UKAS MEDICAL 9027

The Newcastle upon Tyne Hospitals NHS Foundation Trust

Newcastle Highly Specialised Mitochondrial Diagnostic Service

Wellcome Centre for Mitochondrial Research, 4th Floor Cookson Building, The Medical School,
Newcastle University, Framlington Place, Newcastle upon Tyne, NE2 4HH
Telephone: 0191-2824375
www.mitoresearch.org.uk e-mail: tnu-tr.newcastle-mitochondria@nhs.net

REFERRAL FORM

Patient & Contact Details

Name: …… DoB: …… NHS No: ……..
Address:……
…… Post Code: ……..…… Sex: M/F
Referring Hospital: ……Hosp. No: ……
Referring Consultant: …… Specialty: ……
Other Consultants: …… Specialty: ……
Address for correspondence: ……
……
Tel: …… Fax: …… E-mail: ……
GP name and address: ……
……GP Tel: ……
Name of person who collected sample (if known)……
Date of collection……Date of referral for mitochondrial studies: ……

☐ **Please tick box to confirm consent obtained and enclose a copy of this with the referral form**

Sample details

Urgent ☐ **Routine** ☐

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 below)

Molecular Genetic Investigations:

m.3243A>G	☐	*MTATP6/8* Gene Seq.	☐	MERRF	☐	Common *POLG*	☐
LHON	☐	m.1555A>G	☐	SNHL	☐	Full *POLG* analysis	☐
mtDNA full genome sequencing (NGS)			☐	mtDNA rearrangement (long range PCR)			☐
mtDNA depletion (real-time PCR)			☐	Nuclear Complex II 6 Gene Analysis			☐

Other ☐……
Variant Screen (specify) ☐ ……
Familial Variant Screen (specify) ☐ ……
Exome Confirmation (specify) ☐ ……
Nuclear Gene Analysis (specify) ☐ ……

Targeted NGS Gene Panels where appropriate:

mtDNA maintenance/depletion 18 Gene NGS Panel ☐ Nuclear Complex I 49 Gene NGS Panel ☐

Clinical Details

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

Family history:

……

……

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:

……

……

……

……

……

Local report on muscle biopsy:

……

……

……

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:

……

……

……

……