Diamond–Blackfan anemia

Diamond-Blockfan anemia is a congenital aplastic anemia. It is a rare, congenital, pure red cell aplasia or hypoplasia. It usually arises sporadically; hereditary forms have autosomal dominant inheritance. The etiology involves mutations in ribosomal proteins (RPs), e.g. RPs 5, 11, 17, 19. These mutations result in increased adenosine deaminase activity in affected erythrocytes, which is normally involved in the degradation of purine nucleotide.

Clinical symptoms

- Conspicuous pallor (first appears around 6 months of age).
- Hepatosplenomegaly (about 40% of patients) without icterus.
- Renal hypoplasia.
- Thumb anomalies (so-called triphalangeal thumb).
- Growth failure with normal mental development.
- Increased risk of haematological and other malignancies.

Diagnosis

- Macrocytic[1] anemia.
- Significant decrease in reticulocytes (below 0,010).
- Medium bone marrow cellularity with a significant decrease to the absence of erythroid precursors (isolated erythroblasts have a megaloblastoid appearance).
- Elevated erythropoetin levels.
- Leukocytes and Thrombocytes show no abnormalities in their numbers.

Treatment

- Spontaneous remission (between 8-13 months of life).
- Corticoids.
- Substitution with erythrocyte mass - usually the disease has a progressive course that does not respond to corticosteroid therapy; however, iron chelators must be administered at the same time (to remove excess iron from the body and thus prevent tissue damage from this metal).
- Bone marrow or stem cell transplantation.

Sources

Related articles

- Aplastic anemia
- Anaemia

External links

- Velký lékařský slovník: Diamondova-Blackfanova anémie (http://lekarske.slovniky.cz/lexikon-pojem/diamondova-blackfanova-anémie-zkr-dba-3)

Bibliography

- PENKA, Miroslav. *Hematologie. I: Neonkologická hematologie.* 1. edition. Grada, 2001. ISBN 80-247-0023-9.

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