Open Peer Review on Qeios

Pierpont syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pierpont syndrome. ORPHA:487825

Pierpont syndrome is a rare subcutaneous tissue disorder characterized by axial hypotonia after birth, prolonged feeding difficulties, moderate to severe global developmental delay, seizures (in particular absence seizures), fetal digital pads, distinctive plantar fat pads anteromedial to the heels, deep palmar and plantar grooves. Additionally, distinct craniofacial dysmorphic features, notably a broad face with high forehead, high anterior hairline, narrow palpebral fissures that take on a crescent moon shape when smiling, broad nasal bridge and tip with anteverted nostrils, mild midfacial hypoplasia, long, smooth philtrum, thin upper lip vermillion, small, widely spaced teeth and flat occiput/microcephaly/brachycephaly, are also chararteristic. Over time, fat pads may become less prominent and disappear.