Open Peer Review on Qeios

Pancytopenia due to IKZF1 mutations

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pancytopenia due to IKZF1 mutations. ORPHA:317473

Pancytopenia due to IKZF1 mutations is a rare syndrome with combined immunodeficiency characterized by a variable clinical presentation ranging from asymptomatic individuals to potentially life-threatening, recurrent bacterial infections associated with progressive loss of serum immunoglobulins and B cells.