

Full name:	Disorders of Sex Development - Primary Ovarian Insufficiency - Hypogonadotropic Hypogonadism - UGent
Abbreviation:	DSD-POI-HH
Type of panel:	<u>in-house</u>
Version number:	v5
Laboratory:	<u>Centrum Medische Genetica - UZ Gent</u>
Created:	13 Jun 2019 - 14:12
Changed:	16 Dec 2022 - 10:59

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>AARS2</u>	99.98	1	
<u>AKR1C2</u>	91.19	1	
<u>AKR1C4</u>	100.00	1	
<u>AMH</u>	100.00	1	
<u>AMHR2</u>	99.96	1	
<u>ANOS1</u>	99.96	1	
<u>AR</u>	99.76	1	
<u>ARX</u>	95.36	1	
<u>ATRX</u>	99.44	1	
<u>AXL</u>	99.97	1	
<u>BMP15</u>	99.98	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>BMP4</u>	100.00	1	
<u>BNC1</u>	100.00	1	
<u>CBX2</u>	100.00	1	
<u>CHD7</u>	99.99	1	
<u>CLPP</u>	99.99	1	
<u>CREBBP</u>	99.97	1	
<u>CYB5A</u>	99.98	1	
<u>CYP11A1</u>	99.99	1	
<u>CYP11B1</u>	100.00	1	
<u>CYP17A1</u>	100.00	1	
<u>CYP19A1</u>	99.99	1	
<u>CYP21A2</u>	99.85	1	
<u>DHCR7</u>	99.97	1	
<u>DHH</u>	100.00	1	
<u>DHX37</u>	99.98	1	
<u>DMRT1</u>	99.99	1	
<u>DMXL2</u>	99.86	1	
<u>DUSP6</u>	99.99	1	
<u>EIF4ENIF1</u>	100.00	1	
<u>EP300</u>	99.97	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ESR1</u>	100.00	1	
<u>ESR2</u>	99.96	1	
<u>FANCM</u>	99.83	1	
<u>FEZF1</u>	100.00	1	
<u>FGF17</u>	99.81	1	
<u>FGF8</u>	100.00	1	
<u>FGF9</u>	100.00	1	
<u>FGFR1</u>	100.00	1	
<u>FGFR2</u>	99.99	1	
<u>FIGLA</u>	99.95	1	
<u>FLRT3</u>	100.00	1	
<u>FOXL2</u>	99.97	1	
<u>FSHB</u>	100.00	1	
<u>FSHR</u>	99.99	1	
<u>GATA4</u>	99.99	1	
<u>GDF9</u>	99.98	1	
<u>GNRH1</u>	99.96	1	
<u>GNRHR</u>	99.94	1	
<u>GUCY2C</u>	99.97	1	
<u>HDAC8</u>	99.74	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>HESX1</u>	99.77	1	
<u>HFM1</u>	89.74	1	
<u>HHAT</u>	99.93	1	
<u>HOXA13</u>	99.94	1	
<u>HS6ST1</u>	99.99	1	
<u>HSD17B3</u>	99.77	1	
<u>HSD17B4</u>	99.71	1	
<u>HSD3B2</u>	100.00	1	
<u>IL17RD</u>	99.94	1	
<u>INSL3</u>	98.02	1	
<u>KHDRBS1</u>	99.35	1	
<u>KISS1</u>	99.97	1	
<u>KISS1R</u>	100.00	1	
<u>LARS2</u>	99.96	1	
<u>LEP</u>	100.00	1	
<u>LEPR</u>	92.52	1	
<u>LHB</u>	99.93	1	
<u>LHCGR</u>	99.94	1	
<u>LHX1</u>	99.96	1	
<u>LHX3</u>	99.99	1	

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<u>LHX4</u>	99.99	1	
<u>MAMLD1</u>	99.84	1	
<u>MAP3K1</u>	99.93	1	
<u>MCM8</u>	98.78	1	
<u>MCM9</u>	99.76	1	
<u>MEIOB</u>	99.98	1	
<u>MID1</u>	99.90	1	
<u>MRPS22</u>	99.87	1	
<u>MSH4</u>	89.29	1	
<u>MSH5</u>	100.00	1	
<u>NANOS3</u>	100.00	1	
<u>NBN</u>	99.93	1	
<u>NOBOX</u>	99.95	1	
<u>NR0B1</u>	99.98	1	
<u>NR2F2</u>	100.00	1	
<u>NR5A1</u>	99.92	1	
<u>NSMF</u>	100.00	1	
<u>NUP107</u>	97.46	1	
<u>OTUD4</u>	99.83	1	
<u>PATL2</u>	99.99	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PCSK1</u>	99.99	1	
<u>PNPLA6</u>	99.99	1	
<u>POF1B</u>	99.34	1	
<u>POLR2C</u>	99.99	1	
<u>POLR3A</u>	99.97	1	
<u>PORCN</u>	99.98	1	
<u>PROK2</u>	98.65	1	
<u>PROKR2</u>	100.00	1	
<u>PROP1</u>	99.88	1	
<u>PSMC3IP</u>	99.82	1	
<u>RNF216</u>	99.99	1	
<u>RSPO1</u>	99.99	1	
<u>RXFP2</u>	99.99	1	
<u>SEMA3A</u>	99.42	1	
<u>SEMA7A</u>	99.90	1	
<u>SOHLH1</u>	100.00	1	
<u>SOHLH2</u>	100.00	1	
<u>SOX10</u>	100.00	1	
<u>SOX2</u>	100.00	1	
<u>SOX3</u>	100.00	1	

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<u>SOX8</u>	99.99	1	
<u>SOX9</u>	100.00	1	
<u>SPIDR</u>	99.96	1	
<u>SPRY4</u>	100.00	1	
<u>SRD5A2</u>	100.00	1	
<u>SRY</u>	51.98	1	
<u>STAG3</u>	98.99	1	
<u>SYCE1</u>	99.99	1	
<u>TAC3</u>	99.94	1	
<u>TACR3</u>	99.99	1	
<u>TP63</u>	99.97	1	
<u>TSPYL1</u>	100.00	1	
<u>TWNK</u>	100.00	1	
<u>WDR11</u>	99.91	1	
<u>WNT4</u>	99.94	1	
<u>WT1</u>	99.99	1	
<u>WWOX</u>	100.00	1	
<u>ZFPM2</u>	100.00	1	
<u>ZNRF3</u>	99.94	1	