

**ANALYTE:**  
**PTCH1**

<b>NAME:</b>	patched 1
<b>SYMBOL:</b>	PTCH1
<b>VERSION OF ORPHANET:</b>	2023-06-22 14:14:43
<b>SYNONYMS:</b>	BCNS
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">Ensembl</a> <a href="#">Genatlas</a> <a href="#">HGNC</a> <a href="#">OMIM</a> <a href="#">Reactome</a> <a href="#">SwissProt</a>
<b>CREATED:</b>	13 May 2019 - 01:01
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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## RELATED CONTENT

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### Related Genetic Tests

- [Brain malformations \(gene panel\)](#)
- [Congenital malformation \(gene panel - 1721 genes\)](#)
- [Congenital malformation gene panel](#)
- [Dermatogenetic panel, severe, rare and hereditary genodermatoses \(gene panel - 394 genes\)](#)
- [Early onset epileptic encephalopathy \(gene panel - 845 genes\)](#)
- [Epilepsy gene panel](#)
- [Gorlin syndrome \(PTCH1; SUFU genes\)](#)
- [Gorlin syndrome \(gene panel\)](#)
- [Hereditary cancer \(gene panel\)](#)
- [Intellectual disability & Epilepsy \(gene panel\)](#)
- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability \(virtual gene panel\)](#)
- [Maffucci syndrome \(gene panel\)](#)
- [Malformations of cortical development \(235 genes\)](#)
- [Medulloblastoma \(3 genes\)](#)
- [Medulloblastoma \(gene panel\)](#)
- [Microphthalmia / Anophthalmia / Coloboma-Anterior Segment Dysgenesis \(MAC-ASD\) \(gene panel\)](#)
- [Neurodevelopmental disorders \(1300 genes\)](#)
- [Neurodevelopmental disorders gene panel](#)
- [Onco-endocrine pathologies \(gene panel\)](#)
- [Overgrowth & vascular anomalies \(gene panel\)](#)
- [Pediatric oncopredisposition \(gene panel\)](#)
- [Skin disorders \(gene panel\)](#)
- [Sturge-Weber syndrome \(gene panel\)](#)
- [cleft lip with/without cleft palate \(virtual gene panel\)](#)

## Related Diseases

- [Alobar holoprosencephaly](#)
- [Gorlin syndrome](#)
- [Lobar holoprosencephaly](#)
- [Microform holoprosencephaly](#)
- [Midline interhemispheric variant of holoprosencephaly](#)
- [Monosomy 9q22.3](#)
- [Schilbach-Rott syndrome](#)
- [Semilobar holoprosencephaly](#)
- [Septopreoptic holoprosencephaly](#)

## Related Gene Panels

- [Brain malformations \(34 genes\) - ULB](#)
- [Cleft lip and palate / dysmorphic facial features / craniofacial anomalies \(255 genes\) - UCL](#)
- [Congenital malformation \(1721 genes\) - ULB](#)
- [Congenital malformation gene panel - VUB](#)
- [Dermatogenetic / severe, rare and hereditary genodermatoses \(394 genes\) - ULB](#)
- [Early onset epileptic encephalopathy \(845 genes\) - ULB](#)
- [Epilepsy gene panel - VUB](#)
- [Gorlin syndrome \(2 genes\) - KUL](#)
- [Gorlin syndrome \(3 genes\)](#)
- [Hereditary predisposition to cancer \(47 genes\) - IPG](#)
- [Intellectual disability & Epilepsy - UGent](#)
- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability/Epilepsy \(1091 genes\) - ULG](#)
- [Maffucci syndrome \(65 genes\) - KUL](#)
- [Malformations of cortical development \(235 genes\) - VUB](#)
- [Medulloblastoma \(3 genes\) - KUL](#)
- [Medulloblastoma \(3 genes\) - UCL](#)

- Microphtalmia/Anophthalmia/Coloboma - Anterior Segment Dysgenesis - UGent
- 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
- Pediatric oncopredisposition - UGent
- Skin disorders - UGent
- Sturge-Weber syndrome (65 genes) - KUL

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