DISEASE:
Corpus callosum agenesis-neuronopathy syndrome

NAME:	Corpus callosum agenesis-neuronopathy syndrome
DESCRIPTION:	A rare neurodegenerative disorder characterized by severe progressive sensorimotor neuropathy beginning in infancy with resulting hypotonia, areflexia, amyotrophy and variable degrees of dysgenesis of the corpus callosum. Additional features include mild-to-severe intellectual and developmental delays, and psychiatric manifestations that include paranoid delusions, depression, hallucinations, and 'autistic-like' features. Affected individuals are usually wheelchair restricted in the second decade of life and die in the third decade of life. The disease is inherited as an autosomal recessive trait.
ORPHACODE:	1496
SYNONYMS:	Andermann syndrome Charlevoix disease
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	SLC12A6
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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