Reticular dysgenesis

Reticular dysgenesis (De Vaal's disease, hematopoietic hypoplasia, OMIM: 267500) is one of the rarest forms of severe combined immunodeficiency. **Congenital agranulocytosis** and **lymphopenia** associated with thymic and lymph node hypoplasia are characteristic. Granulocytes and lymphocytes are missing in the peripheral blood and bone marrow, the impairment of differentiation is already **at the level of stem cells** (and affects the myeloid and lymphoid lineages). The disease is inherited autosomal recessively, due to a mutation in the **AK2** gene (Adenylate Kinase 2) on chromosome 1 (1p34).

Links

- ws:Retikulární dysgeneze

Related articles

- Primary immunodeficiency
- Severe combined immunodeficiency

Source

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

m