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Paraplegia-intellectual disability-hyperkeratosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Paraplegia-intellectual disability-hyperkeratosis syndrome. ORPHA:2824

Paraplegia-intellectual disability-hyperkeratosis syndrome is characterized by intellectual deficit, spasticity in the lower limbs (spastic paraplegia), pes cavus deformity of both feet, an abnormal gait, and palmar and plantar hyperkeratosis. It has been reported in four brothers. The mother of the affected boys had normal intelligence, plantar hyperkeratosis and a strong facial resemblance to her retarded sons. Her three daughters were normal. This syndrome most likely an X-linked recessive condition.