

# Borderline Result: Blood Spot Screen Result Notification



## Reduced Biotinidase (BTD)

### Next Steps

This week, you should take the following recommended actions:

- **Contact** family to notify them of the newborn screening result and arrange for collection of a repeat newborn screen.
- **Order** a repeat newborn screen. Do not order a serum biotinidase assay.

If repeat screening is abnormal, additional follow-up is needed. In this situation, a Newborn Screening Program genetic counselor will contact you to discuss the next steps.

If repeat screening is normal, no additional follow-up or treatment is required.

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. Educate family about signs, symptoms, and when to contact you with concerns.

### Possible Explanations for Result

Screening result may be due to the child having biotinidase deficiency; especially partial deficiency.

Additionally, screening result can be impacted by transfusion and environmental factors such as heat and humidity.

### Clinical Summary

**Most borderline results have normal repeat screens.**

Biotinidase deficiency is caused by a deficiency in the enzyme, BTD. As a result of this deficiency, the body is unable to recycle the vitamin, biotin. This can lead to a buildup of potentially toxic compounds. There are two forms of biotinidase deficiency: profound and partial.

A borderline result is unlikely to result in a diagnosis of profound biotinidase deficiency. Partial biotinidase deficiency is the milder form with most patients exhibiting cutaneous symptoms, particularly during times of stress, such as during infection.

Should follow up of this screen result in a diagnosis of biotinidase deficiency, treatment consists of lifelong biotin supplementation. If treated early, complications of biotinidase deficiency can be prevented or improved.

# Borderline Result:

## Blood Spot Screen Result Notification



## Borderline Newborn Screening Result

### What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby had a borderline result.

### What does a borderline result mean?

A borderline result means that the test was between normal and abnormal. There are several reasons why results can fall in the borderline range, including:

- Normal changes in baby's body as they adjust to being born
- Babies who are born early (premature)
- Babies who have difficult or stressful deliveries
- The blood spots being exposed to heat and humidity
- Certain treatments for either mom or baby

Most of the time, it is unclear why the result was borderline.

### What happens next?

Your baby's doctor will order a repeat newborn screen. Most babies who have borderline results have normal repeat screens. However, because the disorders on the newborn screen are serious, it is important to repeat the screen and make sure your baby doesn't have a health problem.

### How will I get my baby's repeat screen results?

Once your baby's blood spots arrive at the screening laboratory, testing begins immediately but can take several days. The test results will be provided to your baby's doctor or clinic.

### What if the repeat screen is normal?

If the results are normal, no special testing or treatment will be needed. No more medical care in relation to the borderline result will be needed. If you have questions, please speak with your baby's doctor.

### What if the repeat screen is borderline or abnormal again?

If the repeat screen is borderline or abnormal, your doctor will let you know about additional testing or treatment your baby could need.

## Resources

**Genetics Home Reference:**  
<http://ghr.nlm.nih.gov>

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

**Baby's First Test:**  
[www.babysfirsttest.org](http://www.babysfirsttest.org)