RBC Band 3 Protein Reduction in Hereditary Spherocytosis

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Hereditary spherocytosis is a genetic disorder that causes congenital hemolytic anemia through the production of structurally compromised red blood cells (RBCs). The severity of the disease varies widely; most cases are classified as moderate and present with symptoms such as anemia, jaundice, and splenomegaly. Depending on the level of severity, a diagnosis of hereditary spherocytosis can rule out other causes of hemolytic anemia.[1]

Featured ARUP Testing

RBC Band 3 Protein Reduction in Hereditary Spherocytosis 2008460

Method: Qualitative Flow Cytometry

Use to confirm a diagnosis of hereditary spherocytosis when hemolytic anemia and spherocytes are present.

Disease Overview

Prevalence

1/2,000 in individuals of northern European descent[2]

Genetics

Genes

ANK1, *EPB42*, *SLC4A1*, *SPTA1*, *SPTB*

Inheritance

- Autosomal dominant: 75%[1]
- Autosomal recessive: 25%[1]

Penetrance

Variable

Structure/Function

- Chromosomal location: 17q21.31
- Provides structure for the red cell cytoskeleton

Test Interpretation

Sensitivity/Specificity

Clinical sensitivity: 93%

Analytic sensitivity/specificity: unknown

Results

Test Result	Interpretation

[a]Three other rare disorders may be associated with a positive result for this test: congenital dyserythropoietic anemia type 2, Southeast Asian ovalocytosis, and hereditary pyropoikilocytosis.

EMA, eosin-5-maleimide

Test Result	Interpretation
Normal	Normal staining of band 3 protein with EMA does not suggest hereditary spherocytosis
Abnormal[a]	Decreased staining of band 3 protein with EMA provides evidence for hereditary spherocytosis
Equivocal	Insufficient evidence to support or negate a diagnosis of hereditary spherocytosis Unclear whether the level of band 3 protein staining is normal

[a]Three other rare disorders may be associated with a positive result for this test: congenital dyserythropoietic anemia type 2, Southeast Asian ovalocytosis, and hereditary pyropoikilocytosis.

EMA, eosin-5-maleimide

Limitations

Recent transfusion (within the last three months) may affect test results; correlation with clinical and other laboratory findings is strongly recommended.

References

1. Narla J, Mohandas N. Red cell membrane disorders. *Int J Lab Hematol*. 2017;39 Suppl 1:47-52.

2. Zamora EA, Schaefer CA. Hereditary spherocytosis. 2021. In: StatPearls. StatPearls Publishing; 2021.

Related Information

Hemolytic Anemias

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