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

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/