DISEASE:
MASA syndrome

NAME:	MASA syndrome
DESCRIPTION:	A X-linked, clinical subtype of L1 syndrome, characterized by mild to moderate intellectual disability, delayed development of speech, hypotonia progressing to spasticity or spastic paraplegia, adducted thumbs, and mild to moderate distension of the cerebral ventricles.
ORPHACODE:	2466
SYNONYMS:	Intellectual disability-aphasia-shuffling gait-adducted thumbs syndrome
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	L1CAM
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