composite of related mental abilities which, in combination, represent a general intelligence factor.

The subtests of the SSAIS-R cover a wide field and involve a variety of aspects of behaviour that are representative of intelligence. This makes it possible to calculate a general intelligence score, as well as an analysis of the performance on the different subtests.

The total score for the subtests of the intelligence scale represents the broader, underlying factor of general intelligence, which in turn represents intellectual ability that continuously develops under specific environmental influences.

Description

The SSAIS-R consists of nine subtests involving verbal and non-verbal tasks, based upon the item content. Scores are provided for a Verbal, a Non-verbal and a Full Scale, while the skills measured by the different subtests include learning ability, general knowledge, judgement, concentration, spatial perception, basic perceptual and concept-forming abilities and visual motor skills.

The *Verbal scale* consists of the following subtests: Vocabulary, Comprehension, Similarities, Number Problems, Story Memory, and Memory for Digits (optional). The *Non-verbal scale* consists of the following subtests: Pattern completion, Block Designs, Missing Parts, Form Board, and Coding (optional).

A fact that is often overlooked when dealing with the SSAIS-R is that cognitive functioning (intellectual functioning) forms an intricate part of the *personality structure* as a whole, therefore these intricate parts can never be separated or dealt with in isolation.

1.2 The Quick Neurological Screening Test – Revised Edition (QNST-II)

Aim

The Quick Neurological Screening Test- Revised Edition aims at identifying persons as young as five years old, who experience learning difficulties. It is thus a quick and reliable way to identify possible neurological interference in learning.

Rationale

The Quick Neurological Screening Test – Revised Edition is based on research findings concerning the soft neurological signs that may accompany learning disabilities. This instrument thus alerts special education professionals to physical problems (in dexterity, visual tracking, spatial orientation, tactile perceptual abilities, and motor skills) that often co-occur with learning disabilities.

Description

The Quick Neurological Screening Test – Revised Edition consists of a series of 15 observed tasks that might assist in identifying children and adults with below par neurological integration. It requires the examinee to perform a series of motor tasks adapted from neurological paediatric examinations and from neuropsychological and developmental scales. These non-threatening tasks sample maturity of motor development, skill in controlling large and small muscles, motor planning and sequencing, sense of rate and rhythm, spatial organization, visual and auditory perceptual skills, balance and cerebella-vestibular function, and attention disorders. Scores are easily recorded during test administration (Mutti, Sterling & Spalding, 1998:7). The QNST-II is thus an excellent way to screen individuals for suspected learning disabilities. In addition, the QNST might be used to verify attention span, distractibility, impulsivity, non-verbal concept formation, including perceptual organization, spatial visualization and orientation, and visual-motor integration.

1.3 The Wisconsin Card Sorting Test (WCST)

Aim

While the Wisconsin Card Sorting Test was originally developed to assess abstract reasoning ability, in recent times it has been increasingly employed as a clinical neuropsychological instrument. Much of its current popularity among clinicians stems from its reported specific sensitivity to brain dysfunction affecting the frontal lobes (Heaton, Chelune, Talley, Kay & Curtiss, 1993:1). Thus the WCST can be used as a measure of executive function among school-age children.

Rationale

The Wisconsin Card Sorting Test requires strategic planning, organised searching, utilizing environmental feedback to shift cognitive sets, directing behaviour toward achieving a goal, and modulating impulsive responding. Therefore, with the use of the WCST the effects of frontal lobe lesions on executive functioning can be explored.

Description

The WCST can be considered a measure of 'executive function', requiring the ability to develop and maintain an appropriate problem-solving strategy across changing stimulus conditions in order to achieve a future goal. It consists of four stimulus cards and 128 response cards that depict figures of varying forms, colours and numbers of figures.

1.4 The Neale Analysis of Reading Ability – 2nd Revised Edition (NARA-II)

Aim

The Neale Analysis of Reading Ability consists of a set of graded reading passages for assessing rate of reading, reading accuracy and comprehension of oral reading. It can be used to assess reading progress objectively, and to provide structured diagnostic observations of an individual's reading behaviour.

Rationale

In testing accuracy, the Neale Analysis of reading Ability classifies all miscues and unsuccessful attempts as errors. It allows each type of error to be identified and counted, which gives valuable information about the reading strategies that an individual employs. The Neale Analysis of reading Ability can thus be employed to analyse an individual's phonic awareness and the application of such knowledge, graphic knowledge, word recognition, grammatical knowledge and contextual understanding of the reading matter. Furthermore, the comprehension questions give evidence of the individual's understanding of each narrative, concentrating mainly upon literal understanding and straightforward inference and deduction (Neale, 1997:48).

Description

The Neale Analysis of Reading Ability – Revised can be used with children aged 6 to 12 years and provides reading ages and standardized scores.

1.5 Children's Personality Questionnaire (CPQ)

Aim

The Children's Personality Questionnaire aims at screening with a view to providing individual attention and guidance to those children who need help with emotional and/or behavioural problems, or children whose unusual temperamental sensitivity indicates the need for careful handling (Du Toit & Madge, 19881). Scores are scaled on a continuum ranging from 1 to 10, but no particular positive or negative meanings can be attached to specific low sten scores (1 to 4) or high sten scores (7 to 10), since all of these have different meanings. Sten scores of 5 and 6 usually indicate sound adjustment, but different factors combine in specific ways to reflect a quite different meaning.

The CPQ scales measure temperament – a person's characteristic style of thinking, perceiving, and acting over a relatively long period of time and in a wide range of different situations. These personality traits are manifested in a set of attitudes, preferences, social and emotional reactions, and habits. In short, the CPQ scales measure the following:

Factor A: Affectothymia versus Sizothymia: The warm-cool social orientation
Factor B: The ability to discern relationships (Intelligence)
Factor C: Adaptation to the environment
Factor D: Phlegmatic temperament versus irritability
Factor E: Submissiveness versus control in human relations
Factor F: Sombre (serious) versus exuberant orientation
Factor G: The content and action of moral values
Factor H: Timidity versus courage in human temperament
Factor I: Thinking versus feeling – contrasting modes of evaluating experience
Factor J: Adventurous and sociable versus emotionally cautious orientation
Factor N: Self-presentation in social situations
Factor O: Self-confidence and resilience versus guilt proneness
Factor Q3: Investment in maintaining a socially approved self-image
Factor Q4: Relaxed verus tense temperament

Rationale

The CPQ is used to assess the preceding fourteen main, relatively independent dimensions (factors) of the personality structure, and which dimensions result in individual differences among children. Each factor is represented on a bipolar continuum, and results are expressed as sten scores. Specific low and high sten scores have no specific meaning, as different factors combine in unique ways to reflect a specific individual's personality functioning (Du Toit & Madge, 1988:2).

Description

The CPQ is a standardized pencil-and-paper measure of personality, based on the psychoanalytic theory adapted by Cattell, and originally developed by the Institute for Personality Testing, Illinois, USA. The CPQ was adapted and standardized for administration with South African and Namibian children between the ages eight and thirteen years (Du Toit & Madge, 1988:2). The norm population (as sample population) consisted of 3 805 children within this age range. Measures of reliability and validity were reported to be satisfactory, in keeping with measures for the American equivalent of the CPQ, and within an acceptable range for personality tests that are based on factor analysis, instead of on homogeneity (Du Toit & Madge, 1988:11).

1.6 The Children's Apperception Test (CAT)

Aim

In constructing the CAT, the aim was to depict scenes that would elicit material relevant to important situations and problems in the child's life (feeding, rivalry, aggression, loneliness, interactions with parental figures, etc.). The effectiveness of these cards in identifying important problem areas was enhanced through the use of background props, i.e. human settings for most of the ten cards. The CAT might be used as a projective technique with children between the ages three and ten years of age. However, this age range is not fixed, and should cognitive impairment be suspected, the CAT might be administered with older children at lower levels of mental and social functioning (Bellak & Hurvich, 1990:1).

Rationale

Bellak advocates the use of his analysis blank, which is a check-sheet for scoring eleven important variables. These variables, used for interpretive purposes are: main theme; main hero; qualities of figures; identification; external circumstances; objects and figures omitted; nature of anxieties; punishment for crime; significant conflicts; outcomes, and maturational level. One analysis blank is filled in for each story. A formal scoring system is not used, but the analysis blanks serve as basic data for interpretation (Crumbaugh, 1990:55).

Owing to their unsophisticated verbal skills, children often fail to give clear-cut and cohesive stories. Children also use symbols idiosyncratically and give very clear wish-fulfilment themes. These qualities of children's stories are readily found in test protocols and should not be given interpretative significance (Crumbaugh, 1990:55).

Bellak's interpretative approach is framed in psychoanalytical theory. He emphasizes the thematic pull of each card and the child's response to these themes. For example, Card 1 is assumed to encourage themes related to the oral phase of development, such as feeding problems and relationship with mother. The child's protocol is analyzed in order to learn about his or her structure, defences, and ways of reacting to or handling the problems of growth expected to occur under a psychoanalytical model. In his books, Bellak provides numerous interpretative examples (Crumbaugh, 1990:56).

Description

This CAT employs an original projective technique for presenting children with situations of concern. For the purposes of this research project, the CAT – Animal version was selected. This version of the CAT consists of ten picture cards of animals within a variety of social contexts through which the child becomes involved in conflicts, identities, roles, family structures, etc. (Crumbaugh, 1990:53).

1.7 The Draw-a-Person (D-A-P) Technique

Aim

Florence Goodenough (1926) was the first to make effective use of the draw-a-person technique. She initially used it as a measure of children's intelligence (basing conclusions on the number of details drawn), but it became apparent that it could be more accurately related to personality traits (Crumbaugh, 1990:161). Since then the draw-a-person technique has gained popularity as a graphic projection technique to assess personality.

Rationale

'It is generally held that the human figure drawing is a reflection of the self-image and body concept' (Crumbaugh, 1990:162). Thus, self-conflicts and conflicts with the social environment might be revealed by the draw-a-person technique.

Description

Projective drawings are not ordinarily quantified, though some examiners have attempted to interpret the number of details drawn, and others have measured overall size, line depth, size of the various elements of the figure (feet or hands, for example), and other details. Most clinicians simply 'eye-ball' the drawings and make rather intuitive ('ideographic') and holistic interpretations, but they also have their own empirical concepts of the meaning and weight of all major details (Crumbaugh, 1990:162).

The only accurate way to interpret projective drawings, as in the case of all other indicators of personality, is to integrate the 'signs' in a holistic fashion and to offer meaning to each sign according to its relationship to that totality. An accurate interpretation of projective drawings requires clinical experience, and a thorough clinical knowledge of the examinee serves to confirm or deny these interpretations (Crumbaugh, 1990:163). On the basis of longitudinal studies, various researchers have published indicators of disturbed affect and certain personality traits that are currently widely used by clinicians to interpret projective drawings, e.g. Machover (1949), Murstein (1965), Hammer (1975), Holzberg and Wexler (1950), Burns and Kaufman (1970; 1972), Crumbaugh (1980; 1990) and Woody (1980).

1.8 The Kinetic Family Drawing (K-F-D) Test

Aim

Burns and Kaufman (1970; 1972) modified the D-A-P technique to include (a) the addition of instructions to draw each family member *doing* something, and (b) the creation of an elaborate system of scoring and interpretation. The family drawing can be viewed as an unstructured projective technique that may reveal the child's feelings in relation to those he regards as most important and whose formative influence is most powerful. It thus aims to provide valuable expressions of feelings and transactional patterns within the family. The emphasis on action in the drawings greatly enriches the psychodynamic findings (Crumbaugh, 1990:173).

Rationale

The K-F-D should be interpreted in the light of the family background, age, sex, intellectual level, and current behavioural status of the child at home and at school, as well as in conjunction with other projective data. It is thus less effective when it is interpreted in isolation.

Description

Administration is the same as for the D-A-P, except that instead of being asked to draw a person, the examinee is instructed to draw each family member *doing* something, and to label (or otherwise indicate) which family member each figure represents. There is little objective scoring, and the interpretation parallels that of the D-A-P, with the addition of the dynamics of the family relationships (Crumbaugh, 1990:172). These relationships, especially in the case of children, may often be the key to the psychodynamics of the examinee. Careful use of post-drawing inquiry is likely to reveal the basic constellations of conflict among family members. For example, the absence of a family member such as a sibling in the drawing immediately suggests (when the examiner has already taken basic demographic data to establish the existence of the sibling) a conflict with this member. 'The relative size of the drawings is an obvious tip-off to the feelings toward each family

member. Absence of the subject's own representation may indicate feelings of alienation and withdrawal from the family' (Crumbaugh, 1990:172).

The references for the above work can be found in Chapter 3.

ANNEXURE C

TRANSCRIPTION OF VERBATIM RESPONSES TO THE CAT CARDS

Card 1: Chicks seated around a table

Verbatim response: *The chicken is eating the food. They are happy. They have three chickens and there is another chicken there. That's the chicken's babies and that is the mom. The chickens are looking at each other. They are feeling very happy that they have food. And they lived happily ever after.*

Card 2: Bears pulling a rope

Verbatim response: *They are wolves. There are two bears and a third bear. Like Goldie Locks. They pulling the rope, they fighting over the rope. The baby says, "I want the rope" the other one said, "I want it", the other one said, "Let go of the rope, I'm taking it". Then the baby had a good idea, the baby bear had a good idea. They cut three strings of the rope. Then they could all have the rope themselves. And they lived happily ever after.*

Card 3: A lion with a pipe

Verbatim response: *The lion is sitting down. He is a boy lion and he has a mane. And the lion is sitting down on the chair and he is smoking. And then this rat comes, the lion wants to kill that rat and eat it. And then he said, "I want to eat you rat". And then the rat said, "You are a lion and I am too little for you to eat". "Ah yea", said the lion, "I am going to eat you all up. If you don't do something for me I won't eat… if you do something for me I won't eat you up". And then the rat said, "I will do something for you". And then the rat gave the lion cake. And then the lion said "Thank you". And then there was a very tall cat that wanted to eat the mouse. The lion roared and the cat and the cat ran away. And then they said, the lion said, "Thanks for helping me", and the rat said "thank you for helping, not eating me". The end.*

Card 4: Kangaroos

Verbatim response: *The baby Kangaroos… The mom kangaroo wanted some picnic, a picnic basket and she took her babies to go to the market with her for things.. And the and the the other baby said, "Mom, do we have to go to the park? It is fun here said the kangaroo". Then they went to the park. "Off to the park" said the kangaroo". They were riding on the bicycle, then they heard a gun shot. Then they ran, they were very scared, and then they, that's all. The mom got shot… I don't like sad stories. The mom died, and then the babies had to suffer and they did not know what to do. And there dad also died from a gunshot… Then the kangaroos ran up to the… They thought in their hearts, deep inside, "What are they going to do?" Along came a friend and he says, "What is wrong with you two babies? Are you lost? Are you hungry? "Yes" said the babies. "My mom got shot and we don't know what to do. "Why don't you come live with me and they played with each other and lived happily ever after with their friend. That's all.*

Card 5: A darkened room with two baby bears

Verbatim response: *The two babies are sleeping in the dark night. No one lived with, no one lived there by the fox. The foxes were asleep. And when the mom came.. and they came back.. the mom came back from shopping. And then, the babies thought that their mom was going to leave them there by the house that they can play. Then they all stayed at the house, they all played. Stuck in the bed and they fell asleep. The end. The mom came back and she fell asleep and then the others fell asleep and the dad also.*

Card 6: A cave with two bears

Verbatim response: *A bear.. One day a big bear, three big bears were lying down in a cave and there were lots of rocks around them and the baby bear, the baby bear want to go play. The mommy and dad said, "you cant play now, there is too much stuff to do". What there was to do, there was just, um, washing to do. And then they played again, and they slept and the baby bear, while the mom and dad were sleeping, the baby bear went out the house to go and play. The end. And the baby got lost. The little bear wrote a letter, "please feed me I am hungry, my mom is gone, I lost her". Signed baby bear. And then he*

came to a big house and they knocked on the door and out came a ugly old witch and the, he gave the witch the letter. The witch said, "ok, you can come into the cottage", and then she said, "you can have tea" and the bear said, thought that the witch is not actually nice, the witch is trying to be nice but gets mean in the end. The bear felt that she was a nice girl, even though it was a witch. The parents are feeling very sad that the baby is gone, and then the dad said, we got to look for the baby then, that's an idea. Then the mom said, "you right, let's go look for her". And then they looked and looked and then the letter dropped, "please feed me, I am hungry" signed baby bear. The bear read it, and, and then they new it was the, the baby. Then they say, then there was another letter, "I am in a cottage down there by the nice girl". Then they came to the house and said, "what are you doing with our baby?" The baby was lost and hungry. There is actually three witches, then the other witch said, are you the, the parents of this baby bear said the three witches?, yes. And then the baby saw that, that's mine and I am going to eat them for dinner. Then the bear, thought it was a witch, and then the bear went out of the cottage, and then they went home and lived happily ever after with the baby.

Card 7: A tiger and a monkey

Verbatim response: *I love tigers. I have a toy tiger at home. A doll. The lion scares the monkey and the monkey ran up the tree and the lion said, "I want to eat you monkey". The monkey said, "you not eating me". Do you want a banana, said the… tiger. Then the monkey said, "I love bananas, can I have one?". Then he ate the banana and threw the banana peel on the floor. "Come and catch me" said the monkey. The lion ran and he slipped and he fell on the banana peel. Then the lion got angry and he wanted, and he roared at him. "That was just a joke" said the lion. That has worms in it. The banana had worms in it. And then he said, well I still like worms. Then he ate it. The end.*

Card 8: Monkeys sitting on a sofa

Verbatim response: *The monkeys were having coffee and tea. There was the daddy monkey, the aunty monkey and the baby monkey last of all. "Can I play?" said mom. "Can I play?" said the baby to the mom. "Yes you can". And then the aunty said, "there will be no shouting or screaming or running, or eating all the bananas in my fridge. And then the monkey said, "fine, I won't" then he ran up to the fridge and he took the banana. He didn't listen and feel what he was told and then he ate all the bananas. He didn't know it was for later for everybody. And then what happened was that the mom came up later to go and get the bananas. Then they saw the bananas were not there and then the monkey said, "why are you so fat monkey?" They new he had bananas in his stomach. Why did you eat the bananas out from the tree.. I mean from the fridge. The monkey said, "I was hungry and.." "Who told you so that you must not eat it" "Aunty, but she always shouts at me when I do that stuff". The parents said, "parents do shout at you when.." and the baby said, "I am sorry guys I wont do it again". Then he got bananas from all the parents and they said your sorry is accepted. And they lived happily ever after.*

Card 9: Darkened room seen through open door-rabbit looks through door

Verbatim response: *One day there was a rabbit in the bed, he lied down on the bed and almost fell asleep in the bed. And then the mom left the door open and forgot to close the door. And then the, the rabbit got scared and said "I can't sleep with the door open". Then the mom came up.. and she closed the door… that's all. The rabbit said goodnight mom and they all went back to sleep.*

Do you no what? It is almost my birthday soon.

Card 10: Baby dog lying across knees of adult dog

Verbatim response: *I've got a cousin she loves dogs, she won't stop moaning about a dog... Her name is Caitlin. She is a little bigger, than Caitlin in class. The dog didn't listen to the mom. The dog, the dog ran up to the fridge and he wanted a bone without the mom's permission and then he just grabbed the bone. Then he ran up to his mom with the bone and then he ate it. "Where did you get the bone?" " From the fridge" "then she*

gave the dog a smack. The dog was crying. Look…. And then, the dog said I am sorry and he wanted another one and said, "mom, can I please have another one. Then the mom got a bone for him. "Thank you mom" said the dog. That was all. He is feeling sad that his mom hit him. My mom hits me when I drink pills out of the fridge. It is just like this. You go eat, you go and eat this cake when your mom says no permission, and you go get the cake out of the fridge and eat it, then your mom will shout at you. That never happens to me, I listen to my mom.