Inherited congenital spastic tetraplegia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Inherited congenital spastic tetraplegia. ORPHA:210141

Inherited congenital spastic tetraplegia is a rare, genetic, neurological disease characterized by non-progressive, variable spastic quadriparesis in multiple members of a family, in the absence of additional factors complicating pregnancy or birth (e.g. perinatal asphyxia, congenital infection). Additional clinical features include congenital hypotonia, intellectual disability, and developmental delay. Dysphagia, dysarthria, exotropia, nystagmus, seizures and brain atrophy with ventriculomegaly may be also present.