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Mesomelic dwarfism-cleft palate-camptodactyly syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Mesomelic dwarfism-cleft palate-camptodactyly syndrome. ORPHA:2631*

Mesomelic dwarfism-cleft palate-camptodactyly syndrome is characterised by mesomelic shortening and bowing of the limbs, camptodactyly, skin dimpling and cleft palate with retrognathia and mandibular hypoplasia. It has been described in a brother and sister born to consanguineous parents. Transmission is autosomal recessive.