Cryptorchidism-arachnodactyly-intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cryptorchidism-arachnodactyly-intellectual disability syndrome. ORPHA:1548

Cryptorchidism-arachnodactyly-intellectual disability syndrome is a rare, multiple congenital anomalies syndrome characterized by psychomotor delay, severe intellectual deficit, severe muscle hypoplasia (with absence of subcutaneous fatty tissue), generalized contractures, craniofacial dysmorphic features (dolichocephaly, esotropia, ears of unequal size, high palate), chest and spinal deformities (i.e. sternum shifted to side, kyphoscoliosis), pulmonary anomalies (unilateral hypoplastic bronchial system), arachnodactyly, and genital abnormalities (cryptorchidism, hypospadias, testicular agenesis). Repeated respiratory tract infections and atelectasis are also associated. There have been no further descriptions in the literature since 1970.