MRC Centre for Neuromuscular Diseases

University College London Hospitals NHS
NHS Foundation Trust

National Commissioning Group (NCG) For Highly Specialised Services

UCLH QUEEN SQUARE NCG CLINICAL AND DIAGNOSTIC SERVICE FOR RARE MITOCHONDRIAL DISEASES IN ADULTS AND CHILDREN

Genetic analysis request form

Name: ……

DoB:…….Age at Referral:……..

M/F:…….

Address:……

……Postcode:……..

Hospital. No: ……. NHS number:…….....

Hospital:…….

Referring Consultant: …….

Clinican's phone no:……Email:…….

Referral date:……..

Consent for genetic analysis

It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test and that the sample may be stored for future testing related to specific diagnosis for the patient. In signing this form the clinician confirms that they have obtained consent for testing and storage. The patient should be advised that the sample may be used anonymously for quality assurance, research and training purposes. Please advise us of any restrictions. This laboratory follows the recommendations laid down by the Joint Committee on Medical Genetics guidance document "Consent and Confidentiality in Genetic Practice September 2011".

CLINICIAN NAME:…… SIGNATURE:…….

Sample provided (please circle): Blood Muscle Liver Fibroblasts Other

Date of sample collection: ……

RESULTS OF INVESTIGATIONS IF AVAILABLE

CK: ……

Lactic Acid: Serum: ……CSF:……..

Imaging MRI or CT: …….

Muscle biopsy result:……

EEG result ……

EMG/NCV result……

Clinical details

FAMILY HISTORY Y/N			
Age at onset:……		Maternal inheritance suspected	
This is proband		Parental consanguinity:	
This is affected relative		Family history/pedigree:	
This is unaffected relative			

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SUSPECTED TYPICAL MITOCHONDRIAL PHENOTYPE? Y/N			
CPEO		MELAS	
KSS		MERRF	
PEARSON		LEIGH	
LHON		CARDIOMYOPATHY	
ENCEPHAOLPATHY		ALPERS	
SANDO		MINGIE	
NARP/MILS		MIDD	
ISOLATED MYOPATHY		DEAF DYSTONIA	

IF NOT TYPICAL PHENOTYPE WHICH FEATURES ARE PRESENT? Y/N					
Delayed milestones		Dementia		Stroke-like episode	
Seizures		Encephalopathy		Deafness	
Dystonia		Myoclonus		Retinopathy	
Optic. atrophy		Ptosis		Nystagmus	
Ataxia.		Myopathy		Hypotonia	
Respiratory failure		Renal		Hepatic	
Fatigue		Constipation		Diabetes	
Dysphagia		Anaemia		Migrane	
Cardiomyopathy		Myopathy		Learning Diff	
Growth failure		Myalgia			

SAMPLE REQUIREMENTS

The standard samples sent for analysis are fresh blood in EDTA (ideally 2x6ml), frozen muscle or extracted DNA. If sending DNA extracted by another laboratory, please indicate the original sample type. Other tissues may be accepted after discussion with the laboratory.

AVAILABLE ANALYSIS

For blood: The 3 'common' mitochondrial DNA mutations (m.3243A>G, m.8344A>G and m.8993T>G/C) will be analysed as a preliminary screen. Analysis of large scale rearrangements will be performed in blood samples from patients under the age of 20 years at referral. Sequencing of the complete mitochondrial genome is available in blood if clinically appropriate following discussion with the mitochondrial clinic and/or laboratory.

For muscle: Complete sequence analysis of the mitochondrial genome and analysis of large scale rearrangements will undertaken for cases with strong clinical suspicion and/or muscle biopsy and/or biochemical respiratory chain abnormalities. If mitochondrial depletion is suspected and analysis for mtDNA copy number is required a tissue DNA sample will be forwarded to the mitochondrial NCG service in Oxford.

Nuclear genes: Analysis of nuclear genes involved in mitochondrial disease is available at the Oxford and Newcastle laboratories. Gene-specific panels are available and clinical advice can be sought from the Mitochondrial clinic. Blood DNA can be forwarded from the Neurogenetics laboratory as appropriate.

Please send samples to:
Neurogenetics Department
6th Floor, Institute of Neurology,
Queen Square House
Queen Square, London
WC1N 3BG

Website: www.uclh.nhs.uk/neurogeneticslab
General Enquires: ucl-tr.NHNNgenetics@nhs.net
Tel: 020 344 84250