DISEASE:
Sialidosis type 1

NAME:	Sialidosis type 1
DESCRIPTION:	Sialidosis type 1 (ST-1) is a very rare lysosomal storage disease, and is the normosomatic form of sialidosis (see this term), characterized by gait abnormalities, progressive visual loss, bilateral macular cherry red spots and myoclonic epilepsy and ataxia, that usually presents in the second to third decade of life.
ORPHACODE:	812
SYNONYMS:	Cherry-red spot-myoclonus syndrome Lipomucopolysaccharidosis Normomorphic sialidosis
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	NEU1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Lysosomal Storage Disease (gene panel)
- Metabolic diseases with hepatic disorders (20 genes)
- Neuraminidase deficiency / Sialidosis

Related Laboratories

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

Related Analytes

- neuraminidase 1

Related Gene Panels

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