Blackfan Diamond Anemia

Blackfan Diamond anemia is a congenital aplastic anemia. It is rare, congenital erythroid aplasia or hypoplasia. Sporadic forms are more usual, hereditary forms suggest autosomal dominant mode of inheritance. In ethiology, this anemia is caused by mutations in ribosomal proteins (RP), e.g. PRS 5,11,17,19. The result of these mutations is increased activity of adenosin deaminase in defect erythrocytes. Adenosin deaminase normally helps with degradation of purine nucleotids.

Clinical signs

- Significant paleness (first visible around 6th month of life)
- Hepatosplenomegaly (in circa 40% of patients) without icterus
- Kidney hypoplasia
- Anomaly of fingers (triphalangeal thumb)
- Growth disorder with normal mental development
- Higher risk of hematological and other malignancies

Diagnosis

- Normocytic normochromic anemia
- Significant decrease in reticulocyte count (under 0,010)
- Mean cellular concentration in bone marrow with significant decrease of erythroid precursors (isolated erythroblasts have an apperance of megaloblasts)
- Increased levels of erythropoetin
- Leucocytes and thrombocytes are without any numerical anomaly

Treatment

- Spontaneous remission (between 8th-13th year of life)
- Corticoids
- Erythrocyte mass transfusion - if the disease is progressive, patient does not respond to corticoid therapy; it is important to administer iron chelators (to eliminate abundant iron and prevent tissue damage caused by iron)
- Bone marrow or stem cell transplant

Links

Related Articles

- Aplastická anémie
- Anémie

External Links

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

Sources

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