

**Genetic Diagnostic and Advisory Service for Mitochondrial Diseases**  
**Request Form**

<b>Patient and Requester Details</b>							
<b>NHS No:</b>		<b>Sex:</b>		<b>Date of Birth:</b>			
<b>Surname:</b>		<b>Address (including postcode):</b>					
<b>Forename:</b>							
<b>Ethnicity:</b>		<b>Clinician:</b>					
<b>Hospital:</b>		<b>Hospital No:</b>					
<b>Email:</b>							
<b>Reporting address:</b>							
<b>Sample Information</b>							
<b>Sample provided:</b>	Blood	Muscle*	Liver*	Fibroblasts	Other:		
<b>Date sample taken:</b>		<b>*Biopsy or post mortem?:</b>		Biopsy		Post mortem	
<b>Other samples available:</b>	Blood	Muscle	Liver	Fibroblasts	Other:		
<b>Clinical Diagnoses (tentative):</b>							
<b>CPEO/KSS:</b>		<b>MELAS/MERRF:</b>					
<b>Pearson:</b>		<b>Infantile leigh:</b>					
<b>LHON:</b>		<b>Cardiomyopathy:</b>					
<b>Multisystem disease:</b>		<b>Alpers:</b>					
<b>Neuropathy such as SANDO:</b>		<b>Movement disorder such as MIRAS:</b>					
<b>MtDNA depletion syndrome:</b>		<b>Other:</b>					
<b>Clinical Details</b>							
<b>Age at onset:</b>		<b>Family history:</b>					
		<b>Consanguinity:</b>					
<b>Developmental delay:</b>		<b>Myopathy/Muscle weakness:</b>			<b>Respiratory failure:</b>		
<b>Dementia:</b>		<b>Hypotonia:</b>			<b>Feeding problems:</b>		
<b>Seizures:</b>		<b>Muscle fatigue:</b>			<b>Haematological:</b>		
<b>Encephalopathy:</b>		<b>Other muscle (e.g. contractures, pain):</b>			<b>Hepatic:</b>		
<b>Dystonia:</b>		<b>CPEO:</b>			<b>Renal:</b>		
<b>Myoclonus:</b>		<b>Ptosis:</b>			<b>Deafness:</b>		
<b>Movement disorder:</b>		<b>Nystagmus:</b>			<b>Diabetes:</b>		
<b>Neuropathy:</b>		<b>Cataracts:</b>			<b>Other Endocrine:</b>		
<b>Ataxia:</b>		<b>Retinopathy:</b>					
<b>Stroke-like episodes:</b>		<b>Optic disc pallor:</b>					
<b>Results of Investigations</b>							
<b>Raised CK:</b>		<b>Imaging MRI or CT:</b>		Normal	Leigh	Other:	
<b>Lactic Acid (in serum):</b>	Normal	Raised		<b>Lactic Acid in CSF:</b>		Normal	Raised
<b>Muscle histology:</b>	RRFs	Low COX	Nonspecific abnormal		Normal		
<b>Muscle respiratory chain enzyme analysis:</b>			Abnormal		Normal		
<b>Liver respiratory chain enzyme analysis:</b>			Abnormal		Normal		
<b>Relevant correspondence and other information</b> (Include additional clinical features)							

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