UKAS MEDICAL
9027

NHS
North East and Yorkshire
Genomic Laboratory Hub

NHS Highly Specialised Service for Rare Mitochondrial Disorders
Wellcome Centre for Mitochondrial Research, 4th Floor The Medical School, Newcastle University,
Framlington Place, Newcastle upon Tyne, NE2 4HH www.newcastle-mitochondria.com
Tel: 0191-2824375 **email**: tnu-tr.newcastle-mitochondria@nhs.net

REFERRAL FORM

Patient & Contact Details

Name: …….. DoB: …….. NHS No: ……

Address: ……

…… Post Code: ……

Patient ethnicity: …… Sex: M/F

Referring Hospital: …….. Hosp. No: ……..

Referring Consultant: …….. Specialty: ……..

Other Consultants: …….. Specialty: ……..

Address: ……..

Tel: …….. Secure E-mail (preferably nhs.net): ……..

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Please note all reports will be issued to all referring clinicians by secure email where possible

Name of person who collected sample (if known): ……..

Date of collection: ……Date of referral for mitochondrial studies: ……..

Sample details

Sample Type(s) and Date:

Blood	☐	Buccal	☐	Urine	☐
Muscle (specify)	☐ ……			Fibroblasts	☐
Other (specify)	☐ ……				

If the sample is muscle, please state if it has been obtained from:

Open biopsy ☐ Needle biopsy ☐ Post-mortem ☐ Endomyocardial biopsy ☐

Tests Requested

Biochemistry/Immunohistochemistry ☐ **Histochemistry ☐**

Genetics ☐ (Specify on Page3) **Other ☐** ……

If <u>urgent</u> testing is required, please contact the laboratory by phone or email to confirm

Details of previous or ongoing genetic tests requested elsewhere (required):

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Clinical Details

Proband	☐	Parental consanguinity	☐
Affected relative	☐	Maternal inheritance	☐
Unaffected relative	☐	Age at onset: ……	

Family history: ……

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Classical Clinical Phenotype? Y/N

If **yes**, then which of the following?

Pearson's syndrome	☐	LIMM	☐	NARP/MILS	☐
KSS	☐	MNGIE	☐	LHON	☐
CPEO	☐	MIDD	☐	Deaf/Dystonia	☐
CPEO (+)	☐	SNHL	☐	Leigh syndrome	☐
MELAS	☐	HCM	☐	Alpers' syndrome	☐
MERRF	☐	Pure Myopathy	☐		

If **no**, then which of the following clinical features are present?

Stroke/S-L Episodes	☐	Dev Delay	☐	Deafness	☐
Encephalopathy	☐	Hypotonia	☐	Anaemia	☐
Seizures	☐	Dystonia	☐	Renal dis	☐
Migraine	☐	Central apnoea	☐	Optic atrophy	☐
Diabetes	☐	Dysphagia	☐	Retinopathy	☐
Endocrinopathy	☐	Constipation	☐	Nystagmus	☐
Growth failure	☐	Liver disease	☐	Fatigue	☐
Cardiomyopathy	☐	Myopathy	☐	Dementia	☐
Failure to thrive	☐	Myalgia	☐	Learning Diff	☐

Further clinical details:

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Local report on muscle biopsy:

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Clinical Investigations

Bl. Lactate: ……mmol/l	CSF Lactate: ……mmol/l	Serum CK: ……iu
ECG abnormal: Y/N	EEG abnormal: Y/N	Echo abnormal: Y/N

Brain MRI/CT findings:

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Molecular Genetic Investigations:

R42 LHON ☐

R64 MELAS or MIDD - m.3243A>G ☐

R65 Aminoglycoside exposure posing risk to hearing – m.1555A>G ☐

R299 mtDNA rearrangement (long range PCR) ☐

R301 mtDNA depletion (real-time PCR) ☐

R315 *POLG*-related disorder ☐

R350 MERRF common pathogenic variants ☐

R351 NARP or maternally inherited Leigh syndrome ☐

R394 MNGIE - *TYMP* Gene Sequencing ☐

R395 Thiamine metabolism dysfunction syndrome 2 – *SLC19A3* Gene Sequencing ☐

R396 Mitochondrial complex V deficiency, *TMEM70* type ☐

R397 Maternally inherited cardiomyopathy - m.4300A>G ☐

NGS:

R63 Possible mitochondrial disorder – nuclear genes ☐

R300 mtDNA full genome sequencing (NGS) ☐

R316 Pyruvate dehydrogenase (PDH) deficiency ☐

R317 Mitochondrial Liver Disease, including transient infantile liver failure ☐

R352 Mitochondrial DNA maintenance disorder ☐

R353 Mitochondrial disorder with complex I deficiency ☐

R354 Mitochondrial disorder with complex II deficiency ☐

R355 Mitochondrial disorder with complex III deficiency ☐

R356 Mitochondrial disorder with complex IV deficiency ☐

R357 Mitochondrial disorder with complex V deficiency ☐

Familial Testing:

R240 Diagnostic testing for known pathogenic variant (specify) ☐……..……..

R242 Predictive testing for known familial variant (specify) ☐……..…..

R244 Carrier testing for known familial variant (specify) ☐…….

R246 Carrier testing at population risk for partners of known carriers of autosomal recessive disorders (specify gene) ☐……..

R375 Family follow up testing to aid variant interpretation ☐……

Other ☐……...

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