



Accredited laboratory

Oxford Radcliffe Hospitals



NHS Trust

Genetics Laboratories, The Churchill, Old Road, Headington, Oxford, OX3 7LJ
Head of Laboratory: Dr Anneke Seller 01865 225594



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:.....Referral date:.....

Sample provided (please circle): 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):

- CPEO/KSS MELAS/MERRF
PEARSON INFANTILE LEIGH
LHON CARDIOMYOPATHY
MULTISYSTEM DISEASE ALPERS
SANDO MIRAS
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/ataxia: Y/N..... Myopathy/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 biochemistry: 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, 01865 221007 for clinical queries.