Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome. ORPHA:314394

Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome is a rare, genetic, primary bone dysplasia disorder characterized by severe pre- and post-natal short stature, facial dysmorphism (incl.dolicocephaly, long triangular face, tall forehead, down-slanting palpebral fissures, prominent nose, long philtrum, small ears), early-onset or postpubertal sparse, short hair and hypoplastic fingernails. Small hands with tapering fingers, bracydactyly and fifth-finger clinodactyly, as well as a high-pitched voice are also associated.