CPA Accredited laboratory

Oxford Radcliffe Hospitals NHS
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
- ☐ PEARSON
- ☐ LHON
- ☐ MULTISYSTEM DISEASE
- ☐ SANDO
- ☐ OTHER:.....
- ☐ MELAS/MERRF
- ☐ INFANTILE LEIGH
- ☐ CARDIOMYOPATHY
- ☐ ALPERS
- ☐ MIRAS

CLINICAL DETAILS:

Age at onset:.....

Family history: Y/N.....

MR/delayed milestones/DEMENTIA: Y/N.....
Fits/encephalopathy: Y/N.....
Dystonia/myoclonus: Y/N.....
Optic disc pallor: Y/N.....
Nystagmus/ataxia: Y/N.....
Respiratory failure: Y/N.....
Hepatic: Y/N.....
Diabetes: Y/N.....

Stroke-like episodes: Y/N.....
Deafness: Y/N.....
Retinopathy: Y/N.....
CPEO/ptosis: Y/N.....
Myopathy/hypotonia: Y/N.....
Renal: Y/N.....
Haematological: 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?).....
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PLEASE RETURN COMPLETED FORM TO: DR ANNEKE SELLER (at the above address).
Tel 01865 225594 for laboratory queries, 01865 221007 for clinical queries.