X-linked severe congenital neutropenia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked severe congenital neutropenia. ORPHA:86788

X-linked severe congenital neutropenia is an immunodeficiency syndrome characterized by recurrent major bacterial infections, severe congenital neutropenia, and monocytopenia. It has been described in five males spanning three generations of one family. It is transmitted as an X-linked recessive trait and is caused by mutations in the WAS gene, encoding the WASP protein.