**Mitochondrial Diseases Flagship**

**Optimal Clinical Data**

*Version 1, 24.9.19*

Australian Genomics

| Sequencing Method | | | |
|---|---|---|---|
| Sample randomisation: sequencing method | | ☐ WES ☐ WGS | |
| **Clinical information** | | | |
| Date of first medical presentation: | | | |
| Date of first specialist clinic consult: | | | |
| Name of specialist service: | | | |
| Date of presumed diagnosis (based on biochemical/histological studies performed: | | | |
| Differential diagnoses (please give multiple diagnoses, multiple diagnoses help the laboratory identify the disease gene): | | | |
| Presenting features triggering referral | | | |
| **Age of onset of presenting symptoms** | | | |
| ☐ prenatal<br>☐ Neonatal (Birth to 28 days)<br>☐ Infantile (> 28 days month to 1 year)<br>☐ Childhood (>1 to 5 years) | | ☐ Juvenile (>5-16 years)<br>☐ Early adult (>16-40 years)<br>☐ Late adult (>40 years) | |
| **Evolution of symptoms** | | | |
| ☐ Rapidly progressive (< 6months)<br>☐ Non-progressive | | ☐ Slowly progressive (> 6 months)<br>☐ acute-episodic (relapsing-remitting) | |
| Symptoms at onset: | | | |
| Previous genotype data: | | | |
| **Syndromic phenotype** | | | |
| ☐ Leigh disease<br>☐ MERRF<br>☐ Kearns-Sayre<br>☐ Alpers | | ☐ MELAS<br>☐ Pearson-Lammi<br>☐ MNGIE<br>☐ Others: | |
| **Muscle weakness** | | | |
| Muscle weakness | ☐ Yes ☐ No | | |
| i. Facial | ☐ Yes ☐ No | vi. Prox. LL | ☐ Yes ☐ No |
| ii. Ptosis | ☐ Yes ☐ No | vii. Intrinsic hand muscles | ☐ Yes ☐ No |
| iii. Ophthalmoplegia | ☐ Yes ☐ No | viii Diaphragmatic / chest wall | ☐ Yes ☐ No |
| iv. Axial/paraspinal | ☐ Yes ☐ No | ix. Global weakness | ☐ Yes ☐ No |
| v. Prox. UL | ☐ Yes ☐ No | | |
| **Muscle atrophy/hypotrophy** | | | |
| Muscle atrophy/hypotrophy | ☐ Yes ☐ No | ☐ Not assessed | |
| i. Facial expression normal | ☐ Yes ☐ No | vi. Prox. LL | ☐ Yes ☐ No |
| ii.Axial/ paraspinal | ☐ Yes ☐ No | vii. Distal UL | ☐ Yes ☐ No |
| iii.Masseter | ☐ Yes ☐ No | viii. Distal LL | ☐ Yes ☐ No |
| iv. Glossal | ☐ Yes ☐ No | ix. intrinsic hand muscles | ☐ Yes ☐ No |
| v. Prox. UL | ☐ Yes ☐ No | | |
| **Fasciculations** | | | |
| Fasciculations: | ☐ Yes ☐ No | | |
| - if present, pattern: | | | |
| Short comment: | | | |
| Muscle pain | ☐ Yes ☐ No | | |
| Muscle cramps<br>- If yes:<br>☐ at rest<br>☐ with exercise | ☐ Yes ☐ No | | |

| Muscle fatigue | ☐ Yes ☐ No | | |
|---|---|---|---|
| - If yes:<br>☐ Independently ambulatory for 500m – 1000m without fatigue<br>☐ Independently ambulatory for <500m<br>☐ Dependent ambulation due to neuromuscular disease [wheelchair] | | | |
| General exercise intolerance | ☐ Yes ☐ No | | |
| - Comment: | | | |
| Rhabdomyolysis/myoglobinuria | ☐ Yes ☐ No | | |
| Malignant hyperthermia | ☐ Yes ☐ No | | |
| **Joint hyperlaxity/hypermobility** | | | |
| Joint hyperlaxity/hypermobility | ☐ Yes ☐ No | | |
| i. Neck | ☐ Yes ☐ No | v. Knees | ☐ Yes ☐ No |
| ii. Elbows | ☐ Yes ☐ No | vi. Ankles. | ☐ Yes ☐ No |
| iii. Fingers | ☐ Yes ☐ No | vii. Wrist | ☐ Yes ☐ No |
| iv. Hip | ☐ Yes ☐ No | viii. Cutaneous laxity | ☐ Yes ☐ No |
| ix. Beighton score: __________(0-9 see Appendix 1) | | | |
| x. Comment: | | | |
| **Joint contractures** | | | |
| Joint contractures | ☐ Yes ☐ No | | |
| i. Arthrogryposis multiplex congenita | ☐ Yes ☐ No | | |
| ii. Neck | ☐ Yes ☐ No | | |
| iii. Shoulders | ☐ Yes ☐ No | | |
| iv. Elbows | ☐ Yes ☐ No | | |
| v. Wrists | ☐ Yes ☐ No | | |
| vi. Fingers | ☐ Yes ☐ No | | |
| vii. Hip | ☐ Yes ☐ No | | |
| viii. Knees | ☐ Yes ☐ No | | |
| ix. Ankles | ☐ Yes ☐ No | | |
| x. Toes | ☐ Yes ☐ No | | |
| xi. Comment: | | | |
| **Oropharyngeal dysfunction (see definition – Appendix 2)** | | | |
| Oropharyngeal dysfunction | ☐ Yes ☐ No | | |
| i. Dysarthria | ☐ Yes ☐ No | | |
| ii. Dysphagia | ☐ Yes ☐ No | | |
| iii. Dysarthria and dysphagia | ☐ Yes ☐ No | | |
| iv. Dysphonia | ☐ Yes ☐ No | | |
| v. Bulbar palsy | ☐ Yes ☐ No | | |
| vi. Pseudobulbar palsy | ☐ Yes ☐ No | | |
| vii. Comment: | | | |
| **Respiratory abnormalities** | | | |
| Respiratory abnormalities | ☐ Yes ☐ No | | |
| i. Assisted ventilation | ☐ Continuous ☐ Intermittent | | |
| ii. Episodic hyperventilation | ☐ Yes ☐ No | | |
| iii. Episodic apnoea<br>- If yes:<br>☐ central<br>☐ obstructive | ☐ Yes ☐ No | | |
| iv. Comment: | | | |
| **Tone** | | | |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| i. Hypotonia | ☐ Yes ☐ No | | | | | | | |
| Comment: | | | | | | | | |
| ii. Hypertonia (spasticity) | ☐ Yes ☐ No | | | | | | | |
| Comment: | | | | | | | | |
| iii. Hypertonia (dystonia) | ☐ Yes ☐ No | | | | | | | |
| Comment: | | | | | | | | |
| **Deep tendon reflexes** | | | | | | | | |
| | LEFT | | | | RIGHT | | | |
| i. Biceps | ☐ Hypo | ☐ Normal | ☐ Hyper | ☐ Not Elicited | ☐ Hypo | ☐ Normal | ☐ Hyper | ☐ Not Elicited |
| ii. Triceps | ☐ Hypo | ☐ Normal | ☐ Hyper | ☐ Not Elicited | ☐ Hypo | ☐ Normal | ☐ Hyper | ☐ Not Elicited |
| iii. Patellar | ☐ Hypo | ☐ Normal | ☐ Hyper | ☐ Not Elicited | ☐ Hypo | ☐ Normal | ☐ Hyper | ☐ Not Elicited |
| iv. Ankle | ☐ Hypo | ☐ Normal | ☐ Hyper | ☐ Not Elicited | ☐ Hypo | ☐ Normal | ☐ Hyper | ☐ Not Elicited |
| v. Plantars | ☐ Up | ☐ Down | ☐ Absent | | ☐ Up | ☐ Down | ☐ Absent | |
| vi. Comment: | | | | | | | | |
| **Extrapyramidal features** | | | | | | | | |
| Myoclonus | ☐ Yes ☐ No | | | | | | | |
| Pattern: | | | | | | | | |
| Choreathethosis | ☐ Yes ☐ No | | | | | | | |
| Pattern: | | | | | | | | |
| Tremor | ☐ Yes ☐ No | | | | | | | |
| Pattern: | | | | | | | | |
| Parkinsonism | ☐ Yes ☐ No | | | | | | | |
| Pattern: | | | | | | | | |
| **Cerebellar dysfunction** | | | | | | | | |
| Cerebellar dysfunction | ☐ Yes ☐ No | | | | | | | |
| Trunk axial ataxia | ☐ Yes ☐ No | | | | | | | |
| Pattern: | | | | | | | | |
| Appendicular dysmetria | ☐ Yes ☐ No | | | | | | | |
| Pattern: | | | | | | | | |
| **Other neurological abnormalities** | | | | | | | | |
| Neuro-cognitive abnormalities | ☐ Yes ☐ No ☐ Not Assessed | | | | | | | |
| i. Global development delay | ☐ Yes ☐ No | | | | | | | |
| ii. Delayed early motor milestones | ☐ Yes ☐ No | | | | | | | |
| iii. Delayed social development | ☐ Yes ☐ No | | | | | | | |
| iv. Delayed speech and language development | ☐ Yes ☐ No | | | | | | | |
| v. Intellectual disability | ☐ Yes ☐ No ☐ Not Assessed | | | | | | | |
| a. Severity | ☐ Mild ☐ Moderate ☐ Severe ☐ Profound | | | | | | | |
| vi. Loss of skills/regression<br>- age of onset: | ☐ Yes ☐ No | | | | | | | |
| vii. Comment: | | | | | | | | |
| **Neuro-psychiatric disease** | ☐ Yes ☐ No ☐ Not Assessed | | | | | | | |
| Comment: | | | | | | | | |
| **Neuro-vascular features** | | | | | | | | |
| Neuro-vascular features | ☐ Yes ☐ No | | | | | | | |
| i. Stroke-like episodes<br>Specify:<br>☐ vascular pattern ☐ non-vascular pattern ☐ stroke like episode with resolution | ☐ Yes ☐ No | | | | | | | |
| Pattern: | | | | | | | | |
| ii. Migraine | ☐ Yes ☐ No | | | | | | | |

| | |
|---|---|
| Pattern: | |
| iii. Headache other | ☐ Yes ☐ No |
| Pattern: | |
| **Seizures** | |
| i. Seizures | ☐ Yes ☐ No |
| ii. If Yes, seizure type ☐ Generalised ☐ Focal ☐ Other<br>- Other please specify: | |
| iii. Comment: | |
| **Brainstem/ cranial neuropathy** | |
| Brainstem/ cranial neuropathy | ☐ Yes ☐ No |
| Pattern: | |
| Spinal cord pathology | ☐ Yes ☐ No |
| Pattern: | |
| Peripheral neuropathy | ☐ Yes ☐ No |
| - If yes:<br>☐ Motor neuropathy<br>☐ Sensory neuropathy | |
| Pattern: | |
| **i. Walking** | |
| ☐ Normal ☐ Level surfaces ☐ Stick/s ☐ Crutches<br>☐ Walker/frame ☐ Wheel chair but transfers ☐ Non-ambulant | |
| Day to day variability of gait | ☐ Yes ☐ No |
| **Functional mobility scale (1-6):** | |
| (5m):<br>(50m):<br>(500m): | |
| **Hearing Loss** | |
| Sensorineural deafness | ☐ Yes ☐ No ☐ Not Assessed |
| if present: | ☐ congenital ☐ later onset |
| if present: | ☐ unilateral ☐ bilateral |
| Conductive deafness | ☐ Yes ☐ No ☐ Not Assessed |
| if present: | ☐ unilateral ☐ bilateral |
| Comment: | |
| **Ophthalmological abnormalities** | |
| Ophthalmological abnormalities | ☐ Yes ☐ No ☐ Not Assessed |
| i. Cortical blindness | ☐ Yes ☐ No ☐ Not Assessed |
| ii. Optic atrophy/neuropathy | ☐ Yes ☐ No ☐ Not Assessed |
| iii. Retinitis pigmentosa | ☐ Yes ☐ No ☐ Not Assessed |
| iv. Cataracts | ☐ Yes ☐ No ☐ Not Assessed |
| v. Corneal clouding | ☐ Yes ☐ No ☐ Not Assessed |
| vi. Glaucoma | ☐ Yes ☐ No ☐ Not Assessed |
| vii. Nystagmus | ☐ Yes ☐ No ☐ Not Assessed |
| **Skeletal deformities** | |
| Skeletal deformities | ☐ Yes ☐ No |
| i. Scoliosis | ☐ Yes ☐ No |
| ii. Lordosis | ☐ Yes ☐ No |
| iii. Kyphosis | ☐ Yes ☐ No |
| iv. Pectus excavatum | ☐ Yes ☐ No |
| v. Pectus carinatum | ☐ Yes ☐ No |

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|---|---|
| vi. Brachydactyly | ☐ Yes ☐ No |
| vii. Comment: | |
| **Cardiac** | |
| Cardiomyopathy | ☐ Yes ☐ No ☐ Not Assessed |
| - If present | ☐ Dilated ☐ Hypertrophic ☐ Left ventricular non-compaction<br>☐ Endocardial fibro-elastosis |
| Cardiac conduction defect | ☐ Yes ☐ No ☐ Not Assessed |
| If present specify: | |
| Structural heart defect | ☐ Yes ☐ No ☐ Not Assessed |
| Details: | |
| **Endocrinological abnormalities** | |
| Endocrinological abnormalities | ☐ Yes ☐ No ☐ Not Assessed |
| i. Diabetes mellitus | ☐ Yes ☐ No ☐ Not Assessed |
| ii. Diabetes insipidus | ☐ Yes ☐ No ☐ Not Assessed |
| iii. Hypopituitarism | ☐ Yes ☐ No ☐ Not Assessed |
| iv. Hypothyroidism | ☐ Yes ☐ No ☐ Not Assessed |
| v. Hypoparathyroidism | ☐ Yes ☐ No ☐ Not Assessed |
| vi. Adrenal insufficiency | ☐ Yes ☐ No ☐ Not Assessed |
| v. Hypogonadism | ☐ Yes ☐ No ☐ Not Assessed |
| vi. Growth hormone deficiency | ☐ Yes ☐ No ☐ Not Assessed |
| vii. SIADH | ☐ Yes ☐ No ☐ Not Assessed |
| viii. Comment: | |
| **GI Tract abnormalities** | |
| GI Tract abnormalities | ☐ Yes ☐ No ☐ Not Assessed |
| i. gastro-oesophageal reflux | ☐ Yes ☐ No |
| ii. Nasogastric tube feeding | ☐ Yes ☐ No |
| iii. Pseudo-obstruction | ☐ Yes ☐ No |
| iv. Malabsorption | ☐ Yes ☐ No |
| v. Constipation | ☐ Yes ☐ No |
| vi. Clinical evidence of hepatitis | ☐ Yes ☐ No |
| vii. Comment (hepatitis): | |
| vii. Comment: | |
| **Renal abnormalities** | |
| Renal abnormalities | ☐ Yes ☐ No ☐ Not Assessed |
| i. Hypertension | ☐ Yes ☐ No ☐ Not Assessed |
| ii. Renal impairment | ☐ Yes ☐ No ☐ Not Assessed |
| iii. Fanconi RTA | ☐ Yes ☐ No ☐ Not Assessed |
| iv. Focal segmental glomerulosclerosis | ☐ Yes ☐ No ☐ Not Assessed |
| v. Nephrotic syndrome | ☐ Yes ☐ No ☐ Not Assessed |
| vi. Other renal abnormality | |
| vii. Comment: | |
| **Haematological abnormalities** | |
| Haematological abnormalities | ☐ Yes ☐ No ☐ Not Assessed |
| i. Anaemia | ☐ Yes ☐ No ☐ Not Assessed |
| a. Type | ☐ Fe deficiency ☐ Sideroblastic ☐ Other: |
| Other (please specify) | |
| ii. Neutropenia | ☐ Yes ☐ No ☐ Not Assessed |
| iii. Cyclical | ☐ Yes ☐ No ☐ Not Assessed |

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|---|
| iv. Thrombocytopenia ☐ Yes ☐ No ☐ Not Assessed |
| v. Comment: |
| **Prenatal** |
| i. Pregnancy ☐ Normal ☐ Abnormal |
| a) Gestation (weeks completed): |
| b) Details: |
| c) Foetal movements ☐ Normal ☐ Abnormal |
| d) Foetal dysmorphology ☐ Yes ☐ No |
| e) Polyhydramnios ☐ Yes ☐ No |
| f) IUGR ☐ Yes ☐ No |
| ii. Labour ☐ Normal ☐ Abnormal |
| a) Specify (e.g. CTG abnormalities? Meconium stained liquor? ) |
| b) Type of delivery: ☐ NVD ☐ C-section ☐ Vacuum extraction ☐ Other |
| Other (please specify): |
| **Neonatal** |
| i. Ventilatory support ☐ Yes ☐ No |
| a) Improved over time? ☐ Yes ☐ No |
| ☐ Normalised ☐ Ongoing ventilatory support |
| Comment: |
| ii. Feeding assistance ☐ Yes ☐ No |
| b) Improved over time? ☐ Yes ☐ No |
| iii. Delayed head/neck control ☐ Yes ☐ No |
| iv. Apgar |
| 1 minute ☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 ☐ 8 ☐ 9 ☐ 10 |
| 5 minutes ☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 ☐ 8 ☐ 9 ☐ 10 |
| v. Seizures ☐ Yes ☐ No |
| vi. Hypoglycaemia(< 2.6 mmol/L) ☐ Yes ☐ No |
| vii. General Comments |
| **Medications:** |
| How medications are currently being administered? ☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 ☐ 8 ☐ 9 ☐ 10 |
| i. Medication 1: |
| ii. Medication 2: |
| iii. Medication 3: |
| iv. Medication 4: |
| v. Medication 5: |
| vi. Medication 6: |
| vii. Medication 7: |
| viii. Medication 8: |
| ix. Medication 9: |
| x. Medication 10: |
| xi. Comment: |
| **Findings on laboratory examinations - please enter most recent test results.** |
| Laboratory tests performed ☐ Yes ☐ No |
| i. Highest lactate _____ mmol/L _____ normal range Date of test: |
| ii. Venous/Cap lactate _____ mmol/L _____ normal range Date of test: |
| iii. Venous pyruvate _____ umol/L _____ normal range Date of test: |

| | | | | |
|---|---|---|---|---|
| iv. | Plasma alanine | _____ umol/L | _____ normal range | Date of test: |
| v. | CSF lactate | _____ mmol/L | _____ normal range | Date of test: |
| vi. | CSF pyruvate | _____ umol/L | _____ normal range | Date of test: |
| vii. | CSF alanine | _____ umol/L | _____ normal range | Date of test: |
| viii. | CSF glycine | _____ umol/L | _____ normal range | Date of test: |
| ix. | CSF protein | _____ g/L | _____ normal range | Date of test: |
| x. | CSF folate | _____ nmol/L | _____ normal range | Date of test: |
| xi. | Serum FGF21 | _____ U/L | _____ normal range | Date of test: |
| xii. | Serum GDF15 | _____ U/L | _____ normal range | Date of test: |
| xiii. | Lactate: Pyruvate ratio | | | |
| xiv. | Comment: | | | |
| **Paired tests** | | | | |
| i. | Venous lactate & CSF lactate | ☐ Yes ☐ No | | |
| ii. | Venous pyruvate & CSF pyruvate | ☐ Yes ☐ No | | |
| iii. | Plasma alanine & CSF alanine | ☐ Yes ☐ No | | |
| Urine amino acids and organic acids screen | | ☐ Normal | ☐ Abnormal | ☐ Not Assessed |
| | Findings: | | | |
| Serum CK | _____ U/L | _____ normal range | Date of test: | |
| **Liver function tests** | | | | |
| Liver function tests performed | | ☐ Yes | ☐ No | |
| i. | AST | _____ U/L | _____ normal range | Date of test: |
| ii. | ALT | _____ U/L | _____ normal range | Date of test: |
| iii. | Gamma-GT | _____ U/L | _____ normal range | Date of test: |
| iv. | ALP | _____ U/L | _____ normal range | Date of test: |
| v. | NH4 | _____ U/L | _____ normal range | Date of test: |
| vi. | PT | _____ U/L | _____ normal range | Date of test: |
| vii. | APPT | _____ U/L | _____ normal range | Date of test: |
| viii. | INR | _____ U/L | _____ normal range | Date of test: |
| ix. | Albumin | _____ U/L | _____ normal range | Date of test: |
| x. | Blood glucose | _____ U/L | _____ normal range | Date of test: |
| | Blood glucose ☐ Fasting | ☐ Random | ☐ Postprandial | |
| **Audiology** | | | | |
| Audiology testing | | ☐ Yes ☐ No | ☐ Not Assessed | |
| | i. Comment: | | | |
| Audiology report upload: | | | | |
| **Electrocardiography** | | | | |
| i. | Electrocardiography | ☐ Normal | ☐ Abnormal | ☐ Not Assessed |
| ii. | Abnormality | | | |
| iii. | Comment: | | | |
| **Spirometry** | | | | |
| Spirometry | | ☐ Yes ☐ No | ☐ Not assessed | |
| Age at test: | | | | |
| | i. $FEV_1$ values: | | | |
| | ii. FVC values: | | | |
| | iii. $FEV_1$/FVC ratio: | | | |
| Peak cough flow: | | ☐ Yes ☐No | | |
| Age at test: | | | | |

| i. Cough PEF value: | | | |
|---|---|---|---|
| Electroencephalogram - (can enter up to 3 EEGs on REDCap) | | | |
| Electroencephalogram (EEG) | ☐ Normal | ☐ Abnormal | ☐ Not Assessed |
| i. Date of test 1: | | | |
| Findings: | | | |
| Upload EEG report 1: | | | |
| ii. Date of test 2: | | | |
| Findings: | | | |
| Upload EEG report 2: | | | |
| iii. Date of test 3: | | | |
| Findings: | | | |
| Upload EEG report 3: | | | |
| MRI (can enter up to 5 Brain MRIs on REDCap) | | | |
| Brain MRI | ☐ Yes | ☐ No | ☐ Not Assessed |
| i. Date of test 1: | | | |
| Findings: | | | |
| Upload Brain MRI report 1: | | | |
| ii. Date of test 2: | | | |
| Findings: | | | |
| Upload Brain MRI report 2: | | | |
| iii. Date of test 3: | | | |
| Findings: | | | |
| Upload Brain MRI report 3: | | | |
| iv. Date of test 4: | | | |
| Findings: | | | |
| Upload Brain MRI report 4: | | | |
| v. Date of test 5: | | | |
| Findings: | | | |
| Upload Brain MRI report 5: | | | |
| Spinal MRI (can enter up to 5 Spinal MRIs on REDCap) | | | |
| Spinal MRI | ☐ Yes | ☐ No | ☐ Not Assessed |
| i. Date of test 1: | | | |
| Findings: | | | |
| Upload Spinal MRI report 1: | | | |
| ii. Date of test 2: | | | |
| Findings: | | | |
| Upload Spinal MRI report 2: | | | |
| iii. Date of test 3: | | | |
| Findings: | | | |
| Upload Spinal MRI report 3: | | | |
| iv. Date of test 4: | | | |
| Findings: | | | |
| Upload Spinal MRI report 4: | | | |
| v. Date of test 5: | | | |
| Findings: | | | |
| Upload Spinal MRI report 5: | | | |
| Proton MRS lactate peak<br>Site tested: | ☐ Yes | ☐ No | ☐ Not Assessed |
| Muscle MRI (can enter up to 5 Muscle MRIs on REDCap) | | | |

| Muscle MRI | ☐ Yes ☐ No Date of test: ☐ Not Assessed |
|---|---|
| i. Date of test 1: | |
| Findings: | |
| Muscle MRI 1 | |
| ii. Date of test 2: | |
| Findings: | |
| Muscle MRI 2 | |
| iii. Date of test 3: | |
| Findings: | |
| Muscle MRI 3 | |
| iv. Date of test 4: | |
| Findings: | |
| Muscle MRI 4 | |
| v. Date of test 5: | |
| Findings: | |
| Muscle MRI 5 | |
| Findings: | |
| P31 MRS<br>Site tested: Result: | ☐ Yes ☐ No ☐ Not Assessed |
| **Electromyography** | |
| Electromyography | ☐ Yes ☐ No ☐ Not Assessed |
| Myopathic ☐ Yes ☐ No | iii. Spontaneous activity ☐ Yes ☐ No |
| i. Mixed ☐ Yes ☐ No<br>v. Specify muscle: | iv. Complex discharges ☐ Yes ☐ No |
| vi. Short comment: | |
| **Nerve conduction studies** ☐ Yes ☐ No ☐ Not Assessed | |
| Nerve conduction studies | ☐ Yes ☐ No ☐ Not Assessed |
| i. Motor: | ☐ Demyelinating ☐ Axonal ☐ Mixed. ☐ Normal ☐ Not Assessed |
| ii. Sensory: | ☐ Demyelinating ☐ Axonal ☐ Mixed. ☐ Normal. ☐ Not Assessed |
| iii. Short comment: | |
| **Evoked potentials** | |
| i. Auditory evoked potentials<br>Short comment:<br>Upload report (auditory)<br>ii. Visual evoked potentials<br>Short comment:<br>Upload report (visual)<br>iii. Electroretinogram<br>Short comment: | ☐ Yes ☐ No ☐ Not Assessed<br><br><br>☐ Yes ☐ No ☐ Not Assessed<br><br><br>☐ Yes ☐ No ☐ Not Assessed |
| **Nerve biopsy** | |
| Nerve biopsy | ☐ Yes ☐ No ☐ Not Assessed |
| Short comment: | |
| **Skeletal muscle biopsy** | |
| Skeletal muscle biopsy | ☐ Yes ☐ No ☐ Not Assessed |
| Site of biopsy: | |
| Medications during biopsy: | ☐ Yes ☐ No |
| Medications: | |

| | |
|---|---|
| i. Ragged red fibres | ☐ Yes ☐ No |
| ii. Ragged blue fibres | ☐ Yes ☐ No |
| iii. COX-negative fibres | ☐ Yes ☐ No |
| iv. Reduced COX staining | ☐ Yes ☐ No |
| v. SDH positive | ☐ Yes ☐ No |
| vi. Steatosis | ☐ Yes ☐ No |
| EM abnormalities | ☐ Yes ☐ No |
| findings: | |
| MRC enzymology | ☐ Yes ☐ No ☐ Not Assessed |
| Muscle findings: | |
| Other comments: | |
| **Heart muscle biopsy** | |
| Heart muscle biopsy | ☐ Yes ☐ No ☐ Not Assessed |
| Medications during biopsy: | ☐ Yes ☐ No |
| Medications: | |
| i. Ragged red fibres | ☐ Yes ☐ No |
| ii. Ragged blue fibres | ☐ Yes ☐ No |
| iii. COX-negative fibres | ☐ Yes ☐ No |
| iv. Reduced COX staining | ☐ Yes ☐ No |
| v. SDH positive | ☐ Yes ☐ No |
| vi. Steatosis | ☐ Yes ☐ No |
| EM abnormalities | ☐ Yes ☐ No |
| findings: | |
| MRC enzymology | ☐ Yes ☐ No ☐ Not Assessed |
| Muscle findings: | |
| Other comments: | |
| **Liver biopsy** | |
| Liver biopsy | ☐ Yes ☐ No ☐ Not Assessed |
| Medications during biopsy: | |
| Medications | |
| i. Fibrosis | ☐ Yes ☐ No |
| ii. Cirrhosis | ☐ Yes ☐ No |
| iii. Steatosis | ☐ Yes ☐ No |
| EM abnormalities | ☐ Yes ☐ No |
| Findings: | |
| MRC enzymology | ☐ Yes ☐ No ☐ Not Assessed |
| Muscle findings: | |
| Other comments: | |
| **Echocardiography** | |
| Echocardiography | ☐ Normal ☐ Abnormal ☐ Not assessed |
| Abnormality: | |
| Upload report: | |
| **Previous Genetic Testing** | |
| Previous Genetic Testing | ☐ Yes ☐ No |
| Specify genetic testing: | |

| DNA Source | ☐ Blood<br>☐ Buccal | ☐ Tissue-muscle<br>☐ Saliva | ☐ Tissue- liver<br>☐ Urine | ☐ tissue-other:<br>☐ Other: |
|---|---|---|---|---|
| Specify other tissue: | | | | |
| Specify other DNA source: | | | | |