Opis choroby *

Definicja

Nonspherocytic haemolytic anaemia due to hexokinase deficiency is characterised by severe hemolysis, appearing in infancy. Seventeen affected families have been reported so far. Transmission is autosomal recessive. Mutations have been described in HK1, the gene that encodes red blood cell-specific hexokinase-R.

Dane

Klasyfikacja
Choroba

Kod ORPHA
90031

Kod OMIM
235700

Kod ICD10
D55.2

Kod ICD11
3A10.Y

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*Źródło

orphanet