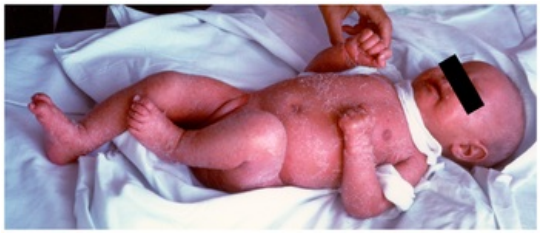
Omenn syndrome

Omenn syndrome (Familial reticuloendotheliosis with eosinophilia, Severe combined immunodeficiency with hypereosinophilia, OMIM: 603554 (https://omim.org/entry/603554)) is an autosomal recessive genetic disorder caused by the mutations of the genes **RAG1** (recombination-activating gene 1, 11p13), **RAG2** (recombination-activating gene 2, 11p13) or **Artemis** (DNA cross-link repair protein 1C; DCLRE1C, 10p). The syndrome is characterised by the **skin and intestinal mucosa (endothelium) infiltration** by the activated oligoclonal T-lymphocytes. Besides the **eosinophilia** (which is caused by the T-lymphocytes producing the IL-4 and IL-5 interleukins) in patients also occurs **skin disorders** of different ranges (erythroderma), **hepatosplenomegaly** and severe **diarrhea**.



Omenn syndrome in 5 months old child.

Links

Related articles

- Primary immunodeficiency
- Severe combined immunodeficiency
- Erythroderma

Source

- ŠÍPEK, Antonín. *Geneticky podmíněné poruchy imunitního systému* [online]. The last revision 9. 6. 2006, [cit. 16. 12. 2009]. <http://www.genetika-biologie.cz/primarni-imunodeficience>.

Bibliography

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