

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

*Źródło

orphanet