



Accredited Medical Laboratory
Reference No: 3118

Newcastle Highly Specialised Mitochondrial Diagnostic Service

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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:

..... Tel:

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 Fibroblasts Urine

Sk. Muscle (Quads) Sk. Muscle (Site) (.....) Cardiac Muscle

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

Open biopsy Needle biopsy Post-mortem Endomyocardial biopsy

Tests Requested

Histochemistry

Biochemistry

Genetics (Specify)

Mutation Screen

Mito Gene Sequence

m.3243A>G *MTATP6/8* Gene Seq. mtDNA rearrangement (long range PCR)

MERRF *MTND* Gene Seq. mtDNA depletion (real-time PCR)

LHON *MTCYB* Gene Seq.

SNHL mtDNA Genome Seq.

Nuclear Gene Sequence

POLG *RRM2B* *SLC25A4* *TK2*

PEO1 *DGUOK/MPV17* *POLG2* *SUCLA2/SUCLG1*

TRMU *RARS2* *DARS2* *MTO1*

AARS2 *AGK* *NFU1* *BCS1L*

Nuclear Complex II Gene Analysis *TMEM70* *ATPAF2*

Other (Please specify)

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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