Becker Muscular Dystrophy (BMD) – OMIM 300376; Duchenne Muscular Dystrophy (DMD) – OMIM 310200

Background

Muscular dystrophy describes a large number of inherited conditions where muscles weaken. These diseases are progressive and different muscle groups are affected depending upon the type of muscular dystrophy, usually leading to some sort of disability. Duchenne muscular dystrophy (DMD) is the most prevalent, affecting 1 in 3500 males. It is a serious x-linked, recessive disorder caused by mutations in the *DMD* gene (OMIM 300377). It includes DMD-associated dilated cardiomyopathy, where the heart muscle is involved. Symptoms usually appear during infancy, the disorder is rapidly progressive and life expectancy is affected. Becker muscular dystrophy (BMD) is a rarer, milder form. It is characterized by later onset skeletal muscle weakness, resulting from different mutations in the *DMD* gene.

Recommended Clinical Referral Criteria

- Diagnostic cases, at risk family members, and to determine carrier status in females
- X-linked, recessive inheritance
- Duchenne muscular dystrophy (DMD)
  - Progressive symmetrical muscle weakness, usually proximal > distal, often with calf hypertrophy
  - Presentation before 5 years
- Becker muscular dystrophy (BMD)
  - Progressive symmetrical muscle weakness, usually proximal > distal, often with calf hypertrophy
  - Later onset (teens)
  - Preservation of neck flexor muscle strength

Molecular Analysis

Mutation screen: MLPA dosage analysis of *DMD* gene to detect gross / multi-exon deletions and duplications; where clinical diagnosis is confirmed the clinical sensitivity is 65% (DMD) and 85% (BMD)
Family follow-up: Testing for known familial mutations in *DMD* gene

Test (Price available on request)	**TAT (calendar days)**
Diagnostic dosage analysis (*DMD*) (R37.2)	42
Family follow-up	42
Predictive testing in neonate	14
Prenatal testing	3
NIPD for Duchenne and Becker muscular dystrophy (R310)	Non-invasive testing is available at an external laboratory. Available from 8 weeks gestation following NIPD fetal sexing result that indicates a single male fetus. Please contact the laboratory to discuss prior to sending samples.
Mutation screening (*DMD*) gene (R37.1)	This service is provided by an external laboratory as part of the National genomic test directory.

Contact Details

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Sample Requirements

Blood–5ml in EDTA (1ml neonate/infant);
Please contact lab prior to sending a prenatal sample.
Please label samples with three identifiers and date of collection.
All samples must be accompanied by request form
Consent for testing & DNA storage is assumed when request is received

Links

Orphanet
http://www.orpha.net/
OMIM
http://www.omim.org/
Genetic Test Registry
http://www.ncbi.nlm.nih.gov/gtr/
Support
Muscular Dystrophy Campaign
www.muscular-dystrophy.org