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.\(^1\)

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\(^1\)

Penetration

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>
</tbody>
</table>

\(^1\)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

<table>
<thead>
<tr>
<th>Test Result</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<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>

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^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

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### 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