

**GENETIC TEST:****Disorders of sex development - Primary Ovarian insufficiency - Hypogonadotropic Hypogonadism (gene panel)**

|                          |                                                                                                           |
|--------------------------|-----------------------------------------------------------------------------------------------------------|
| <b>FULL NAME:</b>        | Disorders of sex development - Primary Ovarian insufficiency - Hypogonadotropic Hypogonadism (gene panel) |
| <b>DESCRIPTION:</b>      | DSD-POI-HH gene panel                                                                                     |
| <b>TEST TYPE:</b>        | Clinical                                                                                                  |
| <b>TEST SPECIALTY:</b>   | Molecular Genetics                                                                                        |
| <b>TEST PURPOSE:</b>     | Post-natal Diagnosis                                                                                      |
| <b>SPECIMEN:</b>         | Peripheral (whole) blood on EDTA                                                                          |
| <b>METHOD CATEGORY:</b>  | Sequence analysis: entire coding region<br>Whole Exome Sequencing (WES)<br>Deletion/duplication analysis  |
| <b>METHOD TECHNIQUE:</b> | Next Generation Sequencing (NGS)                                                                          |
| <b>RIZIV CODE:</b>       | 565493-565504                                                                                             |

|                                   |                                                                                |
|-----------------------------------|--------------------------------------------------------------------------------|
| <b>EQA:</b>                       | <ul style="list-style-type: none"> <li>Disorders of Sex Development</li> </ul> |
| <b>TURNAROUND TIME (MAXIMUM):</b> | 6 months                                                                       |
| <b>CREATED:</b>                   | 29 Jul 2019 - 17:52                                                            |
| <b>CHANGED:</b>                   | 31 Jan 2023 - 13:56                                                            |

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## RELATED CONTENT

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### Related Laboratories

- [Centrum Medische Genetica - UZ Gent](#)

### Related Analytes

- [alanyl-tRNA synthetase 2, mitochondrial](#)
- [aldo-keto reductase family 1 member C2](#)
- [aldo-keto reductase family 1 member C4](#)
- [anti-Mullerian hormone](#)
- [anti-Mullerian hormone receptor type 2](#)
- [anosmin 1](#)
- [androgen receptor](#)
- [aristaless related homeobox](#)
- [ATRX chromatin remodeler](#)
- [AXL receptor tyrosine kinase](#)
- [bone morphogenetic protein 15](#)
- [bone morphogenetic protein 4](#)
- [basoenuclin zinc finger protein 1](#)
- [chromobox 2](#)
- [chromodomain helicase DNA binding protein 7](#)
- [caseinolytic mitochondrial matrix peptidase proteolytic subunit](#)
- [CREB binding protein](#)
- [cytochrome b5 type A](#)
- [cytochrome P450 family 11 subfamily A member 1](#)
- [cytochrome P450 family 11 subfamily B member 1](#)

- cytochrome P450 family 17 subfamily A member 1
- cytochrome P450 family 19 subfamily A member 1
- cytochrome P450 family 21 subfamily A member 2
- 7-dehydrocholesterol reductase
- desert hedgehog signaling molecule
- DEAH-box helicase 37
- doublesex and mab-3 related transcription factor 1
- Dmx like 2
- dual specificity phosphatase 6
- eukaryotic translation initiation factor 4E nuclear import factor 1
- E1A binding protein p300
- estrogen receptor 1
- estrogen receptor 2
- FA complementation group M
- FEZ family zinc finger 1
- fibroblast growth factor 17
- fibroblast growth factor 8
- fibroblast growth factor 9
- fibroblast growth factor receptor 1
- fibroblast growth factor receptor 2
- folliculogenesis specific bHLH transcription factor
- fibronectin leucine rich transmembrane protein 3
- forkhead box L2
- follicle stimulating hormone subunit beta
- follicle stimulating hormone receptor
- GATA binding protein 4
- growth differentiation factor 9
- gonadotropin releasing hormone 1
- gonadotropin releasing hormone receptor
- guanylate cyclase 2C
- histone deacetylase 8
- HESX homeobox 1
- helicase for meiosis 1

- hedgehog acyltransferase
- homeobox A13
- heparan sulfate 6-O-sulfotransferase 1
- hydroxysteroid 17-beta dehydrogenase 3
- hydroxysteroid 17-beta dehydrogenase 4
- hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 2
- interleukin 17 receptor D
- insulin like 3
- KH RNA binding domain containing, signal transduction associated 1
- KiSS-1 metastasis suppressor
- KISS1 receptor
- leucyl-tRNA synthetase 2, mitochondrial
- leptin
- leptin receptor
- luteinizing hormone subunit beta
- luteinizing hormone/choriogonadotropin receptor
- LIM homeobox 1
- LIM homeobox 3
- LIM homeobox 4
- mastermind like domain containing 1
- mitogen-activated protein kinase kinase kinase 1
- minichromosome maintenance 8 homologous recombination repair factor
- minichromosome maintenance 9 homologous recombination repair factor
- meiosis specific with OB-fold
- midline 1
- mitochondrial ribosomal protein S22
- mutS homolog 4
- mutS homolog 5
- nanos homolog 3
- nibrin
- NOBOX oogenesis homeobox
- nuclear receptor subfamily 0 group B member 1
- nuclear receptor subfamily 2 group F member 2

- nuclear receptor subfamily 5 group A member 1
- NMDA receptor synaptonuclear signaling and neuronal migration factor
- nucleoporin 107
- OTU deubiquitinase 4
- PAT1 homolog 2
- proprotein convertase subtilisin/kexin type 1
- patatin like phospholipase domain containing 6
- POF1B actin binding protein
- RNA polymerase II subunit C
- RNA polymerase III subunit A
- porcupine O-acyltransferase
- prokineticin 2
- prokineticin receptor 2
- PROP paired-like homeobox 1
- PSMC3 interacting protein
- ring finger protein 216
- R-spondin 1
- relaxin family peptide receptor 2
- semaphorin 3A
- semaphorin 7A (John Milton Hagen blood group)
- spermatogenesis and oogenesis specific basic helix-loop-helix 1
- spermatogenesis and oogenesis specific basic helix-loop-helix 2
- SRY-box transcription factor 10
- SRY-box transcription factor 2
- SRY-box transcription factor 3
- SRY-box transcription factor 8
- SRY-box transcription factor 9
- scaffold protein involved in DNA repair
- sprouty RTK signaling antagonist 4
- steroid 5 alpha-reductase 2
- sex determining region Y
- STAG3 cohesin complex component
- synaptonemal complex central element protein 1

- [tachykinin precursor 3](#)
- [tachykinin receptor 3](#)
- [tumor protein p63](#)
- [TSPY like 1](#)
- [twinkle mtDNA helicase](#)
- [WD repeat domain 11](#)
- [Wnt family member 4](#)
- [WT1 transcription factor](#)
- [WW domain containing oxidoreductase](#)
- [zinc finger protein, FOG family member 2](#)
- [zinc and ring finger 3](#)

## Related Gene Panels

- [Disorders of Sex Development - Primary Ovarian Insufficiency - Hypogonadotropic Hypogonadism - UGent](#)

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