

Newborn Screening ACT Sheet [Pathogenic Variant in Dystrophin (*DMD* Gene) and elevated creatine kinase muscle isoform (CK-MM)] Duchenne and Becker Muscular Dystrophy

Differential Diagnosis: Duchenne Muscular Dystrophy (DMD), Becker Muscular Dystrophy (BMD), and DMD-associated dilated cardiomyopathy (DMD associated-DCM) forms of dystrophinopathy.

Condition Description: The finding of a pathogenic variant in the *DMD* gene following a highly elevated level of the muscle isoform of creatine kinase (CK-MM) is associated with dystrophinopathies. These are degenerative neuromuscular diseases caused by a defect in the dystrophin (*DMD*) gene. DMD and BMD have similar signs and symptoms but are considered to be the same underlying disease that differs in age of onset and severity. DMD/BMD is an X-linked recessive disorder that primarily affects males although females may be affected. Dystrophinopathies occur in approximately 1 in 5,000 males.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status.
- Provide the family with basic information about risk for DMD/BMD.
- Elicit family history of signs and symptoms of neuromuscular disease.
- Consult with the neuromuscular disease specialist or comprehensive MDA clinic.
- Refer for genetic counseling.
- Report findings to state newborn screening program.

Diagnostic Evaluation: Molecular genetic testing may establish a diagnosis. Molecular testing results should be confirmed if they were obtained as part of the newborn screening algorithm. Additional evaluation includes physical, cardiac, neurological, and neuromuscular testing.

Clinical Considerations: Subtle signs and symptoms may appear between the first and second years of life, specifically proximal leg weakness. Individuals with DMD often present with speech delay and skeletal muscle weakness that is progressive from early childhood. Enlargement of the calf muscles, toe walking, and progressive skeletal muscle weakness are common. Life-threatening dilated cardiomyopathy may arise in adolescence. Some individuals with DMD have cognitive impairment and behavioral abnormalities. BMD typically presents at a later age with slower rate of progression than DMD although life-threatening cardiomyopathy may also develop. Treatments to slow disease progression and slow loss of function will include physical therapy management and may include steroids. Gene therapies may be available and additional therapies are being developed. Ongoing multi-specialty care is necessary. Females who are carriers of pathogenic *DMD* variants may be symptomatic due to cardiac or skeletal muscle involvement and also may have cognitive impairment

Additional Information:

[Gene Reviews](#)
[Genetics Home](#)

Referral (local, state, regional and national):

[Testing](#)
[Find Genetic Service](#)
[Muscular Dystrophy Association Clinics](#)
[PPMD Certified Duchenne Care Centers Network](#)
[Duchenne/Becker and Carrier Testing](#)

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

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 Reviews

<https://www.ncbi.nlm.nih.gov/books/NBK1119/>

Genetics Home Reference

<https://ghr.nlm.nih.gov/gene/DMD>

OMIM

<https://www.omim.org/entry/310200>

Referral (local, state, regional and national):

Testing

<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=DMD>

Find Genetic Services

<https://clinics.acmg.net>

Muscular Dystrophy Association Clinics

<https://www.mda.org/care/mda-care-centers>

PPMD Certified Duchenne Care Centers Network

<https://www.parentprojectmd.org/care/find-a-certified-duchenne-care-center/>

No Charge for Duchenne/Becker and Carrier Testing – PPMD's DecodeDuchenne

<https://www.parentprojectmd.org/about-duchenne/decode-duchenne/>

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