Oxford Regional Genetics Laboratories
Oxford University Hospitals NHS Foundation Trust
The Churchill Hospital
Oxford OX3 7LE
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NHS
Central & South
Genomic Laboratory Hub
Oxford Genetics Laboratories

Genetic Diagnostic and Advisory Service for Mitochondrial Diseases
Request Form

Patient and Requester Details

NHS No:		**Sex:**		**Date of Birth:**	
Surname:		**Address (including postcode):**			
Forename:					
Ethnicity:		**Clinician:**			
Hospital:		**Hospital No:**			
Email:					
Reporting address:					

Sample Information

Sample provided:	Blood ☐	Muscle* ☐	Liver* ☐	Fibroblasts ☐	Other:	
Date sample taken:		***Biopsy or post mortem?:**		Biopsy ☐	Post mortem ☐	
Other samples available:	Blood ☐	Muscle ☐	Liver ☐	Fibroblasts ☐	Other:	

Clinical Diagnoses (tentative)

CPEO/KSS :	☐	**MELAS/MERRF:**	☐
Pearson :	☐	**Infantile leigh:**	☐
LHON :	☐	**Cardiomyopathy:**	☐
Multisystem disease:	☐	**Alpers:**	☐
Neuropathy such as SANDO :	☐	**Movement disorder such as MIRAS:**	☐
MtDNA depletion syndrome:	☐	**Other:**	

Clinical Details

Age at onset:		**Family history:**	☐		
		Consanguinity:	☐		
Developmental delay:	☐	**Myopathy/Muscle weakness:**	☐	**Respiratory failure:**	☐
Dementia:	☐	**Hypotonia:**	☐	**Feeding problems:**	☐
Seizures:	☐	**Muscle fatigue:**	☐	**Haematological:**	☐
Encephalopathy:	☐	**Other muscle (e.g. contractures, pain):**	☐	**Hepatic:**	☐
Dystonia:	☐	**CPEO:**	☐	**Renal:**	☐
Myoclonus:	☐	**Ptosis:**	☐	**Deafness:**	☐
Movement disorder:	☐	**Nystagmus:**	☐	**Diabetes:**	☐
Neuropathy:	☐	**Cataracts:**	☐	**Other Endocrine:**	☐
Ataxia:	☐	**Retinopathy:**	☐		
Stroke-like episodes:	☐	**Optic disc pallor:**	☐		

Results of Investigations

Raised CK:		**Imaging MRI or CT:**	Normal ☐ Leigh ☐	Other:	
Lactic Acid (in serum):	Normal ☐	Raised ☐	**Lactic Acid in CSF:**	Normal ☐	Raised ☐
Muscle histology:	RRFs ☐	Low COX ☐	Nonspecific abnormal ☐	Normal ☐	
Muscle respiratory chain enzyme analysis:			Abnormal ☐	Normal ☐	
Liver respiratory chain enzyme analysis:			Abnormal ☐	Normal ☐	

Relevant correspondence and other information (Include additional clinical features)

In submitting this sample the clinician confirms that consent has been obtained for testing and storage. Anonymised stored samples may be used for quality control procedures including validation of new genetic tests.

Please return completed form (and accompanying samples) to the above address.

Lab queries: mito.oxfordgenetics@ouh.nhs.uk Website: www.ouh.nhs.uk/geneticslab Admin Office: 01865 226001
Clinical queries: Dr Victoria Nesbitt (mitohelp@ouh.nhs.uk, mobile: 07880071103, admin: 01865 225 899)