Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Persistently Elevated CK-MM

Differential Diagnosis

A persistently elevated muscular creatine-kinase level on newborn screening is primarily associated with:

- Duchenne muscular dystrophy (DMD)—Prevalence of 1 in 3,500-5,000 for males, less common for females
- Becker muscular dystrophy (BMD)—Prevalence of 1 in 10,000 for males, less common for females

Other disorders to consider:

- Congenital muscular dystrophies/neuromuscular disorders (ex. Limb-Girdle muscular dystrophy)

False Positives

Possible. Other causes of high CK-MM include traumatic birth, injury, medical procedures, and early newborn screening sample collection.

Next Steps

<u>This week,</u> you should take the following recommended actions:

- Consult a neuromuscular specialist familiar with DMD for guidance on clinical follow-up. Contact information for the specialists can be found on the resource list provided.
- Contact family to notify them of the newborn screening result and next steps, as MDH has not notified them.
- Arrange referral and help family coordinate follow-up.

If you have questions about the newborn screening result or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

Clinical Summary

DMD, and related condition BMD, are X-linked conditions caused by mutations of the dystrophin gene. Therefore, they affect mostly males. Most females (carriers) are unaffected or have milder symptoms. Dystrophin is a protein that protects muscle cells from damage. Without it, muscle tissues fail to regenerate and are replaced by scar tissue and fat. This leads to extreme muscle fragility as the individual ages.

DMD and BMD are variable, progressive muscle disorders. DMD has an earlier onset and more severe symptoms. Noticeable signs of DMD appear in early childhood, usually between two and three years of age, starting with difficulty moving, walking, and running. Other common symptoms in childhood are large calf muscles paired with thin thighs (pseudohypertrophy), and slight learning disabilities. Males with DMD often lose the ability to walk and use a wheelchair by age twelve. By their early twenties, individuals with DMD develop cardiomyopathy. Heart and respiratory problems worsen with age, and typically become life-threatening. Individuals with BMD often do not develop these symptoms until later in life.

DMD/BMD cannot be cured, but early interventions can help slow the disease and improve quality of life. Management may include:

- Oral glucocorticoid treatment
- New gene therapies may be available, such as possible exon-skipping and gene replacement
- Regular screenings to identify heart/muscle issues
- Supportive therapies like tailored physical therapy
- Establishing with a team of doctors that specialize in DMD, including a neuromuscular specialist

DEPARTMENT OF HEALTH

Minnesota Newborn Screening Program | www.health.state.mn.us/newbornscreening
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To obtain this information in a different format:
contact health.newbornscreening@state.mn.us
or call (800) 664-7772 ***interpreters available***

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