Reticular dysgenesis

GENERAL INFORMATION

Description:

Reticular dysgenesis is a rare form of severe immunodeficiency that is usually fatal unless a successful stem cell transplant is performed. It is characterized by congenital agranulocytosis, lymphopenia, and lymphoid and thymic hypoplasia with absent cellular and humoral immunity functions. In 1959, de Vaal and Seynhaeve first described this disorder as RD. To date, fewer than 30 cases have been described.

Alternative names:

- Reticular dysgenesia
- Reticular dysgenesis
- Congenital aleukia
- Aleukocytosis
- Severe combined immunodeficiency with leukopenia
- De vaal disease
- Hematopoietic hypoplasia, generalized
- SCID with leukocyte deficiency

Classification:

- Combined B and T cell immunodeficiencies
  - T⁻B⁻ Severe combined immunodeficiency (SCID)

Inheritance:

Autosomal recessive

OMIM:

- %267500 Reticular dysgenesia

Cross references:

Incidence:

1 in 3-5 million people

CLINICAL INFORMATION

Description:

Reticular dysgenesis is a very rare autosomal recessive form of SCID, which generally leads to early death. The disorder usually manifests early in the neonatal period, with the sign of sepsis as the first day of life, mainly caused by severe leukopenia. The disease is characterized by lack or very reduced levels of B and T cells, thrombocytes involved in blood clotting, and granulocytes that form the majority of blood leukocytes, due to the failed maturation of not only lymphoid but also of myeloid precursor cells.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Reticular dysgenesis, ORPHANET
- Reticular dysgenesis, eMedicine

Therapeutic options:

- The only curative therapy is stem cell transplantation. Other recommendations include intravenous gamma-globulin infusion, irradiation of all blood products, antibiotherapy.
- Reticular dysgenesis, eMedicine
- National Marrow Donor Program

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies 2001-2004

GENE INFORMATION

Names:

Localization:

Other gene-based resources:

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- March of Dimes Birth Defects Foundation
- NIH/National Institute of Allergy and Infectious Diseases
- European Society for Immunodeficiencies

Other information sources:

- Chronic neutropenia, Haema 2002
- Reticular dysgenesis, Faculte de Medecine Broussais-Hotel-Dieu