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Hyperphosphatasia-intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Hyperphosphatasia-intellectual disability syndrome. ORPHA:247262

A rare, congenital disorder of glycosylation-related bone disorder characterized by hypotonia, severe developmental delay, intellectual disability, seizures, increased serum alkaline phosphatase, short distal phalanges with hypoplastic nails, and dysmorphic facial features. In some cases, cleft palate, megacolon, anorectal malformations, and congenital heart defects have been reported.