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Proximal myopathy with extrapyramidal signs

INSFRM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Proximal myopathy with extrapyramidal signs</u>. ORPHA:401768

Proximal myopathy with extrapyramidal signs is a rare, hereditary non-dystrophic myopathy characterized by proximal muscle weakness, delayed motor development, learning difficulties, and progressive extrapyramidal motor signs including chorea, dystonia and tremor. Variable additional features have been reported - ataxia, microcephaly, ophthalmoplegia, ptosis, and optic atrophy.

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