Severe Combined Immunodeficiency due to NHEJ1 Deficiency

National Cancer Institute

Source

National Cancer Institute. Severe Combined Immunodeficiency due to NHEJ1 Deficiency. NCI Thesaurus. Code C162695.

An autosomal recessive condition caused by mutation(s) in the NHEJ1 gene, encoding non-homologous end-joining factor 1. It is characterized by severe combined immunodeficiency that is T-cell negative, B-cell negative, NK-cell positive. Microcephaly, growth retardation, and sensitivity to ionizing radiation are also characteristic of this disease.