# Belgian Genetic Tests database

## ANALYTE:
## PYCR1

| | |
|---|---|
| **NAME:** | pyrroline-5-carboxylate reductase 1 |
| **SYMBOL:** | PYCR1 |
| **VERSION OF ORPHANET:** | 2023-06-22 14:14:43 |
| **SYNONYMS:** | P5C |
| **XREF(S):** | Orphanet<br>Ensembl<br>Genatlas<br>HGNC<br>OMIM<br>Reactome<br>SwissProt |
| **CREATED:** | 13 May 2019 - 01:01 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

Source URL: http://gentest.healthdata.be/analyte/1649

# RELATED CONTENT

## Related Genetic Tests

- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Cutis Laxa / Geroderma osteodysplasticum (gene panel)
- Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel - 394 genes)
- Intellectual disability & Epilepsy (gene panel)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Mitochondrial disorders (gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)

## Related Diseases

- Autosomal recessive cutis laxa type 2B
- Geroderma osteodysplastica
- PYCR1-related De Barsy syndrome

## Related Gene Panels

- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB

- Cutis Laxa / Geroderma osteodysplasticum - UGent
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent
- mitochondrial disease, nuclear based (343 genes) - VUB

---