FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome. ORPHA:404451

FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome is a rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by delayed motor development, intellectual disability, dysarthria, pseudobulbar signs, cryptorchidism, and syndactyly associated with a FLBN1 gene point mutation. Macular degeneration and signs of brain atrophy and spinal cord compression have also been reported.