

ANALYTE:
PTPN11

NAME:	protein tyrosine phosphatase non-receptor type 11
SYMBOL:	PTPN11
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	BPTP3 PTP2C SH-PTP2 SH2 domain-containing protein tyrosine phosphatase 2 SHP-2 SHP2
XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
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- [Congenital malformation \(gene panel - 1721 genes\)](#)
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- RASopathy (gene panel)
- Rendu-Osler-Weber disease (4 genes)
- Short Stature (gene panel)
- Short stature/ Growth retardation/ (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skin disorders (gene panel)
- Stroke (gene panel)
- Sturge-Weber syndrome (gene panel)
- Venous malformation (3 genes)
- « Inherited bone marrow failures syndromes » with or without organ dysfunction

Related Diseases

- Juvenile myelomonocytic leukemia
- Metachondromatosis
- Noonan syndrome
- Noonan syndrome with multiple lentigines

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Congenital structural heart defects - UGent
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB
- Early onset epileptic encephalopathy (845 genes) - ULB
- Epilepsy gene panel - VUB
- Growth retardation/short stature (genepanel) - UZA

- [Hematologic Familiar Forms - ULG](#)
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