DISEASE:
Primary familial polycythemia

NAME:	Primary familial polycythemia
DESCRIPTION:	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.
ORPHACODE:	90042
SYNONYMS:	Congenital erythrocytosis due to erythropoietin receptor mutation Congenital polycythemia due to erythropoietin receptor mutation Familial erythrocytosis PFCP Primary congenital erythrocytosis Primary familial and congenital polycythemia
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	EPOR
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Erythrocytoses, polycythémies, thrombocytoses et neutropénies congénitales (gene panel)
- Primary familial erythrocytosis or Primary familial congenital polycythemia

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- Centre de Génétique Humaine - CHU Sart-Tilman

Related Analytes

- erythropoietin receptor

Related Gene Panels

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