



Accredited laboratory
Reference: 0745



Genetics Laboratories, The Churchill, Old Road, Headington, Oxford, OX3 7LJ
Head of Laboratory: Dr Anneke Seller ☎ 01865 225594 www.oxfordradcliffe.nhs.uk/geneticslab

GENETIC DIAGNOSTIC AND ADVISORY SERVICE FOR MITOCHONDRIAL DISEASES
(NCG RARE MITOCHONDRIAL DISORDERS SERVICE FOR ADULTS AND CHILDREN,
FOR ENGLAND AND SCOTLAND – OXFORD CENTRE)

Name:.....DoB:..... Age at referral:..... M/F:.....

Address:.....

Hosp. no: Referring Consultant:.....Hospital:.....

Clinician's phone no:.....NHS no:.....Referral date:.....

Sample provided (please circle – if extracted DNA, please indicate source):

Blood Muscle Liver Fibroblasts Other (specify).....

Date sample taken: **For muscle/liver, is sample biopsy or post mortem?:**

Other samples available (please circle): Blood Muscle Liver Fibroblasts Other (specify).....

CLINICAL DIAGNOSES (tentative):

- | | |
|---|--|
| <input type="checkbox"/> CPEO/KSS | <input type="checkbox"/> MELAS/MERRF |
| <input type="checkbox"/> PEARSON | <input type="checkbox"/> INFANTILE LEIGH |
| <input type="checkbox"/> LHON | <input type="checkbox"/> CARDIOMYOPATHY |
| <input type="checkbox"/> MULTISYSTEM DISEASE | <input type="checkbox"/> ALPERS |
| <input type="checkbox"/> SANDO | <input type="checkbox"/> MIRAS |
| <input type="checkbox"/> MTDNA DEPLETION SYNDROME | <input type="checkbox"/> OTHER:..... |

CLINICAL DETAILS:

Age at onset:.....	Family history: Y/N.....
MR/delayed milestones/dementia: Y/N.....	Stroke-like episodes: Y/N.....
Fits/encephalopathy: Y/N.....	Deafness: Y/N.....
Dystonia/myoclonus: Y/N.....	Retinopathy: Y/N.....
Optic disc pallor: Y/N.....	CPEO/ptosis: Y/N.....
Nystagmus: Y/N.....	Myopathy: Y/N.....
Ataxia: Y/N.....	Hypotonia: Y/N.....
Respiratory failure: Y/N.....	Renal: Y/N.....
Hepatic: Y/N.....	Haematological: Y/N.....
Diabetes: Y/N.....	Other Endocrine?.....
Fed by PEG/mild feeding problems?.....	
Cardiac: cardiomyopathy/abnormal ECG/abnormal ECHO?.....	
Other:.....	

RESULTS of INVESTIGATIONS:

Raised CK: Y/N.....
Lactic Acid: Normal/Raised in Serum:.....Normal/Raised in CSF:.....
Imaging MRI or CT: Normal/Leigh/Other:.....
Muscle biopsy: RRFs/Low COX/Nonspecific abnormal/Normal:.....
Muscle/Liver respiratory chain enzyme analysis: Abnormal/Normal:.....

Relevant correspondence/other information: include unusual features, e.g. scoliosis/contractures (or perhaps include this with each feature?).....

PLEASE RETURN COMPLETED FORM TO: Dr Anneke Seller (at the above address).
Tel 01865 225594 for laboratory queries. Contact Prof J Poulton, Joanna.poulton@obs-gyn.ox.ac.uk, for clinical queries.