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Short-limb skeletal dysplasia with severe combined immunodeficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Short-limb skeletal dysplasia with severe combined immunodeficiency. ORPHA:935*

Short-limb skeletal dysplasia with severe combined immunodeficiency is an extremely rare type of SCID (see this term) characterized by the classical signs of T-B- SCID (severe and recurrent infections, diarrhea, failure to thrive, absence of T and B lymphocytes) (see this term), associated with skeletal anomalies like short stature, bowing of the long bones and metaphyseal abnormalities of variable degree of severity.