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Hypomyelination with brain stem and spinal cord involvement and leg spasticity

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Hypomyelination with brain stem and spinal cord involvement and leg spasticity.

ORPHA:363412

Hypomyelination with brain stem and spinal cord involvement and leg spasticity is a rare, genetic, leukodystrophy disorder characterized by diffuse hypomyelination in the supratentorial brain white matter, brain stem and spinal cord. Patients usually present nystagmus, lower limb spasticity, hypotonia, and motor developmental delay, as well as MRI signal abnormalities involving the corpus callosum, anterior brainstem, pyramidal tracts, superior and inferior cerebellar peduncles, dorsal columns and/or lateral corticospinal tracts.