Opis choroby *

Definicja

A rare endocrine disease characterized by neonatal hypoglycemia, prolonged cholestatic jaundice, and seizures. Typical are low plasma ACTH and cortisol levels in the absence of structural pituitary defects, and sometimes low partial growth hormone deficiency is associated.

Dane

Klasyfikacja
Choroba

Kod ORPHA
199296

Kod OMIM
201400

Kod ICD10
E23.6

Kod ICD11
5A74.Y

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*Źródło

orphanet