Severe congenital Kostmann's neutropenia

Severe congenital neutropenia, or **Kostmann's 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 severe neutropenia and severe bacterial infections.[1] The genetic basis is a mutation in the HAX1 gene (1q21.3). There are other forms of severe congenital neutropenia, such as autosomal dominant inherited (mutations of the GFI1-1p22 gene or the ELA2-19p13.3 gene).

Pathogenesis

- Disruption of the signal transduction cascade via the G-CSF causes the absence of all stages of maturation from promyelocytes,[1]
- Phagocytosis is impaired.

Clinical picture

- **Already in the first days of life - fever, infection of the skin and umbilical cord, stomatitis,**
- **Infections tend to become generalized,**
- Most common agents: *Staphylococcus aureus*, *E. coli*, *Pseudomonas aeruginosa*.[1]

Diagnostics

- Blood count + differential: **deep neutropenia,**
- Bone marrow: near-complete **absence of promyelocytes and myelocytes** at normal myeloid lineage.[1]

Therapy

- Recombinant **G-CSF** (long-term side effects: osteoporosis, bone fibrosis, splenomegaly),
- Alternatively, a bone marrow transplant.[1]

References

Related articles

- Primary immunodeficiency
- Neutropenia

Source

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

1. MUNTAU, Ania Carolina. *Pediatrie.* 4. vydání. Praha : Grada, 2009. s. 251-252. ISBN 978-80-247-2525-3.

Literature

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- BARTŮŇKOVÁ, Jiřina. *Imunodeficience.* 1. vydání. Praha : Grada, 2002. 228 s. ISBN 80-247-0244-4.

1. MUNTAU, Ania Carolina. *Pediatrie.* 4. vydání. Praha : Grada, 2009. s. 251-252. ISBN 978-80-247-2525-3.