Cervical hypertrichosis-peripheral neuropathy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cervical hypertrichosis-peripheral neuropathy syndrome. ORPHA:2218

Cervical hypertrichosis peripheral neuropathy is a rare syndrome characterized by the association of congenital hypertrichosis in the anterior cervical region with peripheral sensory and motor neuropathy. It has been described in three members of the same family and in one unrelated boy. Associated features in the familial cases include retinal anomalies, spina bifida, kyphoscoliosis and hallux valgus, while that in the non-familial case includes developmental delay. An autosomal recessive mode of inheritance is suggested. There have been no further descriptions in the literature since 1993.