

REFERRAL FORM

NEWCASTLE MITOCHONDRIAL NCG DIAGNOSTIC LABORATORY

Rare Mitochondrial Disorders Service for Adults and Children

Mitochondrial Research Group, 4th Floor, The Medical School, Newcastle University,
Framlington Place, Newcastle upon Tyne, NE2 4HH.

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Patient & contact details

Name: DoB: Sex: M/F

Address:

..... Post Code:

Referring Hospital: Hosp. No:

Referring Consultant: Specialty:

Other Consultants: Specialty:

Address for correspondence:

Tel: Fax: email:

GP name and address:

..... Tel:

Date of referral for mitochondrial studies.:

Sample details

PLEASE READ THE ATTACHED INSTRUCTIONS FOR SAMPLE PREPARATION & TRANSPORT.

Sample Type(s) and Date:

Blood Fibroblasts

Sk. Muscle (Quads) Urine

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

MELAS

MERRF

LHON

SNHL

MITO GENE SEQUENCE

MTATP6/8 GENE SEQ

MTND GENE SEQ

MTCYB GENE SEQ

WHOLE GENOME SEQ

NUCLEAR GENE SEQUENCE

POLG1

SLC25A4

PEO1

TK2

DGOUK

mtDNA rearrangement (LRPCR ± Southern blot)

mtDNA depletion (RealTime PCR)

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?

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

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

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

Further clinical details:
.....
.....

Local report on muscle biopsy:
.....

Clinical Investigations

Bl. Lactate.....mmol/L CSF Lactate.....mmol/L Serum CKIU.
ECG abnormal **Y/N** EEG abnormal **Y/N** Echo abnormal **Y/N**

Brain MRI/CT findings:
.....