Hereditary Spherocytosis Diagnosis & Treatment

Hereditary spherocytosis is a disorder of RBC membrane proteins, leading to small, round RBCs. This an intrinsic hemolytic anemia. Patients are diagnosed via the osmotic fragility test, and treatment includes folic acid supplementation, and eventually splenectomy.



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Labs

Increased MCHC
Up-arrow M.C. He-man-globe
MCHC, or mean corpuscular hemoglobin concentration, is used to measure the concentration of hemoglobin in a given volume of packed RBCs. It is elevated in hereditary spherocytosis, as the cell is dehydrated and the membrane surface area is decreased.

Spherocytes
Sphere-cells
As spectrin and ankyrin, which interact with the RBC membrane, are commonly defective, the RBC membrane is no longer anchored. Small, round, spherical RBCs are formed, which have no central pallor.

Normocytic Anemia
Normal-sized-cells Anemone
RBCs need to be flexible to pass through the spleen (cords of Billroth). As spherocytes are less flexible and abnormally shaped, they bottleneck in the spleen. They are then phagocytosed, leading to hemolysis. Thus, this disease is characterized as a normocytic (normal sized) anemia, which is intrinsic and extravascular.

Diagnosis

Eosin-5-Maleimide (EMA) Binding Test
EMA-Emu
The Eosin-5-Maleimide Binding Test is the gold standard confirmatory test for the diagnosis of hereditary spherocytosis. This test involves exposing the patient's RBCs to a fluorescent dye that stains their surface. This fluorescence can then be measured by flow cytometry. The brightness of spherocytic RBCs is expected to be about 2/3rds that of normal. As small spherocytes have less surface area than a normal biconcave disk-shaped RBC, less surface area and thus less binding is expected.

Osmotic Fragility Test
Water-gun Bursting blood cell
When the Eosin-5-Maleimide Binding Test cannot be used, the osmotic fragility test may be performed. This test is easily performed in resource-poor settings, though it has a high false negative rate. In this test, spherocytes easily rupture in hypotonic solutions due to increased membrane permeability to salt and water.

Glycerol Lysis Test
Glitter-roll Laser
The glycerol lysis test is another useful test that may be used in the diagnosis of hereditary spherocytosis. In this test, the patients' RBCs are exposed to a basic glycerol solution. The extent of RBC hemolysis is then measured. A modified version of this test, the "pink test", can also be used.

Treatment

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Folic Acid

Flicking Acidic-lemon

As hereditary spherocytosis is an anemia, folic acid is supplemented to patients. Folic acid helps develop healthy RBCs. Furthermore, patients are creating more RBCs continuously, due to increased demand (as spherocytes are being destroyed). Thus, a folate deficiency may occur due to increased bone marrow demand.

Splenectomy

Chopped-off Spleen

Splenectomy, or surgical removal of the spleen, is indicated for moderate to severe cases of hereditary spherocytosis. Patients post-splenectomy are more susceptible to encapsulated bacterial infections, and on lab examination may show Howell-Jolly bodies.

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