Genetic Testing ACT Sheet

Duchenne & Becker Muscular Dystrophy

Condition Description: Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) are clinically distinct X-linked disorders that represent ends of a spectrum of degenerative neuromuscular diseases caused by mutations in the *dystrophin* gene. Both are characterized by progressive muscle wasting and proximal muscle weakness but vary in age of onset, rate of progression and severity.

Major Indications for Ordering Dystophin Testing: *Dystrophin* gene testing is considered for: a) males with clinical features of DMD or BMD, b) females at risk for being a carrier, c) pregnancies of females known to carry a *dystrophin* mutation.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Inform individual of the test result.
- Refer for genetic evaluation and counseling.

Diagnostic Evaluation: Evaluation includes physical, cardiac, neurological and neuromuscular testing. Molecular genetic testing can establish a diagnosis without muscle biopsy. Virtually all males with DMD and about 85% of males with BMD have identifiable *dystrophin* mutations. In cases where *dystrophin* mutations are identified, testing of the mother should be considered.

Clinical Considerations: Individuals with DMD often have muscle weakness that is progressive from early childhood. Pseudohypertrophy of the calf muscles and associated "toe walking' are common. Life-threatening dilated cardiomyopathy may arise in adolescence. Some individuals with DMD have cognitive impairment and behavioral abnormalities. BMD typically presents later in life with slower rate of progression, though life-threatening dilated cardiomyopathy may also develop. Females who are carriers of *dystrophin* mutations may be symptomatic due to cardiac or skeletal muscle involvement. About 2/3 of cases are inherited, the remainder resulting from spontaneous new mutations.

Additional Information:

Gene Tests/Gene Clinics
Genetics Home Reference
Muscular Dystrophy Association

Referral (local, state, regional and national):

Testing
Clinical Services
Find Genetic Services

ACMG
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LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Tests/Gene Clinics
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=dbmd

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=duchenneandbeckermusculardystrophy

Muscular Dystrophy Association
http://www.mda.org/home.htm

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/53738?db=genetests

Clinical Services
http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

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