Opis choroby *

Definicja

Primary familial polycythemia is an inherited hematological disorder resulting from mutations in the erythropoietin (EPO) receptor and is characterized by an elevated absolute red blood cell mass caused by uncontrolled red blood cell production in the presence of low EPO levels.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital erythrocytosis due to erythropoietin receptor mutation
PFCP
Pierwotna rodzinna i wrodzona policytemia
Pierwotna wrodzona erytrocytoza
Rodzinna erytrocytoza
Wrodzona erytrocytoza z powodu mutacji receptora erytropoetyny
Wrodzona policytemia z powodu mutacji receptora erytropoetyny
Congenital polycythemia due to erythropoietin receptor mutation
Familial erythrocytosis
PFCP
Primary congenital erythrocytosis
Primary familial and congenital polycythemia

Kod ORPHA
90042

Kod OMIM
133100

Kod ICD10
D75.0

Kod ICD11
3A80.0

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*Źródło

orphanet