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Radioulnar synostosis-developmental delay-hypotonia syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Radioulnar synostosis-developmental delay-hypotonia syndrome. ORPHA:3270*

Radioulnar synostosis-developmental delay-hypotonia syndrome, also known as Der Kaloustian-McIntosh-Silver syndrome, is an extremely rare syndrome with synostosis described in about 4 patients to date with clinical manifestations including congenital unilateral radioulnar synostosis, generalized hypotonia, developmental delay, and dysmorphic facial features (long face, prominent nose and ears).