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

Patient & Contact Details

Patient Name:……

DoB: …… NHS No: ……

Address:……

…… Post Code: ……

Patient ethnicity:…… Sex: M/F …….

Referring Hospital: …… Hosp. No: ……

Referring Consultant: …… Specialty: ……

Other Consultants: …… Specialty:……

Address for correspondence: ……

Tel:…… E-mail (preferably nhs.net……

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 details

Sample Type(s) and Date:

Please see sample requirement information on page 2

Blood Buccal Urine* Muscle (specify) Fibroblasts

Other (specify) ……

If the sample is muscle, please state if it has been obtained from:

Open biopsy Needle biopsy Post-mortem Endomyocardial biopsy

Clinical Details

This is: Proband Affected relative Unaffected relative

If affected: Age at onset: ……

Family history of: Parental consanguinity Maternal inheritance

Local report on muscle biopsy:

……

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Clinical Investigations

Bl. Lactate …… mmol/l CSF Lactate …… mmol/l Serum CK …… iu

ECG abnormal Y/N EEG abnormal Y/N Echo abnormal Y/N

Brain MRI/CT findings:

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Classical Clinical Phenotype? Y/N

If yes, then which of the following?

Pearson's syndrome		LIMM		NARP/MILS	
KSS		MNGIE		LHON	
CPEO		MIDD		Deaf/Dystonia	
CPEO (+)		SNHL		Leigh syndrome	
MELAS		HCM		Alpers' syndrome	
MERRF		Pure Myopathy			

If no, then which of the following clinical features are present?

Stroke/S-L Episodes		Dev Delay		Deafness	
Encephalopathy		Hypotonia		Anaemia	
Seizures		Dystonia		Renal dis	
Migraine		Central apnoea		Optic atrophy	
Diabetes		Dysphagia		Retinopathy	
Endocrinopathy		Constipation		Nystagmus	
Growth failure		Liver disease		Fatigue	
Cardiomyopathy		Myopathy		Dementia	
Failure to thrive		Myalgia		Learning Diff	

Further clinical details:

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Molecular Genetic Investigations:

R42 LHON
R64 MELAS or MIDD - m.3243A>G
R299 mtDNA rearrangement (long range PCR)
R301 mtDNA depletion (real-time PCR)
R350 MERRF common pathogenic variants
R351 NARP or maternally inherited Leigh syndrome
R397 Maternally inherited cardiomyopathy - m.4300A>G
NGS:
R63 Possible mitochondrial disorder – nuclear genes
R300 mtDNA full genome sequencing (NGS)
R352 Mitochondrial DNA maintenance disorder
Familial Testing:
R240 Diagnostic testing for known pathogenic variant (specify) ……..
R242 Predictive testing for known familial variant (specify) ……..
R244 Carrier testing for known familial variant (specify) …….
R246 Carrier testing at population risk for partners of known carriers of autosomal recessive disorders (specify gene) ……
R375 Family follow up testing to aid variant interpretation ……
Other ……..
……

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.
*Urine should be ~50mls early morning sample and should arrive in lab within 48hrs
Other tissues may be accepted after discussion with the laboratory.

Address to:

North Thames GLH, Rare & Inherited Disease Genomic Laboratory
Specimen Reception,
Level 5 Barclay House,
37 Queen Square,
London WC1N 3BH

Opening hours: Monday to Friday 9.00am to 5.30pm (please ensure samples arrive by 5pm)