

**ANALYTE:**  
**NF1**

|                             |  |
|-----------------------------|--|
| <b>NAME:</b>                | neurofibromin 1  |
| <b>SYMBOL:</b>              | NF1  |
| <b>VERSION OF ORPHANET:</b> | 2023-06-22 14:14:43  |
| <b>SYNONYMS:</b>            | Watson disease<br>neurofibromatosis<br>von Recklinghausen disease  |
| <b>XREF(S):</b>             | <a href="#">Orphanet</a><br><a href="#">Ensembl</a><br><a href="#">Genatlas</a><br><a href="#">HGNC</a><br><a href="#">OMIM</a><br><a href="#">Reactome</a><br><a href="#">SwissProt</a> |
| <b>CREATED:</b>             | 13 May 2019 - 01:01  |
| <b>CHANGED:</b>             | 26 Oct 2023 - 23:49  |

## RELATED CONTENT

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### Related Genetic Tests

- [Congenital malformation \(gene panel - 1721 genes\)](#)
- [Congenital malformation \(gene panel\)](#)
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- [Dermatogenetic panel, severe, rare and hereditary genodermatoses \(gene panel - 394 genes\)](#)
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- [Epilepsy \(gene panel\)](#)
- [Epilepsy gene panel](#)
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- [Intellectual disability \(virtual gene panel\)](#)
- [Maffucci syndrome \(gene panel\)](#)
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- [Neurodevelopmental disorders \(gene panel\)](#)
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- [Neurofibromatosis type 1 / Legius syndrome \(2 genes\)](#)
- [Neurofibromatosis type I](#)
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- [Paraganglioma-pheochromocytoma \(gene panel\)](#)
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- [Skin disorders \(gene panel\)](#)
- [Stroke \(gene panel\)](#)
- [Sturge-Weber syndrome \(gene panel\)](#)
- « Inherited bone marrow failures syndromes » with or without organ dysfunction

## Related Diseases

- [17q11 microdeletion syndrome](#)
- [17q11.2 microduplication syndrome](#)
- [Alveolar rhabdomyosarcoma](#)
- [Embryonal rhabdomyosarcoma](#)
- [Hereditary pheochromocytoma-paraganglioma](#)
- [Juvenile myelomonocytic leukemia](#)
- [Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion](#)
- [Neurofibromatosis-Noonan syndrome](#)
- [Pleomorphic rhabdomyosarcoma](#)

## Related Gene Panels

- [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](#)
- [Epilepsy, seizures - IPG](#)
- [Hematologic Familiar Forms - ULG](#)
- [Hereditary predisposition to cancer \(47 genes\) - IPG](#)

- [Intellectual Disability \(104 genes\) - IPG](#)
- [Intellectual disability & Epilepsy - UGent](#)
- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability \(gene panel\) - UZA](#)
- [Maffucci syndrome \(65 genes\) - KUL](#)
- [Neurodevelopmental disorders \(1300 genes\) - ULB](#)
- [Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders \(1162 genes\) - VUB](#)
- [Onco-endocrine pathologies \(50 genes\) - UCL](#)
- [Overgrowth & vascular anomalies \(65 genes\) - KUL](#)
- [Paraganglioma and pheochromocytoma \(29 genes\) - UCL](#)
- [Paraganglioma-pheochromocytoma \(8 genes\) - KUL](#)
- [Pediatric oncopredisposition - UGent](#)
- [Rare epilepsy with developmental delay \(gene panel\) - UZA](#)
- [Skeletal dysplasia \(394 genes\) - VUB](#)
- [Skeletal dysplasia \(genepanel\) - UZA](#)
- [Skeletal dysplasia - UGent](#)
- [Skin disorders - UGent](#)
- [Stroke - UGent](#)
- [Sturge-Weber syndrome \(65 genes\) - KUL](#)
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