Arachnodactyly-intellectual disability-dysmorphism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Arachnodactyly-intellectual disability-dysmorphism syndrome. ORPHA:1130

A rare multiple congenital anomalies/dysmorphic syndrome characterized by facial dysmorphism (brachycephaly, long, narrow, triangular face, prominent forehead, hypertelorism, flat philtrum, microstomia, thin lips, hypoplastic maxilla), marfanoid habitus with arachnodactyly, and moderate to severe intellectual disability. Additional features may include clinodactyly, triphalangeal thumbs, hammer-shaped toes, hyperextensible joints, hypotonia, hyperreflexia and underdeveloped musculature. Delayed external genitalia development, as well as seizures and mitral regurgitation have been reported in some cases. There have been no further descriptions in the literature since 1995.