

Rare Disease Testing Service – Application for mutation confirmation (page 1)

Patient (Proband) Details

Forename	Surname	Other Names	Sex M F
Address (please include a Post Code)		Date of Birth (dd/mm/yyyy)	
Local Patient Identifier (your reference)		NHS No.	
Is a DNA sample available?	Yes	No	
Has the DNA sample been extracted under a research or diagnostic protocol?	Research	Diagnostic	
Can a fresh blood sample be obtained?	Yes	No	
Family Tree/Pedigree attached	Yes	No	

Family Details – Father

Forename	Surname	Date of Birth (dd/mm/yyyy)
Fresh Blood/DNA sample available?	Yes	No

Family Details - Mother

Forename	Surname	Date of Birth (dd/mm/yyyy)
Fresh Blood/DNA sample available?	Yes	No

Disease & Mutation Information

Disease Name	OMIM No.		
Mode of Inheritance			
Autosomal Dominant Recessive	X-linked Dominant Recessive	Mitochondrial	Other (specify)
Class of Mutation			
Point (single base substitution insertion/deletion of a known number of nucleotides)	*Deletion/Duplication (insertion/deletion of unknown number of nucleotides)	*Dynamic (repeat expansion)	*Methylation
Mutation Description (Please provide a concise description or standard name for each mutation to allow us to unequivocally identify it. We must be given a reference sequence or GenBank accession number against which each mutation has been named)			
GenBank Acc No.	Reference Sequence Attached	Yes	No

*Services for these classes of mutations are not currently available. We can however accept samples for banking pending future testing for these mutation types.

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Consultant Contact Details – for reporting

Consultant Name

Consultant Address

Tel no:

Fax no:

Email:

Further information (for audit and resource planning)

Information on the impact of the disease – Is this condition

	Yes	No
Lethal?		
Associated with physical disability?		
Associated with intellectual disability?		
Associated with both physical & intellectual disability?		
Untreatable?		
Associated with life-long treatment?		
Associated with treatment with a high morbidity and mortality?		
Late onset?		
Associated with reduced penetrance?		

Information on the incidence of the disease

<10 cases reported in literature
10-50 cases reported in the literature
50-100 cases reported in the literature
Incidence if known

Information on the demand for testing – is this request

For carrier testing in family members?
For pre-natal diagnosis?
For diagnosis in family members?
How many further tests are anticipated?
Is testing available outside the UK?
If Yes, where?
If known what is the cost per case?

When completed this form should be printed off and sent to:

Nicola Marchbank/Andrew Wallace, Rare Disease Testing Service,
Regional Molecular Genetics Laboratory
Regional Genetics Service, St Mary's Hospital,
Oxford Road, Manchester M13 9WL
Fax: 0161-276-6606 Tel: 0161-276-6122