

Opis choroby *

Definicja

Zellweger-like syndrome without peroxisomal anomalies is an extremely rare mitochondrial disorder characterized by facial dysmorphism similar to that seen in Zellweger syndrome (see this term), such as frontal bossing, high forehead, upslanting palpebral fissures, hypoplastic supraorbital ridges, and epicanthal folds, and in addition, pale skin, profound hypotonia, developmental delay, and minor metabolic anomalies. No peroxysomal defects, however, have been reported. Transmission is thought to be autosomal recessive.

Dane

Klasyfikacja

Choroba

Synonimy

Ahn-Lerman-Sagie syndrome
Zespół Ahna, Lermana i Sagie'a

Kod ORPHA

50812

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

5C53.2Y

*Źródło

orphanet