RBC Band 3 Protein Reduction in Hereditary Spherocytosis

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

Tests to Consider

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

Related Test

Osmotic Fragility, Erythrocyte 2002257

Method: Spectrophotometry

Functional testing of RBC sensitivity to osmotic stress

Disease Overview

Prevalence

1/2,000 in individuals of northern European descent2

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%

Analytical sensitivity/specificity: unknown
Results

<table>
<thead>
<tr>
<th>Test Result</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>Normal staining of band 3 protein with EMA does not suggest hereditary spherocytosis</td>
</tr>
<tr>
<td>Abnormal*</td>
<td>Decreased staining of band 3 protein with EMA provides evidence for hereditary spherocytosis</td>
</tr>
<tr>
<td>Equivocal</td>
<td>Insufficient evidence to support or negate a diagnosis of hereditary spherocytosis</td>
</tr>
<tr>
<td></td>
<td>Unclear whether the level of band 3 protein staining is normal</td>
</tr>
</tbody>
</table>

*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


Related Information

Hemolytic Anemias