Opis choroby *

Definicja

Transient neonatal multiple acyl-CoA dehydrogenase deficiency describes a very rare condition where a maternal riboflavin deficiency causes an infant to present with manifestations similar to those seen in multiple acyl-CoA dehydrogenase (MAD) deficiency (see this term) such as poor suck, metabolic acidosis and hypoglycemia, but that resolves completely with oral riboflavin. In the one patient described haploinsufficiency of the human riboflavin transporter (hRFT1) was described in the mother.

Dane

Klasyfikacja
Choroba

Synonimy
Transient neonatal MAD deficiency
Przejściowa noworodkowa acyduria glutarowa typu 2
Przejściowa noworodkowa kwasica glutarowa typu 2
Przejściowy noworodkowy MADD
Przejściowy noworodkowy Niedobór MAD
Transient neonatal MADD
Transient neonatal glutaric acidemia type 2
Transient neonatal glutaric aciduria type 2

Kod ORPHA
329942

Kod OMIM
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Kod ICD10
E71.3

Kod ICD11
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*Źródło

orphanet