

# Borderline Result:

## Blood Spot Screen Result Notification



### Elevated CK-MM

#### Next Steps

You should take the following recommended actions:

- **Contact** family to notify them of the newborn screening result.
- **Order** serum total CK for collection after child is two weeks of age. This allows enough time for elevated CK-MM levels related to the birth process to normalize.
- **Fax** total CK results to MDH at (651) 215-6285.

If total CK is abnormal, a neuromuscular specialist should be contacted. Contact information can be found on the resource list provided.

If total CK is normal (using age-appropriate cutoffs), no additional follow-up is required. **Most borderline results are expected to resolve.**

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.

#### Possible Explanations for Result

An elevated muscular creatine-kinase (CK-MM) on a newborn screen sample collected during the first few days of life is most likely due to the birthing process. This is especially true if baby had a birth injury, was in the NICU, or had an early newborn screening sample collection. CK-MM is expected to normalize with time in these cases.

Rarely, this screening result is due to a genetic muscle disorder, such as Duchenne muscular dystrophy (DMD) or Becker muscular dystrophy (BMD). CK-MM is expected to be persistently elevated in affected individuals.

#### Review with Family

Discuss this result with the family as MDH has **not** notified them. Share the follow-up plan with them.

#### Clinical Summary

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 two and three years of age, starting with difficulty moving, walking, and running. 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 symptoms until later in life. These disorders typically affect males, with females usually being asymptomatic or having milder symptoms.

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
- Regular screenings to identify heart/muscle issues early
- Supportive therapies like tailored physical therapy
- Establishing with a team of doctors that specialize in DMD, including a neuromuscular specialist