DISEASE:
Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome

NAME:	Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome
DESCRIPTION:	A rare autosomal ichthyosis syndrome with prominent neurologic signs characterized by the association of congenital ichthyosis with global developmental delay, intellectual disability, infantile-onset seizures, and spastic tetraplegia. Brain imaging may show delayed myelination and cerebral atrophy. Marked intrafamilial variability has been reported.
ORPHACODE:	352333
SYNONYMS:	Congenital ichthyosis-intellectual disability-spastic tetraplegia syndrome ELOVL4-related neuro ichthyosis
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ELOVL4
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