

# Positive Result:

## Blood Spot Screen Result Notification



### Persistently Elevated CK-MM: Risk for Duchenne Muscular Dystrophy

#### What was found on the newborn screen?

The newborn screen sample that was collected found your baby has high levels of a muscle enzyme called creatine-kinase (CK-MM).

#### What does this mean?

High CK-MM in the blood can be a sign of muscle damage. This can be due to an injury (such as after difficult birth or medical procedure) or a muscle condition like Duchenne muscular dystrophy (DMD). Therefore, while there is a chance that your baby could have a muscle condition based on this screening result, further testing is needed.

#### What happens next?

Your baby's doctor will help arrange for more testing with specialists familiar with muscle conditions. This will give more information about if your baby has DMD.

#### What causes Duchenne muscular dystrophy?

DMD and a related milder condition called Becker muscular dystrophy (BMD) are muscle disorders. They occur when children, usually boys, are unable to make enough of a specific protein that protects muscles from damage. Without it, the body has trouble replacing lost muscle tissue, which results in severe muscle weakness and muscle loss over time.

#### What health problems can it cause?

DMD/BMD is different for each child. The first signs of DMD usually appear between two and three years of age, starting with difficulty moving, walking, and running. Children may also have slight learning disabilities. Children with DMD usually lose the ability to walk and use a wheelchair by age twelve. By their early twenties, individuals with DMD often develop weak hearts (cardiomyopathy). Heart and respiratory problems worsen with age, and can become life-threatening. BMD has similar symptoms, but they usually are more mild and develop later than DMD.

#### What treatment options are available?

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

- Medication taken by mouth
- New gene therapies may be available that replace or partially repair the body's instructions for making the specific muscle protein (dystrophin)
- Screenings to find heart and muscle issues early
- Tailored physical therapy

Children with DMD should see a team of doctors that specialize in the different aspects of DMD, including a neuromuscular specialist. They will help determine the treatment options and timing that are best for a child.

#### Resources



Scan here for more information about newborn screening in Minnesota or visit our website:  
[www.health.state.mn.us/people/newbornscreening](http://www.health.state.mn.us/people/newbornscreening)

MedlinePlus:  
[www.medlineplus.gov/genetics](http://www.medlineplus.gov/genetics)

Save Babies Through Screening Foundation:  
[www.savebabies.org](http://www.savebabies.org)

Newborn Screening Info Center:  
[www.newbornscreening.hrsa.gov](http://www.newbornscreening.hrsa.gov)