Newborn Screening ACT Sheet

[No Pathogenic Variant in Dystrophin (DMD) Gene after elevated creatine kinase muscle isoform (CK-MM)]

Genetic Neuromuscular Diseases

Differential Diagnosis: CK-MM associated neuromuscular condition with residual risk of a DMD/BMD dystrophinopathy.

Condition Description: The absence of a pathogenic variant being detected in the DMD gene following a highly elevated level of the muscle isoform of creatine kinase (CK-MM) is associated with a number of neuromuscular conditions. Additional genes for other neuromuscular conditions that may explain elevated CK-MM will need to be tested next to identify secondary conditions associated with elevated CK-MM. There remains a residual risk of a dystrophinopathy.

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 residual risk for DMD/BMD and the risk for other neuromuscular conditions for which additional genes will be tested. Other newborn screen results may be available for some of the conditions (e.g., Pompe, SMA).
- 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 of a neuromuscular condition. Additional evaluation includes physical, cardiac, neurological, and neuromuscular testing.

Clinical Considerations: Signs and symptoms of a neuromuscular disease may be apparent depending on the underlying molecular diagnosis. Follow-up and treatment must be conducted in close collaboration with the center. Treatments including targeted gene therapies may be available for some of the neuromuscular diseases being tested.

Additional Information:
Gene Reviews
Genetics Home
OMIM

Referral (local, state, regional and national):
Testing
Find Genetic Service
Muscular Dystrophy Association Clinics
PPMD Certified Duchenne Care Centers Network

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

- Genetics Home Reference

- OMIM
  [https://www.omim.org/entry/310200](https://www.omim.org/entry/310200)

**Referral (local, state, regional and national):**

- Testing

- Find Genetic Services
  [https://clinics.acmg.net](https://clinics.acmg.net)

- Muscular Dystrophy Association Clinics

- PPMDCertified Duchenne Care Centers Network