Xp21 MUSCULAR DYSTROPHY (DUCHENNE & BECKER MUSCULAR DYSTROPHY)  
- OMIM #310200, #300376

INTRODUCTION
Xp21 muscular dystrophies or dystrophinopathies are muscle diseases caused by mutations in the DMD gene, which encodes the dystrophin protein. Duchenne muscular dystrophy (DMD) is the most severe form, it usually presents in early childhood, and is characterised by rapidly progressive muscle weakness and loss of ability to walk by age 12 years. Becker muscular dystrophy (BMD) is milder, and patients remain ambulatory into their 20s. Inheritance is X-linked recessive, and so usually only males are affected. Rarely, females can be affected, due to skewed X-inactivation. The incidence of DMD is approximately 1 in 3,000 male births, and the incidence of BMD is approximately 1 in 20,000 male births. The DMD gene has 79 exons and spans 2.4Mb genomic DNA. Exonic deletions and duplications account for approximately 67% of mutations in DMD, and approximately 85% of mutations in BMD.

TESTING AND REFERRALS
Diagnostic:
- Clinically affected patients
- From Clinical Genetics, Paediatrics, Neurology
- All diagnostic referrals must be accompanied by a serum CK result – if requesting analysis in the absence of elevated CK levels, please contact the laboratory to discuss

Carrier:
- Female relatives of clinically affected patients
- From Clinical Genetics only

Prenatal:
- At risk of having an affected child
- From Clinical Genetics and / or Prenatal Diagnosis
- Prenatal testing must be discussed with the laboratory and arranged in advance

STRATEGY & TECHNICAL INFORMATION
Diagnostic cases:
- Analysis for deletions and duplications of one or more exons of the DMD gene by MLPA.
- Patients with a confirmed diagnosis of DMD/BMD in whom no deletion or duplication is identified can be sent to the DNA Laboratory, Guy’s Hospital, for further point mutation analysis of the DMD gene.

Carrier tests:
- Testing for the known familial mutation by MLPA or sequencing, as appropriate.
- Linkage analysis, by fluorescent PCR, to indicate carrier status in cases where the mutation is not known but the diagnosis is certain.

Prenatal diagnosis:
- Fetal sexing by Amelogenin/SRY analysis of CVS/annio DNA.
- Direct mutation analysis, as appropriate, by MLPA or sequencing.
- Linkage analysis by fluorescent PCR to exclude maternal contamination.

TARGET REPORTING TIMES
Diagnostic test: 10 days
Carrier test: 10 days
Linkage analysis: 10 days
Prenatal testing (includes maternal contamination check): 3 days

N.B. Details are correct for the date of printing only – last updated 19/07/2012