DISEASE:
Severe Canavan disease

NAME:	Severe Canavan disease
DESCRIPTION:	Severe Canavan disease (CD) is a rapidly progressing neurodegenerative disorder characterized by leukodystrophy with macrocephaly, severe developmental delay and hypotonia.
ORPHACODE:	314911
SYNONYMS:	Infantile Canavan disease Neonatal Canavan disease
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ASPA
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Canavan disease (hot spot mutation - p.Glu285Ala, p.Tyr231*)
- Jewish mutation panel (Tay Sachs, Fanconi, Dysautonomia, Canavan) (4 genes; 7 hot spot mutations)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- aspartoacylase

Related Gene Panels

- Hot spot mutation among Jewish (4 genes, 7 mutations) - UZA