

Open Peer Review on Qeios

L1 syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>L1</u> syndrome. ORPHA:275543

L1 syndrome is a mild to severe congenital X-linked developmental disorder characterized by hydrocephalus of varying degrees of severity, intellectual deficit, spasticity of the legs, and adducted thumbs. The syndrome represents a spectrum of disorders including: X-linked hydrocephalus with stenosis of the aqueduct of Sylvius (HSAS), MASA syndrome, X-linked complicated hereditary spastic paraplegia type 1, and X-linked complicated corpus callosum agenesis (see these terms).

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