Oxford Regional Genetics Laboratories
Churchill Hospital
Old Road, Headington
Oxford OX3 7LE

UKAS MEDICAL 8694

Oxford University Hospitals
NHS Foundation Trust
Director of Laboratory: Carolyn Campbell, FRCPath
www.ouh.nhs.uk/geneticslab oxford.mitogenetics@nhs.net ☎ +44 (0)1865 226001

GENETIC DIAGNOSTIC AND ADVISORY SERVICE FOR MITOCHONDRIAL DISEASES
NHS Highly Specialised Services (HSS) for Rare Mitochondrial Disorders – 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):

- ☐ CPEO/KSS
- ☐ PEARSON
- ☐ LHON
- ☐ MULTISYSTEM DISEASE
- ☐ NEUROPATHY such as SANDO
- ☐ MTDNA DEPLETION SYNDROME
- ☐ MELAS/MERRF
- ☐ INFANTILE LEIGH
- ☐ CARDIOMYOPATHY
- ☐ ALPERS
- ☐ MOVEMENT DISORDER such as MIRAS
- ☐ OTHER:.....

CLINICAL DETAILS:

Age at onset:.....

Family history: Y/N.....
Consanguinity: Y/N.....

Developmental delay: Y/N.....
Dementia: Y/N.....
Seizures: Y/N.....
Encephalopathy: Y/N.....
Dystonia: Y/N.....
Myoclonus: Y/N.....
Movement disorder : Y/N.....
Neuropathy: Y/N.....
Ataxia: Y/N.....
Stroke-like episodes: Y/N.....

Haematological: Y/N.....
Hepatic: Y/N.....
Renal: Y/N.....
Deafness: Y/N.....
Diabetes: Y/N.....

CPEO: Y/N.....
Ptosis: Y/N.....
Retinopathy: Y/N.....
Optic disc pallor: Y/N.....
Nystagmus: Y/N.....
Cataracts: Y/N.....

Myopathy/Muscle weakness: Y/N.....
Hypotonia: Y/N.....
Muscle fatigue: Y/N.....
Other muscle: Y/N.....
(e.g. contractures, cramps, pain)
Respiratory failure: Y/N.....
Feeding problems (e.g. PEG fed): Y/N.....

Other Endocrine: Y/N.....

Cardiac: Cardiomyopathy/Abnormal ECG/Abnormal ECHO.....

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 histology: RRFs/Low COX/Nonspecific abnormal/Normal:.....
Muscle respiratory chain enzyme analysis: Abnormal/Normal:.....
Liver respiratory chain enzyme analysis: Abnormal/Normal:.....

Relevant correspondence/other information: (include additional clinical features)

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PLEASE RETURN COMPLETED FORM (AND ACCOMPANYING SAMPLES TO):
Carl Fratter (at the above address). Tel 01865 226001 or email oxford.mitogenetics@nhs.net for laboratory queries.
Contact Dr Victoria Nesbitt, mitohelp@ouh.nhs.uk, mobile 07880071103, for clinical queries.