

# Barts Present:

## Blood Spot Screen Result Notification



### Hemoglobin Barts Present

#### Next Steps

You should take the following recommended actions:

- **Discuss** the result with the family at the next well-child visit.
- **Collect** follow-up testing. Between 4-6 months of age, a hemoglobin electrophoresis, CBC, and reticulocyte count should be performed.
- **Consult** with pediatric hematologist for clinically abnormal results.

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 at the next well-child visit. Share your follow-up plan with them. Educate family about signs, symptoms, and when urgent treatment may be needed.

MDH mailed information about the result to the family, so they may have some familiarity with the implications of the result.

#### False Positives

This result was found using hemoglobin gel electrophoresis. Sometimes a shadow can be on the gel that may look like a hemoglobin Barts band with low concentration, similar to what is seen in silent carriers.

#### Differential Diagnosis

Hemoglobin Barts is primarily associated with:

- Alpha thalassemia — more common in people from Southeast Asia, Mediterranean, Africa, Middle East, India, and Central Asia

#### Clinical Summary

Alpha thalassemia is a blood disorder that impacts the production of normal hemoglobin. Instead of the normal four copies of the alpha globin gene, affected individuals have fewer copies. Hemoglobin Barts is present in the newborn when one or more of the four alpha globin genes are missing.

| Alpha Globin Gene Deletions | Clinical Outcome                               |
|-----------------------------|--|
| 0                           | Normal   |
| 1                           | Silent carrier                                 |
| 2                           | Alpha thalassemia trait                        |
| 3                           | Hemoglobin H disease                           |
| 4                           | Alpha thalassemia major (a.k.a. fetal hydrops) |

#### Silent Carrier:

Red cell indices are normal and Barts hemoglobin is not seen after 6 months of age. There are no clinical implications and no treatment is needed.

#### Alpha Thalassemia Trait:

Most have mildly abnormal red cell indices and are mildly anemic, but no treatment is needed.

#### Hemoglobin H Disease:

Most patients are anemic and develop splenomegaly, though clinical symptoms vary. Care by pediatric hematology is recommended.

#### Alpha Thalassemia Major:

Often identified prenatally due to hydrops fetalis. Because of severe anemia, hepatosplenomegaly, cardiac defects, and genitourinary abnormalities, individuals are often stillborn or die soon after birth. Infants have survived, however, with early detection and chronic transfusion therapy.