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

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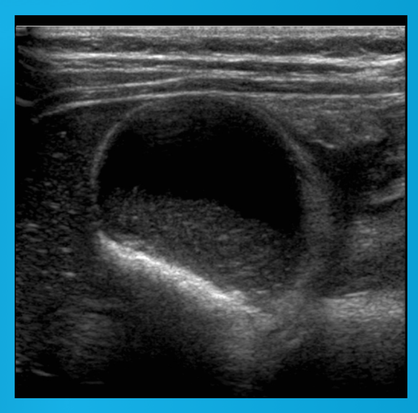
Introduction

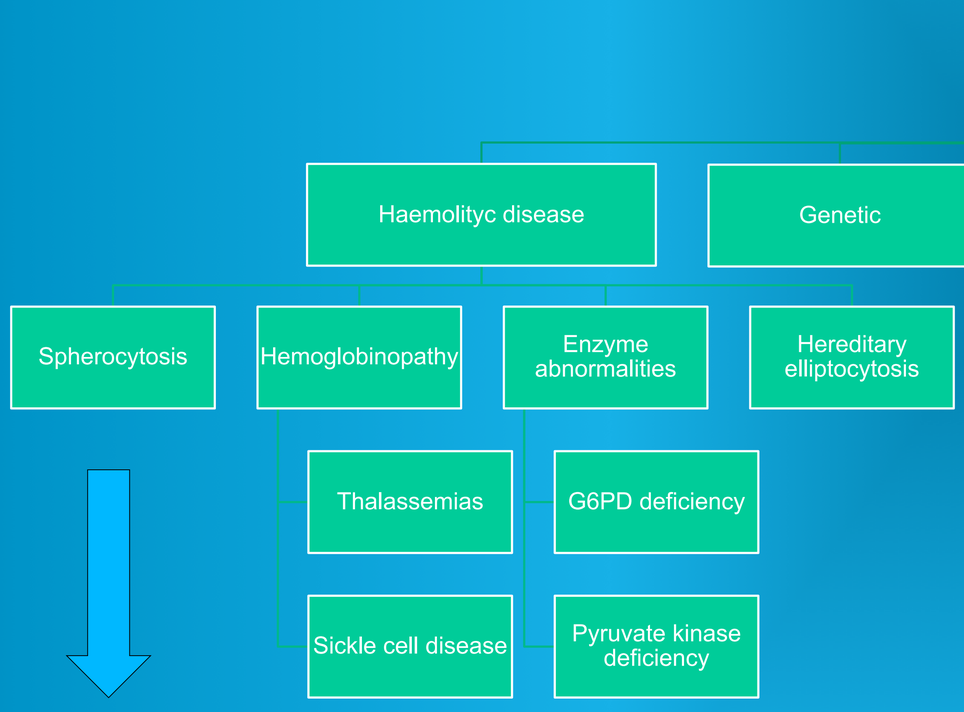
Hereditary spherocytosis is one of the most common cause for hemolytic anemia and is due to a red cell membrane defect. The incidence is likely to be underestimate, as mild cases are often not diagnosed.

Case: E., 9y

- **Presenting for vomiting, jaundice and anorexia without fever or trauma**
- Clinical examination: Mucocutaneous 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 splenomegaly
  - Normal pancreas and kidneys
- **Evolution:** **acute pancreatitis** with elevated lipase and abdominal ultrasound demonstrated a protruding pancreas without identified lithiasis
- **Treatment:** analgesia, enteral nutrition because of major anorexia, IV fluid and antibiotics.
- **Follow-up:**
  - cholecystectomy after recovery
  - positive screening for hereditary spherocytosis

	18/07/2015	19/07/2015	20/07/2015	22/07/2015	27/07/2015
HB	15,3 g/dl	13,8 g/dl	14,2 g/dl	14,2 g/dl	12 g/dl
MCH	31,2 pg	31,4 pg	31,1 pg	31,5 pg	31,2 pg
MCV	84,7 µ^3	86,3 µ^3	88,4 µ^3	88,2 µ^3	83,6 µ^3
Réticulocytes	5,2%	5,58%	5,26%		
Haptoglobine	0 mg/dl	2 mg/dl	1 mg/dl	44 mg/dl	44mg/dl
LDH	298 U/l	222 U/l	220 U/l	236 U/l	223 U/l
Bilirubine totale	19,09 mg/dl	14,48 mg/dl		6,9 mg/dl	2,34 mg/dl
Lipase		155 U/l	7342 U/l	4444 U/l	198 U/l
TGO	248 U/l	151 U/l	176 U/l	98 U/l	40 U/l
TGP	635 U/l	454 U/l	373 U/l	239 U/l	73 U/l





Hereditary spherocytosis

• Defect in one of the six genes that encode for the protein involved in the red cell membrane: Ankryn, spectrin, pallidin, band 3, band 4,1, RhAG → red cell membrane instability

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

Mild HS 20-30%	Moderate HS 60-75 %	Severe HS 5%
No anemia, modest reticulocytosis Little way of jaundice and splenomegaly	Anemia Reticulocytes ↑, bilirubin↑	Marked hemolysis and anemia Bilirubin ↑, splenomegaly
Disorder may not be detected	Detected in infancy or childhood	Early detection Regular need for transfusion

•**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 gallsatones
- Genetic counseling

Conclusion

•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.