CHAPTER 11

Medical Terms and Definitions

1. Introduction

As early as the mid-1980's, at a time when American courts have started to become aware of the importance of wrongful life litigation, there were at least between 60 and 90 foetal health impediments detectable before birth.[1] According to Bey-Berkson,[2] this very availability of accurate genetic testing procedures together with legal support provided by the landmark decision of **Roe v Wade**[3] could basically be earmarked as the two main events that brought about this novel form of litigation.[4]

In this section various genetic disorders commonly associated with wrongful life will be briefly discussed in order to inform the reader of this very intricate part of the whole debate. It is of paramount importance that the reality and consequential suffering caused by these diseases and aberrations should be thoroughly considered before a final decision concerning wrongful life is made.

The genetic terminology, procedures and processes mentioned in this chapter could assist the reader without any medical background, in understanding unfamiliar medical jargon associated with and encountered in wrongful life cases. Note, however, that only a very superficial reference to basic genetic concepts are made. As this is primarily a legal work, no attempt has been made in any way to give a complete account of foetal development and genetic testing. The sole purpose of this chapter is to familiarise the reader with some relevant medical aspects.

It is therefore submitted that, in a study where the legal viewpoint to challenges created by the advancement of medical technology and the science of genetics is discussed, it is permissible

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1 1988. Wrongful Life, Wrongful Birth: A Pathfinder for a Legal and Moral Dilemma. **Legal Reference Services Quarterly** (8:1), 63.

2 *ibid.*

3 in which case the American Supreme Court recognized a woman's right to an abortion on demand - see ch 3.

4 *ie* wrongful life litigation in general.

to consider the very basic foundation upon which the majority of the actions[5] are based.

> "Genetic screening should enable people to escape their fate by giving them the freedom to make an informed choice and adopt a chosen course of action which they regard as acceptable."[6]

2. Foetal development

2.1 Brief summary of gestational development[7]

Slabbert[8] gives a brief account of the normal human foetal development: Gestational development consists of various moments or phases. Shortly after insemination, the spermatozoon penetrates the woman's ovum, and if so, fertilisation will take place, a process of approximately twenty-four hours' duration in which the haploid chromosome sets of the respective gametes commingle to form a single celled embryo with a normal complement of forty-six chromosomes.

After more or less one week, implantation of the embryo may take place in the uterine wall. If the latter dos not happen, then no pregnancy occurs. By ten weeks, all human organs are present in rudimentary form and inchoate electrical activity can be detected in the embryo's brain cells. The foetal brain develops rapidly between the nineteenth and thirtieth week, with the cerebral cortex starting to mature both structurally and functionally at the twenty-second to the twenty-fourth week. The foetal lungs also become capable of respiration at this time. At this point the foetus can, with the assistance of medical technology, survive *ex utero*.

Natural birth usually occurs around the thirty-eighth week of pregnancy. The emerging infant is connected to the umbilical cord which, until parturition, remains its only source of oxygen.

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5 a large percentage of both wrongful birth and wrongful life actions originate because of the transmission of hereditary anomalies - see *infra* (for a discussion on hereditary diseases frequently found in these actions) and also ch 7 and 8.

6 Anon. 1994. Genetic screening. **Report: Health Council of the Netherlands: Committee Genetic Screening** (Publication no. 1994/22E) - see also ch 5, on lack of proper medical/ genetic information as basis for wrongful life litigation.

7 Sadler, T.W. (previous fn) discusses foetal growth and also provides illustrations thereof - aslo see end of ch.

8 1997. The fetus and embryo: Legal status and personhood. **Tydskrif vir die Suid-Afrikaanse Reg** (2), 238.

Only after the cord is cut, the infant starts breathing on its own.[9]

Sadler[10]reports that the age and level of foetal development is vitally important to the susceptibility to teratogenesis[11] and illustrates this occurrence graphically.[12] He[13] describes the occurrence of abnormalities with reference to the various developmental stages, namely abnormal zygotes,[14] abnormal blastocysts[15] and abnormal foetal growth.[16]

Shepherd[17] points out an interesting consideration with regard to the occurrence of birth defects as a result of environmental factors (teratogens) and reports that arguments have gone up stating that such outside influences break the causal link between the defendant-physician's negligence and the eventual loss suffered by the plaintiff.

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9 see ch 3 on the various theories of when a foetus attains personhood.

10 *op cit* p 123.

11 "Despite the rapid development of the field of teratology, our knowledge of congenital malformations in humans has increased relatively little. At present it is estimated that approximately 10 percent of all known human malformations are caused by **environmental factors** and another 10 percent by genetic and chromosomal factors; the remaining 80 percent are presumably caused by the intricate interplay of several genetic and environmental factors." *op cit* p 110.

12 see end of ch.

13 *op cit* p 31.

14 "16 percent of all oocytes coming in contact wit sperm are not cleaving, either because they are nog properly penetrated by sperm or the mitotic mechanism is not functioning. Another 15 percent are lost during the first week at cleavage and blastula stages. Since many abnormal zygotes are lost during the early stages of development, this process is often considered as a "self-cleaning" process, whereby abnormal embryos are eliminated without the mother being aware of it (spontaneous abortion)." *op cit* p 31.

15 "In selected fertile women under optimal conditions for pregnancy, 15 percent of oocytes fail to become fertilized and 10 to 15 percent start cleavage, but fail to implant. Of the 70 to 75 percent that implant, only 58 percent will survive until the second week and 16 percent of those will be abnormal. Hence, at the time when the first expected menstruation is missed, only 42 percent of the eggs exposed to sperm are surviving. Of this percentage, a number of cases will be aborted during subsequent weeks and a number will be abnormal at the time of birth." *op cit* p 46.

16 "Considerable variability exists in fetal length and weight and sometimes these values do not correspond with the calculated age of the fetus in months or weeks. Most factors influencing length and wight are genetically determined, but it is now known than environmental factors also play an important role. It is generally accepted that severe malnutrition as well as heavy smoking leads to reduced fetal growth. Similarly, placental insufficiency may cause severe growth retardation." *op cit* p 86.

17 1996. Sophie's choices: Medical and Legal responses to suffering. **Notre Dame Law Review** (72:1), 103.

"The Committee also notes that many diseases are multi-factorial in causation, meaning that environmental factors may interact with one family's set of genes but not with another's. Additionally, the various genes themselves may interact with each other, and this 'multiple gene action' is impossible to predict using a separate analysis of each single gene. "In such cases, definitive predictions will rarely, if ever, be possible, and it will be impossible to group individuals into two district categories - those at no (or very low) risk and those at high risk."[18]

3. Background on hereditary anomalies and foetal development

3.1 Concise summary of transfer of hereditary disease[19]

Genes are coded messages that instruct the growing body in how to develop and function. There are many thousands of different genes,[20] which come in matching pairs; one of each pare of genes comes in the egg from the mother, and the other comes in the sperm from the father. When an adult produces an egg or a sperm, one member of each pair of genes is copies into the gamete (the egg or sperm).

Genes are too small to be visible even under the microscope. However, genes are organized into physical structures called chromosomes, which are visible under the microscope. Each chromosome can be thought of as a string of beads, with each bead representing a gene. There are twenty-three pars of chromosomes in each cell in the body, with one of each pair being packaged into each egg or sperm.

Twenty-two of the chromosome pairs are the sex chromosomes, X and Y. A man has one X chromosome and one Y chromosome; a woman has two X chromosomes. The Y chromosome makes a man male, but does little else. Therefore if an X chromosome has a faulty gene on it, and if it is present in a male, that person is likely to show signs of the gene fault. Woman who carry a faulty gene on one of their two X chromosomes are much less likely to be affected by it, because they have a second (spare) copy of the gene on the other X chromosome that will usually mask it. The X chromosome carries many genes that are quite unrelated to sex, so that the sex-linked disorders that are much more common in males than females include some

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18 *op cit* p 113.

19 Clarke, A. 1994. **Genetic Counselling - Practice and Principles: Professional Ethics** Routledge, 23.

20 the genetic information consists of between 50 000 and 100 000 genes.

types of muscular dystrophy (including the severe Duchenne type)[21], haemophilia,[22] colour-blindness and many more. Such disorders may arise as new mutations in a gene, or females in the family may be unaffected carriers. In that case, affected males will have carrier mothers, and the males will be linked in the family tree through healthy women.

Faulty copies of genes are common - everyone has at least one such gene fault (mutant gene). Most such gene faults cause no problem,[23] because a single intact copy of most genes is enough to get by on without any difficulty. However, if a person inherits a faulty copy of the same gene from both parents, then that child will have no intact copy of the gene, and may have a genetic disease such as cystic fibrosis[24] or a haemoglobin disorder such as haemophilia, sickle-cell anaemia[25] or a thalassaemia.[26] Because faulty copies of such genes do not usually manifest problems, these gene defects are termed *recessive*. Parents must be carriers of the faulty gene, and other relatives may be; brothers and sisters may be also affected, or other relatives if a carrier in the family has a partner who is also a carrier.

Some genes are sufficiently important that both copies must be intact for the person to avoid a genetic disease. Such gene faults give rise to *dominant* gene defects, which can be transmitted from one generation to the next (to 50 per cent of the children on average).

Chromosomes may also be involved in genetic disease. First, an egg or sperm may be produced that contains the wrong number of chromosomes. Many such conceptions miscarry early pregnancy, but some survive to be born. *Down syndrome*[27] occurs when a child inherits three copies of chromosome 21 instead of the normal two. Anomalies of the sex chromosome are common, and in live-born infants they are not usually associated with major physical or developmental problems.

Another type of chromosomal problem arises when a rearrangement occurs - either with two small chromosomes joining together to form a single chromosome, or when two chromosomes

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21 see *infra*.

22 see *infra*.

23 while most mutations are quite harmless, some of them affect functional characteristics - if it arises in somatic cells, the mutation is not passed on to the offspring.

24 see *infra*.

25 see *infra*.

26 see *infra*.

27 see *infra*.

exchange segments. Such rearrangements may cause no immediate problem, but if a person carrying such a rearranged set of chromosomes has a child, the child may be at some risk of a serious developmental disorder. The *balanced chromosomal rearrangement* may cause no difficulty because all the genes are present, just arranged differently. However, there may be a risk of such a person handing on to his or her children an *unbalanced complement* of chromosomes, that contains an incorrect set of genes. The rearrangement may also be handed on in the balanced form, so that several members of a family may carry such a rearrangement before it is recognized as the cause of physical or developmental problems in a child.

3.2 Hereditary disorders in general[28]

Gene mutations are caused by molecular "errors" in DNA,[29] which are partly caused by environmental factors. Such "errors" arise during the cell division. The error may consist of the substitution of one or more building blocks (point mutation), loss of (part) of a gene (deletion) or of larger rearrangements such as insertions, duplications or the repetition of a given sequence of building blocks (repeat).

Dependent upon the moment when the disorder manifest itself, it is possible to discriminate between *congenital abnormalities* such as *spina bifida*,[30] harelip, club foot, *Down's syndrome* and *hereditary diseases* occurring later in life such as such as some forms of Alzheimer's disease,[31] Huntington's disease,[32] some cancers,[33] cardiovascular diseases and several psychiatric illnesses.

Genetic disorders can be classified in a number of different ways. One commonly used system is to distinguish between *chromosomal abnormalities* and *gene mutations*. This is based on the presence or absence of visible, morphological abnormalities of the chromosomes.

3.2.1 Chromosomal abnormalities

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28 Anon. 1994. *op cit* p 29.

29 "deoxyribonucleic acid": chemical compound whose structure is such that it is capable of storing genetic information.

30 see *infra*.

31 see *infra*.

32 see *infra*.

33 see *infra*.

Chromosomal abnormalities are taken to mean morphological abnormalities or chromosomes which can be seen with the aid of a light microscope. These features usually arise during the development of the sex cells or during the first few divisions of the fertilised egg cell. The older the pregnant woman, the greater the chance of a numerical chromosome abnormality occurring in the foetus. The chances of structural chromosome abnormalities are increased if one or both parents have been exposed to external influences such as radiation or cytostatic drugs. At least half of all spontaneous abortions are caused by chromosomal abnormalities in the foetus. The use of modern techniques has shown that, despite of this natural selection, there are chromosome abnormalities in 0.92% of live births.

3.2.2 Gene mutations

Gene mutations are variations in the structure of a gene, and they can give rise to hereditary disorders. An abnormality in a single gene which (partly) causes a disorder is described as a monogenetic abnormalities can be further classified into autosomal dominant, autosomal recessive and sex-linked disorders. The hereditary component for the development of the abnormality is determined by the interplay of various abnormal hereditary traits. Congenital abnormalities arising in this way occur in 2.5% to 4% of live births in the Netherlands, an annual total of 5000 - 8000 individuals.

Andrews[34] writes that the frightening fact about some of these diseases and conditions is that a seemingly healthy couple could be at relatively high risk of bearing a child with serious genetic anomaly, without even knowing it. This would be the case if both parents have a single gene recessive disorder. The adults themselves would suffer no ill results from hosting the singular recessive gene, but this condition would increase the risk of having a child affected by this disease to 25%.

3.3 Outline of hereditary anomalies

Eriksson *et al*[35] give a broad outline of hereditary anomalies. He reports that although monogenetic hereditary disorders occur seldom,[36] the vast number of different aberrations has the cumulative result of affecting approximately 1% of a total population.

It is reported that in the majority of cases, multi-factorial diseases affect about 1 out of every

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34 1992. Torts and the Double Helix: Malpractice Liability for Failure to Warn of Genetic Risks. **Houston Law Review** (29:1), 149.

35 1985. **Over erfelijkheid - aangeboren afwijkingen erfelijkheids voor lichting begeleiding.** Bosch & Kenning, 11.

36 1-3 out of every 1000 children born.

1000 people, examples of these being: lip, structural, heart, feet and back abnormalities, different forms of mental retardation as well as psychological illnesses. Certain of these diseases occur relatively frequently such as hereditary heart aberration in 1 out of every 200 and schizophrenia in 1 out of every 100. The various classes of hereditary abnormalities are summarised:[37]

- congenital,[38] but not hereditary[39]- for example infectious diseases contracted from the mother (AIDS);
- combination of hereditary diseases with environmental/ external factors - affecting 4% of all newborns with anomalies such as heart misformation;
- chromosome abnormalities - affecting ½ % of all newborns, 5% of all babies born dead, and 60% of all premature miscarriages;
- hereditary diseases with a Mendelian pattern - consisting of about 3500 variations affecting 1% of all newborns - these could be sub-classified into three categories: dominant (50% chance of repetition); recessive (25% chance of repetition); sex orientated (50% chance of repetition for male children where the mother is the carrier).

Sadler[40] gives interesting global figures on the prevalence of congenital malformations and states that a survey comprising of 20 million births showed that malformations occurred, according to birth certificates in 0,83%, according to hospital and clinical records in 1,26%, and according to intensive examinations by pediatricians in 4,5%of newborns. He writes that the incidence was the highest in the United States of America (8,76%) and the lowest in Germany (2,2%). He writes at the same place:

> "Summarizing, it is probable that 2 to 3 percent of all live-born infants show one or more significant congenital malformation at birth, and that at the end of 1 year this figure is doubled by discovery of malformations indiscernible at birth."

3.4 Genetic disease in developing countries

It is reported[41] that health standards, with a few exceptions, are lower in developing countries

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37 *op cit* p 14.

38 *"aangeboren".*

39 *"erfelijk".*

40 1985. **Langman's Medical Embryology** Williams & Wilkins (5th edition), 109.

41 Anon. 1999. **Report on Epidemiological transition- Genetic disorders and birth defects in developing countries.** Department of Pathology, University of Pretoria.

than the industrialised world. Eighty percent of the world's population live in the World's developing nations where 90% of the 140 million births in 1997 occurred.[42]

Compounding this situation is the AIDS pandemic which is currently ravaging many countries, particularly in Sub-Saharan Africa and South East Asia. In some instances it has already reduced life expectancy by more that a decade.[43]

In each country, the approach to the management of genetically determined disorders and birth defects will depend on local frequencies of the individual conditions, the health burden they represent, the resources available for their care and prevention, and the health care infrastructure.[44]

It is accepted that the prevalence of genetic disorders and birth defects, which are recognisable in 2-3% of all newborns, varies according to geographic, ethnic, socio-cultural and socio-economic characteristics of a population.[45] Factors that predispose to higher prevalences of these disorders in developing countries include:

- traditional consanguineous marriages resulting in a higher frequency of autosomal recessive conditions;
- advance parental age, resulting from continued child bearing into the upper end of the reproductive lifespan;[46]
- socio-economic factors. The increased risk of birth defects in families of low socio-economic status has long been recognized;[47]
- inadequate health care prior to and during pregnancy.[48]

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42 (UNICEF, 1999).

43 (World Bank, 1997).

44 (WHO Technical Report 865).

45 (ERMO Technical Publication 24, 1997, World Atlas of birth defects, 1998).

46 advanced maternal age (35 years and older) is associated with high frequencies of chromosomal abnormalities - also relevant is autosomal dominant mutations in men of advance paternal age (55 years and older).

47 at least in part this is due to inadequate pre and post conceptional nutrition, including deficient intakes of micronutrients (folic acid and other vitamins, iodine).

48 which can be predisposed to an increased frequency of congenital infections such as syphilis and *rubella*, or birth defects consequent to inadequate control of diabetes and the unsupervised intake of drugs and traditional medicines.

3.5 Genetic research terminology[49]

Allele: One of the various forms of a gene.

Amnion: Extra-embryonic membrane that lines the chorion and encloses the embryo-foetus in the so-called amniotic fluid.

Alphafoetoprotein: A protein which is produced by the foetus.

Blastocyst: A hollow ball of cells, filled with fluid, that forms about four days after fertilization and prior to the beginning of the process of implantation.

Carrier status: The presence within the genetic material of one mutated and one normal allele of a gene associated with a recessively inherited disease.

Chorion: Outermost cellular extra-embryonic membrane.

Chorionic villi: Finger-like projections growing from the external surface of the chorion that contribute to the formation of the placenta.

Chromosomes: Linear threads of DNA that transmit genetic information through genes spaced along their entire length.[50]

Cleavage (mitosis): The process whereby the cells divide and thereby multiply to become similar identical daughter cells during early embryo development.

Diploid: Having two sets of chromosomes, usually one paternal and one maternal, twice the haploid number (in humans 46).

DNA: Chemical compound whose structure is such that it is capable

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[49] Ford, N.M. 1988. **When did I begin? Conception of the human individual in history, philosophy and science.** Cambridge University Press, 210; Anon. 1994. *op cit* p 147.

[50] see *supra* - in the human somatic cell there are normally two sets of 23 chromosomes including the two (XX or XY) that determine the sex of the individual, each gamete normally contains only one set of 23 chromosomes.

of storing genetic information. Deoxyribonucleic acid, the primary constituent of chromosomes and the basis of the genetic code and inherited traits.

Embryo: Refers to the newly formed organism in its first stages of growth, these being the stages characterised by the multiplication and differentiation of the fertilised egg-cell after its implantation, until blood circulation is established between the new organism and the maternal body.[51]

Epiblast: Also called primitive or primary embryonic ectoderm. The non-endodermal part of the inner cell mass of the blastocyst.

Foetus: Refers to the embryo after two months' growth in the uterus, when the blood circulation is established and the general anatomy of the growing organism is formed. This stage of development thus follows the embryonic period and continues until birth or abortion. The transition from embryo to foetus occurs about eight weeks after fertilisation and seven weeks after implantation.[52]

Gamete: A mature reproductive cell, usually haploid eg. a sperm or ovum.

Gene: The portion of a DNA strand within a chromosome which contains the genetic information for a single trait.

Genome: The complete set of hereditary factors, as contained in the haploid assortment of chromosomes. Frequently used broadly to refer to the complete genetic material for any cell or organism.

Genotype: The hereditary or genetic constitution of an individual or of a cell, usually referring only to the nuclear material.

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51 Slabbert *op cit* p 234.

52 Slabbert, *op cit* p 236.

Heterozygote: The two alleles for a given gene (on both chromosomes where the gene is located) differ from one another.

Homozygote: The two alleles for a given gene (on both chromosomes where the gene is located) are identical to one another.

Meiosis: Division of a diploid nucleus into four nuclei, each with half the number of the chromosomes of the parent nucleus and with a mix of both maternal and paternal chromosome sets, resulting in both sperm and egg with 23 genetically unique chromosomes each.

Monogenetic: Associated with a single gene.

Mutation: An abnormality in the structure of a gene or chromosome,, or in the number of chromosomes, or the process by which such abnormalities arise.

Neonatal: The period shortly after birth (until a few weeks of age).

Oocyte: The immature female germ cell. It is called the *ovum* when it matures after the penetration of the sperm during fertilisation and the completion of the second meiotic division.

Prenatal diagnosis: Refers to a variety of medical techniques used to detect the presence or absence of a possible disease or defect in the foetus. Specific techniques of prenatal diagnosis include: amniocentesis, ultrasonography, fetoscopy, chorion villus biopsy and maternal serum alpha-fetoprotein screening.[53]

Prenatal: The time between the start of a pregnancy and birth.

Prenatal screening: Attempts to identify, either before or after conception, women who have a high risk of bearing an abnormal child.[54] Such women can be identified through the taking of a medical

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53 Anon. 1987. Wrongful Birth Actions: The Case against Legislative Curtailment. **Harvard Law Review** (100), 2021 - see *infra*.

54 *ibid.*

history during their initial visit to the doctor. Signs of increased risk include, for example, advanced maternal age, previous offspring with a chromosomal aberration, family history of birth defects, and exposure to teratogenic agents during pregnancy.

Primitive streak: A piling up of cells on the caudal end of the embryonic disc, providing the earliest evidence of the embryonic axis and the formation of the embryo proper.

Proembryo: The developing cells produced by the division of the zygote before the formulation of the embryo proper at the appearance of the primitive streak.

Zygote: The fertilised egg - the single cell that is formed when the two haploid sets of chromosomes in the pronuclei of the male and female gametes come together at syngamy. Also used loosely to refer to the early embryo during the first few weeks.

3.6 Status of pre-born

Strauss[55] refers to the decision of **S v Collop**[56] and states that in terms of this decision, there is legally no distinction between a zygote, an embryo or a foetus in South African law.

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55 1991. **Doctor, patient and the law** JL van Schaik (3rd edition).

56 1981 (1) SA 150 (A).

4. Detection techniques[57]

4.1 Background

Lupton[58] reports on the Human Genome Project, which is a 15 year effort to draw the first detailed map of every gene in a human's DNA composition. He writes that through this and other similar projects genetic engineers worldwide are decoding life's molecular secrets and trying to use that knowledge to reverse the natural course of disease.

> "This is however just the start of a process. The growing ability to manipulate genes could eventually change everything in our society - what we eat, what we wear, how we live, how we die and most important of all, how we procreate."[59]

Lupton[60] conveys with regard to the accuracy of genetic tests that "genetic diagnosis is complex and the vast amount of new information streaming in daily from genomic studies will undoubtedly lead to premature conclusions".

Eriksson *et al*[61] confirm that knowledge with regard to genetics and cell biology has increased vastly, which knowledge has made possible various new techniques to detect an increasing number of abnormalities at ever earlier stages. A number of the differing test available[62] are DNA testing, egg nuclei tests, blood tests, urine tests and also chromosomal testing.

4.2 Application of tests

Sadler[63] summarises the value and use of the various tests, which could also be effectively used in combination:

> "Several approaches are now available to the perinatologist for assessing the growth

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57 Anon. 1994. *op cit* p 33.

58 1996. Genetic engineering: The legal implications. **Tydskrif vir Suid-Afrikaanse Reg**, 56.

59 *ibid.*

60 1996. *op cit* p 62.

61 *op cit* p 15.

62 some of which is discussed below.

63 *op cit* p 87.

and development of the fetus in utero. In combination, these techniques are designed to detect malformations, chromosomal abnormalities, and overall growth of the fetus."

It is reported[64] that these techniques are not used on a routine basis,[65] but are generally reserved for so-called high risk pregnancies, such as:

- late maternal age (35 years and older);
- history of neural tube defects in the family;
- previous birth of a child with a chromosomal abnormality;
- chromosomal abnormalities in either of the parents; and
- mothers who are carriers of X-linked recessive disorders.

Andrews[66] distinguishes between various types or classes of anomalies that can be detected by modern genetic testing. He classifies them as:

- less serious abnormalities and impairments;[67]
- serious genetic diseases and conditions that are treatable after birth[68] and
- latent hereditary conditions and defects only manifesting much later in life.[69]

He[70] consequently asks whether a general legal duty[71] rests on medical practitioners who have patients that fall in any of these categories to urge them to undergo genetic testing. To what extent will they be held accountable and where does the limit lie concerning the various groups of patients and/or family members to be informed?[72]

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64 *ibid.*

65 except ultrasonography.

66 *op cit* p 590.

67 mild hereditary abnormalities, *eg* poor eyesight, slight deafness or a tendency towards heart problems *etc.*

68 *eg* the condition of phenylketonuria, which causes retardation in children if a special diet is not followed.

69 *eg* Huntington's disease.

70 Andrews, *ibid.*

71 to inform or to take responsibility for their patient's and their patient's children health and well-being.

72 see ch 5 on informed consent.

4.3 Various tests

4.3.1 Diagnosis test

The initial test carried out in the context of early detection is referred to as a "diagnosis test." This is usually a relatively simple test whose results indicate whether the test subject has a greater than normal chance of possessing the trait in question. If this is indeed the case, then further diagnostic work is carried out. However, the definitive diagnosis can still be girded with an error margin. This general model of diagnosis does not necessarily apply in all cases. Detection sometimes takes place by means of a combination of diagnostic tests which may or may not be carried out in sequence. Sometimes detection is achieved directly, by means of the definitive diagnostic test.

4.3.2 Chromosome testing

Major chromosome abnormalities[73] can be detected by examining chromosomes within cell nuclei, under a light microscope. The test requires either a small amount of blood or, if carried out during the prenatal phase, either chorionic villi[74] (tissue projecting from the placenta) or cells from a sample of amniotic fluid.[75] If the cells to be tested come from a blood sample, then the test will take several days. Amniotic fluid for this test should be withdrawn between the fifteenth and eighteenth week of pregnancy. This test takes about two weeks since the cells first have to reach the appropriate stage of cell division. Chorionic villi to be used in this test are taken between the eleventh and thirteenth week of the pregnancy.[76] Since these villi generally contain a sufficient number of cells in the appropriate stage of cell division, the test results can be available within one week. The time gained is of importance in that it shortens the period of uncertainty. If necessary, a termination of pregnancy can usually be carried out up to the thirteenth week of pregnancy.

Where a choice has to be made between using chorionic villi or amniotic fluid as a source of cell material for chromosome testing, various factors (besides the time involved) have to be taken into consideration. Two such factors are cytogenetic reliability and the risks[77] to the

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73 *eg* abnormal number, large piece missing or additional pieces onto another chromosome.

74 *ie* **chorionic villi biopsy test**.

75 *ie* **amniocentesis test**.

76 Eriksson *et al op cit* p 65 reports that a chorion biopsy or "*vlokkentest*" could be performed, as early as from 8-9 weeks of pregnancy.

77 Eriksson *et al op cit* p 78 states that an amniocentesis or "*vruchtwaterpunktie*" generally has no detrimental consequence on the unborn child and that the risk for spontaneous miscarriage is about 0,5%.

foetus which are inherent to the procedure. If the time factor is not critical then the question of whether to opt for using chorionic villi[78] or amniotic fluid[79] involves a delicate weighing up of the benefits and drawbacks. It is also possible to draw blood from the unborn child (by umbilical puncture) for the purpose of testing for chromosome abnormalities and other disorders. However, there are only a limited number of situations in which this is indicated.

Chromosome testing can be used for the diagnosis of a number of diseases. When carried out by experienced staff, this type of testing is highly specific and extremely sensitive, which gives it considerable predictive value. While the test usually only involves the individual concerned, very infrequently it is necessary for other members of the family to be tested (usually the parents).

Fain[80] mentions the chorionic villi biopsy is less risky[81] to the mother and child and will probably be the preferred method used in future. Fain conveys that obstetricians generally feel that public discussion on amniocentesis testing has tended to de-emphasize the real risks inherent in foetal testing such as harm to the foetus, possible miscarriage, bleeding, while over-emphasizing the benefits and accuracy thereof.

4.3.3 Biochemical examination

In some cases, monogenetic gene mutations may either block the synthesis of certain enzymes or lead to the production of enzymes with abnormal structures, either situation will disrupt metabolic processes. The resultant diseases can be detected by checking whether certain products of normal metabolic processes are present in body fluids such as blood and urine (metabolite studies). About one hundred rare clinical pictures can be detected by this means. With several clinical pictures, it is possible to find out directly whether the correct form of the enzyme is present (enzyme diagnosis). In addition to patients, (healthy) carriers can be identified in this way. The test is restricted to the individual concerned. However, if an abnormality is detected, this will often lead to the testing of other family members, who may request testing to see if they are carriers.

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78 Erikkson *et al op cit* p 80 reports that a chorionic biopsy could be executed on 8-9 weeks, about 8 weeks earlier than an amniocentesis, while te results are available within days (saving another 2 weeks).

79 Eriksson *et al op cit* p 79 conveys that the results from an amniocentesis is available after
2½ - 3 weeks.

80 1987. Wrongful Life: Legal and Medical Aspects. **Kentucky Law Journal** (75), 585.

81 than an amniocentesis.

It has been known for 10 to 15 years that measurement of the amount of alphafoetoprotien in the blood of a pregnant woman can indicate a heightened chance of neural tube defects or of chromosome abnormalities in the foetus. The original triple test involves measuring the levels of alphafoetoprotien, hCG[82] and oestriol. These are used in combination with the duration of the pregnancy, and the age and weight of the mother, to calculate the probability that the woman in question is carrying a foetus with either *Down's syndrome* or an open neural tube defect. The best results are obtained by measurement of the levels of alphafoetoprotein and of the free beta chain of hCG, in combination with the age of the expectant mother. The testis carried out between the fifteenth and the eighteenth week of pregnancy.

Another form of biochemical testing which can reveal the presence of a risk factor is the determination of the amounts of cholesterol and triglycerides in the blood. Increased levels of these substances indicate a risk of cardiovascular diseases which is statistically greater than normal.

The measurement of proteins produced by cancer cells is another form of biochemical testing which can give an early indication that the person concerned is suffering from some form of cancer.

4.3.4 DNA testing

A small amount of DNA is generally sufficient for DNA testing and any nucleated body cells can be used for this purpose. With the technique of PCR (polymerase chain reaction), a method of replicating DNA, it is even possible to perform test on DNA from just a few cells, or even from a single cell. This method can be important for prenatal testing using foetal cells isolated from the mother's blood, or which have been taken from an embryo produced by test tube fertilisation (so-called re-implantation diagnosis). DNA testing can be subdivided into so-called linkage-testing and the direct detection of mutations.

4.3.5 Testing with ultrasonography

From the twelfth week of pregnancy, ultrasonography can be used to detect structural and functional abnormalities in the unborn child. The ultrasonography test which is generally carried out is primarily aimed at testing the vitality, growth and position of the foetus, the position of the placenta and the detection or exclusion of multiple pregnancies. If this type of test reveals indications of foetal abnormality, follow-up testing with advanced ultrasonography techniques is required to establish the precise nature of the abnormality involved. Such follow-up testing requires exceptional expertise and special equipment. It is also carried out if other observations have revealed a heightened risk of abnormalities which can be detected by

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82 human chorionic gonadotrophin.

ultrasonography. This is the case, for example, if the foetus exhibits abnormal growth or if a previous pregnancy resulted in a child with an abnormality of the brain, heart, kidneys, urinary ducts, skeleton, et cetera, which is detectable by ultrasonography.[83] In well equipped centres, where there is a heightened risk of particular structural abnormalities, a specificity of 98% and a sensitivity of 93% have been achieved.

4.4 Time of diagnosis[84]

The time of genetic testing is relevant to the various options open to (prospective) parents.[85] A basic distinction can be made between prenatal diagnosis[86] and diagnosis in later life.[87]

4.4.1 Diagnosis prior to conception

The aim of diagnosis prior to conception is to gain insight into the chances of hereditary diseases occurring in the offspring and to do so at a moment when all possible options with regard to procreation are still open. These options include choosing to avoid having offspring of one's own; acceptance of the risk possibly in combination with the use of prenatal diagnosis (now being possible for an increasing number of diseases); the use of donor insemination or of in-vitro fertilisation (using donor sperm cells or egg cells, or combined with pre-implantation diagnosis and the adoption of a child.

Diagnosis prior to conception is currently possible when any of the following occur in a family: an X-linked hereditary disorder, some autosomal recessive and some autosomal dominant disorders, a familial chromosome translocation or frequent spontaneous abortion. Genealogical investigation and the collaboration of members of the applicant's family are often essential in this regard. The drawbacks are a degree of intrusion by medical science into the process of procreation, the possibly irksome repercussions which the knowledge gained might have for other members of the family or for access to employment and private insurance, and the psychological burden of being faced with difficult choices.

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83 Eriksson *et al op cit* p 84 - or "echoscopie".

84 Anon. 1994. *op cit* p 39.

85 Anon. 1994. *op cit* p 65.

86 "With prenatal diagnosis, the unborn child's right not to know can be frustrated if parents decide to continue the pregnancy in a situation involving a severe, untreatable disorder which only manifests itself in later life."

87 "The hereditary nature of disorders means that any information obtained will also be of interest to other members of the family."

Regarding diagnosis prior to conception, one could cite clinical pictures such as cystic fibrosis (CF), fragile X syndrome,[88] hereditary haemoglobinopathies, Tay-Sachs disease[89] and infantile spinal hereditary muscular dystrophy (Werdnig-Hoffmann disease). These are severe disorders which become apparent at birth or shortly afterwards and for which there is no cure.

4.4.2 Prenatal diagnosis

The aim of prenatal diagnosis is to create courses of action for those involved. This is achieved via the early detection of couples with a heightened risk of having children with a hereditary of foetal disorders. The term "prenatal diagnosis" implies that a pregnancy is already under way. Prenatal diagnosis is chiefly used to detect neural defects, such as *Down's syndrome* and congenital anatomical abnormalities. In addition, prenatal diagnosis is also carried out for other disorders such as those cited for diagnosis prior to conception, and for parents with a heightened risk (usually evident from the family's medical history) of having offspring with a severe hereditary disorder.

The earliest time when prenatal diagnosis can be made, actually precedes implantation of the fertilised egg cell. Testing a foetus during pregnancy can be carried out either via ultrasonography or using cellular material from the foetus or from the future placenta (chorion). Cellular material can be obtained by taking chorionic villus biopsies, amniotic fluid or foetal blood. The invasive nature of such tissue sampling means that this type of testing involves a (small) risk of losing the pregnancy or of premature birth. The most common indication for the testing of chorionic villi or of cells derived from amniotic fluid is a heightened risk of *Down's syndrome* due to the age of the mother. Such testing may also be carried out if there is prior knowledge of other heightened risks of a foetal organic defect.

At the prenatal stage, tests are carried out either on the mother alone or on both parents. Diagnosis of the mother involves, first and foremost, factors which are known to be able to affect pregnancy (high blood pressure, diabetes, antibodies against *rubella*, blood group and blood group antibodies). In addition, the triple test[90] can be used to establish whether or not there is a heightened risk of having a child with *Down's syndrome* or an open neural tube defect. If it emerges that there is indeed a heightened risk, then this diagnosis can be followed by concentrated diagnostic testing of the foetus, either by means of ultrasonography or by using cells from the amniotic fluid. Furthermore, in the prenatal stage, the tests described in association with diagnosis prior to conception can be carried out on one or both parents.

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88 see *infra*.

89 see *infra*.

90 see *supra*.

The termination of pregnancy should be viewed in the light of the fact that, if prenatal diagnosis was not available, some of the pregnancies would never have been initiated, as the parents involved would have refrained from having (further) offspring.

4.4.3 Neonatal diagnosis

The aim of neonatal diagnosis is the prevention of (or timely intervention in) hereditary disorders, by means of timely diagnosis, genetic counselling, provision of information, treatment and counselling. With some untreatable hereditary disorders, the possibility of timely genetic counselling is mentioned. For diseases in which invalidity can be prevented by timely intervention following birth, the best option is neonatal diagnosis. This may take the form of biochemical/ endocrionological testing or DNA testing. Just a few drops of blood (obtained by means of the "heel prick") are all that is required for either test.

4.4.4 Diagnosis later in life

Furthermore, the person involved can attempt to prevent expression of the genetic predisposition, by the avoidance of certain environmental factors. The number of clinical pictures in which it is possible to detect the presence of a hereditary component will increase as more genetic information becomes known.

4.5 Effect of genetic counselling

Frets[91] gives statistics on the varying ways couples reacted in making a reproductive decision upon receiving genetic counselling:

> "Of the 164 couples under study, 137 had made a reproductive decision: 109 (66%) had decided to have (more) children, while 28 (17%) had decided to refrain from having children. Eighteen couples (12%) were undecided at the time of the follow-up. (The remaining 9 couples were excluded from the statistical analysis.)"[92]

It is obvious that genetic counselling does not preclude couples from having children, it rather gives them the "go-ahead" as confirmation is given that the child will be born healthy.

5. Related aspects

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91 1990. The Reproductive Decision after Genetic Counselling. **Proefschrift - Erasmus Universiteit, Rotterdam.**

92 *op cit* p 80.

5.1 Sterilization

Podewils[93] reports that voluntary sterilization is the most effective and popular method of birth control in the United States,[94] as a 1988 contraception study showed that 28.3% of all American women chose sterilization.

Nicholson[95] describes the various sterilization procedures: Performance of a vasectomy[96] consists of cutting out a portion of each of the two ducts which carry sperm from the testicle to the ejaculatory duct. The cut ends are then tied surgically. After about six weeks, which is the time needed for the existing sperm to be ejected or to deteriorate within the reproductive system, the patient is normally rendered sterile.[97]

The corresponding operation performed on a female is a salpingectomy, or tubectomy. This procedure can be accomplished in one of two ways. The first method requires the surgical removal of the fallopian tube. The second involves cutting the fallopian tubes and tying the ends to prevent uniting of sperm and egg. Following successful removal or severing of the tube, the patient can no longer conceive a child.[98]

Hampton[99] reports that *tubal cauterization* is a process by which the fallopian tubes are blocked by burning them with instruments inserted through one or two small incisions in the abdomen, whereas in a tubal ligation procedure, the tubes are actually cut.

A vasectomy or tubal ligation may fail for one of two reasons. The possibility exists that nature may defeat the purpose of the operation. [In the case of a vasectomy, a process known as recanalization, although rare, sometimes occurs following the operation. It cannot be

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93 1993. Traditional Tort Principles and Wrongful Conception Child-rearing Damages. **Boston University Law Review** (73), 407.

94 U.S. Bureau of the census, statistical abstract of the United States - 1992.

95 1984. Damages: Recovery of Damages in Actions for Wrongful Birth, Wrongful Life and Wrongful Conception. **Washburn Law Journal** (23), 309.

96 male sterilization.

97 *op cit* p 312.

98 *op cit* p 313.

99 1986. The Continuing Debate over Recover ability of the Costs of Child - rearing in 'Wrongful Conception' Cases: Searching for Appropriate Judicial Guidelines. **Family Law Quarterly** (20), 47.

prevented, nor predicted,[100] by the physician.[101] Or an operation may fail due to the physician's negligence.[102]

5.2 Statistics

Habets[103] gives interesting statistics regarding failed sterilization in the Netherlands. He reports that failed sterilization procedures not only make up a substantial part of the total of annual medical injury claims, but are also responsible for a sizable portion of the total amount claimed. He indicates that claims of between ƒ20 000 and 30 000 are common, while 20% of sterilization-claims fall betweenƒ10 000 en ƒ50 000 and 13% above ƒ50 000.

It is reported[104] that the failure rate of sterilization procedures[105] for both men and women is estimated at respectively 0.3 to 0.5%. In 1993, 14742 women and 2341 men were sterilised in the Netherlands.

Habets[106] gives an enlightening account of the outcome of failed sterilization procedures in the Netherlands. A study was undertaken of 30 claims instituted against a medical insurer due to failed sterilization: 17 have been rejected first hand by the insurance company in question, while 11 were acknowledged. The remaining two patients, one instituted a civil claim, the other's actions were unknown.[107]

Of the 11 successful claims, a total amount of ƒ232 000 was awarded for patrimonial damages, the highest award being ƒ103 000. The total amount paid out for non-patrimonial damages was

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100 **Ball v Mudge**, 64 Wash. 2d 247, 249, 391 P.2d 201, 204 (1964).

101 *ibid.*

102 in a female, regeneration of fallopian tubes after a tubal ligation may negate the effect of cutting the tubes - see **Custodio v Bauer**, 251 Cal. App. 2d 303, 304, 59 Cal.Rptr. 463, 463 (1967).

103 1995. Schadeclaims naar aanleiding van mislukte sterilisaties: een dossieronderzoek. **Tijdschrift voor Gezondheidsrecht** (5), 266.

104 Habets *op cit* p 267.

105 depending on the proficiency of the physician.

106 1995. Schadeclaims naar aanleiding van mislukte sterilisaties: een dossieronderzoek. **Tijdschrift voor Gezondheidsrecht** (5), 270.

107 it is interesting to note the trend of directing one's plight of compensation towards an insurer, instead of towards the courts - Habets *op cit* p 272: *"..aangetoond worden de meeste medische aansprakelijkstellingen zonder rechterlijke tussenkomst afgehandeld."*

ƒ142 000, with highest claim of ƒ45 000.

Up to November 1993 a total amount of ƒ316 000 was paid out for both patrimonial and non-patrimonial damages. The highest claim award amounted to ƒ75 000, which included payment for legal fees. Other awards were for ƒ54 000, ƒ50 000, ƒ30 000 en ƒ17 500, while the remaining five were given ƒ10 000.

Habets[108] reports that in 50% of cases were a "second-look operation" were performed[109] it was clear that improper sterilization techniques were performed.[110] These statistics are in line with a 1993 report of the Dutch Association for Obstetrics and Gynaecology[111] which indicated that 50% of all failed sterilization procedures could be attributed to chance, such as when spontaneous re-canalisation takes place or when sterilization clips become loose and 50% due to physician negligence.

5.3 Genetic engineering

Singer[112] reports that we can distinguish two major purposes for genetic engineering in humans: to remove defects not present in normal members of the species;[113] and to produce individuals with more desirable qualities than would be the case with normal reproduction.[114]

It is suggested[115] that as remedying defects is at the core of medicine, "the ethical and policy issues do not seem appreciably different from those involved in the development of any new diagnostic or therapeutic techniques".

The medical model is no help to us in sorting out these characteristics; hence we can agree on eliminating defects much more easily than we can agree on a moral basis for enhancing

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108 *op cit* p 271.

109 such a second operation, performed specifically to inquire whether the initial procedure was properly performed, can only be done in cases of female sterilization.

110 and liability accordingly acknowledged.

111 *"Nederlandse Vereniging voor Obstetrie en Gynaecologie".*

112 1985. **Making Babies - The New Science of Ethics of Conception.** Charles Scribner's Sons, 164.

113 therapeutic genetic engineering.

114 eugenic genetic engineering.

115 *op cit* p 165.

someone's genetic constitution above what would normally be expected.

> "Whatever we call it, this form of genetic engineering can be justified on the same basis as cases that are indisputably therapeutic. It does not matter whether the outcome is a life that is or is not better than the statistical norm; the essential element is that no one disputes that, other things being equal, it is better to live longer, in good health, than to die earlier. The acceptability of genetic engineering depends not on whether it falls under the label "therapeutic" rather than "eugenic", but on the ends toward which the engineering is directed. When the goal is something that would indisputably improve the human condition, safe and successful genetic engineering would be a good thing."[116]

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116 *op cit* p 166.

6. Congenital anatomical abnormalities[117]

6.1 Genetic diseases/ anomalies found in wrongful life litigation

Two to three per cent of births involve children with severe congenital abnormalities other than neural tube defects and *Down's syndrome*. Congenital abnormalities are responsible for one quarter of all prenatal mortality. Ninety per cent of congenital abnormalities affect the children of parents who had no heightened risk in that regard. Many prenatal disorders can be detected using ultrasonography (more than 200).

Congenital abnormalities can affect various organ systems, such as the cardiovascular system, the central nervous system, the sex organs and urinary ducts, the gastro-intestinal system of the skeleton. They may be determined purely by genetics, caused by exogenous factors (infections, medicines) or by combinations of the two. Knowledge of the exact cause also provides information about the chance of prevention or of repetition. Many such abnormalities are lethal. Here is listed[118] and briefly discussed some of the most commonly found genetic anomalies that afflict real people and that have lead to actual wrongful life litigation in the past:[119]

Alzheimer's disease:

> Dementia, which is characterised by progressive memory disorders, deterioration of cognitive functions and (often) personality changes, leads to the disruption of patients' ability to function. As a result, during the course of their illness, patients become gradually more dependent on others to take care of them. Dementia is not a disease of the brain in the strict sense of the word, but rather a syndrome. Alzheimer's disease is one of the most important causes of dementia syndrome in elderly people. *Down's syndrome*[120] patients frequently develop Alzheimer's disease in later life.

Anencephaly:

> In 1988, the total number of new cases of anencephaly in the Netherlands[121]

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117 Anon. 1994. *op cit* p 127 *ao*, Sadler *op cit* p 110 *ao*, relevant court cases and varying other medical sources, incl the internet.

118 in alphabetical order.

119 disease/ condition, description and case reference (where possible).

120 see *infra*.

121 anencephaly is seen in the United States at a frequency of 1:1 000 births.

was estimated to be 75 per annum.[122] Anencephaly is due to failure of the anterior neural tube to close properly during very early intrauterine life. Mothers with infants having neural tube defects have high levels of alpha feto-protein.

In anencephaly, the infant is born with no cerebral hemispheres, only a rudimentarybrainstem and brainstem, and no calvarium (skull bone). Often there are associated malformations such as cleft lip and palate, abnormalities of hands and feet and other internal malformations. Usually associated with bulging, frog-like, eyes.[123]

These babies may survive outside the womb for varying amounts of time, anywhere between minutes and weeks. Although the condition is always fatal and there are no techniques to correct the problem.[124]

Cancer of the colon:

Within the clinical picture of cancer of the colon, there is a clearly described hereditary disorder called polyposis coli or familial adenomatous polyposis. This disorder is autosomal dominant.

Congenital hypothyroidism:

Congenital hypothyroidism is caused by a deficiency of thyroxin (T4). The thyroid itself may be defective or the problem may lie with those organs which stimulate the thyroid to produce thyroxin (congenital thyrotropin deficiency syndrome) via thyroid stimulating hormone. A greatly reduced T4 level can lead to severe mental retardation, behavioural disorders and motor disorders. In the past, it was not usually possible to make a diagnosis until the condition was at a relatively advanced stage (since the symptoms are relatively unspecific and only develop gradually). However, if this disorder is detected in time, treatment with hormone preparations is almost entirely effective in preventing mental retardation. Only in the case of severe T4 deficiency do mild motor disorders occur.

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122 neural tube defects - disruption of the closure of the neural tube during embryonic development can lead to anencephaly or spina bifida, dependent upon the location of the closure defect.

123 http://155.37.5.42/TMGEN/X2021000.htm.

124 http://geocities.com/HotSprings/Spa/2147/anencephaly.html.

Cri-du-chat Syndrome:

Cri-du-chat syndrome was first described as a hereditary congenital syndrome associated with deletion of part of the short arm of chromosome 5. The syndrome is characterized in young children by microcephaly, round face, hypertelorism, micrognathia, epicanthal folds, low-set ears, hypotonia, and severe psychomotor and mental retardation. One of the most characteristic features in new born children is a high pitched cat-like cry that is usually considered diagnostic for the syndrome. Although the majority of patients die in early childhood, some survive into adulthood and exhibit an IQ below 20, a loss of hypertelorism and epicanthic folds and development of a thin narrow face with prominent nasal bridge.

The cri-du-chat syndrome appears to be one of the most common human deletion syndromes with an incidence varying between 1 in 20 000 to 1 in 50 000 births. The frequency in populations of profoundly retarded patients (IQ less than 20) is approximately 1%.[125]

Cri-du-chat syndrome, also known as 5p-Syndrome (five p minus) or "Cat Cry Syndrome", is a rare genetic disorder characterized at birth by a high pitched cry, low birth weight, poor muscle tone, and other anomalies.[126]

Johnson v Yeshiva Univ.[127]

Cystic Fibrosis:

Cystic fibrosis is an autosomal recessive disease which results in damage to the respiratory system and to digestive functions. The clinical picture usually manifests itself at a very early age. The frequency of patients at birth is 1 in 3 600.[128] Such individuals receive an abnormal genetic trait from both parents. Although the severity of the disease can vary, it often leads to frequent hospital admissions and periods of treatment. Generally it is a serious handicap which forms a great burden for the parents and family. Thanks to intensive therapy and support, the average life expectancy of such patients (dependent upon when they were born) has gradually increased from

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125 http://www3.ncbi.nlm.nih.gov/htbin-post/Omim/dispmim.

126 http://www.geocities.com/CapitolHill/1703/CRI.html.

127 396 N.Y. 2d 818, 364 N.E. 2d 1340, N.Y.S. 2d 647 (1977).

128 about 3% of the population of the Netherlands carry a harmful mutation in the gene which is involved in this disease.

25 to 40. Thus 50% of such patients will have died before attaining that age.

Fain[129] reports that cystic fibrosis is the most common genetic disorder among Caucasians. This dangerous disease is discovered in about 2000 new cases per year in United States of America. Approximately 5% of all Americans are carriers.

Schroeder v Perkel[130]

Downs's Syndrome:

Down's syndrome (trisomy 21) is associated with severe mental handicap and my also be combined with characteristic abnormalities in some organ systems (e.g. the heart and the proximal part of the duodenum). Many patients develop Altzheimer's disease[131] after reaching the age of forty. Most cases of *Down's syndrome* (96%) involve a separate, extra chromosome 21 (non-hereditary form). In the remaining 4% of cases, a chromosome translocation is involved, with (part of) the extra chromosome 21 being attached to another chromosome. Both abnormalities can be detected by examination of the chromosomes in cellular material taken from the foetus. In 1% of all cases, a balanced chromosome translocation is found in one of the parents (hereditary form). Life expectancy is highly dependent on medical policy with regard to any additional congenital abnormalities.

Azzolino v Dingfelder[132], **Berman v Allan**[133], **Becker v Schwartz**[134], **James G v Casceta**[135], **Hickman v Group Health Plan Inc**[136], **Phillips v United States**[137] and **Andalon v Superior Court.**[138]

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129 *ibid.*

130 432 A. 2d 834 N.J. (1981).

131 see *supra.*

132 337 S.E. 2d 528, 537 N.C. (1985), *cert denied*, 475 U.S. 835 (1986).

133 80 N.J. 421, 404, A. 2d 8 (1979).

134 46 N.Y. 2d 401, 386 N.E. 2d 807 N.Y.S. 2d 895 (1978).

135 332, S.E. 2d 872 Va. (1985).

136 396 N.W. 2d 10 Minn. (1986).

137 575 F. Supp. 1309 D.S.C. (1983).

138 208, Cal. Rptr. 899, 901 (1984).

Duchenne and Becker muscular dystrophy:

Duchenne muscular dystrophy and Becker muscular dystrophy are sex-linked hereditary diseases with a progressive course, which are associated with mutations of the same gene on the X chromosome. Onset is usually between the ages of two and four years. The earliest symptom is difficulty with walking resulting from weakness in the pelvic girdle and the thigh muscles. The disease gradually spreads to the arm, neck and respiratory muscles. The cardiac musculature is often affected as well. Such children become confined to a wheelchair at around the age of eleven and they ultimately die, aged about 20, from respiratory or cardiac insufficiency. More than one third of such children are also mentally handicapped. One third of cases do not involve a mutation which has been passed down through the family. In these instances, a denovo mutation (occurring in the mother in two thirds of cases) is responsible for the disease.

Nelson v Krusen[139]

Familial hypercholesterolaemia:

Hyperlipidaemia (an excessive level of cholesterol or triglycerides in various lipoprotein fractions in the blood) is common within the population and is one of the most important risk factors in relation to cardiovascular diseases. It has been estimated that one third of the population will die from cardiovascular disease. While the genetic background to hyperlipidaemia is still poorly understood, research has indicated that a large number of genes are involved. It is possible to distinguish between monogenetic and polygenetic forms of the condition. One important monogenetic form is familial hypercholesterolaemia.

Fetal Hydantoin Syndrome:

In a retrospective study the frequency of major malformations such as heart abnormalities, facial clefts, and microcephaly was high when drugs such as phenytoin and trimethadione were used[140] by the mothers. Specifically diphenylhydantoin produces a broad spectrum of abnormalities, including craniofacial defects, nail and digital hypoplasia, growth abnormalities and mental deficiency. These defects constitute a distinct pattern of dysmorphogenesis, namely fetal hydantoin syndrome.

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139 678 S.W. 2d 918 Tex. (1984).

140 for epilepsy - the drug *in casu* was "Dilatin".

Harbeson v Parke-Davis Inc.[141]

Fragile X syndrome:

Fragile X syndrome is the most common cause of familial impaired mental development (familial mental retardation). It displays X chromosome transmission with several unusual features: about 35% of the female carriers display mild to moderate mental retardation and it is even possible for a healthy man to transmit the mutation to carrier females. Besides the impaired mental development, there are also physical abnormalities and behavioural problems. Boys with the syndrome generally attend schools for children with extreme learning difficulties. While they generally continue to live at home, they sometimes have to be removed in connection with anxiety attacks or temper tantrums. In adulthood they reside in surrogate family units, other institutions for the mentally handicapped, or with their parents. Some of them attend a sheltered workshop during the day. Women with the syndrome are often less severely mentally handicapped than affected men and, dependent upon their level of disability, they live either independently or in supervised accommodation.

Haemophilia:

Haemophilia A is a recessive, X-linked bleeding disorder affecting approximately 1/5000 males due to a deficiency of blood coagulation Factor VIII. The severity of the condition varies from severe (<1% normal clotting activity), moderate (2-5%) to mild (5-30%). Further complications may arise due to the presence of inhibitors (antibodies) to replacement factor VIII.[142]

Haemophilia is a blood condition in which an essential clotting factor is either partly or completely missing. This causes a person with haemophilia to bleed for longer than normal. Cuts and grazes are not great problems as a little pressure and a plaster are usually enough to stop bleeding. The main problem is internal bleeding into joints, muscles and sort tissues. Haemophilia is a genetically inherited condition. It is usually carried in the female genes, but generally only affects males. About a third of new diagnoses are where there is no previous family history. It is a lifelong condition, appears world-wide and occurs in all racial groups. About 10 000 people are affected in the

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141 98 Wash. 2d 460, 656 P. 2d 483 (1983).

142 http://www.leedsdna.demon.co.uk/haema.htm.

UK.[143] A hereditary disease occurring almost exclusively in males, and about 1 in 6 500 male Australians are affected.[144]

Siemienic v Lutheran Gen. Hosp.[145]

Hearing deficiency:

Turpin v Sortini.[146]

Hereditary forms of breast cancer:

In the Netherlands, more than 7 000 women are diagnosed with breast cancer each year. Of these, about 20% ultimately die from the effects of this disease. About 350 to 560 of the 7 000 patients who develop breast cancer each year have a genetic predisposition to the disease.

Huntington's disease:

Huntington's disease, which is transmitted as autosomal dominant, is associated with degeneration of the nervous system. The disorder usually becomes manifest during adult life, although some cases occur earlier and some later. Following the appearance of the first symptoms, the disease progresses gradually for 15 to 25 years with the loss of mental and physical functions as well as personality changes until the patient finally dies. All carriers of the abnormal gene have an almost 100% chance of developing the disease before they reach old age. By the time the disease makes its appearance, patients have usually had children, and have thereby passed the disease on. The disease is incurable. Carriers of the abnormal gene occur with a frequency of 1 in 5 000 among the general population.

Hydrocephalus:

An accumulation of cerebro-spinal fluid within the skull. The condition is characterized by an enlarged head and often results in mental retardation. Early treatment can save approximately half of the children born with this condition, but of those who survive approximately two-thirds will suffer some

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143 http://www.haemophilia.org.uk/whatis.html.

144 http://www.australiahealth.com/Health%20Conditions/haemophi.htm.

145 134 Ill. App. 3d 823, 480 N.E. 2d 1277 (1985).

146 182 Cal. Rptr. 337 (1982).

level of brain damage.[147]

Edmonds v Western Pennsylvania Hospital Radiology Assocs.[148]

Infantile spinal muscular atrophy:[149]

This autosomal recessive disorder is the most dramatic form of a group of disorders of the anterior horn cells in the spinal cord and in part of the brain stem. Muscle weakness and hypotonia are characteristic features of all forms, but they can differ in time of onset and in severity. In the infantile form, these symptoms appear before the child reaches an age of 6 months, and they are sometimes even apparent at birth. This results in an arrest in motor development. The motor milestone of sitting up is never achieved. Besides muscular weakness in the limbs and trunk, swallowing difficulties also occur. Treatment is only aimed at combatting the symptoms and most affected children die of pneumonia before the age of two.

Klinefelter's Syndrome:

The clinical features of this syndrome, found only in males, are sterility, testicular atrophy, hyalinization of the seminiferous tubules, and usually gynecomastia. Th incidence is about 1 in 500 males in the normal population. Among mentally defective subjects, the incidence is as high as 1 in 100 males. On the basis of statistical evidence, it is believed that non-disjunction of the XX homologs is the most common causative event. Occasionally, however, patients with Klinefelter's syndrome have 28 chromosomes, that is, 44 autosomes and 4 sex chromosomes (XXXY).[150]

Larsen's Syndrome:

Larsen Syndrome[151] is a multi-system genetic disorder that is present at birth. It is characterized by multiple bone dislocations and abnormalities, an extremely high arch of the foot, non-tapering cylindrically shaped fingers, and an unusual facial appearance. In some cases, short stature, heart problems,

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147 Silverman, A.M. 1993. Constitutional Law: Pennsylvania's Wrongful birth statute's impact on abortion rights: state action and undue burden- Edmonds v Western Pennsylvania Hospital Radiology Assocs. **Temple Law Quarterly** (66:4), 1088, with reference to **Black's Medical Dictionary** 1987 (35th Edition), 345.

148 607 A.2d 1083 Pa. Super. Ct. (1992).

149 also Werdnig-Hoffmann's disease.

150 Sadler *op cit* p 120.

151 also Desbuquois syndrome or Sinding-Larsen-Johansson disease.

cleft palate or lips, deafness, or mental retardation may also occur.[152] **Moores v Lucas.**[153]

MCAD:[154]

MCAD deficiency (deficiency of the mid-chain acylco-enzyme A dehydrogenase) is an autosomal recessive disease of fatty acid metabolism. The disorder is linked with a significant chance that metabolism will become disordered, resulting in increasing lethargy (particularly when fasting or feverish) and ultimately in coma and death. This clinical picture is held to be responsible for 2% of all cases of cot death. The symptoms usually develop at an age of 5 to 24 months. The chance of death is highest between the ages of 15 to 26 months.

Multiple birth defects:[155]

The child had one to few vessel umbilical cords, a short cord, *ventriculomegaly*, absent right leg, imperforate anus, one testicle, one kidney, a vertebrate anomaly in the lumbar sacral region, *hydrocephaly*, a large fluid-filled sac extending off the right aspect of the sacrum consistent with *meningocele (spina bifida).*[156] **Keel v Banach,**[157] **Bruggeman v Schimke.**[158]

Myotonic dystrophy:

This autosomal dominant disorder is the most commonly occurring muscular dystrophy in adults. Besides muscular dystrophy there may also be abnormalities in various other organs. Those carrying the genetic abnormality are certain to develop the disease. There is great variation in terms of severity and time of onset. With the congenital or infantile form, the

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152 http://www.stepstn.com/cgi-win/nord.exe? proc=Redirect&type= rdb_ sum&id=497.htm.

153 405 Fla. App. So. 2d 1022 (1981).

154 a disease involving fatty acid metabolism.

155 Faircloth, R.C. 1994. Keel v Banach: Alabama gives life to Wrongful birth actions. Should we sue for malpractice? **Cumberland Law Review** (24:3), 547.

156 Haavi Morreim, E. 1988.The Concept of Harm Reconceived: A different look at Wrongful Life. **Law and Philosophy** (7), 5.

157 624 So. 2d 1022 Ala. (1993).

158 718 P.2d 635 Kan. (1986).

symptoms of muscular weakness following birth are often so severe that the child dies within a few days. This form only occurs when the abnormal gene is inherited via the mother. If the onset of the disease occurs during childhood there is some muscular weakness, although the clinical picture is predominantly one of mental retardation. With the adult type, the initial symptoms of the disease manifest themselves between the ages of 12 and 50. In general, muscle weakness is gradually progressive. Affected men in whom disease onset occurs during early adulthood are generally sterile. Life expectancy is determined by the occurrence of acute cardiac arrest and respiratory disorders, usually as a complication during general anaesthesia. The late onset type usually begins during late middle age (above age 50) and often manifests itself primarily in the form of cataracts.

Neurofibromatosis:[159]

Is characterized by developmental changes in the nervous system, muscles, bones and skin. The condition is both congenital and *heredofamilial* (inherited by more than one member of a family). There is no known treatment or cure for this disease.

Ellis v Sherman,[160] **Speck v Finegold**.[161]

Phenylketonuria:

Phenylketonuria is a congenital autosomal recessive disorder. The disease is caused by a defective enzyme. If left untreated, the disease will cause irreversible damage to the central nervous system of affected children, and a severe mental handicap. If the disorder is detected on time, a special long-term diet can prevent damage from occurring. This disorder has a frequency of 1 in 18 000 for newborns.

Polycystic kidney disease:

Description: Inherited disorders characterized by the development and growth of cysts in the kidneys; lined by epithelium, filled with flued or semi-solid debris; accounts for 5-10% of patients with end stage renal disease.[162]

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159 A.H.S. 1983. Torts. **Journal of Family Law** (21:1),168.

160 515 A. 2d 1327 Pa. (1986).

161 497 Pa. 77, 439 A. 2d 110 (1981).

162 http://www.5mcc.com/5MCC/SUMMARY/0719.html.

Developmental disorder of the kidneys discovered in developing foetuses (by ultrasound) or in the newborn period (enlarged kidneys). Most cases that go to term die shortly after birth from respiratory difficulties due to the enlarged kidneys that have resulted in developmental pulmonary hypoplasia. This disease is negligent invariably fatal. Those cases that survive infancy may subsequently develop hepatic portal fibrosis, portal hypertension and splenomegaly.[163]

Autosomal dominant polycystic kidney disease affects about 1 in 1 000 persons in Caucasian populations and may be 2.5 to 15 times more frequent than other common hereditary disorders such as cystic fibrosis and sickle cell disease.

The disease is characterized by cyst formation in ductal organs particularly the kidney and the liver and by gastrointestinal, cardiovascular and other abnormalities. As offspring of an affected parent has a 50% risk of inheriting the disease, precise and factual genetic counselling can be provided. The impact of this measure may however be limited as far as the application of pre-natal diagnosis procedures is concerned.[164]

Park v Chessin.[165]

Quadrigeminal arachnoid cysts:[166]

A four part, fluid filled cyst between the layers of the leptomeinges lined with arachnoid membrane and most commonly occurs in the sylvian fissure of the brain.

Rh disease:

During the birthing process, blood cells from the unborn child can escape into the mother's bloodstream. These cells are recognized as foreign if they are a different blood type from the mother and a natural rejection process will ensue with the formation of antibodies. The process is known as red cell alloimmunization. This event typically occurs after the delivery of a baby at

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163 http://155.37.42?TMGEN/71026740.htm.

164 http://www.seychelles.net/smdj/orig3.htm.

165 60 A.D. 2d 80, 400 N.Y.S. 2d 110 (1977).

166 Silverman *ibid* with reference to **Dorland's Illustrated Medical Dictionary** 1988. (27th Edition), 421.

the end of pregnancy, but other pregnancy-related events such as elective abortion of spontaneous miscarriage can result in antibody formation. Although the pregnancy in which the alloimmunization firts occurs results in an unaffected child, future children are at substantial risk. In these subsequent pregnancies, newly formed antibodies in the pregnant patient can cross to the unborn child and attach to its red blood cells producing a low blood count (anemia) and in the word case scenario, foetal death. These antibodies can be measured in a woman's bloodstream by a test called an indirect Coombs or antibody titer. In general, the foetus of each subsequent pregnancy exhibits more severe effects that in the previous pregnancy. The foetal and newborn effects of red cell alloimmunization are known as hemolytic disease of the newborn.

In more than 98% of cases, the red blood cell incompatibility involves the Rhesus or Rh D antigen so the disease is known as Rhesus disease or Rh disease. Rhesus disease is approximately one case per 1000 live born infants.[167]

Continental Casualty Co. v Empire Casualty Co.[168]

Rubella syndrome:

Rubella is a viral disease characterized by slight fever, rash and swollen glands. Most cases are mild.

The effect: *Rubella* infection is dangerous because of its ability to damage an unborn baby.[169] Infection of a pregnant woman may result in a miscarriage, stillbirth or the birth of an infant with abnormalities which may include deafness, cataracts, heart defects, liver and spleen damage and mental retardation. Congenital *rubella syndrome* occurs among at least 25 percent of infants born to women who have had *rubella* during the first trimester of pregnancy.[170]

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167 http://www.med.unc.edu/obgn/rh.htm.

168 713 P. 2d 384 Colo. App. (1985) - *in casu* the child suffered from a "stroke and brain damage due to *erythroblastosis*, a hemolytic disease which befell an infant when physicians had not properly treated RH factor problems in his mother's prior pregnancies.

169 congenital *rubella syndrome.*

170 webmaster@healthanswers.com.

The disease is potentially serious because of the ability to produce defects in a developing foetus if the mother is infected during early pregnancy. As many as 10 to 15% of women in their childbearing years are susceptible in infection. Congenital *rubella syndrome* occurs in 25% or more infants born to women who acquired *rubella* during the first trimester of pregnancy. Defects are rare if the infection occurs after the 20th week of pregnancy. One or more defects may occur in an infected foetus and include deafness, cataracts, microcephaly, mental retardation, congenital heart defects and other defects. A miscarriage or stillbirth may occur.[171]

Symptomes:

- history of mother having *rubella* while pregnant;
- skin rash at birth (purpura, petechiae);
- low birth weight;
- small head size (microcephaly);
- bulging fontanelle;
- lethargy;
- irritability;
- hearing loss;
- deafness;
- seizures;
- abnormal muscle tone;
- cloudy corneas or white appearance to pupil (leukocoria);
- motor-mental retardation;
- mental retardation;
- simian crease.

Robak v United States[172], **Procanic v Cillo**[173], **LaPoint v Shirley**[174], **Blake v Cruz**[175], **Eisbrenner v Stanley**[176], **Gleitman v Cosgrove**[177], **Steward v**

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171 adam.com.

172 658 F. 2d 471 (1981).

173 97 N.J. 339, 478 A. 2d 755 (1984).

174 409 F. Supp. 118 Tex. (1976).

175 698 P. 2d 315, 319 Idaho (1984).

176 106 Mich. App. 351, 308 N.W. 2d 209 Mich Ct. App. (1981).

177 49 N.J. 22, 227 A. 2d 689, 22 ALR 3d 1411 (1967).

Long Island College Hospital[178], **Jacobs v Theimer**[179] and **Dumer v St Michael's Hospital.**[180]

Sickle cell anaemia:

Sickle cell disorder is the name for several related but different inherited disorders associated with the sickling of the red blood cell. Sickle cell disorder alters the shape of the red blood cells from their usual round appearance to something which resembles a sickle, or half moon. Sickle cell anaemia is usually the most severe type of sickle cell disorder where the majority of the haemoglobin inherited is sickle. Sufferers may experience "crises", bouts of pain. Anaemia, infections or jaundice. Sickle cell anaemia does not affect a person's intelligence, except in extremely rare cases.[181]

About 1 in 12 black Americans carries the gene for the sickle cell trait (that is, they have the ability to produce children with sickle cell anemia, but have no symptoms of the disease). If both parents carry the trait, the chance of having a child with sickle cell anemia is one out of four, or 25 percent (this trait occurs only in the black population).

According to Fain,[182] approximately 200 000 deaths per year occur in the United States alone because of only two common[183] haemoglobin disease, namely: *Sickle cell anaemia*[184] and *thalassemia*

Signs and symptoms:

- pain, ranging rom mild to severe, in the chest, joints, back or abdomen;
- swollen hands and feet;
- jaundice;

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178 58 Misc. 2d 452, 296 N.Y.S. 2d 41 (1968).

179 519 S.W. 2d 846 Tex. (1975).

180 69 Wis. 2d 766, 233 NW 2d 372 (1976).

181 http://www.sicklecellsociety.org.

182 *op cit* p 585.

183 there are an estimated 100 million carriers in the world for these disorders.

184 *the sickle cell anaemia*-condition entails that due to the presence of an abnormal type of haemoglobin (S) in red blood cells, the frequency of the gene that causes the disease is high in Mediterranean and African populations.

- repeated infections, particularly pneumonia or meningitis;
- kidney failure;
- gallstones (at an early age);
- strokes (at an early age).

Dorlin v Providence Hosp.[185]

Tay-Sachs disease:

The Tay-Sachs baby may appear well at birth, but suffers from blindness, paralysis, feeding problems and seizures as the disease progresses. Death occurs between three and five years of age and the last years of life are spent in a vegetative state. There is no cure or hope of recovery for the child with this disease.

Tay-Sachs disease[186] is an autosomal recessive disorder in which the enzyme hexoseaminidase A is absent. This results in a disruption in the breakdown of fatty substances (gangliosides) and accumulation of these substances in the brain cells. The clinical picture is characterised by disrupted development of the brain and muscle functions. Children with Tay-Sachs disease initially show normal development, but the disease manifest itself at around the age of 6 months. Children with the disease usually do not live beyond the age of four. In some cases the initial symptoms only occur at around the second to the third year of life. Such children usually survive longer, until the fifth to the tenth year of life. The disease process, which causes severe mental handicap, deafness and blindness, is untreatable.

Gildiner v Thomas Jefferson University Hospital[187], **Curlender v Bio-Science Laboratories**[188], **Howard v Lecher**[189], **Goldberg v Ruskin**[190] **Naccash v Burger.**[191]

Thalassaemia:

The name thalassaemia covers several autosomal recessive clinical pictures,

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185 118 Mich. App. 831, 325 N.W. 2d 600 (1982).

186 Bey-Berkson, *op cit* p 68.

187 451 F. Supp. 692 Pa. (1978).

188 106 Cal. App. 3d 811 (1980).

189 42 NY 2d 109, 366 N.E. 2d 64 (1977).

190 113, Ill. 2d 482, 499 N.E. 2d 406 (1986).

191 290 S.E. 2d 825 Va. (1982).

which involve faulty synthesis of the red blood pigment, haemoglobin. Haemoglobin is the protein which is responsible for the take-up and release of oxygen by red blood cells. It is constructed from two pairs of protein chains, namely two alpha and two gamma chains. The gradual production of adult haemoglobin commences even during foetal development. The major shift from one form of haemoglobin to the other occurs during the first few months of life. Some forms of thalassaemia can be extremely severe. In the case of alpha thalassaemia major, the alpha chains cannot be synthesised, thereby preventing the production either of foetal or adult haemoglobin. This leads to the death of the child during pregnancy or shortly thereafter. It can also lead to severe pregnancy-related complications for the mother. With beta thalassaemia major the defect blocks the production of the beta chains. At birth, the child has normal levels of haemoglobin but an extremely severe anaemia develops soon after birth (the production of gamma chains stops naturally, but no beta chains can be produced).

Bani-Esraili v Wald.[192]

Triple X Syndrome:

Patients with triple X syndrome are infantile, with scanty menses and some degree of mental retardation. They have two sex chromatin bodies in their cells and are therefore sometimes called "superfemale". The triple X syndrome results from fertilization of an XX oocyte and an X-containing sperm. Some of the patients are of proven fertility and, surpisingly, the offspring have been uniformly normal.[193]

Trisomy 9:

Trisomy 9[194] is a rare chromosomal disorder in which parto or all of the short arm (p) of chromosome 9 is present three times rather than twice in the cells of the body. Chromosome 9, Trisomy 9p is characterized by mental retardation, characteristic physical abnormalities of the head and facial (craniofacial) area, and/or skeletal malformations. In about 25 percent of infants with this disorder, heart defects are present at birth (congenital). Affected infants may also experience growth retardation and a significant

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192 127, Misc. 2d 202, 485 N.Y. 2d 708 (1985).

193 Sadler *op cit* p 122.

194 also Trisomy 9P Syndrome (Partial), Chromosome 9, Partial Trisomy 9P, Chromosome 9, Complete Trisomy 9P Chromosome 9, Rethore Syndrome (obsolete).

delay in the acquisition of language and communication skills. In some cases of Chromosome 9, Trisomy 9p occurs because of a balanced chromosomal rearrangement between two chromosomes of one of the parents; other cases are the result of a spontaneous (de novo) genetic change early in embryonic development that occurs for unknown reasons. Chromosomal analysis is necessary for definite diagnosis.[195]

Gallagher v Duke Univ.[196]

Trisomy 13-15:

The main abnormalities of this syndrome are mental retardation, congenital heart defects, deafness, cleft lip and palate, and eye defects such as microphthalmia, anophthalmia, and coloboma. The incidence of this abnormality is about 0.2 per 1000 newborns. Most of the infants die by the age of 3 months.[197]

Trisomy 17-18:

Patients with this chromosomal arrangement show the following features suggesting a distinct clinical entity: mental retardation, congenital heart defects, low-set esrs, and flexion of fingers and hands. In addition, the patients frequently show micrognathia, renal anomalies, syndactyly, and malformations of the skeletal system. The incidence of this condition is about 0.3 per 1000 births. The infants usually die by the age of 2 months.[198]

Turner's Syndrome:

This condition, found in women with an unmistakably female appearance, is characterized by the absence of the ovaries (gonadal dysgenesis). Other abnormalities frequently found are webbed neck, lymphedema of the extremities, skeletal deformities, and mental retardation. Despite the female appearance of these patients, almost all of their cells are sex chromatin-negative. In addition, the cells have only 45 chromosomes with an XO chromosomal complement. Genetic analysis has shown that this syndrome is usually caused by non-disjunction in the male gamete during meiosis.[199]

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195 http://www.trisomy.org.

196 638 F. Supp. 979 N.C. (1986).

197 Sadler *op cit* p 120.

198 Sadler *op cit* p 118.

199 Sadler *op cit* p 121.

6.2 The future of genetic science

It is apt that this chapter is concluded with a sense of hope for possible future sufferers from birth defects. Lupton[200] convincingly argues in favour of medical advancement in general and the development of new genetic tests and techniques specifically:

> "To turn our backs on the potential offered by germ cell therapy would be short-sighted, given the magnitude of the problems facing millions of inhabitants of this planet. I do not believe that we should give up our efforts to treat genetic afflictions or to improve the quality of life of millions of people because the device we use lies on the boundary of moral and ethical acceptability."[201]

It should be noted that the development of safe genetic treatment of foetuses would create totally new causes of action. An obvious action would, for example, arise where a handicapped child is born while prenatal treatment of the condition was possible.[202]

Collins[203] reports on the modern medical advances that has made the treatment of foetuses possible:

"The therapeutic treatment of foetuses before birth is one of the fastest growing and most significant new areas in medicine. Doctors have drained flued from foetuses' vital organs and have removed a foetus from the womb and returned it after treatment. The risks are high and the techniques experimental, but the doctors who perform foetal surgery have just begun to examine the possibilities of foetal treatment. Given the stage of development of the prenatal life and the wrongful death statutes in many states, doctors may not be liable for negligence in foetal surgery.

Since the treatment of a prenatal life will probably be possible from preconception, conception or implantation in the future, some guidelines for liability and a standard of care will have to be developed by the courts and legislatures. Although it can be argued that providing a remedy for the death of a life after implantation my discourage prenatal treatment, it is more

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200 1992. Genetic engineering: Does it merely facilitate the process of evolution. **Tydskrif vir Hedendaagse Romeins-Hollandse Reg**, 79.

201 *op cit* p 87.

202 these possibilities will doubtlessly be the topic of future studies.

203 1984. An Overview and Analysis: Prenatal Torts, Preconception Torts, Wrongful Life, Wrongful Death, and Wrongful Birth: Time for a new framework. **Journal of Family Law** (22), 677.

likely that such a wrongful death statute would only discourage indiscriminate and unrealistic foetal surgery. A doctor's liability, of course, would be based on the plaintiff's proof of wrongful conduct and causation."[204]

204 *op cit* p 690.