

East Midlands Regional Molecular Genetics Laboratory

City Hospital campus
Hucknall Road
Nottingham
NG5 1PB

Tel: 0115 9691169 ext. 55207

nuhnt.moleculargenetics@nhs.netwww.nuh.nhs.uk**Service information: Xp21 dystrophinopathy (Duchenne Muscular Dystrophy (DMD)/Becker Muscular Dystrophy (BMD))****Gene/Locus:** *DMD* gene (Xp21) **OMIM:** #310200 (DMD), #300376 (BMD), *300377 (*DMD* gene)**Referrals:** Paediatricians, Neurologists, Clinical Geneticists, other relevant specialities.**Testing:** Diagnostic testing in patients affected with DMD, BMD or *DMD*-associated cardiomyopathy, carrier testing in women with a family history of Xp21 dystrophinopathy.**Target Reporting Times:**

- Diagnostic 28 calendar days
- Carrier Testing/Linkage analysis 28 calendar days on referral from Clinical Genetics
- Urgent Testing* 3 calendar days

Please contact the laboratory for urgent testing, e.g. prenatal diagnosis or carrier testing in pregnant women.*Test Details:**

- **MLPA (multiplex ligation-dependent probe amplification) analysis of the *DMD* gene** to detect exonic deletions and duplications (MRC Holland P034/P035 kits) is used for diagnostic testing and for direct carrier testing.
- Linkage analysis by PCR of **microsatellite markers** can be used to determine carrier risks for women in families where a causative mutation has not been detected. Please contact the laboratory to discuss samples required. These types of referral are accepted from Clinical Genetics only

Service Details:

MLPA analysis of the *DMD* gene is predicted to detect pathogenic mutations in 72% of Xp21 dystrophinopathy cases (Gatta et al Hum Genet (2005), 117: 92-98; White et al Hum Mutation (2006), 27(9), 938-945). If no mutation is detected by MLPA, but a diagnosis of Xp21 dystrophinopathy is still suspected, DNA can be sent to another laboratory to test for point mutations in the *DMD* gene. The cost for point mutation testing is the responsibility of the referring clinician. In addition, histopathological studies to determine the level of dystrophin protein present in the patient's muscle tissue can be offered by a local histopathology department, which may aid diagnosis.

Sample Requirements:

- **EDTA** blood sample (1-4 ml), labelled with patient's **full name, date of birth and NHS number**, or genomic DNA ($\geq 20 \mu\text{l}$ at $\geq 50 \text{ ng}/\mu\text{l}$).
- Samples should be accompanied by a **fully** completed referral card which should include the patient's full name, date of birth and NHS number.
- Please also include details of the test, relevant clinical details and full details of the referring clinician and centre.
- Maternal samples are required in cases of prenatal diagnosis to exclude maternal cell contamination.
- Relevant familial samples are required for linkage analysis.

Consent:

Please note that in submitting a sample, it is the responsibility of the clinician to ensure that consent has been taken i) for testing, ii) for storage, and iii) for the use of this sample and the information generated to be shared with the patient's relatives and their health professionals. Following testing, a sample of the patient's DNA may also be used anonymously to validate new tests and for internal quality control purposes.

Date modified – Jan 2016. If printed, this document is only valid on the day of printing