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
Choroba

Synonimy
Infantile dysmorphic sialidosis
Dziecięca sialidoza dysmorficzna

Kod ORPHA
87876

Kod OMIM
256550

Kod ICD10
E77.1

Kod ICD11
5C56.21

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*Źródło

orphanet