
DISEASE:
Metachromatic leukodystrophy, late infantile form

NAME:	Metachromatic leukodystrophy, late infantile form
DESCRIPTION:	A subtype of Metachromatic leukodystrophy characterized by rapidly progressive psychomotor regression with an onset before 30 months of age after a period of apparently normal development. Manifestations developing during the course of the disease are impaired feeding and swallowing due to pseudobulbar palsies, seizures, painful spasms, muscle weakness, ataxia, paralysis, dementia, and loss of speech, vision, and hearing, quickly resulting in complete loss of motor and cognitive skills, and decerebration. Death occurs within the first decade of life.
ORPHACODE:	309256
SYNONYMS:	Arylsulfatase A deficiency, late infantile form MLD, late infantile form
XREF(S):	<u>Orphanet</u> <u>ICD-10</u>
ANALYTE(S):	<u>PSAP</u> <u>ARSA</u>
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