Acute pancreatitis as initial presentation for hereditary spherocytosis: a case report

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Introduction
Hereditary spherocytosis is one of the most common cause for hemolytic anemia and is due to a red cell membrane defect. The incidence is likely to be underestimated, as mild cases are often not diagnosed.

Case: E., 9y

• Presenting for vomiting, jaundice and anorexia without fever or trauma
• Clinical examination: Muscutaneous jaundice, hepatomegaly and diffusely painful abdomen
• Laboratory findings:
  • Hepatic cytolysis
  • Hyperbilirubinemia (both conjugated and unconjugated)
  • Hemolysis
  • No anemia
  • Negatives infectious serologies
• Ultrasound:
  • Normal liver, a slightly enlarged gallbladder with biliary sludge but the biliary ducts were not enlarged
  • Small splenomegally
  • Normal pancreas and kidneys
• Evolution: acute pancreatitis with elevated lipase and abdominal ultrasound demonstrated a protruding pancreas without identified lithias
• Treatment: analgesia, enteral nutrition because of major anorexia, IV fluid and antibiotics.
• Follow-up:
  • Cholecystectomy after recovery
  • Positive screening for hereditary spherocytosis

Differential diagnosis of biliary lithiasis in children

<table>
<thead>
<tr>
<th>Haemolytic disease</th>
<th>Genetic</th>
<th>Systemic</th>
<th>Medications</th>
<th>Dietary</th>
<th>Surgery</th>
<th>Bilary dyssinesis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spherocytosis</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Hemoglobinopathy</td>
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<td>Enzyme abnormalities</td>
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<td>Thalassemias</td>
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<td>Sickle cell disease</td>
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<td>Pyruvate kinase</td>
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Hereditary spherocytosis

• Defect in one of the six genes that encode for the protein involved in the red cell membrane: Ankryn, spectrin, pallidin, band 1, band 4.1, RHAG → red cell membrane instability

• Variable clinical severity: symptom-free carrier → severe hemolysis

• Suspecting diagnosis:
  • Anemia, jaundice, and splenomegaly
  • Positive family history of hemolytic anemia.
  • Routine blood counts; anemia and reticulocytosis, low MCV, increased MCHC
  • Peripheral blood smear: spherocytes.
  • Negative direct and indirect antiglobulin tests

• Specific laboratory investigations: in atypical cases
  • Osmotic fragility testing
  • Ektacytometry
  • Acidified glycerol lysis test
  • EMA Binding Test
  • Cryohemolysis test

• Treatment and follow-up
  • Supportive care: folic acid in severe HS, transfusion if needed
  • Splenectomy; benefits > risks in moderate and severe HS
  • Cholecystectomy for symptomatic gallstones
  • Genetic counseling

Hereditary spherocytosis is a common cause for hemolytic anemia but clinical presentation can be misleading. We suggest that this diagnosis should be evocated in children presenting with acute unexplained jaundice that can be associated with acute pancreatitis.