Severe congenital Kostmann neutropenia

Severe congenital neutropenia or **Kostmann syndrome** or **infantile agranulocytosis** (SCN3, OMIM: 610738 (https://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=610738)) is an AR-inherited disease characterized by marked neutropenia and severe bacterial infections[1]. The genetic basis is a mutation of the HAX1 gene (1q21.3). There are also other forms of severe congenital neutropenia, for example autosomal dominantly inherited (mutations of the GFI1 gene - 1p22 or the ELA2 gene - 19p13.3).

Pathogenesis

- Disruption of the signaling cascade through the G-CSF pathway causes the absence of all stages of maturation from promyelocytes,
- Phagocytosis is impaired.

Clinical picture

- **Already in the first days of life – fever, skin and umbilical cord infections, stomatitis,**
- **Infections tend to generalize,**
- The most common causative agents: *Staphylococcus aureus*, *E. coli*, *Pseudomonas aeruginosa*.

Diagnostics

- Blood count + differential: **profound neutropenia**,
- Bone marrow: almost complete **absence of promyelocytes and myelocytes** with a normal number of myeloid lineage.

Treatment

- Recombinant **G-CSF** (long-term side effects: osteoporosis, bone fibrosis, splenomegaly),
- Event. bone marrow transplantation.

Links

Related Articles

- Primary immunodeficiency
- Neutropenia in children

Source

- - ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. The last revision 9. 6. 2006, [cit. 23. 12. 2009]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.

References

1. MUNTAU, Ania Carolina. *Pediatrie.* 4. edition. Grada, 2009. pp. 251-252. ISBN 978-80-247-2525-3.

Used literature

- BARTŮŇKOVÁ, Jiřina. *Imunodeficience.* 1. edition. Grada, 2002. 228 pp. ISBN 80-247-0244-4.