Supplementary Data: ICGNMD REDCap Database Instrument

ICGNMD Study Database

Consent obtained	◯ Consent by subject ◯ Consent by parent/guardian, advocate, or next of kin ◯ Other
Consent obtained - other	_____
Consent taken by (must be authorised member of study team)	_____ (Full name)
Consent Method Used	◯ In person (face to face) ◯ Postal Consent* ◯ Telephone Consent* ◯ Online Consent* (*Only where permitted under local ethics)

After determination of eligibility and consent, assign and record ICGNMD study ID

ICGNMD study ID	_____ (IC_[site code]_[participant number]; IC_XXX_XXXXX, e.g. IC_UTH_00001)
Other local study ID (optional)	_____ (Other local study ID for participant (in addition to ICGNMD study ID))
Date consent form signed	_____ (YYYY-MM-DD)

Participant type

Participant type	◯ Affected proband ◯ Affected relative ◯ Unaffected relative ◯ Unrelated control subject (Clinical status)
Proband study ID (family grouping code)	_____ (The ICGNMD study ID of the Proband, e.g. IC_UTH_00001)
Proband initials	_____ (For data validation)
Proband year of birth	_____ (For data validation)

Relation

Relationship to Proband

- ◯ Mother
- ◯ Father
- ◯ Son
- ◯ Daughter
- ◯ Grandchild
- ◯ Brother - non-identical
- ◯ Sister - non-identical
- ◯ Identical twin
- ◯ Paternal aunt/uncle
- ◯ Maternal aunt/uncle
- ◯ Paternal grandparent
- ◯ Maternal grandparent
- ◯ Paternal cousin
- ◯ Maternal cousin
- ◯ Niece/nephew
- ◯ Other

Relationship to Proband - other

_____

Diagnosis

Diagnostic category

- ◯ Genetic motor neuron disease
- ◯ Spinal muscular atrophy
- ◯ Genetic peripheral neuropathy
- ◯ Congenital myasthenic syndrome
- ◯ Congenital muscular dystrophy or myopathy
- ◯ Duchenne or Becker muscular dystrophy
- ◯ Facioscapulohumeral muscular dystrophy
- ◯ Myotonic dystrophy type I
- ◯ Limb girdle muscular dystrophy
- ◯ Distal myopathy
- ◯ Adult onset myopathy
- ◯ Metabolic myopathy
- ◯ Mitochondrial disease
- ◯ Skeletal muscle channelopathy
- ◯ Undetermined genetic neuromuscular or neurological disorder

(Clinical diagnosis most likely, in opinion of researcher)

Confirmed diagnosis

_____
(Orphanet)

Other confirmed diagnosis - 1

_____
(Orphanet)

Other confirmed diagnosis - 2

_____
(Orphanet)

Confirmed diagnoses - freetext

_____
(Offline entry)

Age clinically diagnosed - years

_____

Age clinically diagnosed - months

_____

Pre-existing samples available

Pre-existing samples available for genetic analysis
◯ No
◯ Yes
◯ Unknown

DNA
◯ No
◯ Yes
◯ Unknown

Blood
◯ No
◯ Yes
◯ Unknown

Skin tissue/fibroblast
◯ No
◯ Yes
◯ Unknown

Muscle tissue
◯ No
◯ Yes
◯ Unknown

Buccal
◯ No
◯ Yes
◯ Unknown

Saliva
◯ No
◯ Yes
◯ Unknown

Urine
◯ No
◯ Yes
◯ Unknown

New samples collected

New samples collected at study visit
◯ No
◯ Yes
◯ Unknown

DNA
◯ No
◯ Yes
◯ Unknown

Blood
◯ No
◯ Yes
◯ Unknown

Skin tissue/fibroblast	◯ No ◯ Yes ◯ Unknown
Muscle tissue	◯ No ◯ Yes ◯ Unknown
Buccal	◯ No ◯ Yes ◯ Unknown
Saliva	◯ No ◯ Yes ◯ Unknown
Urine	◯ No ◯ Yes ◯ Unknown

Demographics

Current age - years	_____
Current age - months	_____
Sex	◯ Male ◯ Female (Biological sex at birth)
Proband year of birth	_____

Ethnicity (self-reported)

Country of birth	◯ Brazil ◯ India ◯ South Africa ◯ Turkey ◯ United Kingdom ◯ Zambia ◯ Other
Country of birth - other	_____
Region/province of birth	_____
Main language	_____

Ethnicity

_____
(Please describe the ethnic origin)

Ethnicity - subgroup

_____
((if applicable))

Father's ethnicity

_____
(Please describe the paternal ethnic origin)

Father's ethnicity - subgroup

_____
((if applicable))

Mother's ethnicity

_____
(Please describe the maternal ethnic origin)

Mother's ethnicity - subgroup

_____
((if applicable))

Brazil

Ethnicity

◯ White
◯ Amerindians
◯ Pardo-Brazilians
◯ African-Brazilians
◯ Asian-Brazilians
◯ Other

Ethnicity - other

_____
(Please describe the ethnic origin)

Ethnicity - subgroup

_____
((if applicable))

Father's ethnicity

◯ White
◯ Amerindians
◯ Pardo-Brazilians
◯ African-Brazilians
◯ Asian-Brazilians
◯ Other

Father's ethnicity - other

_____
(Please describe the paternal ethnic origin)

Father's ethnicity - subgroup

_____
((if applicable))

Mother's ethnicity	◯ White ◯ Amerindians ◯ Pardo-Brazilians ◯ African-Brazilians ◯ Asian-Brazilians ◯ Other
Mother's ethnicity - other	_____ (Please describe the maternal ethnic origin)
Mother's ethnicity - subgroup	_____ ((if applicable))

India

Ethnicity - tribe	_____
Ethnicity - other	_____ (Please describe the ethnic origin)
Ethnicity - subgroup	_____ ((if applicable))
Father's ethnicity - tribe	_____
Father's ethnicity - other	_____ (Please describe the paternal ethnic origin)
Father's ethnicity - subgroup	_____ ((if applicable))
Mother's ethnicity - tribe	_____
Mother's ethnicity - other	_____ (Please describe the maternal ethnic origin)
Mother's ethnicity - subgroup	_____ ((if applicable))

South Africa

Ethnicity

◯ Black African
◯ Mixed ancestry
◯ White
◯ Indian/Asian
◯ Other

Ethnicity - other

_____
(Please describe the ethnic origin)

Ethnicity - subgroup

_____
((if applicable))

Father's ethnicity

◯ Black African
◯ Mixed ancestry
◯ White
◯ Indian/Asian
◯ Other

Father's ethnicity - other

_____
(Please describe the paternal ethnic origin)

Father's ethnicity - subgroup

_____
((if applicable))

Mother's ethnicity

◯ Black African
◯ Mixed ancestry
◯ White
◯ Indian/Asian
◯ Other

Mother's ethnicity - other

_____
(Please describe the maternal ethnic origin)

Mother's ethnicity - subgroup

_____
((if applicable))

Turkey

Ethnicity

◯ Turkish
◯ Kurdish
◯ Other

Ethnicity - other

_____
(Please describe the ethnic origin)

Ethnicity - subgroup

_____
((if applicable))

Father's ethnicity

◯ Turkish
◯ Kurdish
◯ Other

Father's ethnicity - other

_____
(Please describe the paternal ethnic origin)

Father's ethnicity - subgroup

_____
((if applicable))

Mother's ethnicity

◯ Turkish
◯ Kurdish
◯ Other

Mother's ethnicity - other

_____
(Please describe the maternal ethnic origin)

Mother's ethnicity - subgroup

_____
((if applicable))

United Kingdom

Ethnicity

◯ White British
◯ White Irish
◯ Gypsy or Irish Traveller
◯ Any other White background
◯ White and Black Caribbean
◯ White and Black African
◯ White and Asian
◯ Any other Mixed / Multiple ethnic background
◯ Indian
◯ Pakistani
◯ Bangladeshi
◯ Chinese
◯ Any other Asian background
◯ African
◯ Caribbean
◯ Any other Black / African / Caribbean background
◯ Arab
◯ Any other ethnic group
◯ Unknown

Ethnicity - other

_____
(Please describe the ethnic origin)

Ethnicity - subgroup

_____
((if applicable))

Father's ethnicity

◯ White British
◯ White Irish
◯ Gypsy or Irish Traveller
◯ Any other White background
◯ White and Black Caribbean
◯ White and Black African
◯ White and Asian
◯ Any other Mixed / Multiple ethnic background
◯ Indian
◯ Pakistani
◯ Bangladeshi
◯ Chinese
◯ Any other Asian background
◯ African
◯ Caribbean
◯ Any other Black / African / Caribbean background
◯ Arab
◯ Any other ethnic group
◯ Unknown

Father's ethnicity - other

_____
(Please describe the paternal ethnic origin)

Father's ethnicity - subgroup

_____
((if applicable))

Mother's ethnicity

◯ White British
◯ White Irish
◯ Gypsy or Irish Traveller
◯ Any other White background
◯ White and Black Caribbean
◯ White and Black African
◯ White and Asian
◯ Any other Mixed / Multiple ethnic background
◯ Indian
◯ Pakistani
◯ Bangladeshi
◯ Chinese
◯ Any other Asian background
◯ African
◯ Caribbean
◯ Any other Black / African / Caribbean background
◯ Arab
◯ Any other ethnic group
◯ Unknown

Mother's ethnicity - other

_____
(Please describe the maternal ethnic origin)

Mother's ethnicity - subgroup

_____
((if applicable))

Zambia

Ethnicity

◯ Black African
◯ Mixed ancestry
◯ White
◯ Indian/Asian
◯ Other

Ethnicity - other

_____
(Please describe the ethnic origin)

Ethnicity - subgroup

_____
((if applicable))

Father's ethnicity

◯ Black African
◯ Mixed ancestry
◯ White
◯ Indian/Asian
◯ Other

Father's ethnicity - other

_____
(Please describe the paternal ethnic origin)

Father's ethnicity - subgroup

_____
((if applicable))

Mother's ethnicity

◯ Black African
◯ Mixed ancestry
◯ White
◯ Indian/Asian
◯ Other

Mother's ethnicity - other

_____
(Please describe the maternal ethnic origin)

Mother's ethnicity - subgroup

_____
((if applicable))

Known genes in VCF format - please use GRCh38 (e.g. 1 230710048 rs699 A G)

Known gene mutation - VCF

_____
(1 230710048 rs699 A G)

Known gene mutation - OMIM

_____
(Optional)

Second known gene mutation - VCF

_____
(1 230710048 rs699 A G)

Second known gene mutation - OMIM

(Optional)

Third known gene mutation - VCF

(1 230710048 rs699 A G)

Third known gene mutation - OMIM

(Optional)

Fourth known gene mutation - VCF

(1 230710048 rs699 A G)

Fourth known gene mutation - OMIM

(Optional)

Fifth known gene mutation - VCF

(1 230710048 rs699 A G)

Fifth known gene mutation - OMIM

(Optional)

Known mutation(s) - freetext

Family history

Family history of core disease

- ◯ Absent
- ◯ Present
- ◯ Unknown

Family history of core disease - details

Consanguinity

- ◯ Absent
- ◯ Present
- ◯ Unknown

Consanguinity - description

(E.g. 1st, 2nd, 3rd cousin)

Known mutation(s) in family - freetext

(Offline entry)

Upload pedigree

(Online tool for drawing pedigree: https://www.progenygenetics.com/online-pedigree/)

Inheritance

Inheritance

◯ Autosomal dominant
◯ Autosomal recessive
◯ X-linked dominant
◯ X-linked recessive
◯ Matrilineal
◯ Familial (unspecified inheritance)
◯ Idiopathic/sporadic
◯ Unknown

Symptoms

First symptom noticed by subject or parents/carers

_____

First symptom noticed by subject or parents/carers - freetext

_____
(Offline entry)

Age of onset symptoms - years

_____
(If present at birth, enter 0 as value)

Age of onset symptoms - months

_____
(If present at birth, enter 0 as value)

Disease type

Progression

◯ Progressive
◯ Non-progressive
◯ Other or unspecified
◯ Unknown

Global pace of progression

◯ Days
◯ Months
◯ Years

Characteristics

◯ Typical
◯ Atypical
◯ Other or unspecified
◯ Unknown

Persistence

◯ Chronic
◯ Episodic/intermittent
◯ Other or unspecified
◯ Unknown

Complexity

◯ Pure (uncomplicated)
◯ Complicated
◯ Other or unspecified
◯ Unknown

Disease type - comments

Present phenotypic features (HPO) - please enter at least five

Present feature - 1

Present feature - 2

Present feature - 3

Present feature - 4

Present feature - 5

Present feature - 6

Present feature - 7

Present feature - 8

Present feature - 9

Present feature - 10

Present feature - 11

Present feature - 12

Present feature - 13

Present feature - 14

Present feature - 15

Present feature - 16

Present feature - 17

Present feature - 18

Present feature - 19

Present features - freetext

(Offline entry)

Absent phenotypic features (HPO) - please enter at least five

Absent feature - 1

Absent feature - 2

Absent feature - 3

Absent feature - 4

Absent feature - 5

Absent feature - 6

Absent feature - 7

Absent feature - 8

Absent feature - 9

Absent feature - 10

Absent feature - 11

Absent feature - 12

Absent feature - 13

Absent feature - 14

Absent feature - 15

Absent feature - 16

Absent feature - 17

Absent feature - 18

Absent feature - 19

Absent features - freetext

(Offline entry)

Comments

Any other comments on core data

Other Medical History

Handedness

Handedness	◯ Left-handed ◯ Right-handed ◯ Both hands/Ambidextrous ◯ Unknown (The hand used predominately; not necessarily the hand used for writing with exclusively)

Smoking history

Smoking status	◯ Current tobacco/cigarette smoker (including non-daily smoker) ◯ Ex-smoker ◯ Never smoker (not smoked for more than a year in total) ◯ Unknown
Frequency of smoking	◯ Non-daily ◯ Daily
Age smoking regularly	_____ (At what age (years) did the subject start smoking regularly?)
Cigarette consumption / day	_____ (If the subject smoked a pipe, rolling tobacco, cigar or similar then ** g of tobacco = 1 cigarette)
Age quit smoking	_____ (At what age (years) did the subject quit smoking?)
Smoking - comments	_____ (Any other comments on smoking history. Also insert any comments about (i) chewing tobacco and/or (ii) betel leaf/areca nut use here.)

Alcohol history

Does the participant currently consume alcohol? (within the past year)	◯ No ◯ Yes ◯ Unknown
Has the participant consumed alcohol in the past? (prior to the past year)	◯ No ◯ Yes ◯ Unknown

At what age did the participant start consuming alcohol?	_____
At what age did the participant stop consuming alcohol (if applicable)?	_____
How often does the participant consume alcohol? If the subject no longer consumes alcohol, how often did they previously consume alcohol?	◯ 4 or more times per week ◯ 2-3 times per week ◯ 2-4 times per month ◯ Monthly or less ◯ N/A
How many units does the participant consume on a typical day when drinking? If the participant no longer consumes alcohol, how many units did they previously consume?	◯ 1-2 ◯ 3-4 ◯ 5-6 ◯ 7-9 ◯ 10+ ◯ N/A
How often does the participant have 6 or more alcoholic beverages on one occasion? If the participant no longer consumes alcohol, how often did they previously consume 6 or more alcoholic beverages on one occasion?	◯ Daily or almost daily ◯ Weekly ◯ Monthly ◯ Less than monthly ◯ Never
Has the participant ever been hospitalised for any alcohol-related problem? (e.g. oesophageal varices, delirium, tremors, cirrhosis, etc.)	◯ No ◯ Yes ◯ Unknown
Hospitalisation for alcohol - description	_____ (Please describe)

Medication

Medication(s) taken	_____ (Start/end dates. Generic drug name(s) and mean daily dosage(s) in mg.)
Describe other treatment given	_____

Prenatal, perinatal, or early developmental history

Prenatal, perinatal, or early developmental history available	◯ No ◯ Yes

Prenatal history

In-utero movements
- ◯ Normal
- ◯ Reduced
- ◯ Unknown

Polyhydramnios
- ◯ No
- ◯ Yes
- ◯ Unknown

Maternal risk factors

_____

Perinatal history

Gestation period in weeks

_____

Birth - delivery
- ◯ Normal
- ◯ C-section (elective)
- ◯ C-section (emergency)
- ◯ Assisted (ventouse/forceps)

APGAR score(s) available?
- ◯ No
- ◯ Yes
- ◯ Unknown

APGAR - 1 minute - score

_____
((0-10))

APGAR - 5 minutes - score

_____
((0-10))

APGAR - 10 minutes - score

_____
((0-10))

Birth length in centimetres

_____

Birth weight in kg

_____

Birth head circumference in centimetres

_____

Neonatal complications
- ◯ No
- ◯ Yes
- ◯ Unknown

Neonatal complications - type

- ☐ Contractures
- ☐ Fractures
- ☐ Poor feeding
- ☐ Neonatal encephalopathy
- ☐ Required ventilatory support
- ☐ Other

Neonatal encephalopathy - severity

- ◯ Mild
- ◯ Moderate
- ◯ Severe

Infancy and early childhood developmental history

Motor development normal or abnormal

- ◯ Normal
- ◯ Abnormal
- ◯ Unknown

Age at sitting - months

_____
(If never developed, enter 0)

Age at crawling - months

_____
(If never developed, enter 0)

Age at walking - months

_____
(If never developed, enter 0)

Speech and hearing development

- ◯ Normal
- ◯ Abnormal
- ◯ Unknown

Age at first words - months

_____
(If never developed, enter 0)

Age using two-word phrases - months

_____
(If never developed, enter 0)

Social development

- ◯ Normal
- ◯ Abnormal
- ◯ Unknown

Age overcoming stranger anxiety - months

_____
(If never developed, enter 0)

Age waving bye bye - months

_____
(If never developed, enter 0)

Past medical history

Past medical history

(Including any other medical conditions and details of any environmental toxin exposure)

Past surgical history

Past surgical history

Comments

Any other comments on other medical history

Clinical Assessment

Examination normal or abnormal	◯ Normal ◯ Abnormal ◯ Unknown
Have any aspects of patient examination been video recorded or photographed?	◯ No ◯ Yes ◯ Unknown
If yes, which aspects	☐ Gait ☐ Abnormal movements ☐ Seizures ☐ Cranial nerve examiation ☐ Limb examination - motor ☐ Limb examination - sensory ☐ Other
Please describe other aspect(s)	_____

Cardiac dysfunction

Cardiac dysfunction	◯ No ◯ Yes ◯ Unknown
Cardiac dysfunction - type	☐ Conduction defects ☐ Cardiomyopathy ☐ Other
Cardiac dysfunction - type - other	_____
Cardiomyopathy - type	◯ Dilated cardiomyopathy ◯ Hypertrophic cardiomyopathy ◯ Other
Cardiomyopathy type - other	_____
Age at onset of cardiac dysfunction - years	_____ (If present at birth, enter 0 as value)
Age at onset of cardiac dysfunction - months	_____ (If present at birth, enter 0 as value)
Cardiac intervention	◯ No ◯ Yes ◯ Unknown

Cardiac intervention - type	☐ Pacemaker ☐ ICD ☐ Cardiac transplant ☐ Other
Cardiac intervention - type - other	_____
Age at intervention - years	_____
Age at intervention - months	_____

Respiratory dysfunction

Respiratory dysfunction	◯ No ◯ Yes ◯ Unknown
Respiratory dysfunction - type	☐ Neuromuscular ☐ Cardiac ☐ Pulmonary ☐ Central ☐ Other
Respiratory dysfunction - other	_____
Age at onset of respiratory dysfunction - years	_____ (If present at birth, enter 0 as value)
Age at onset of respiratory dysfunction - months	_____ (If present at birth, enter 0 as value)
FVC tested	◯ No ◯ Yes ◯ Unknown
FVC tested - value - 1	_____ (% of predicted value)
FVC tested - age in years - 1	_____ (Years)
FVC tested - position - 1	◯ Sitting ◯ Lying
FVC tested - value - 2	_____ (% of predicted value)

FVC tested - age in years - 2

_____
(Years)

FVC tested - position - 2

◯ Sitting
◯ Lying

FVC tested - value - 3

_____
(% of predicted value)

FVC tested - age in years - 3

_____
(Years)

FVC tested - position - 3

◯ Sitting
◯ Lying

PEFR tested

◯ No
◯ Yes
◯ Unknown

PEFR tested - value - 1

_____
(% of predicted value)

PEFR tested - age in years - 1

_____
(Years)

PEFR tested - position - 1

◯ Sitting
◯ Lying

PEFR tested - value - 2

_____
(% of predicted value)

PEFR tested - age in years - 2

_____
(Years)

PEFR tested - position - 2

◯ Sitting
◯ Lying

PEFR tested - value - 3

_____
(% of predicted value)

PEFR tested - age in years - 3

_____
(Years)

PEFR tested - position - 3

◯ Sitting
◯ Lying

Artificial ventilation	◯ No ◯ Yes ◯ Unknown (Ever used)
Daily duration of artificial ventilation	◯ All day ◯ Day only ◯ Night only ◯ Only with infections ◯ Unknown
Artificial ventilation - type	◯ NIV ◯ CPAP ◯ Tracheostomy ◯ Other
Artificial ventilation type - other	_____
Artificial ventilation started age - years	_____
Artificial ventilation started age - months	_____
Artificial ventilation stopped age - years	_____ (If applicable)
Artificial ventilation stopped age - months	_____ (If applicable)

Seizures

Seizures	◯ No ◯ Yes ◯ Unknown
Seizures - type	◯ Focal onset ◯ Generalised onset ◯ Unknown onset
Seizures - age at onset - years	_____ (If present at birth, enter 0 as value)
Seizures - age at onset - months	_____ (If present at birth, enter 0 as value)

Movement disorder

Movement disorder
- ◯ No
- ◯ Yes
- ◯ Unknown

Movement disorder - type
- ☐ Cerebellar ataxia
- ☐ Chorea
- ☐ Dystonia
- ☐ Tremor
- ☐ Other

Movement disorder type - other

_____

Movement disorder - distribution
- ☐ UL proximal
- ☐ UL distal
- ☐ LL proximal
- ☐ LL distal
- ☐ Ocular
- ☐ Facial
- ☐ Oromandibular
- ☐ Cervical

Movement disorder - age at onset - years

_____
(If present at birth, enter 0 as value)

Movement disorder - age at onset - months

_____
(If present at birth, enter 0 as value)

Autonomic dysfunction

Autonomic dysfunction
- ◯ No
- ◯ Yes
- ◯ Unknown

Autonomic dysfunction - type
- ☐ Hyperhidosis
- ☐ Constipation
- ☐ Cardiac dysfunction
- ☐ Bladder dysfunction
- ☐ Erectile dysfunction
- ☐ Other

Autonomic dysfunction type - other

_____

Autonomic dysfunction - age at onset - years

_____
(If present at birth, enter 0 as value)

Autonomic dysfunction - age at onset - months

_____
(If present at birth, enter 0 as value)

Cognitive

Cognitive dysfunction	◯ No ◯ Yes ◯ Unknown
Cognitive dysfunction - type	☐ Global developmental delay (< 5 years) ☐ Intellectual disability (> 5 years) ☐ Learning disabilities ☐ Dementia ☐ Behavioural change ☐ Other
Learning disabilities - type	☐ Dyscalculea ☐ Dyspraxia ☐ Dyslexia
Cognitive dysfunction type - other	_____
Cognitive dysfunction - course	◯ Static ◯ Improving ◯ Regression ◯ Unknown
Cognitive dysfunction - age at onset - years	_____ (If present at birth, enter 0 as value)
Cognitive dysfunction - age at onset - months	_____ (If present at birth, enter 0 as value)
Neuropsychometry performed	◯ No ◯ Yes ◯ Unknown
Neuropsychometry - report	(Upload/view Neuropsychometry report)
Neuropsychiatric disorders	◯ No ◯ Yes ◯ Unknown
Neuropsychiatric disorders- type	☐ Autistic spectrum disorder ☐ ADHD ☐ Psychosis ☐ Depression ☐ Other
Neuropsychiatric disorders - type - other	_____
Neuropsychiatric disorders - age at onset - years	_____ (If present at birth, enter 0 as value)

Neuropsychiatric disorders - age at onset - months

_____
(If present at birth, enter 0 as value)

Gait

Motor ability

◯ Normal for age
◯ Walks without restrictions, with limitations for running and jumping
◯ Walks in most settings, may use physical assistance and climb stairs holding onto a railing
◯ Walks using a hand-held mobility device, use wheeled mobility for long distances
◯ Use methods of mobility that require physical assistance or powered mobility in most settings
◯ Transported in a manual wheelchair in all settings
(Gross motor function)

Diminished motor ability - age at onset - years

_____
(If present at birth, enter 0 as value)

Diminished motor ability - age at onset - months

_____
(If present at birth, enter 0 as value)

Lost ambulation - age at onset - years

_____
(If present at birth, enter 0 as value)

Lost ambulation - age at onset - months

_____
(If present at birth, enter 0 as value)

6 min walk test - distance in metres

_____

Dysmorphic features

Dysmorphic features

◯ No
◯ Yes
◯ Unknown

Dysmorphic features - description

_____

Dysmorphic features - age of onset - years

_____
(If present at birth, enter 0 as value)

Dysmorphic features - age of onset - months

_____
(If present at birth, enter 0 as value)

Dysmorphic features - photo

(Upload/view Photo of dysmorphic features)

Cranial nerves

Visual acuity
◯ Normal
◯ Abnormal
◯ Unknown

Visual acuity - right eye
_____
(E.g. "6/6" (Snellen), "0.0" (LogMAR) or "20/20")

Visual acuity - left eye
_____
(E.g. "6/6" (Snellen), "0.0" (LogMAR) or "20/20")

Visual impairment
◯ No
◯ Yes
◯ Unknown

Visual impairment - type
◯ Sight impaired
◯ Severely sight impaired or blind

Ocular signs
◯ No
◯ Yes
◯ Unknown

Ocular signs - type
☐ Ptosis
☐ Opthalmoparesis
☐ INO
☐ Gaze palsy
☐ Nystagmus
☐ Cataracts
☐ Glaucoma
☐ Optic atrophy
☐ Structural eye abnormalities
☐ Squint
☐ Other

Ocular signs type - other
_____

Ocular signs - age at onset - years
_____
(If present at birth, enter 0 as value)

Ocular signs - age at onset - months
_____
(If present at birth, enter 0 as value)

Facial weakness
◯ No
◯ Yes
◯ Unknown

Facial weakness - age at onset - years
_____
(If present at birth, enter 0 as value)

Facial weakness - age at onset - months	_____ (If present at birth, enter 0 as value)
Bulbar signs	◯ No ◯ Yes ◯ Unknown
Bulbar signs - type	☐ Dysarthria ☐ Dysphagia ☐ Brisk jaw jerk ☐ Tongue fasciculations ☐ Tongue stiff ☐ Tongue weak ☐ Tongue large ☐ Neck weakness ☐ Other
Bulbar signs type - other	_____
Bulbar signs - age at onset - years	_____ (If present at birth, enter 0 as value)
Bulbar signs - age at onset - months	_____ (If present at birth, enter 0 as value)
Percutaneous Endoscopic Gastrostomy (PEG)	◯ No ◯ Yes ◯ Refused ◯ Unknown (Ever used)
Feeding orally with PEG?	◯ No ◯ Yes ◯ Unknown
Percutaneous Endoscopic Gastrostomy (PEG) - age at insertion - years	_____
Percutaneous Endoscopic Gastrostomy (PEG) - age at insertion - months	_____
Percutaneous Endoscopic Gastrostomy (PEG) - age at removal - years	_____ (If applicable)
Percutaneous Endoscopic Gastrostomy (PEG) - age at removal - months	_____ (If applicable)
Nasogastric (NG) tube	◯ No ◯ Yes ◯ Unknown (Ever used)

Nasogastric (NG) tube - age at insertion - years	_____
Nasogastric (NG) tube - age at insertion - months	_____
Nasogastric (NG) tube - age at removal - years	_____ (If applicable)
Nasogastric (NG) tube - age at removal - months	_____ (If applicable)
Hearing loss	◯ No ◯ Yes ◯ Unknown
Hearing loss - type	◯ Conductive ◯ Sensorineural ◯ Unknown
Hearing loss - age of onset - years	_____ (If present at birth, enter 0 as value)
Hearing loss - age of onset - months	_____ (If present at birth, enter 0 as value)

Limbs and trunk

Scapular winging	◯ No ◯ Yes ◯ Unknown
Scapular winging - distribution	◯ Left ◯ Right ◯ Bilateral
Scapular winging - age at onset - years	_____ (If present at birth, enter 0 as value)
Scapular winging - age at onset - months	_____ (If present at birth, enter 0 as value)
Spinal abnormalities	◯ No ◯ Yes ◯ Unknown
Spinal abnormalities - type	☐ Kyphosis ☐ Scoliosis ☐ Hyperlordosis

Spinal abnormalities - age at onset - years

_____
(If present at birth, enter 0 as value)

Spinal abnormalities - age at onset - months

_____
(If present at birth, enter 0 as value)

Contractures

◯ No
◯ Yes
◯ Unknown

Contractures - distribution

☐ UL
☐ LL
☐ Axial/rigid spine

Contractures distribution - UL joints

☐ Left fingers
☐ Right fingers
☐ Left wrist
☐ Right wrist
☐ Left elbow
☐ Right elbow
☐ Left shoulder
☐ Right shoulder

Contractures distribution - LL joints

☐ Left toes
☐ Right toes
☐ Left ankle
☐ Right ankle
☐ Left knee
☐ Right knee
☐ Left hip
☐ Right hip

Contractures - age at onset - years

_____
(If present at birth, enter 0 as value)

Contractures - age at onset - months

_____
(If present at birth, enter 0 as value)

Joint laxity/hypermobility

◯ No
◯ Yes
◯ Unknown

Joint laxity/hypermobility - distribution

☐ UL
☐ LL
☐ Axial

Joint laxity/hypermobility distribution - UL joints

☐ Left fingers
☐ Right fingers
☐ Left wrist
☐ Right wrist
☐ Left elbow
☐ Right elbow
☐ Left shoulder
☐ Right shoulder

Joint laxity/hypermobility distribution - LL joints	☐ Left toes ☐ Right toes ☐ Left ankle ☐ Right ankle ☐ Left knee ☐ Right knee ☐ Left hip ☐ Right hip
Joint laxity/hypermobility - age at onset - years	_____ (If present at birth, enter 0 as value)
Joint laxity/hypermobility - age at onset - months	_____ (If present at birth, enter 0 as value)
Webbing/pterygium	◯ No ◯ Yes ◯ Unknown
Webbing/pterygium - distribution	☐ Hands ☐ Feet
Pes cavus	◯ No ◯ Yes ◯ Unknown
Pes cavus - age at onset - years	_____ (If present at birth, enter 0 as value)
Pes cavus - age at onset - months	_____ (If present at birth, enter 0 as value)
Pes planus	◯ No ◯ Yes ◯ Unknown
Pes planus - age at onset - years	_____ (If present at birth, enter 0 as value)
Pes planus - age at onset - months	_____ (If present at birth, enter 0 as value)
Equinovarus	◯ No ◯ Yes ◯ Unknown
Equinovarus - age at onset - years	_____ (If present at birth, enter 0 as value)

Equinovarus - age at onset - months	_____ (If present at birth, enter 0 as value)
Amputation(s)	◯ No ◯ Yes ◯ Unknown
Amputation - which limb(s) or toe(s)	_____
Ulcers	◯ No ◯ Yes ◯ Unknown
Ulcers - distribution	☐ UL ☐ LL
Ulcers - age at onset - years	_____ (If present at birth, enter 0 as value)
Ulcers - age at onset - months	_____ (If present at birth, enter 0 as value)
Muscle bulk	◯ Normal ◯ Abnormal ◯ Unknown
Muscle atrophy/wasting	◯ No ◯ Yes ◯ Unknown
Muscle atrophy/wasting - distribution	☐ UL proximal ☐ UL distal ☐ LL proximal ☐ LL distal ☐ Axial
Muscle atrophy/wasting - age at onset - years	_____ (If present at birth, enter 0 as value)
Muscle atrophy/wasting - age at onset - months	_____ (If present at birth, enter 0 as value)
Muscle hypertrophy	◯ No ◯ Yes ◯ Unknown

Muscle hypertrophy - distribution	☐ UL proximal ☐ UL distal ☐ LL proximal ☐ LL distal ☐ Neck ☐ Facial ☐ Paraspinal ☐ Scapular
Muscle hypertrophy - age at onset - years	_____ (If present at birth, enter 0 as value)
Muscle hypertrophy - age at onset - months	_____ (If present at birth, enter 0 as value)
Muscle weakness	◯ No ◯ Yes ◯ Unknown
Muscle weakness - distribution	☐ UL proximal ☐ UL distal ☐ LL proximal ☐ LL distal ☐ Axial ☐ Symmetrical ☐ Asymmetrical
Muscle weakness - LL proximal compartment(s)	☐ Anterior compartment ☐ Posterior compartment
Muscle weakness - LL distal compartment(s)	☐ Anterior compartment ☐ Posterior compartment
Muscle weakness - age at onset - years	_____ (If present at birth, enter 0 as value)
Muscle weakness - age at onset - months	_____ (If present at birth, enter 0 as value)
Fatigability	◯ No ◯ Yes ◯ Unknown
Fatigability - type	◯ Exertion ◯ Diurnal
Fatigability - distribution	☐ UL ☐ LL ☐ Ptosis ☐ Extraocular ☐ Cervical ☐ Bulbar

Fatigability - age at onset - years	_____ (If present at birth, enter 0 as value)
Fatigability - age at onset - months	_____ (If present at birth, enter 0 as value)
Myotonia	◯ No ◯ Yes ◯ Unknown
Myotonia - distribution	☐ UL proximal ☐ UL distal ☐ LL proximal ☐ LL distal ☐ Eye closure ☐ Facial ☐ Cervical ☐ Paraspinal
Myotonia - age at onset - years	_____ (If present at birth, enter 0 as value)
Myotonia - age at onset - months	_____ (If present at birth, enter 0 as value)
Peripheral/lower motor neuron signs	◯ No ◯ Yes ◯ Unknown
Peripheral/lower motor neuron signs - type	☐ Reduced tone ☐ Reduced/absent reflexes
Peripheral/lower motor neuron signs reduced tone - distribution	☐ RUL ☐ LUL ☐ RLL ☐ LLL
Peripheral/lower motor neuron signs reduced/absent reflexes- distribution	☐ RUL ☐ LUL ☐ RLL ☐ LLL
Peripheral/lower motor neuron signs reduced/absent reflexes distribution - RUL	☐ Bicep ☐ Supinator ☐ Tricep
Peripheral/lower motor neuron signs reduced/absent reflexes distribution - LUL	☐ Bicep ☐ Supinator ☐ Tricep
Peripheral/lower motor neuron signs reduced/absent reflexes distribution - RLL	☐ Knee ☐ Ankle

Peripheral/lower motor neuron signs reduced/absent reflexes distribution - LLL	☐ Knee ☐ Ankle
Peripheral signs - age at onset - years	_____ (If present at birth, enter 0 as value)
Peripheral signs - age at onset - months	_____ (If present at birth, enter 0 as value)
Pyramidal signs	◯ No ◯ Yes ◯ Unknown
Pyramidal signs - type	☐ Spasticity ☐ Hyper-reflexia
Pyramidal signs spasticity - distribution	☐ RUL ☐ RLL ☐ LUL ☐ LLL ☐ Clonus
Pyramidal signs hyper-reflexia - distribution	☐ RUL ☐ LUL ☐ RLL ☐ LLL
Pyramidal signs hyper-reflexia distribution - RUL	☐ Bicep ☐ Supinator ☐ Tricep ☐ Finger flexors ☐ Hoffman's response
Pyramidal signs hyper-reflexia distribution - LUL	☐ Bicep ☐ Supinator ☐ Tricep ☐ Finger flexors ☐ Hoffman's response
Pyramidal signs hyper-reflexia distribution - RLL	☐ Knee ☐ Ankle ☐ Babinski
Pyramidal signs hyper-reflexia distribution - LLL	☐ Knee ☐ Ankle ☐ Babinski
Pyramidal signs - age at onset - years	_____ (If present at birth, enter 0 as value)
Pyramidal signs - age at onset - months	_____ (If present at birth, enter 0 as value)

Extrapyramidal signs	◯ No ◯ Yes ◯ Unknown
Extrapyramidal signs - type	☐ Rigidity ☐ Bradykinesia ☐ Tremor ☐ Loss of postural reflexes
Extrapyramidal signs - age of onset - years	_____ (If present at birth, enter 0 as value)
Extrapyramidal signs - age of onset - months	_____ (If present at birth, enter 0 as value)
Cerebellar signs	◯ No ◯ Yes ◯ Unknown
Cerebellar signs - type	☐ UL ☐ LL ☐ Gait ☐ Nystagmus ☐ Saccades ☐ Speech ☐ Tremor ☐ Trunk
Cerebellar signs - age at onset - years	_____ (If present at birth, enter 0 as value)
Cerebellar signs - age at onset - months	_____ (If present at birth, enter 0 as value)
Sensory loss	◯ Yes ◯ No ◯ Unknown
Sensory loss - age at onset - years	_____ (If present at birth, enter 0 as value)
Sensory loss - age at onset - months	_____ (If present at birth, enter 0 as value)
Pin prick	◯ Normal ◯ Abnormal ◯ Unknown
Pin prick - level and distribution	_____

Temperature	◯ Normal ◯ Abnormal ◯ Unknown
Temperature - level and distribution	_____
Vibration	◯ Normal ◯ Abnormal ◯ Unknown
Vibration - level and distribution	_____
Joint position sense	◯ Normal ◯ Abnormal ◯ Unknown
Joint position sense - level and distribution	_____
Joint position sense - pseudoathetosis	◯ No ◯ Yes ◯ Unknown

Hammersmith scale test

If the diagnosis is SMA Type 2 or 3, is a Hammersmith Scale result available?	◯ No ◯ Yes ◯ Unknown
Which Hammersmith Scale test was used? The Revised Hammersmith Scale (RHS) is preferred.	◯ Hammersmith Functional Motor Scale (HFMS) ◯ Modified Hammersmith Functional Motor Scale (MHFMS) ◯ Hammersmith Functional Motor Scale Expanded (HFMSE) ◯ Revised Hammersmith Scale (RHS) ◯ Modified Hammersmith Functional Motor Scale Extended (MHFMS-Extend) (http://www.smareachuk.org/smaoutcomemeasures/the-hammersmith-scale-what-is-it)
Hammersmith Scale test score	_____
Upload/view Hammersmith Scale test form	

North Star Ambulatory Assessment (NSAA)

If the diagnosis is DMD and participant is ambulatory and < 18 Years, is North Star Ambulatory Assessment (NSAA) result available?	◯ No ◯ Yes ◯ Unknown
Total NSAA score	_____ (0-34)
Upload/view NSAA test form	

Medical Research Council (MRC) sum score - used with the permission of the Medical Research Council

The total MRC sum score ranges from 0 (total paralysis) to 70 (normal strength). The score is the sum of the MRC score of 7 muscles (4 in the upper limbs and 3 in the lower limbs) on both sides, each muscle graded from 0 to 5.

Grade 5: Normal Strength
Grade 4: Ability to resist against moderate pressure throughout range of motion
Grade 3: Ability to move through full range of motion against gravity. If a subject has a contracture that limits joint movement, the mechanical range will be to the point at which the contracture causes joint restriction
Grade 2: Ability to move through full range of motion with gravity eliminated
Grade 1: A flicker of motion is seen or felt in the muscle
Grade 0: No movement

Upper limbs

	Grade 5	Grade 4	Grade 3	Grade 2	Grade 1	Grade 0
Right shoulder abduction (deltoid)	○	○	○	○	○	○
Right elbow flexion (biceps)	○	○	○	○	○	○
Right wrist extension (wrist extensors)	○	○	○	○	○	○
Right 1st DIO	○	○	○	○	○	○
Left shoulder abduction (deltoid)	○	○	○	○	○	○
Left elbow flexion (biceps)	○	○	○	○	○	○
Left wrist extension (wrist extensors)	○	○	○	○	○	○
Left 1st DIO	○	○	○	○	○	○

Lower limbs

	Grade 5	Grade 4	Grade 3	Grade 2	Grade 1	Grade 0
Right hip flexion (Ileopsoas)	○	○	○	○	○	○
Right knee extension (quadriceps femoris)	○	○	○	○	○	○
Right dorsiflexion foot (tibialis anterior)	○	○	○	○	○	○
Left hip flexion (Ileopsoas)	○	○	○	○	○	○
Left knee extension (quadriceps femoris)	○	○	○	○	○	○
Left dorsiflexion foot (tibialis anterior)	○	○	○	○	○	○

Total MRC sum score _____

CMTNS

Patient name: [subj_first_name] [subj_surname]

Date:

_____
(YYYY-MM-DD)

Evaluator:

_____

Sensory symptoms1

- ◯ None
- ◯ Symptoms below or at ankle bones
- ◯ Symptoms up to the distal half of the calf
- ◯ Symptoms up to the proximal half of the calf, including knee
- ◯ Symptoms above knee (above the top of the patella)

Motor symptoms legs2

- ◯ None
- ◯ Trips, catches toes, slaps feet. Shoe inserts
- ◯ Ankle support or stabilization (AFOs). Foot surgery5
- ◯ Walking aids (cane, walker)
- ◯ Wheelchair

Motor symptoms arms

- ◯ None
- ◯ Mild difficulty with buttons
- ◯ Severe difficulty or unable to do buttons
- ◯ Unable to cut most foods
- ◯ Proximal weakness (affect movements involving the elbow and above)

Pinprick sensibility1,3

- ◯ Normal
- ◯ Decreased below or at ankle bones
- ◯ Decreased up to the distal half of the calf
- ◯ Decreased up to the proximal half of the calf, including knee
- ◯ Decreased above knee (above the top of the patella)

Vibration4

- ◯ Normal
- ◯ Reduced at great toe
- ◯ Reduced at ankle
- ◯ Reduced at knee (tibial tuberosity)
- ◯ Absent at knee and ankle

Strength legs

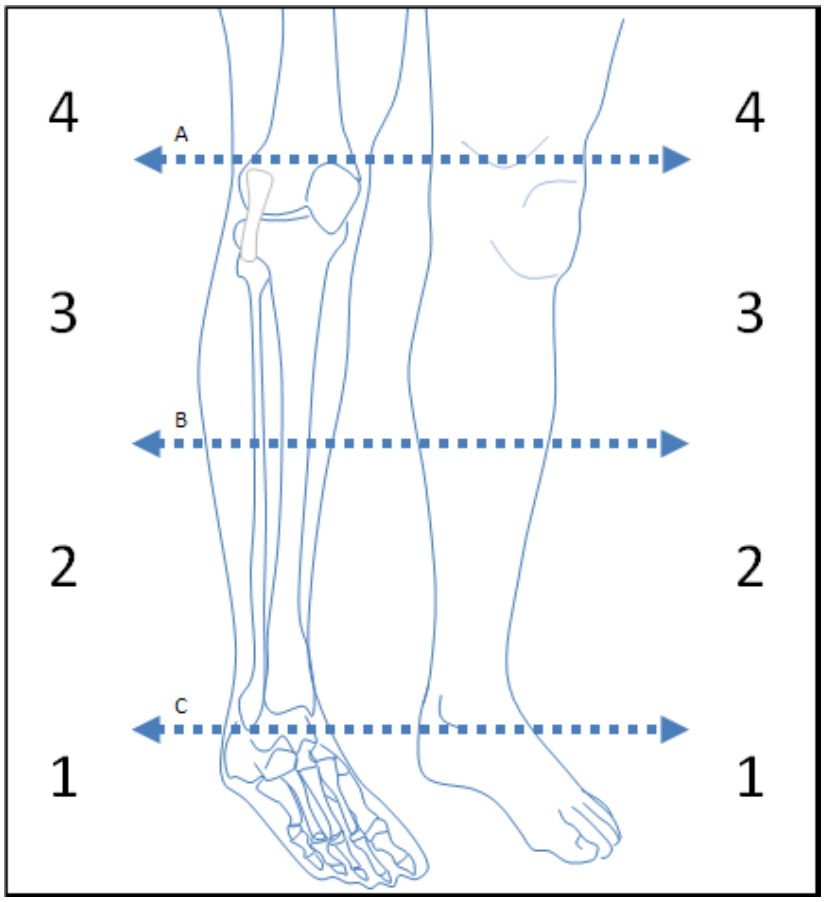
- ◯ Normal
- ◯ 4+,4 or 4- on foot dorsiflexion or plantar flexion
- ◯ < 3 on foot dorsiflexion or < 3 on foot plantar flexion
- ◯ < 3 on foot dorsi and < 3 on plantar flexion
- ◯ Proximal weakness

Strength arms

- ◯ Normal
- ◯ 4+,4 or 4- on intrinsic hand muscles5
- ◯ < 3 on intrinsic hand muscles6
- ◯ < 5 on wrist extensors
- ◯ Weak above elbow

Ulnar CMAP (Median)	◯ >6mV (>4mV) ◯ 4-5.9mV (2.8-3.9) ◯ 2-3.9 mV (1.2-2.7) ◯ 0.1-1.9 mV (0.1-1.1) ◯ Absent (Absent)
Radial SAP amplitude, antidromic	◯ >15 ?V ◯ 10 - 14.9 ?V ◯ 5 - 9.9 ?V ◯ 1 - 4.9 ?V ◯ < 1 ?V

Notes: 1: Use the picture below to discriminate the level of the symptoms; 2: Uses aid most of the time. The patient was prescribed to wear/use or should be wearing/using the aid in the examiners opinion (see written instructions); 3: Abnormal if patient says it is definitely decreased compared to a normal reference point; 4: Use Rydell Seiffer tuning fork. Definition of Normal: > 5; 5: See written instructions for details of eligible foot surgery; 6: Intrinsic hand muscles strength assessment: Test only Abductor Pollicis Brevis (ABP) and First Dorsal Interosseus (FDI), then choose the stronger to give the score.



CMTSS Subtotal	_____
CMTES Subtotal	_____

CMTNS result

CMTNS Total

_____

Patient details

Patient name: [subj_first_name] [subj_surname]

Age: [subj_age_yrs]

Gender: [subj_sex]

Raw scores

1. Functional Dexterity

_____
(seconds)

2. 9 Hole Peg

_____
(seconds)

3. Grip

_____
(Newtons)

4. Plantarflexion

_____
(Newtons)

5. Dorsiflexion

_____
(Newtons)

6. Pinprick

◯ Normal
◯ Decreased below or at ankle bones
◯ Decreased at or below midline of calf
◯ Decreased above calf midline up to and including knee
◯ Decreased above knee (above top of patella)

7. Vibration

◯ Normal
◯ Reduced at first metatarsal bone
◯ Reduced at ankle
◯ Reduced at knee (tibial tuberosity)
◯ Absent at knee and ankle

8. Balance

_____
(Bruininks Oseretsky (0-37))

9. Gait

Foot drop

◯ No
◯ Some
◯ Yes

Difficulty heel walking	◯ No ◯ Some ◯ Yes
Difficulty toe walking	◯ No ◯ Some ◯ Yes
10. Long Jump	_____ (centimetres)
11. 6 Minute Walk	_____ (metres)

CMTPedS result

Total CMTPedS Score	_____ (CMTPedS calculator: https://www.cmtpeds.org/)

Report

Participant CMTPedS report	(Upload/view CMTPedS report)

Comments

Any other comments on clinical assessment	_____

Follow-up comments

Comments from follow-up visits	_____ (For utility; not expected as part of the ICGNMD study (cross-sectional))

Investigations

EMG	
EMG	◯ Normal ◯ Abnormal ◯ Not done
EMG - abnormality	☐ Myopathic ☐ Neurogenic ☐ Neuromuscular transmission disorder ☐ Other
EMG abnormality - other	_____
EMG spontaneous activity	◯ No ◯ Yes ◯ Not done
EMG spontaneous activity - type	◯ Myotonic/pseudomyotonic ◯ Fibrillation ◯ Positive sharp waves ◯ Fasciculations
EMG - report	(Upload/view EMG report)
EMG images available	◯ No ◯ Yes ◯ Unknown
Nerve conduction studies	
Motor nerve conduction studies	◯ Normal ◯ Abnormal ◯ Not done
Motor nerve conduction studies - abnormality	◯ Axonal ◯ Demyelinating ◯ Mixed
Sensory nerve conduction studies	◯ Normal ◯ Abnormal ◯ Not done
Sensory nerve conduction studies - abnormality	◯ Axonal ◯ Demyelinating ◯ Mixed
Nerve conduction studies - report	(Upload/view Nerve conduction study report)

Echocardiogram

Echocardiogram	◯ Normal ◯ Abnormal ◯ Not done
Echocardiogram - report	(Upload/view Echocardiogram report)

MRI

Brain MRI	◯ Normal ◯ Abnormal ◯ Not done
Brain MRI - findings	☐ White matter abnormalities ☐ Lissencephaly ☐ Polymicrogyria ☐ Brainstem abnormalities ☐ Cerebral atrophy ☐ Basal ganglia atrophy ☐ Cerebellar atrophy ☐ Cerebellar dysplasia ☐ Cerebellar cysts ☐ Optic nerve abnormalities ☐ Other
Brain MRI - findings - other	_____
Brain MRI - report	(Upload/view Brain MRI report)
Spine MRI	◯ Normal ◯ Abnormal ◯ Not done
Spine MRI - report	(Upload/view Spine MRI report)
Limbs MRI	◯ Normal ◯ Abnormal ◯ Not done
Limbs MRI - report	(Upload/view Limbs MRI report)
MRI images available	◯ No ◯ Yes ◯ Unknown

Muscle ultrasound

Muscle ultrasound	◯ Normal ◯ Abnormal ◯ Not done
Muscle ultrasound - report	(Upload/view Muscle ultrasound report)

Nerve ultrasound

Nerve ultrasound	◯ Normal ◯ Abnormal ◯ Not done
Nerve ultrasound - report	(Upload/view Nerve ultrasound report)

Muscle biopsy

Muscle biopsy	◯ Normal ◯ Abnormal ◯ Not done
Suggestive of muscular dystrophy	◯ No ◯ Yes ◯ Unknown (Internalized nuclei, fibrosis, fibre size variation, rounded fibers.)
Muscular dystrophy - findings	☐ Alpha dystroglycanopathy ☐ Merosin (laminin 2) deficiency ☐ Collagen VI related myopathy ☐ Myofibrillar myopathy (desmin, myotilin, VCP abnormal staining) ☐ Caveolin deficiency ☐ Emerin deficiency ☐ Calpain deficiency ☐ Dystrophin deficiency ☐ Sarcoglycan deficiency ☐ Dysferlin deficiency ☐ Other
Muscular dystrophy - merosin deficiency	◯ Partial ◯ Complete ◯ Unknown
Muscular dystrophy findings - other	_____
Suggestive of congenital myopathy	◯ No ◯ Yes ◯ Unknown

Congenital myopathy - findings	☐ Central core disease ☐ Centronuclear/Myotubular myopathy ☐ Congenital fibre type disproportion ☐ Multiminicore myopathy ☐ Nemaline myopathy ☐ Other
Congenital myopathy findings - other	_____
Suggestive of mitochondrial disease	◯ No ◯ Yes ◯ Unknown
Mitochondrial disease - findings	☐ Ragged red fibres ☐ COX negative fibres ☐ COX/SDH positive fibres ☐ Other
Mitochondrial disease findings - other	_____
Mitochondrial disease - respiratory chain enzyme analysis	◯ No ◯ Yes ◯ Unknown
Mitochondrial disease respiratory chain enzyme analysis - findings	_____
Mitochondrial disease - respiratory chain enzyme analysis - report	(Upload/view Respiratory chain enzyme analysis report)
Additional histological findings	☐ Inflammatory infiltrates ☐ Myofibrillar changes ☐ Mitochondrial changes ☐ Rimmed vacuoles ☐ Protein aggregates ☐ Neurogenic change
Muscle biopsy - report	(Upload/view Muscle biopsy report)
Muscle biopsy slides available	◯ No ◯ Yes ◯ Unknown

Nerve biopsy

Nerve biopsy
- ◯ Normal
- ◯ Abnormal
- ◯ Not done

Nerve biopsy - report

(Upload/view Nerve biopsy report)

Nerve biopsy slides available
- ◯ No
- ◯ Yes
- ◯ Unknown

Other tissue studies

Other tissue studies
- ◯ No
- ◯ Yes
- ◯ Unknown

Other tissue studies done

_____

Other tissue studies - report

(Upload/view Other tissue studies report)

Creatine phosphokinase (CPK)

CPK tested
- ◯ No
- ◯ Yes
- ◯ Unknown

CPK result - 1

_____

CPK unit - 1

_____

CPK tested - age - years - 1

_____

CPK tested - age - months - 1

_____

CPK result - 2

_____

CPK unit - 2

_____

CPK tested - age - years - 2

_____

CPK tested - age - months - 2

_____

CPK result - 3

CPK unit - 3

CPK tested - age - years - 3

CPK tested - age - months - 3

Additional laboratory tests

Additional laboratory tests available

- ◯ No
- ◯ Yes
- ◯ Unknown

Additional laboratory tests available - type

- ☐ Alb
- ☐ ALP
- ☐ ALT
- ☐ Ammonia (plasma)
- ☐ Blood lactate
- ☐ Cr
- ☐ CrCl
- ☐ CSF lactate
- ☐ Fasting glucose level
- ☐ GGT
- ☐ Glu (random)
- ☐ Hb
- ☐ HbA1c
- ☐ K
- ☐ MCV
- ☐ Na
- ☐ Plt
- ☐ T4
- ☐ TSH
- ☐ Urea
- ☐ WCC
- ☐ Other

Alb

Alb - unit

ALP

ALP - unit

ALT

ALT - unit

Ammonia (plasma)

Ammonia (plasma) - unit

Blood lactate

Blood lactate - unit

Cr

Cr - unit

CrCl

CrCl - unit

CSF lactate

CSF lactate - unit

Fasting glucose level

Fasting glucose level - unit

GGT

GGT - unit

Glu (random)

Glu (random) - unit

Hb

Hb - unit

HbA1c

HbA1c - unit

K

K - unit

MCV

MCV - unit

Na

Na - unit

Plt

Plt - unit

T4

T4 - unit

TSH

TSH - unit

Urea

Urea - unit

WCC

WCC - unit

Lab test - other(s)

_____

HIV and hepatitis status

HIV status

◯ Negative
◯ Positive
◯ Unknown

HIV status - CD4 count

_____

HIV status - viral load

_____

Hep A status

◯ Negative
◯ Positive
◯ Unknown

Hep B status

◯ Negative
◯ Positive
◯ Unknown

Hep C status

◯ Negative
◯ Positive
◯ Unknown

Other known infectious diseases

List any other known infectious diseases

_____

Comments

Any other comments on investigations

_____

Non Icgnmd Genetic Testing

Have any of the following genetic tests been performed? (or are they currently being performed?)

	No	Yes, abnormal result (confirms diagnosis)	Yes, normal result	In progress	Unknown
SMA (SMN1) MLPA	○	○	○	○	○
DMD MLPA	○	○	○	○	○
DMD Sequencing	○	○	○	○	○
CMT1A MLPA (17p duplication analysis)	○	○	○	○	○
Myotonic dystrophy Type 1 testing	○	○	○	○	○
Myotonic dystrophy Type 2 testing	○	○	○	○	○
FSHD testing	○	○	○	○	○
Familial ALS (C9ORF72)	○	○	○	○	○
RYR-1 Screening for South African founder mutations	○	○	○	○	○
CANVAS	○	○	○	○	○

Please give details of result(s)

_____

Optional: Pseudonymised reports can be uploaded here

(Zip if more than one)

Record details of other local testing and local testing limitations here

_____

You can upload de-identified copies of non-ICGNMD genetic reports here (.pdf, .jpg, .png)

Are any of the following diagnostic tests required, but not available locally, for this participant?

- ☐ SMA (SMN1) MLPA
- ☐ DMD MLPA
- ☐ DMD Sequencing
- ☐ CMT1A MLPA (17p duplication analysis)
- ☐ Myotonic dystrophy Type 1 testing
- ☐ Myotonic dystrophy Type 2 testing
- ☐ FSHD testing
- ☐ Familial ALS (C9ORF72)
- ☐ RYR-1 Screening for South African founder mutations
- ☐ CANVAS

Has mitochondrial DNA testing been performed?

	No	Yes, abnormal result (confirms diagnosis)	Yes, normal result	In progress	Unknown
Common mitochondrial point mutations	◯	◯	◯	◯	◯
Whole mitochondrial genome sequencing	◯	◯	◯	◯	◯
mtDNA rearrangement analysis	◯	◯	◯	◯	◯
mtDNA depletion analysis	◯	◯	◯	◯	◯
Other	◯	◯	◯	◯	◯

Other - please specify test

_____

Please give details of result(s)

_____

Has any exome sequencing been performed (targeted exome/ gene panel, or whole exome sequencing)?

	No	Yes	In progress	Unknown
Whole Exome Sequencing as singleton	◯	◯	◯	◯
Whole Exome Sequencing duo	◯	◯	◯	◯
Whole Exome Sequencing trio or larger	◯	◯	◯	◯
Gene Panel	◯	◯	◯	◯
Targeted Exome	◯	◯	◯	◯

Please give details e.g. positive results or family members included. Panel content may also be listed here.

_____

Are any other diagnostic genetic tests outstanding?

◯ No
◯ Yes
◯ Unknown

Please give details

_____

Personal Details

Personal details	
Clinic/lab/local study subject ID	_____ (Primary identifier at host)
Initials	_____
Year of birth	_____

Sample Tracking

SENDER TO COMPLETE

Samples shipped for genetic analysis
◯ No
◯ Yes

Sample type(s) shipped

DNA
◯ No
◯ Yes

Number of DNA sample tubes sent
◯ 1
◯ 2
◯ 3
◯ 4

Blood
◯ No
◯ Yes

Number of blood sample tubes sent
◯ 1
◯ 2
◯ 3
◯ 4

Skin tissue/fibroblast
◯ No
◯ Yes

Number of skin tissue sample tubes sent
◯ 1
◯ 2
◯ 3
◯ 4

Muscle tissue
◯ No
◯ Yes

Number of muscle tissue sample tubes sent
◯ 1
◯ 2
◯ 3
◯ 4

Buccal
◯ No
◯ Yes

Number of buccal sample tubes sent
◯ 1
◯ 2
◯ 3
◯ 4

Saliva
◯ No
◯ Yes

Number of saliva sample tubes sent	◯ 1 ◯ 2 ◯ 3 ◯ 4
Urine	◯ No ◯ Yes
Number of urine sample tubes sent	◯ 1 ◯ 2 ◯ 3 ◯ 4
Date samples sent	_____ (YYYY-MM-DD)
Courier tracking ID	_____
Upload/view photo of sample	(Photo of 1 x sample tube showing ICGNMD Study ID on tube to be shipped)
Upload/view sample manifest	
Sender comments	_____

RECIPIENT TO COMPLETE

UCL OR IN-COUNTRY GENETIC ANALYSIS PROVIDER (TURKEY, INDIA)

Sample received	◯ No ◯ Yes
Date samples received	_____ (YYYY-MM-DD)
Samples received ok?	◯ No ◯ Yes
Issue with samples received	◯ Sample damage/loss ◯ Sample ID unclear ◯ Sample absent from box ◯ Other
Issue with samples received - other	_____
Sample quality	◯ Acceptable ◯ Re-send existing sample ◯ Collect fresh sample

Upload/view photo of received sample

(Photo of 1 x sample tube showing ICGNMD Study ID on tube to be shipped)

Date samples processed

_____
(YYYY-MM-DD)

Processed sample information

Recipient comments

_____

RETURNS

Will some or all of the processed samples be returned to sender?

◯ No
◯ Yes

Date samples returned to sender

_____
(YYYY-MM-DD)

Sample return comments

_____
(Person shipping can add notes here)

Sample return complete comments

_____
(Person at destination can add notes here)

THIRD PARTY PROCESSING

Will some or all of the processed samples be shipped to third party?

◯ No
◯ Yes

Date samples sent to third Party

_____
(YYYY-MM-DD)

Third party processing comments

_____
(Person shipping can add notes for third party recipient here)

Cohort Management

Participant update	
Is the participant dead?	◯ Yes
Age at death - years	_____
Age at death - months	_____
Cause of death - type	◯ NMD-disease related ◯ Non-NMD-disease related
Cause of death - description	_____
Full consent for entry/continued data use post-mortem by the participant or parent/guardian?	◯ No ◯ Yes ◯ Undetermined
Any other comments on participant	_____

Monitoring And Analysis

RELATIVE: ICGNMD genetic testing

Relative affected or unaffected?
- ◯ Affected
- ◯ Unaffected

Relative Test #1

ICGNMD Test #1 done on this relative's sample
- ◯ Reanalyse non-ICGNMD WES/WGS data
- ◯ Single gene test
- ◯ Sanger
- ◯ Array
- ◯ WES
- ◯ WGS

(Always record relative's results separate from proband results)

Date of test

_____
(YYYY-MM-DD)

Summary of this relative's test result

_____

Upload this relative's report here

Optional: upload additional ICGNMD report for this relative

Add another ICGNMD test result for this relative?
- ◯ yes
- ◯ no

Relative Test #2

ICGNMD Test #2 done on this relative's sample
- ◯ Single gene test
- ◯ Sanger
- ◯ Array
- ◯ WES
- ◯ WGS

(Always record relative's results separate from proband results)

Date of test #2

_____
(YYYY-MM-DD)

Summary of relative's test result

_____

Upload relative report here

Optional: upload additional ICGNMD report for this relative	
Add another ICGNMD test result for this relative?	◯ yes ◯ no

Relative Test #2

ICGNMD Test #3 done on this relative's sample	◯ Single gene test ◯ Sanger ◯ Array ◯ WES ◯ WGS (Always record relative's results separate from proband results)
Date of test #3	_____ (YYYY-MM-DD)
Summary of relative's test result	_____
Upload relative report here	
Optional: upload additional ICGNMD report for this relative	

UNRELATED CONTROL PARTICIPANT: ICGNMD Genetic testing

ICGNMD Test done on this unrelated control sample	◯ Single gene test ◯ Sanger ◯ Array ◯ WES ◯ WGS
Date of test	_____ (YYYY-MM-DD)
Any notes for control test data	_____
Upload unrelated control report here	

Genetic Test Planning

Test Planning Note #1

_____

Date of Discussion

_____
(YYYY-MM-DD)

Add another discussion note? ◯ Yes

Test Planning Note #2

_____

Date of Discussion

_____
(YYYY-MM-DD)

Add another discussion note? ◯ Yes

Test Planning Note #3

_____

Date of Discussion

_____
(YYYY-MM-DD)

Add another discussion note? ◯ Yes

Test Planning Note #4

_____
(If >4 test discussions, add here)

Date of Discussion

_____
(YYYY-MM-DD)

Participant already genetically "solved", no ICGNMD test needed?
◯ yes
◯ yes but want to discuss local results with ICGNMD
◯ no
(No ICGNMD report generated if solved locally)

Local tests/checks needed OUTSIDE ICGNMD project (Not funded by ICGNMD)

Any local tests/checks to do outside ICGNMD Project?
◯ yes
◯ no
(May be BEFORE or AT SAME TIME as ICGNMD testing. List any tests NOT FUNDED by ICGNMD award.)

Local tests/checks to do BEFORE ICGNMD test can start
◯ yes
◯ no

Summary of local tests/checks to do BEFORE ICGNMD testing	_____ (Most recent notes at top, with date & initials)
Local tests/checks completed now? (Local team to complete)	◯ Yes, local tests complete: start ICGNMD test ◯ Yes, local tests complete: review & decide next steps ◯ Yes, local tests complete: ICGNMD test NO LONGER NEEDED ◯ No, local work in progress: ICGNMD testing can't start yet (REMEMBER! Update "Non-ICGNMD Genetic Testing" section with local test results)
Local tests/checks to do AT SAME TIME as ICGNMD testing	◯ yes ◯ no
Summary of local tests/checks to do AT SAME TIME AS ICGNMD testing	_____ (Most recent notes at top, with date & initials)
Local checks/test results - summary by local team	_____ (REMEMBER! Update other REDCap sections with local test results)

ICGNMD reanalyse Non-ICGNMD data

Reanalyse existing WES/WGS data with ICGNMD pipeline?	◯ Yes ◯ No
Date of Joint Decision	_____ (YYYY-MM-DD)
Reason for re-analysis of non-ICGNMD data	_____
File format	◯ fastq (Best format) ◯ vcf (OK) ◯ Other (Check first)
File name (must contain ICGNMD Study ID)	_____ (Transferring site must complete)
File(s) transferred to UCL?	◯ Yes ◯ No (Transferring site must complete)
Date file transferred to UCL	_____ (YYYY-MM-DD)

PanelApp filters	
Select 1-5 PanelApp Panels to filter PDF report?	◯ Yes ◯ Not yet (SELECT 1 to 5 PANELS)
Channelopathies	☐ Brain channelopathy ☐ Skeletal muscle channelopathy
Inherited Epilepsy Syndromes	☐ Genetic epilepsy syndromes
Mitochondrial	☐ Mitochondrial Disorders
Motor and Sensory Disorders of the PNS	☐ Hereditary neuropathy ☐ Paediatric motor neuronopathies
Motor Disorders of the CNS	☐ Cerebellar hypoplasia ☐ Early onset dystonia ☐ Hereditary spastic paraplegia ☐ Neurotransmitter disorders ☐ Structural basal ganglia disorders
Neurodegenerative Disorders	☐ Amyotrophic lateral sclerosis/motor neuron disease ☐ Hereditary ataxia - adult onset
Neurodevelopmental disorders	☐ Intellectual disability ☐ Malformations of cortical development ☐ Hereditary ataxia and cerebellar anomalies - childhood onset
Neuromuscular disorders	☐ Arthrogryposis ☐ Congenital muscular dystrophy ☐ Congenital myaesthenic syndrome ☐ Congenital myopathy ☐ Distal myopathies ☐ Limb girdle muscular dystrophy ☐ Rhabdomyolysis and metabolic muscle disorders
Parenchymal brain disorders	☐ Intracerebral calcification disorders
Peroxisomal disorders	☐ Peroxisomal disorders
Connective tissues disorders	☐ Ehlers Danlos syndromes
Specific metabolic abnormalities	☐ Undiagnosed metabolic disorders
List other PanelApp panels here	_____ (Use EXACT panel names as listed at https://panelapp.genomicsengland.co.uk/panels/)
Additional analysis notes, e.g. specific genes to check	_____ (Note: Checking specific genes NOT part of standard pipeline)

Request bespoke ICGNMD analysis?

Select bespoke ICGNMD analyses (in addition to Panels)

◯ Yes
◯ Not yet
(Please only select if essential: bespoke analyses use a lot of ICGNMD time.)

Bespoke ICGNMD analyses

☐ mtDNA analysis
☐ CNV or SV detection
☐ notinuse1
☐ notinuse2
(Please only select if essential: bespoke analyses use a lot of ICGNMD time.)

ICGNMD Report for Reanalysed Non-ICGNMD Data

UCL research report uploaded

_____
(YYYY-MM-DD)

ICGNMD research report (summary PDF with filters)

ICGNMD .csv file (no panel filters)

(If requested) ICGNMD bespoke analysis report

ICGNMD Review of Reanalysis

Date of joint review of result

_____
(YYYY-MM-DD)

Has ICGNMD reanalysis provisionally solved?

◯ Yes: provisionally solved
◯ Yes, confirms other test result
◯ No, has not supported a research finding

Causative variant #1

Condition #1 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #1 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 1 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant results #1 - freetext notes

_____

Variant #1 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Add another variant?

◯ yes
◯ no

Causative variant #2

Condition #2 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #2 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 2 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #2 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Variant #2 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #3

Condition #3 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #3 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 3 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #3 Provisional classification by ICGNMD/ACMG	◯ Pathogenic ◯ Likely Pathogenic ◯ VUS - Suspected Pathogenic ◯ VUS
Variant #3 results - freetext notes	_____
Add another variant?	◯ yes ◯ no

Causative variant #4

Condition #4 [Orphanet]	_____
Free text box if Orphanet not suitable	_____
Caustive gene & variant #4 [OMIM]	_____
Free text box if OMIM not suitable	_____
Known gene mutation 4 - VCF	_____ (Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)
Variant #4 Provisional classification by ICGNMD/ACMG	◯ Pathogenic ◯ Likely Pathogenic ◯ VUS - Suspected Pathogenic ◯ VUS
Variant #4 results - freetext notes	_____

ICGNMD Next steps

ICGNMD testing needed after reanalysis?	◯ Yes ◯ No
Which additional test?	◯ WES ◯ WGS ◯ Sanger seq ◯ Array ◯ Single Gene Test ◯ Reanalyse WES data (If you select a new test, complete that section)

ICGNMD Single Gene Test #1

Go to ICGNMD single gene test #1?
- ◯ Yes
- ◯ No

Date of Joint Decision

_____
(YYYY-MM-DD)

Notes (e.g. justification, test conditions)

_____

Which country will test take place in?
- ◯ UK (or Leiden for some FSHD)
- ◯ India
- ◯ Brazil
- ◯ Other

Single Gene Test #1 Agreed

At UCL or Leiden
- ◯ DM1
- ◯ CMT1a MLPA
- ◯ c9orf72
- ◯ SMA MLPA
- ◯ FSHD1
- ◯ FSHD2
- ◯ OPMD
- ◯ CANVAS
- ◯ Friedreich's Ataxia
- ◯ SCAS
- ◯ Other

Other test or extra detail

_____

In India
- ◯ DM1
- ◯ CMT1a MLPA
- ◯ c9orf72
- ◯ SMA MLPA
- ◯ FSHD1
- ◯ FSHD2
- ◯ OPMD
- ◯ CANVAS
- ◯ Friedreich's Ataxia
- ◯ SCAS
- ◯ DMD MLPA
- ◯ DMD gene seq
- ◯ Other

Other test or extra detail

_____

In Brazil
- ◯ pmp22
- ◯ MPZ
- ◯ GJB1
- ◯ Other

Other test or extra detail

_____

Other (enter single gene test & country)

_____

Same single gene test for relatives AT SAME TIME as proband?

Test relatives as well?
- ◯ Yes
- ◯ No

Optional: Relative #1

Relative #1: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #1 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #2

Relative #2: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #4

Relative #4: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #4 Study ID (IC_ABC_NNNNN)

_____

Relatives: summary

Tick box when relative(s) single gene tests complete and their REDCap records updated?
- ☐ family single gene test 1 complete

(Record relative's results in own REDCap record - not in proband record)

Sample tracking

Samples available at test site (or test co-ordinating site)?
- ◯ yes
- ◯ no
- ◯ not all samples

Notes on sample availability at testing/test co-ordinating site

_____

(Optional) Samples sent to approved 3rd party test lab?
- ◯ yes
- ◯ no

(MTA & Terms of Service must be in place prior to sending to 3rd party)

(Optional) Date samples sent to approved 3rd party

_____
(YYYY-MM-DD)

After-test reporting

Result Quality Satisfactory?
◯ yes
◯ no

Testing site to repeat test on same sample?
◯ yes
◯ no

New sample needed to repeat?
◯ yes
◯ no

Decision = do not repeat test
◯ yes
◯ no

Reason for single gene test failure?

_____

Data file for failed test

(OPTIONAL: upload file to show failed test or low quality outputs)

Single Gene Test Results file (research only)

(Upload/View Single Gene Test report)

Single gene: test provisional results
◯ Supports diagnosis
◯ Partially supports diagnosis
◯ Does not support diagnosis
◯ Inconclusive result

UCL ICGNMD Report for Single Gene Test#1

UCL research report uploaded

_____
(YYYY-MM-DD)

ICGNMD research report

ICGNMD Review of Single Gene Test #1

Date of joint review of result

_____
(YYYY-MM-DD)

Condition [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 1 - VCF	_____ (Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)
Single gene test #1 results - freetext notes	_____
Provisional classification by ICGNMD/ACMG	◯ Pathogenic ◯ Likely Pathogenic ◯ VUS - Suspected Pathogenic ◯ VUS
Has Single Gene Test #1 provisionally solved?	◯ Yes: provisionally solved with this test ◯ Yes, this test confirms other test result ◯ No, this test has not supported a research finding

ICGNMD Next steps

More ICGNMD testing needed after Single Gene Test #1?	◯ Yes ◯ No
Which additional test?	◯ Same test on relatives ◯ WES ◯ WGS ◯ Sanger seq ◯ Array ◯ Different Single Gene Test (If you select a new test, complete that section)

Sample tracking

Relative samples at test site (or test co-ordinating site)?	◯ yes ◯ no ◯ not all samples
Notes on sample availability at testing/test co-ordinating site	_____

Optional: Relative #1

Relative #1: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #1 Study ID (IC_ABC_NNNNN)	_____
Add another relative?	◯ yes ◯ no

Optional: Relative #2

Relative #2: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #4

Relative #4: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #4 Study ID (IC_ABC_NNNNN)

_____

Relatives: summary

Tick box when relative(s) single gene test1 family REDCap records updated?

☐ family single gene test 1 records completed (Record relative's results in own REDCap record - not in proband record)

ICGNMD Review of later relative tests

Date of joint review of additional family single gene tests

_____
(YYYY-MM-DD)

(Proband record only) What has single gene testing of relatives added to proband's ICGNMD research findings?

_____

ICGNMD Single Gene Test #2

Go to ICGNMD single gene test #2?

◯ Yes
◯ No

Date of Joint Decision

_____
(YYYY-MM-DD)

Notes (e.g. justification, test conditions)

_____

Which country will test take place in?

◯ UK (or Leiden for some FSHD)
◯ India
◯ Brazil
◯ Other

Single Gene Test #2 Agreed

At UCL or Leiden

◯ DM1
◯ CMT1a MLPA
◯ c9orf72
◯ SMA MLPA
◯ FSHD1
◯ FSHD2
◯ OPMD
◯ CANVAS
◯ Friedreich's Ataxia
◯ SCAS
◯ Other

Other test or extra detail

_____

In India	◯ DM1 ◯ CMT1a MLPA ◯ c9orf72 ◯ SMA MLPA ◯ FSHD1 ◯ FSHD2 ◯ OPMD ◯ CANVAS ◯ Friedreich's Ataxia ◯ SCAS ◯ DMD MLPA ◯ DMD gene seq ◯ Other
Other test or extra detail	_____
In Brazil	◯ pmp22 ◯ MPZ ◯ GJB1 ◯ Other
Other test or extra detail	_____
Other (enter single gene test & country)	_____

Same single gene test for relatives AT SAME TIME as proband?

Test relatives as well?	◯ Yes ◯ No

Optional: Relative #1

Relative #1: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #1 Study ID (IC_ABC_NNNNN)	_____
Add another relative?	◯ yes ◯ no

Optional: Relative #2

Relative #2: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #4

Relative #4: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #4 Study ID (IC_ABC_NNNNN)

_____

Sample tracking

Samples available at test site (or test co-ordinating site)?	◯ yes ◯ no ◯ not all samples
Notes on sample availability at testing/test co-ordinating site	_____

Relatives: summary

Tick box when relative(s) testing complete and their REDCap records updated	☐ family single gene test 2 complete (Record relative's results in own REDCap record - not in proband record)
(Optional) Samples sent to approved 3rd party test lab?	◯ yes ◯ no (MTA & Terms of Service must be in place prior to sending to 3rd party)
(Optional) Date samples sent to approved 3rd party	_____ (YYYY-MM-DD)

After-test reporting

Result Quality Satisfactory?	◯ yes ◯ no
Testing site to repeat test on same sample?	◯ yes ◯ no
New sample needed to repeat?	◯ yes ◯ no
Decision = do not repeat test	◯ yes ◯ no
Reason for single gene test failure?	_____
Data file for failed test	(OPTIONAL: upload file to show failed test or low quality outputs)
Single Gene Test Results file (research only)	(Upload/View Single Gene Test report)
Single gene: test provisional results	◯ Supports diagnosis ◯ Partially supports diagnosis ◯ Does not support diagnosis ◯ Inconclusive result

UCL ICGNMD Report for Single Gene Test#2

UCL research report uploaded

_____
(YYYY-MM-DD)

ICGNMD research report

ICGNMD Review of Single Gene Test #2

Date of joint review of result

_____
(YYYY-MM-DD)

Condition [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 1 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Single gene test #2 results - freetext notes

_____

Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Has Single Gene Test #2 provisionally solved?

◯ Yes: provisionally solved with this test
◯ Yes, this test confirms other test result
◯ No, this test has not supported a research finding

ICGNMD Next steps

More ICGNMD testing needed after Single Gene Test #2?

◯ Yes
◯ No

Which additional test?

◯ Same test on relatives
◯ WES
◯ WGS
◯ Sanger seq
◯ Array
◯ Different Single Gene Test
(If you select a new test, complete that section)

Sample tracking

Relative samples at test site (or test co-ordinating site)?
- ◯ yes
- ◯ no
- ◯ not all samples

Notes on sample availability at testing/test co-ordinating site

_____

Optional: Relative #1

Relative #1: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #1 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #2

Relative #2: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)	_____
Add another relative?	◯ yes ◯ no

Optional: Relative #4

Relative #4: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #4 Study ID (IC_ABC_NNNNN)	_____

Relatives: summary

Tick box when relative(s) single gene tests complete and their REDCap records updated?	☐ family single gene test 2 records completed (Record relative's results in own REDCap record - not in proband record)

ICGNMD Review of later relative tests

Date of joint review of additional family single gene tests	_____ (YYYY-MM-DD)
(Proband record only) What has single gene testing of relatives added to proband's ICGNMD research findings?	_____

ICGNMD Whole EXOME Sequencing

Go to ICGNMD WES	◯ Yes ◯ No
Date of Joint Decision	_____ (YYYY-MM-DD)
Notes (e.g. justification)	_____
Which country will test take place in?	◯ UK ◯ India ◯ Turkey ◯ Other (UK includes Macrogen (EU))

Details if "Other" (Country & Testing Site)	_____

WES PanelApp filters

Select 1-5 PanelApp Panels to filter PDF report?	◯ Yes ◯ Not yet (SELECT 1 to 5 PANELS)
Channelopathies	☐ Brain channelopathy ☐ Skeletal muscle channelopathy
Inherited Epilepsy Syndromes	☐ Genetic epilepsy syndromes
Mitochondrial	☐ Mitochondrial Disorders
Motor and Sensory Disorders of the PNS	☐ Hereditary neuropathy ☐ Paediatric motor neuronopathies
Motor Disorders of the CNS	☐ Cerebellar hypoplasia ☐ Early onset dystonia ☐ Hereditary spastic paraplegia ☐ Neurotransmitter disorders ☐ Structural basal ganglia disorders
Neurodegenerative Disorders	☐ Amyotrophic lateral sclerosis/motor neuron disease ☐ Hereditary ataxia - adult onset
Neurodevelopmental disorders	☐ Intellectual disability ☐ Malformations of cortical development ☐ Hereditary ataxia and cerebellar anomalies - childhood onset
Neuromuscular disorders	☐ Arthrogryposis ☐ Congenital muscular dystrophy ☐ Congenital myaesthenic syndrome ☐ Congenital myopathy ☐ Distal myopathies ☐ Limb girdle muscular dystrophy ☐ Rhabdomyolysis and metabolic muscle disorders
Parenchymal brain disorders	☐ Intracerebral calcification disorders
Peroxisomal disorders	☐ Peroxisomal disorders
Connective tissues disorders	☐ Ehlers Danlos syndromes
Specific metabolic abnormalities	☐ Undiagnosed metabolic disorders
List other PanelApp panels here	_____ (Use EXACT panel names as listed at https://panelapp.genomicsengland.co.uk/panels/)

Analysis notes, e.g. specific genes to check	_____ (Note: Checking specific genes NOT part of standard pipeline)

Request bespoke ICGNMD analysis?

Select bespoke ICGNMD analyses (in addition to Panels)	◯ Yes ◯ Not yet (Please only select if essential: bespoke analyses use a lot of ICGNMD time.)
Bespoke ICGNMD analyses	☐ mtDNA analysis ☐ CNV or SV detection ☐ notinuse1 ☐ notinuse2 (Please only select if essential: bespoke analyses use a lot of ICGNMD time.)

WES for relatives AT SAME TIME as proband?

Test relatives at same time?	◯ Yes ◯ No (Proband normally has WES first, relatives later)
Justification for testing relatives at same time as proband?	_____

Optional: Relative #1

Relative #1: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #1 Study ID (IC_ABC_NNNNN)	_____
Add another relative?	◯ yes ◯ no

Optional: Relative #2

Relative #2: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #4

Relative #4: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #4 Study ID (IC_ABC_NNNNN)

_____

Relatives: summary

Tick box when relative(s) WES complete and their REDCap records updated	☐ family WES complete (Record relative's results in own REDCap record - not in proband record)

Sample tracking

Samples available at test site (or test co-ordinating site)?	◯ yes ◯ no ◯ not all samples
Notes on sample availability at testing/test co-ordinating site	_____
(Optional) Samples sent to approved 3rd party? E.g. UCL to Macrogen	◯ yes ◯ no (MTA & Terms of Service must be in place prior to sending to 3rd party)
(Optional) Date samples sent to approved 3rd party	_____ (YYYY-MM-DD)
Samples sent to Macrogen? (UCL only)	◯ yes ◯ no

WES reporting

Raw WES data received by UCL?	◯ yes ◯ no
Date WES data received	_____ (YYYY-MM-DD)
UCL record: Data Quality Satisfactory?	◯ yes ◯ no
Testing site to repeat test on same sample?	◯ yes ◯ no
New sample needed to repeat?	◯ yes ◯ no
Decision = do not repeat test	◯ yes ◯ no
Reason for WES failure?	_____
Data file for failed test	(OPTIONAL: upload file to show failed test or low quality outputs)

UCL ICGNMD Report for WES

UCL research report uploaded

_____
(YYYY-MM-DD)

ICGNMD research report (summary PDF with filters)

ICGNMD .csv file (no panel filters)

(If requested) ICGNMD bespoke analysis report

ICGNMD Review of WES

Date of joint review of result

_____
(YYYY-MM-DD)

Has WES provisionally solved?

◯ Yes: provisionally solved with WES
◯ Yes, WES confirms other test result
◯ No, WES has not supported a research finding

Causative variant #1

Condition #1 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #1 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 1 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

WES variant results #1 - freetext notes

_____

Variant #1 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Add another variant?

◯ yes
◯ no

Causative variant #2

Condition #2 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #2 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 2 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #2 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WES variant #2 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #3

Condition #3 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #3 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 3 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #3 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WES variant #3 results - freetext notes

_____

Add another variant?	◯ yes ◯ no

Causative variant #4

Condition #4 [Orphanet]	_____
Free text box if Orphanet not suitable	_____
Caustive gene & variant #4 [OMIM]	_____
Free text box if OMIM not suitable	_____
Known gene mutation 4 - VCF	_____ (Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)
Variant #4 Provisional classification by ICGNMD/ACMG	◯ Pathogenic ◯ Likely Pathogenic ◯ VUS - Suspected Pathogenic ◯ VUS
WES variant #4 results - freetext notes	_____

ICGNMD Next steps

More ICGNMD testing needed after WES review?	◯ Yes ◯ No
Which additional test?	◯ WES on relatives ◯ WGS ◯ Sanger seq ◯ Array ◯ Single Gene Test ◯ Reanalyse WES data (If you select a new test, complete that section)

Sample tracking

Relative samples at test site (or test co-ordinating site)?	◯ yes ◯ no ◯ not all samples
Notes on sample availability at testing/test co-ordinating site	_____

Optional: Relative #1

Relative #1: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #1 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #2

Relative #2: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #4

Relative #4: Relation to proband

◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?

◯ yes
◯ no

Relative #4 Study ID (IC_ABC_NNNNN)

_____

Relatives: summary

Tick box when relative(s) WES complete and their REDCap records updated

☐ family WES complete & REDCap updated (Record relative's results in own REDCap record - not in proband record)

ICGNMD Review of later relative tests

Date of joint review of additional family single gene tests

_____
(YYYY-MM-DD)

(Proband record only) What has later WES of relatives added to proband's ICGNMD research findings?

_____

ICGNMD repeat WES analysis

Reason for repeating WES analysis?

◯ Long time since 1st analysis, check for new variants
◯ New bespoke analysis available
◯ Other

Expand with further details here, e.g specific genes to check

_____
(Note: Checking specific genes NOT part of standard pipeline)

UCL ICGNMD WES Re-analysis

New research report created

_____
(YYYY-MM-DD)

ICGNMD new research report (summary PDF with filters)

ICGNMD new .csv file (no panel filters)

(If requested) new ICGNMD bespoke analysis report

ICGNMD Review of WES Re-analysis

Date of joint review of re-analysis

_____
(YYYY-MM-DD)

Has WES provisionally solved?

◯ Yes: provisionally solved with WES
◯ Yes, WES confirms other test result
◯ No, WES has not supported a research finding

Causative variant #1

Condition #1 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #1 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 1 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

WES variant results #1 - freetext notes

_____

Variant #1 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Add another variant?

◯ yes
◯ no

Causative variant #2

Condition #2 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #2 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 2 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #2 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WES variant #2 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #3

Condition #3 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #3 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 3 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #3 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WES variant #3 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #4

Condition #4 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #4 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 4 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #4 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WES variant #4 results - freetext notes

_____

ICGNMD Whole GENOME sequencing

Go to ICGNMD WGS

◯ Yes
◯ No

Date of Joint Decision

_____
(YYYY-MM-DD)

Notes (e.g. justification)

_____

Which country will test take place in?

◯ UK Macrogen
◯ UK Illumina
◯ India
◯ Turkey
◯ Other

Details if "Other" (Country & Testing Site)

_____

WGS PanelApp filters	
Select 1-5 PanelApp Panels to filter PDF report?	◯ Yes ◯ Not yet (SELECT 1 to 5 PANELS)
Channelopathies	☐ Brain channelopathy ☐ Skeletal muscle channelopathy
Inherited Epilepsy Syndromes	☐ Genetic epilepsy syndromes
Mitochondrial	☐ Mitochondrial Disorders
Motor and Sensory Disorders of the PNS	☐ Hereditary neuropathy ☐ Paediatric motor neuronopathies
Motor Disorders of the CNS	☐ Cerebellar hypoplasia ☐ Early onset dystonia ☐ Hereditary spastic paraplegia ☐ Neurotransmitter disorders ☐ Structural basal ganglia disorders
Neurodegenerative Disorders	☐ Amyotrophic lateral sclerosis/motor neuron disease ☐ Hereditary ataxia - adult onset
Neurodevelopmental disorders	☐ Intellectual disability ☐ Malformations of cortical development ☐ Hereditary ataxia and cerebellar anomalies - childhood onset
Neuromuscular disorders	☐ Arthrogryposis ☐ Congenital muscular dystrophy ☐ Congenital myaesthenic syndrome ☐ Congenital myopathy ☐ Distal myopathies ☐ Limb girdle muscular dystrophy ☐ Rhabdomyolysis and metabolic muscle disorders
Parenchymal brain disorders	☐ Intracerebral calcification disorders
Peroxisomal disorders	☐ Peroxisomal disorders
Connective tissues disorders	☐ Ehlers Danlos syndromes
Specific metabolic abnormalities	☐ Undiagnosed metabolic disorders
List other PanelApp panels here	_____ (Use EXACT panel names as listed at https://panelapp.genomicsengland.co.uk/panels/)
Additional WGS notes, e.g. specific genes to check	_____ (Note: Checking specific genes NOT part of standard pipeline)

Request bespoke ICGNMD analysis?

Select bespoke ICGNMD analyses (in addition to Panels)

◯ Yes
◯ Not yet
(Please only select if essential: bespoke analyses use a lot of ICGNMD time.)

Bespoke ICGNMD analyses

☐ mtDNA analysis
☐ CNV or SV detection
☐ notinuse1
☐ notinuse2
(Please only select if essential: bespoke analyses use a lot of ICGNMD time.)

WGS for relatives AT SAME TIME as proband?

Test relatives at same time?

◯ Yes
◯ No
(Proband normally has wgs first, relatives later)

Justification for testing relatives at same time as proband?

_____

Optional: Relative #1

Relative #1: Relation to proband

◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?

◯ yes
◯ no

Relative #1 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

◯ yes
◯ no

Optional: Relative #2

Relative #2: Relation to proband

◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?

◯ yes
◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?	◯ yes ◯ no

Optional: Relative #3

Relative #3: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #3 Study ID (IC_ABC_NNNNN)	_____
Add another relative?	◯ yes ◯ no

Optional: Relative #4

Relative #4: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #4 Study ID (IC_ABC_NNNNN)	_____

Relatives: summary

Tick box when relative(s) WGS complete and their REDCap records updated	☐ family WGS complete (Record relative's results in own REDCap record - not in proband record)

Sample tracking

Samples available at test site (or test co-ordinating site)?	◯ yes ◯ no ◯ not all samples
Notes on sample availability at testing/test co-ordinating site	_____
(Optional) Samples sent to approved 3rd party? E.g. UCL to Macrogen/Illumina	◯ yes ◯ no (MTA & Terms of Service must be in place prior to sending to 3rd party)

(Optional) Date samples sent to approved 3rd party

_____
(YYYY-MM-DD)

Samples sent to Macrogen/Illumina? (UCL only)

◯ yes
◯ no

WGS reporting

Raw WGS data received by UCL?

◯ yes
◯ no

Date WGS data received

_____
(YYYY-MM-DD)

UCL record: Data Quality Satisfactory?

◯ yes
◯ no

Testing site to repeat test on same sample?

◯ yes
◯ no

New sample needed to repeat?

◯ yes
◯ no

Decision = do not repeat test

◯ yes
◯ no

Reason for WGS failure?

_____

Data file for failed test

(OPTIONAL: upload file to show failed test or low quality outputs)

UCL ICGNMD Report for WGS

UCL research report uploaded

_____
(YYYY-MM-DD)

ICGNMD research report (summary PDF with filters)

ICGNMD .csv file (no panel filters)

(If requested) ICGNMD bespoke analysis report

ICGNMD Review of WGS

Date of joint review of result

_____
(YYYY-MM-DD)

Has WGS provisionally solved?

◯ Yes: provisionally solved with WGS
◯ Yes, WGS confirms other test result
◯ No, WGS has not supported a research finding

Causative variant #1

Condition #1 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #1 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 1 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

WGS variant results #1 - freetext notes

_____

Variant #1 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Add another variant?

◯ yes
◯ no

Causative variant #2

Condition #2 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #2 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 2 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #2 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WGS variant #2 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #3

Condition #3 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #3 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 3 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #3 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WGS variant #3 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #4

Condition #4 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #4 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 4 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #4 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WGS variant #4 results - freetext notes

_____

ICGNMD Next steps

More ICGNMD testing needed after WGS review?

◯ Yes
◯ No

Which additional test?

◯ WGS on relatives
◯ WGS
◯ Sanger seq
◯ Array
◯ Single Gene Test
◯ Reanalyse WGS data
(If you select a new test, complete that section)

Sample tracking

Relative samples at test site (or test co-ordinating site)?

◯ yes
◯ no
◯ not all samples

Notes on sample availability at testing/test co-ordinating site

_____

Optional: Relative #1

Relative #1: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #1 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #2

Relative #2: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband

- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?

- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

- ◯ yes
- ◯ no

Optional: Relative #4

Relative #4: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #4 Study ID (IC_ABC_NNNNN)	_____

Relatives: summary

Tick box when relative(s) WGS complete and their REDCap records updated	☐ family WGS complete & REDCap records updated (Record relative's results in own REDCap record - not in proband record)

ICGNMD Review of later relative tests

Date of joint review of additional family single gene tests	_____ (YYYY-MM-DD)
(Proband record only) What has later WGS of relatives added to proband's ICGNMD research findings?	_____

ICGNMD repeat WGS analysis

Reason for repeating WGS analysis?	◯ Long time since 1st analysis, check for new variants ◯ New bespoke analysis available ◯ Other
Expand with further details here	_____

UCL ICGNMD WGS Re-analysis

New research report created	_____ (YYYY-MM-DD)
ICGNMD new research report (summary PDF with filters)	
ICGNMD new .csv file (no panel filters)	
(If requested) new ICGNMD bespoke analysis report	

ICGNMD Review of WGS Re-analysis

Date of joint review of re-analysis

_____
(YYYY-MM-DD)

Has WGS provisionally solved?

◯ Yes: provisionally solved with WGS
◯ Yes, WGS confirms other test result
◯ No, WGS has not supported a research finding

Causative variant #1

Condition #1 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #1 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 1 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

WGS variant results #1 - freetext notes

_____

Variant #1 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Add another variant?

◯ yes
◯ no

Causative variant #2

Condition #2 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #2 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 2 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #2 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WGS variant #2 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #3

Condition #3 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #3 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 3 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #3 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WGS variant #3 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #4

Condition #4 [Orphanet]

_____

Free text box if Orphanet not suitable

_____

Caustive gene & variant #4 [OMIM]

_____

Free text box if OMIM not suitable

_____

Known gene mutation 4 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #4 Provisional classification by ICGNMD/ACMG

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

WGS variant #4 results - freetext notes

_____

ICGNMD Sanger Sequencing

Go to ICGNMD Sanger Sequencing?

◯ Yes
◯ No
(Complete ONLY if ICGNMD to fund Sanger)

Date of Joint Decision

_____
(YYYY-MM-DD)

Discussion notes

_____
(Most recent at top, include date & initials)

Which country will test take place in?

◯ UK (or Leiden for some FSHD)
◯ India
◯ Brazil
◯ Other

Gene & variant to be tested by Sanger Seq (provide VCF and OMIM terms)

Variant to test in VCF format

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant to test [OMIM]

_____

Free text box if OMIM not suitable

_____

Testing notes (e.g. justification, conditions, protocol refs)

_____

Same Sanger seq for relatives AT SAME TIME as proband?

Test relatives at same time as proband?

◯ Yes
◯ No

Optional: Relative #1

Relative #1: Relation to proband

◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?

◯ yes
◯ no

Relative #1 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

◯ yes
◯ no

Optional: Relative #2

Relative #2: Relation to proband

◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?

◯ yes
◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?

◯ yes
◯ no

Optional: Relative #3

Relative #3: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #3 Study ID (IC_ABC_NNNNN)	_____
Add another relative?	◯ yes ◯ no

Optional: Relative #4

Relative #4: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #4 Study ID (IC_ABC_NNNNN)	_____

Relatives: summary

Tick box when relative(s) Sanger complete and their REDCap records updated	☐ family Sanger test 1 complete (Record relative's results in own REDCap record - not in proband record)

Sample tracking

Samples available at test site (or test co-ordinating site)?	◯ yes ◯ no
Notes on sample availability at testing/test co-ordinating site	_____
(Optional) Samples sent to approved 3rd party test lab?	◯ yes ◯ no (MTA & Terms of Service must be in place prior to sending to 3rd party)
(Optional) Date samples sent to approved 3rd party	_____ (YYYY-MM-DD)

After-test reporting

Result Quality Satisfactory?	◯ yes ◯ no
Testing site to repeat test on same sample?	◯ yes ◯ no
New sample needed to repeat?	◯ yes ◯ no
Decision = do not repeat test	◯ yes ◯ no
Reason for Sanger failure?	_____
Data file for failed test	(OPTIONAL: upload file to show failed test or low quality outputs)
Sanger seq Results file (research only)	(Upload/View Single Gene Test report)
Sanger seq test: results from test site	◯ Supports diagnosis ◯ Partially supports diagnosis ◯ Does not support diagnosis ◯ Inconclusive result

UCL ICGNMD Report for Sanger Sequencing

UCL research report uploaded	_____ (YYYY-MM-DD)
ICGNMD research report	

ICGNMD Review of Sanger sequencing result

Date of joint review of result	_____ (YYYY-MM-DD)
Known gene mutation - VCF	_____ (Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)
Condition [Orphanet]	_____
Free text box if Orphanet not suitable	_____
Caustive gene & variant [OMIM]	_____

Free text box if OMIM not suitable	_____
Sanger seq results - freetext notes	_____
Provisional classification by ICGNMD/ACMG	◯ Pathogenic ◯ Likely Pathogenic ◯ VUS - Suspected Pathogenic ◯ VUS
Has Sanger seq provisionally solved?	◯ Yes: provisionally solved with this test ◯ Yes, this test confirms other test result ◯ No, this test has not supported a research finding

ICGNMD Next steps

More ICGNMD testing needed after Sanger seq?	◯ Yes ◯ No
Which additional test?	◯ Same test on relatives ◯ WES ◯ WGS ◯ Array ◯ Single Gene Test (If you select a new test, complete that section)

Sample tracking

Relative samples at test site (or test co-ordinating site)?	◯ yes ◯ no ◯ not all samples
Notes on sample availability at testing/test co-ordinating site	_____

Optional: Relative #1

Relative #1: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #1 Study ID (IC_ABC_NNNNN)	_____
Add another relative?	◯ yes ◯ no

Optional: Relative #2

Relative #2: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #4

Relative #4: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #4 Study ID (IC_ABC_NNNNN)

_____

Relative Sanger complete and relatives' REDCap records updated?
- ◯ yes
- ◯ no

(Record relative's results in own REDCap record - not in proband record)

ICGNMD Review of later relative tests

Date of joint review of additional family Sanger seq

_____
(YYYY-MM-DD)

(Proband record only) What has Sanger seq of relatives added to proband's ICGNMD research findings?

_____

ICGNMD Array

Go to ICGNMD Array?

◯ Yes
◯ No
(Complete ONLY if ICGNMD to fund array)

Date of Joint Decision

_____
(YYYY-MM-DD)

Reason for Array

◯ CNV analysis
◯ Multigenerational testing/linkage
◯ Other

If "Other", enter reasons here

_____

Which country will test take place in?

◯ UK
◯ India
◯ Other

If Other, enter details here (country & name of test provider)

_____

Array product details (e.g. manufacturer, product code, batch #)

_____

Array for relatives AT SAME TIME as proband?

Test relatives as well?

◯ Yes
◯ No

Optional: Relative #1

Relative #1: Relation to proband

◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?

◯ yes
◯ no

Relative #1 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
◯ yes
◯ no

Optional: Relative #2

Relative #2: Relation to proband
◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?
◯ yes
◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
◯ yes
◯ no

Optional: Relative #3

Relative #3: Relation to proband
◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?
◯ yes
◯ no

Relative #3 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
◯ yes
◯ no

Optional: Relative #4

Relative #4: Relation to proband
◯ mother
◯ father
◯ brother
◯ sister
◯ other

Is this Relative also affected?
◯ yes
◯ no

Relative #4 Study ID (IC_ABC_NNNNN)	_____

Relatives: summary

Tick box when relative(s) array complete and their REDCap records updated?	☐ family array complete (Record relative's results in own REDCap record - not in proband record)

Sample tracking

Samples available at test site (or test co-ordinating site)?	◯ yes ◯ no
Notes on sample availability at testing/test co-ordinating site	_____
(Optional) Samples sent to approved 3rd party test lab?	◯ yes ◯ no (MTA & Terms of Service must be in place prior to sending to 3rd party)
(Optional) Date samples sent to approved 3rd party	_____ (YYYY-MM-DD)

After-test reporting

Result Quality Satisfactory?	◯ yes ◯ no
Testing site to repeat test on same sample?	◯ yes ◯ no
New sample needed to repeat?	◯ yes ◯ no
Decision = do not repeat test	◯ yes ◯ no
Reason for Array failure?	_____
Data file for failed test	(OPTIONAL: upload file to show failed test or low quality outputs)
Array Results file (research only)	(Upload/View Single Gene Test report)

Array: results from test site

◯ Supports diagnosis
◯ Partially supports diagnosis
◯ Does not support diagnosis
◯ Inconclusive result

UCL ICGNMD Report for Array

UCL research report uploaded

_____
(YYYY-MM-DD)

ICGNMD research report

ICGNMD Review of Array

Date of joint review of result

_____
(YYYY-MM-DD)

Has Array provisionally solved or helped to solve?

◯ Yes: provisionally solved with this test
◯ Yes, this test confirms other test result
◯ No, this test has not supported a research finding

Causative variant #1

Known gene mutation - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Condition #1 [Orphanet]

_____

Caustive gene & variant #1 [OMIM]

_____

Array variant #1 results - freetext notes

_____

Provisional classification by ICGNMD

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Add another variant?

◯ yes
◯ no

Causative variant #2

Known gene mutation 2 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Condition #2 [Orphanet]

_____

Caustive gene & variant #2 [OMIM]

_____

Variant #2 Provisional classification by ICGNMD

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Array variant #2 results - freetext notes

_____

Add another variant?

◯ yes
◯ no

Causative variant #3

Condition #3 [Orphanet]

_____

Caustive gene & variant #3 [OMIM]

_____

Known gene mutation 3 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Variant #3 Provisional classification by ICGNMD

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic
◯ VUS

Array variant #3 results - freetext notes

_____

ICGNMD Next steps

More ICGNMD testing needed after Array?

◯ Yes
◯ No

Which additional test?

◯ Array on relatives
◯ WES
◯ WGS
◯ Sanger seq
◯ Single Gene Test
(If you select a new test, complete that section)

Sample tracking

Relative samples at test site (or test co-ordinating site)?
- ◯ yes
- ◯ no
- ◯ not all samples

Notes on sample availability at testing/test co-ordinating site

_____

Optional: Relative #1

Relative #1: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #1 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #2

Relative #2: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #2 Study ID (IC_ABC_NNNNN)

_____

Add another relative?
- ◯ yes
- ◯ no

Optional: Relative #3

Relative #3: Relation to proband
- ◯ mother
- ◯ father
- ◯ brother
- ◯ sister
- ◯ other

Is this Relative also affected?
- ◯ yes
- ◯ no

Relative #3 Study ID (IC_ABC_NNNNN)	_____
Add another relative?	◯ yes ◯ no

Optional: Relative #4

Relative #4: Relation to proband	◯ mother ◯ father ◯ brother ◯ sister ◯ other
Is this Relative also affected?	◯ yes ◯ no
Relative #4 Study ID (IC_ABC_NNNNN)	_____
Relative(s) arrays complete and their REDCap records updated?	◯ yes ◯ no (Record relative's results in own REDCap record - not in proband record)

ICGNMD Review of later relative tests

Date of joint review of additional family Arrays	_____ (YYYY-MM-DD)
(Proband record only) What has Array of relatives added to proband's ICGNMD research findings?	_____

Final ICGNMD Research Summary (UCL: complete at end of pathway)

Reason for issuing final report now	◯ No further testing required ◯ Further testing desirable, but not possible under current project ◯ No ICGNMD report, recruited as "solved" participant
Participant ICGNMD outcome (research only) - Select any that apply	☐ Genetically solved with no ICGNMD input ☐ Outcome supported with ICGNMD intellectual input ☐ Outcome supported by ICGNMD genetic testing ☐ Local AND ICGNMD genetic tests supported outcome ☐ No causative/likely causative variants identified ☐ Most suitable test not available locally or via ICGNMD ☐ Study ended before testing

Which ICGNMD genetic test(s) gave positive research result? (Select any that apply)

- ☐ WGS - proband
- ☐ WES - proband
- ☐ Array - proband
- ☐ Single Gene Test - proband
- ☐ Sanger seq - proband
- ☐ WGS - relatives
- ☐ WES - relatives
- ☐ Array - relatives
- ☐ Single gene test - relatives
- ☐ Sanger seq - relatives
- ☐ Other
- ☐ None

Further details

_____

Final ICGNMD diagnosis (research only)

Final diagnostic category

- ◯ Genetic motor neuron disease
- ◯ Spinal muscular atrophy
- ◯ Genetic peripheral neuropathy
- ◯ Congenital myasthenic syndrome
- ◯ Congenital muscular dystrophy or myopathy
- ◯ Duchenne or Becker muscular dystrophy
- ◯ Facioscapulohumeral muscular dystrophy
- ◯ Myotonic dystrophy type I
- ◯ Limb girdle muscular dystrophy
- ◯ Distal myopathy
- ◯ Adult onset myopathy
- ◯ Metabolic myopathy
- ◯ Mitochondrial disease
- ◯ Skeletal muscle channelopathy
- ◯ Undetermined genetic neuromuscular or neurological disorder

(Diagnosis most likely, after ICGNMD pathway complete. DO NOT COMPLETE FOR PARTICIPANTS RECRUITED AS SOLVED CASES)

Condition #1

_____

((Orphanet) You can add extra diagnoses below if complex (e.g. "double trouble"))

Other Condition #2

_____

(Orphanet)

Other Condition #3

_____

(Orphanet)

Confirmed diagnoses - freetext

_____

Has original "best guess" diagnosis changed with ICGNMD?

- ◯ yes
- ◯ no
- ◯ not clear

Final Causative variant #1

Variant #1 ICGNMD Classification

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic

Causative variant #1 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Condition #1 [Orphanet]

_____

Caustive gene & variant #1 [OMIM]

_____

Variant #1 - freetext notes

_____

Add another variant?

◯ yes
◯ no

Final Causative variant #2

Variant #2 ICGNMD Classification

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic

Causative variant #2 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Condition #2 [Orphanet]

_____

Caustive gene & variant #2 [OMIM]

_____

Variant #2 - freetext notes

_____

Add another variant?

◯ yes
◯ no

Final Causative variant #3

Variant #3 ICGNMD Classification

◯ Pathogenic
◯ Likely Pathogenic
◯ VUS - Suspected Pathogenic

Causative variant #3 - VCF

_____
(Use format (CHR POS ID REF ALT): 1 230710048 rs699 A G)

Condition #3 [Orphanet]

Caustive gene & variant #3 [OMIM]

Variant #3 - freetext notes

Final ICGNMD Report (Research only, not diagnostic)

Upload final ICGNMD report

ICGNMD comments for follow-on research

Select one option

- ◯ Variant(s) already well-known & good fit to phenotype
- ◯ Novel, suspected pathogenic variant in established disease gene
- ◯ Variant(s) known pathogenic but phenotype novel features
- ◯ Novel suspected variant in gene not linked to this disease
- ◯ Other

Details

Participant's data despoited in archive?

- ◯ yes
- ◯ no

Date deposited in EMBL-EBI EGA

(YYYY-MM-DD)

Specify which archive (all that apply)

- ☐ EMBL-EBI EGA
- ☐ RD-Connect
- ☐ Other

If "Other", add details here (location & date archived)

ICGNMD Data Use

Participant's data used in ICGNMD publication?	◯ yes ◯ no
Enter DOI for publication(s) here	_____

Participant links to other studies

Is participant also recruited to another study (e.g. SOLVE-RD)?	◯ yes ◯ no ◯ don't know
Details of other studies participant enrolled in	_____

Non-ICGNMD publications

Has participant been included in publications outside of ICGNMD (using non-ICGNMD data)?	◯ yes ◯ no
Enter DOI for non-ICGNMD-related publication(s) here	_____

Final summary: relatives of proband

Has ICGNMD also provided findings for affected relatives?	◯ yes ◯ no (Do NOT put details about relatives findings here - use relatives' own records)
How many affected relatives received their own ICGNMD research findings? (1-10)	_____
List ICGNMD Study IDs of affected relatives with ICGNMD findings here	_____ (Format IC_ABC_NNNNN, IC_ABC_NNNNN etc)