Fanconi's anemia

Fanconi anemia is a genetic disease with predominantly hematological symptoms.

⚠**Don't get confused with Fanconi's syndrome , which is another disease.**

- Etiologically, mutations in one of the group of genes (FANCA, FANCB, -C, -D1, -D2, -E, -F, -G, -J, -L, -M, -N, -O, -P) . The products of these proteins are involved in DNA repair .
- Significant chromosome instability, probably associated with numerous structural aberrations, which probably contributes to the higher incidence of leukemias in affected individuals, is probably also important.
- The disease is autosomal recessively inherited or X-linked.

Clinical Manifestations

- It usually manifests around the age of 7, sometimes right after birth. The form with minimal physical manifestations, usually diagnosed around the age of 30, is called Estren-Dameshek.
- pancytopenia , more often as a primary capture of partial (thrombo- or leukopenia) before the development of complete bone marrow failure. Over time, cytopenia deepens. Macrocytic erythropoiesis in hypocellular, dysplastic, fat-altered bone marrow, sometimes developed MDS or AML can be detected.
- **growth disorders ,**
- **developmental anomalies of the upper limb or thumb**
- **mental disorders ,**
- **typical dysmorphic facial features - wide nasal root, epicants, micrognathia**
- **hypogonadism in men**
- **deafness**
- **brown skin pigmentation ,**
- eye and **kidney hypoplasia**
- **tendency to malignant transformation** - the most common are leukemia or myelodysplastic syndrome , in adulthood then solid tumors, especially of the head and neck.

Diagnostics

- In peripheral blood leukopenia with thrombocytopenia , anemia is normocytic or slightly macrocytic.
- The bone marrow is initially hyperplastic with dysplasia in erythropoiesis, proliferation of plasma cells and mast cells .
- As the disease progresses, severe marrow hypoplasia occurs.
- Elevated fetal hemoglobin and cytogenetic abnormalities are typical features.
- Chromosome fragility assay with diepoxybutane (DEB) or Mitomycin C (MMC) performed on peripheral cells. blood.

Treatment

- Allogeneic bone marrow transplantation from an HLA-related donor (LD regime Cyclophosphamide + radiotherapy) is the only curative modality, umbilical cord blood cell transplants have also been successfully performed. Until then, androgens in combination with corticoids (balancing the anabolic effect of androgens with the catabolic effect of corticoids) and G-CSF substitution (in chronic application, however, Risk of leukemic clone stimulation and / or stem cell capacity depletion can be used temporarily with benefit).

Links

Related Articles

- Chromosomal instability syndromes
- Hereditary tumor syndromes
  - Neurofibromatosis
  - Wilms' tumor

Source

- ŠÍPEK, Antonín. *Genetic diseases* [online]. Last revision June 8, 2007, [cited. April 17, 2010]. < http://www.genetika-biologie.cz/hereditarni-nadorove-syndromy >.

Reference Litrature

- KLEIBL, Zdeněk and Jan NOVOTNÝ. *Hereditary tumor syndromes.* 1st edition. Prague: Triton, 2003. 31 pp. ISBN 80-7254-357-1 .
- PENKA, Miroslav. *Hematology. I, Neoncological hematology.* 1st edition. Prague: Grada, 2001. ISBN 80-247-0023-9 .
- Griffin P Rodgers, Neal S. Young: The Bethesda Handbook of Clinical Hematology, Third Edition by Lippincott, Williams and Wilkins, 2013, ISBN 987-1-4511-8270-5