

**ANALYTE:  
JAG1**

|                             |                                                                                                                           |
|-----------------------------|---------------------------------------------------------------------------------------------------------------------------|
| <b>NAME:</b>                | jagged canonical Notch ligand 1                                                                                           |
| <b>SYMBOL:</b>              | JAG1                                                                                                                      |
| <b>VERSION OF ORPHANET:</b> | 2023-06-22 14:14:43                                                                                                       |
| <b>SYNONYMS:</b>            | AHD<br>AWS<br>CD339<br>HJ1                                                                                                |
| <b>XREF(S):</b>             | <u>Orphanet</u><br><u>Ensembl</u><br><u>Genatlas</u><br><u>HGNC</u><br><u>OMIM</u><br><u>Reactome</u><br><u>SwissProt</u> |
| <b>CREATED:</b>             | 13 May 2019 - 01:01                                                                                                       |
| <b>CHANGED:</b>             | 22 Jun 2023 - 16:14                                                                                                       |

## RELATED CONTENT

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### Related Genetic Tests

- [Alagille syndrome \(2 genes\)](#)
- [Aneurysm, Thoracic Aortic, familial \(gene panel\)](#)
- [Cholestasis \(gene panel\)](#)
- [Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophthisis / Bardet-Biedl syndromes and kidney cancers \(gene panel\)](#)
- [Congenital malformation \(gene panel - 1721 genes\)](#)
- [Congenital malformation gene panel](#)
- [Congenital structural heart defects \(gene panel\)](#)
- [End-stage renal disease, ESRD \(gene panel\)](#)
- [Heart / Cardio disorders / Cardiopathy \(gene panel\)](#)
- [Hepatology \(gene panel\)](#)
- [Inherited Kidney Diseases \(Gene Panel\)](#)
- [Intellectual disability & Epilepsy \(gene panel\)](#)
- [Intellectual disability \(virtual gene panel\)](#)
- [Nephrogenetics / Nephropathy \(gene panel\)](#)
- [Nephropathies, hereditary \(gene panel\)](#)
- [Neurodevelopmental disorders \(1300 genes\)](#)
- [Neurodevelopmental disorders gene panel](#)
- [Neuropathy \(gene panel\)](#)
- [Peripheral neuropathy \(gene panel\)](#)
- [Primary ciliary dyskinesia \(PCD\) Heterotaxyies \(gene panel\)](#)
- [Renal or urinary tract malformation \(CAKUT\) \(gene panel\)](#)
- [Retinal dystrophy / RETNET \(gene panel\)](#)
- [Skeletal dysplasia \(gene panel\)](#)
- [Stroke \(gene panel\)](#)
- [Thyroid dysgenesis \(38 genes\)](#)

## Related Diseases

- [Alagille syndrome due to 20p12 microdeletion](#)
- [Alagille syndrome due to a JAG1 point mutation](#)
- [Tetralogy of Fallot](#)

## Related Gene Panels

- [Alagille \(2 genes\) -UCL](#)
- [Alagille syndrome \(2 genes\) - UCL](#)
- [Cakut \(congenital anomalies of the kidney and urinary tract-1\) \(69 genes\) - IPG](#)
- [Cholestasis \(40 genes\) - UCL](#)
- [Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophthisis, Bardet-Biedl syndromes and kidney cancers \(146 genes\) - IPG](#)
- [Congenital heart disease \(29 genes\) - VUB](#)
- [Congenital malformation \(1721 genes\) - ULB](#)
- [Congenital malformation gene panel - VUB](#)
- [Congenital structural heart defects - UGent](#)
- [End-stage renal disease \(106 genes\) - IPG](#)
- [Familial Thoracic Aortic Aneurysm \(genepanel\) - UZA](#)
- [Hepatology panel - UGent](#)
- [Heterotaxie PCD - UGent](#)
- [Hypogonadotropic Hypogonadism/Kallmann \(61 genes\) - ULG](#)
- [Intellectual disability & Epilepsy - UGent](#)
- [Intellectual disability \(gene panel\)](#)
- [Nephropathies, hereditary \(219 genes\) - KUL](#)
- [Nephropathy panel - UGent](#)
- [Neurodevelopmental disorders \(1300 genes\) - ULB](#)
- [Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders \(1162 genes\) - VUB](#)
- [Neuropathy \(genepanel\) - UZA](#)
- [Neuropathy panel - UGent](#)
- [Panel Nephro-ULG-V1](#)

- Retinal dystrophy - UGent
- Skeletal dysplasia (394 genes) - VUB
- Stroke - UGent
- Thyroid disgenesis (38 genes) - VUB
- cardiopathy panel - UGent

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