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Severe combined immunodeficiency due to CTPS1 deficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Severe combined immunodeficiency due to CTPS1 deficiency. ORPHA:420573*

Severe combined immunodeficiency (SCID) due to CTPS1 deficiency is a rare primary immunodeficiency disorder due to impaired capacity of activated T- and B-cells to proliferate in response to antigen receptor-mediated activation characterized by early-onset, severe, persistent and/or recurrent viral infections due to Epstein-Barr virus (EBV) and Varicella Zoster virus (VZV, including generalized varicella)), as well as recurrent sino-pulmonary bacterial infections due to encapsulated pathogens.