Normocytic Anemia: Hereditary Spherocytosis

In hereditary spherocytosis, red cells on the blood film are smaller and more dense than normal. Treatment is with folic acid and, where necessary, a splenectomy.

Definition of Hereditary Spherocytosis

In most cases, hereditary spherocytosis is an autosomal-dominantly inherited disease of the red blood cell line, which presents itself in a morphological change of the erythrocytes to so-called spherocytes.

Epidemiology of Hereditary Spherocytosis

With a prevalence of 1 : 3,000, hereditary spherocytosis is the most frequently inherited hemolytic anemia in Central Europe.

Etiology and Pathogenesis of Hereditary Spherocytosis
Generally, spherocyte anemia is autosomal-dominantly inherited. The cause of the disease is a membrane defect. Due to the defect or a lack of certain structure proteins of the cytoskeleton (ankyrin, spektrin or pallidin), cell stability is impaired in such a manner that the typical biconcave shape of the erythrocytes is lost, being replaced by a spherical shape instead. With its shape and morphology, this spherocyte has a negative influence on microcirculation and gets prematurely stuck in the spleen to be degraded there.

**Note:** Mutations in RBC membrane proteins, especially spectrin, that maintain the integrity of the cell membranes. Loss of membrane fragments causes the RBCs to assume a spherical, rather than biconcave, shape. Misshapen cells are cleared in the spleen (extravascular hemolysis), causing anemia.

**Clinic of Hereditary Spherocytosis**

Spherocyte anemia can clinically appear at every age. Like any other anemia, spherocytosis develops the typical signs of anemia. Another very distinct symptom is **recurrent jaundice**. Also, an enlarged spleen can often be palpated (splenomegaly). Often, pigmented gallstones develop.

Aplastic crisis with parvovirus B19 infection – due to decreased RBC life span.
Diagnosis of Hereditary Spherocytosis

A positive family history gives the first important hint. Also, microspherocytes with low diameter are visible in the blood smear. The proportion of reticulocytes in the blood count is usually 5 – 20%. For protection of the findings, osmotic resistance of the erythrocytes can be tested, which has a right-shifted spread at spherocytosis. To exclude auto-immunological events as a cause of hemolysis, the Coombs test should also be performed. Since there is no autoimmune mechanism at spherocytosis, it will be inconspicuous.

Therapy of Hereditary Spherocytosis

The first-resort therapy is a splenectomy. After removal of the spleen, the hemoglobin level usually normalizes since the number of degraded erythrocytes markedly decreases. However, a splenectomy should only be conducted if the state of the patients makes this necessary, as the removal of the organ increases the risk for infectious disease and sepsis.

It is absolutely advised against splenectomy on children under the age of 6 due to the risk for infections. Alternatively, an ectomy can be performed subtotal. In any case, vaccination against pneumococci, meningococci, and haemophilus influenza should be performed before splenectomy!

Complications of Hereditary Spherocytosis

Frequent complications of spherocytosis are gallstone colics due to pigmented gallstones. Also, hemolytic, vasoocclusive crises can occur, which can also lead to infarctions.
References

Begemann, Michael: Praktische Hämatologie, Stuttgart 1999 (11. Auflage)

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