Severe combined immunodeficiency due to complete RAG1/2 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Severe combined immunodeficiency due to complete RAG1/2 deficiency. ORPHA:331206

Severe combined immunodeficiency due to complete RAG1/2 deficiency is a rare, genetic T-B- severe combined immunodeficiency disorder due to null mutations in recombination activating gene (RAG) 1 and/or RAG2 resulting in less than 1% of wild type V(D)J recombination activity. Patients present with neonatal onset of life-threatening, severe, recurrent infections by opportunistic fungal, viral and bacterial micro-organisms, as well as skin rashes, chronic diarrhea, failure to thrive and fever. Immunologic observations include profound T- and B-cell lymphopenia, normal NK counts and low or absent serum immunoglobulins; some patients may have eosinophilia.