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Short ulna-dysmorphism-hypotonia-intellectual disability syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Short ulna-dysmorphism-hypotonia-intellectual disability syndrome. ORPHA:357175*

Short ulna-dysmorphism-hypotonia-intellectual disability syndrome is a rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by mild to severe global development delay, severe intellectual disability, mild hypotonia, a short ulna, hirsutism of the face and extremities, minimal scoliosis, and facial dysmorphism, notably a tall broad forehead, synophrys, hypertelorism, malar hypoplasia, broad nose with thick alae nasi, low-set, small ears, long philtrum, thin upper lip and everted lower lip vermillion.