L1 syndrome (L1CAM-related disorders)

Record Status
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Citation

Authors’ conclusions
L1 syndrome is a genetic disorder caused by variants in the L1CAM gene. The L1CAM gene, which is located on the X chromosome at band q28, encodes the L1 cell adhesion molecule. Since L1 syndrome is inherited in an X-linked manner, the condition primarily affects males (since they have only 1 X chromosome and therefore just 1 copy of the L1CAM gene). Carrier females (who have 2 X chromosomes with 1 normal copy of L1CAM and 1 copy with a variant) are typically unaffected. Estimates of the prevalence of the L1 syndrome range from 1 in 25,000 to 1 in 60,000 male births. The L1CAM gene product is a cell surface protein with important roles in nerve cell development and function. Consequently, variants in the L1CAM gene result in a wide range of neurological symptoms, including hydrocephalus (excess fluid in the brain), intellectual disability, adducted (clasped) thumbs, spastic paraplegia (stiffness of the limbs), agenesis of the corpus callosum (absence of the tissue connecting the 2 hemispheres of the brain), and aphasia (an absence of normal speech). The diagnosis of L1 syndrome encompasses several neurological disorders that were initially believed to be distinct clinical syndromes. However, these conditions, which include X-linked hydrocephalus with stenosis (narrowing) of the aqueduct of Sylvius (HSAS), MASA (mental retardation, adducted thumbs, shuffling gait, and aphasia) syndrome, X-linked complicated corpus callosum agenesis, and hereditary spastic paraplegia type 1 (SPG1), are now known to represent a disease spectrum that results from L1CAM gene variants. At the severe of the spectrum (e.g., HSAS), patients present before birth with significant hydrocephalus and may die in the first few years of life. At the mild end of the spectrum (e.g., SPG1), patients present in infancy or early childhood and may exhibit only mild intellectual disability and limb spasticity. There is no cure for L1 syndrome and no way to prevent or delay symptoms. Disease management is generally based on the symptoms present and may include physical and occupational therapies, medications for spasticity and seizures, surgery to improve function in those with mildly adducted thumbs, and shunt placement for those with hydrocephalus.

Final publication URL
The report may be purchased from: http://www.hayesinc.com/hayes/crd/?crd=16296

Indexing Status
Subject indexing assigned by CRD

MeSH
Humans; Genetic Diseases, X-Linked; Neural Cell Adhesion Molecule L1

Language Published
English

Country of organisation
United States

English summary
An English language summary is available.

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**AccessionNumber**
32014000482

**Date abstract record published**
04/04/2014