

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 Dystrophin 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](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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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	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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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