

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** a repeat newborn screen for after two weeks of age. This allows enough time for elevated CK-MM levels related to the birth process to normalize.

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.

Review with Family

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

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 traumatic birth, injury, was in the NICU, or had an early newborn screening sample collection (before or around 24 hours of life). 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.

Clinical Summary

Most borderline results have normal repeat newborn screens. Children with a borderline result who have a normal repeat screen require no additional follow-up or treatment. If repeat screening is abnormal, a Newborn Screening Program genetic counselor will contact you to discuss the next steps

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