Opis choroby *

Definicja

A rare peroxisomal beta-oxidation disorder characterized by deficiency of peroxisomal D-bifunctional protein, type 1 being caused by deficiency of both dehydrogenase and hydratase activities of the enzyme, and types 2 and 3 by hydratase or dehydrogenase deficiency alone, while type 4 is due to compound heterozygous mutations affecting both units and represents a clinically milder phenotype. Types 1-3 are typically fatal in infancy. Patients present with early onset of generalized hypotonia, seizures, severe global developmental delay, craniofacial dysmorphism (large fontanel, high forehead, hypertelorism, epicanthal folds) and elevated plasma very long chain fatty acids. Variable features include hepatomegaly, polymicrogyria, and cerebral white matter abnormalities, among others.

Dane

Klasyfikacja
Choroba

Kod ORPHA
300

Kod OMIM
261515

Kod ICD10
E71.3

Kod ICD11
5C57.1

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*Źródło

orphanet