Opis choroby *

Definicja

A very rare non-syndromic autosomal recessive pyridoxine-refractory sideroblastic anemia due to a splice defect of glutaredoxin-5 (GLRX5) described in a single patient with adult onset microcytic hypochromic anemia with liver iron overload and type 2 diabetes.

Dane

Klasyfikacja
Choroba

Synonimy
GLRX5-related sideroblastic anemia
Niedokrwistość syderoblastyczna związana z GLRX5

Kod ORPHA
255132

Kod OMIM
616860

Kod ICD10
D64.0

Kod ICD11
3A72.00

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*Źródło

orphanet