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Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome. ORPHA:477993*

Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome is a rare, genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, axial hypotonia, palate abnormalities (including cleft palate and/or high and narrow palate), dysmorphic facial features (including prominent forehead, hypertelorism, downslanting palpebral fissures, wide nasal bridge, thin lips and widely spaced teeth), and short stature. Additional manifestations may include digital anomalies (such as brachydactyly, clinodactyly, and hypoplastic toenails), a single palmar crease, lower limb hypertonia, joint hypermobility, as well as ocular and urogenital anomalies.