

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Choroba	Cherry-red spot-myoclonus syndrome Lipomukopolisacharydoza Zespół skurczów mioklonicznych z wiśniowymi plamkami Lipomucopolysaccharidosis Normomorphic sialidosis

Kod ORPHA	Kod OMIM	Kod ICD10
812	256550	E77.1

Kod ICD11
5C56.21

*Źródło

orphanet