Leukoencephalopathy-metaphyseal chondrodysplasia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Leukoencephalopathy-metaphyseal chondrodysplasia syndrome. ORPHA:83629

The association of leukoencephalopathy and metaphyseal chondrodysplasia has been reported in four men from a three-generation family. Onset manifests by spastic paraplegia at the age of 2, followed by tremor, ataxia, optic atrophy, and spastic tetraparesis. Transmission is X-linked and the gene responsible of the disease may be located at Xq25-q27.