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
Choroba

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

Kod ORPHA
812

Kod OMIM
256550

Kod ICD10
E77.1

Kod ICD11
5C56.21

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*Źródło

orphanet