

Opis choroby *

Definicja

Sialidosis type 2 (ST-2) is a rare lysosomal storage disease, and the severe, early onset form of sialidosis (see this term) characterized by a progressively severe mucopolysaccharidosis-like phenotype (coarse facies, dysostosis multiplex, hepatosplenomegaly), macular cherry-red spots as well as psychomotor and developmental delay. ST-2 displays a broad spectrum of clinical severity with antenatal/congenital, infantile and juvenile presentations.

Dane

Klasyfikacja	Synonimy
Choroba	Infantile dysmorphic sialidosis Dziecięca sialidoza dysmorficzna

Kod ORPHA	Kod OMIM	Kod ICD10
87876	256550	E77.1

Kod ICD11
5C56.21

*Źródło

orphanet