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Absent thumb-short stature-immunodeficiency syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Absent thumb-short stature-immunodeficiency syndrome. ORPHA:2951*

Absent thumb-short stature-immunodeficiency is an exceedingly rare, autosomal recessive immune disease characterized by thumb aplasia, short stature with skeletal abnormalities, and combined immunodeficiency described in three sibships from two possibly related families. The skeletal abnormalities included unfused olecranon and the immunodeficiency manifested with severe chickenpox and chronic candidiasis. No new cases have been reported since 1978.