GENOMIC DIAGNOSTICS LABORATORY
MANCHESTER CENTRE FOR GENOMIC MEDICINE

NHS

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)

Please include a post code

Date of Birth (dd/mm/yyyy)

NHS No.

Local Patient Identifier (your reference)

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		X-linked		Mitochondrial	Other (specify)
Dominant	Recessive	Dominant	Recessive		
☐	☐	☐	☐	☐	☐

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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Rare Disease Testing Service – Application for mutation confirmation (page 2)

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?		

If you have Adobe Acrobat (NOT READER) you may email this form
Alternatively print off the completed form and post or fax to:

Carolyn Gokhale/Andrew Wallace, Rare Disease Testing Service,
Genomic Diagnostics Laboratory, Manchester Centre for Genomic Medicine,
6th Floor, St Mary's Hospital,
Oxford Road, Manchester, M13 9WL.
Fax: 0161-276-6606 Tel: 0161-276-6122 or 0161-276-3265

Submit by Email

Print Form

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