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Hereditary Spherocytosis

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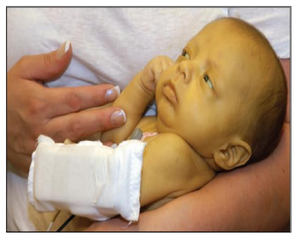
Definition

Hereditary Spherocytosis (HS) is a hemolytic anemia where red blood cell membranes are spherical unlike common red blood cells, which are flat and round. In individuals with HS the spleen does not recognize these amorphous cells and destroys—rather filters—them them, making the individual anemic (MedlinePlus, 2013).
HS is an autosomal dominant, or recessive autosomal inherited blood disease (Huq, Pietroni, Rahman & Alam, 2010)
HS happens in 1 in 2,000 of the Caucasian population in or from Northern Europe (MedlinePlus, 2013).
The proteins involved in HS are ankyrin-1, ά-spectrin, β-spectrin, band 3, and protein 4.2.
Hyperbilirubinemia is a common symptoms in neonates with HS. According to Christensen and Henry (2010) hyperbilirubinemia is sometimes miss as a diagnose for HS (Christensen & Henrym 2010).



Jaundice

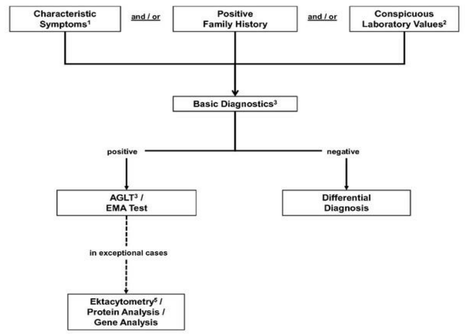
Normal

Symptoms

HS symptoms:
•Anemia and increased bilirubin in newborns
•In older children: varies from fatigue, palor of the skin, enlarged spleen or weakness
•In some cases: gallstones, (Huq, Pietroni, Rahman & Alam, 2010)
•Aplastic crisis due to Pavovirus B19 infection (Bharne, Gowler, Dias, 2012).

Diagnosis

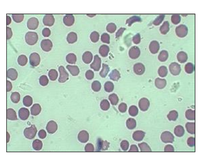
•Peripheral blood smear
•Family history
•Osmotic fragility test
•Acid glycerol lysis time test (AGLT)
•Cryohemolysis test
•Eosin-5'-maleimide (EMA)-binding test.(King & Zanella, 2013).



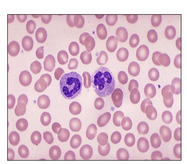
(Eber et al., 2012)

Pathophysiology

Cells in HS have distorted Na+/K+ fluctuations. The membrane proteins in the red blood cells are changed. The normal intracellular K+ and Na+ are reversed in plasma. Spectrin, a structural protein of the membrane skeleton is reduced, which leads to increased loss of the outside area of the cell and makes it spherical instead of a biconcave disc. The spleen does not recognize these new transformed cells and destroys them (King & Zanella, 2013).



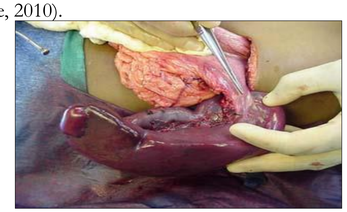
Spherocytes



Normal RBC (King & Zanella, 2013)

Treatments

While in hemolytic crisis: Red blood cell transfusions might be necessary (Biyiki, Gokce, Cakalagaoglu, Turkkan & Alpay, 2010)
Activity restriction is recommended, however some researchers stated that it is not necessary (Biyiki, Gokce, Cakalagaoglu, Turkkan & Alpay, 2010).
Splenectomy is the only therapy to eradicate the anemia. Hollingsworth and Rice, 2010, suggest that partial splenectomy is a better alternative since a total splenectomy would increase risk of infections and other problems. However, a re-growth of the spleen can take place, so close observation is crucial.
Antibiotic prophylactic is recommended until the patient reaches the age of 18 years old.
If after surgery the anemia still continues, the HS diagnosis might be inadequate (Hollingsworth & Rice, 2010).



Partial splenectomy (Hollingsworth & Rice, 2010).

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