DISEASE:
Chronic visceral acid sphingomyelinase deficiency

NAME:	Chronic visceral acid sphingomyelinase deficiency
DESCRIPTION:	A rare autosomal recessive, chronic, acid sphingomyelinase deficiency characterized clinically by onset in childhood with hepatosplenomegaly, growth retardation, interstitial lung disease and absence of neurodegenerative disorders.
ORPHACODE:	77293
SYNONYMS:	Chronic visceral ASMD NPD-B Niemann-Pick disease type B
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	SMPD1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Lysosomal Storage Disease (gene panel)
- Metabolic diseases with hepatic disorders (20 genes)
- Niemann-Pick disease

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- sphingomyelin phosphodiesterase 1

Related Gene Panels

- Cholestasis (40 genes) - UCL
- Lysomal Storage (64 genes) - VUB