# Belgian Genetic Tests database

## ANALYTE:
## NPHP1

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|---|---|
| **NAME:** | nephrocystin 1 |
| **SYMBOL:** | NPHP1 |
| **VERSION OF ORPHANET:** | 2023-06-22 14:14:43 |
| **SYNONYMS:** | JBTS4<br>SLSN1 |
| **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/2053

# RELATED CONTENT

## Related Genetic Tests

- Ataxia (autosomic dominant and recessive / except expansion of triplets) (gene panel - 722 genes)
- Cerebral palsy (gene panel)
- Cholestasis (gene panel)
- Ciliopathy (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Early onset epileptic encephalopathy (gene panel - 845 genes)
- End-stage renal disease, ESRD (gene panel)
- Epilepsy gene panel
- Hepatology (gene panel)
- Hepatorenal disorders (gene panel)
- Inherited Kidney Diseases (Gene Panel)
- Intellectual disability & Epilepsy (gene panel)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Nephrogenetics / Nephropathy (gene panel)
- Nephropathies, hereditary (gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Retinal dystrophy / RETNET (gene panel)

## Related Diseases

- Bardet-Biedl syndrome
- Joubert syndrome with renal defect
- Juvenile nephronophthisis
- Senior-Loken syndrome

## Related Gene Panels

- Ataxia (348 genes) - ULB
- Cerebral palsy (212 genes) - UZA
- Cholestasis (40 genes) - UCL
- Ciliopathy (120 genes) - UGent
- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Early onset epileptic encephalopathy (845 genes) - ULB
- End-stage renal disease (106 genes) - IPG
- Epilepsy gene panel - VUB
- Hepatology panel - UGent
- Hepatorenal disorders (13 genes) - UCL
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Nephropathies, hereditary (219 genes) - KUL
- Nephropathy panel - UGent
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Panel Nephro-ULG-V1
- Retinal dystrophy - UGent

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