

Genetic Diagnostic and Advisory Service for Mitochondrial Diseases Request Form

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

*Please state if karyotypic and/or phenotypic sex differ from given sex.

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