Whole Exome Sequencing
Gene package Disorders of Sex Development (DSD), version 10.2, 25-2-2022

Technical information
DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number variant detection is performed using the BAM multiscale reference method using depth of coverage analysis and dynamical bins in NexusClinical. The detected copy number variants are annotated and filtered with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
AKR1C2	600450	100	100	100	100
AKR1C4	600451	100	100	100	100
AMH	600957	100	100	96.21	76.24
AMHR2	600956	100	100	99.74	94.02
ANOS1	300836	98.60	95.69	92.18	90.22
AR	313700	100	98.66	93.77	83.73
ARX	300382	84.88	79.12	68.56	50.95
ATRX	300032	100	100	99.00	93.02
BMP15	300247	100	100	100	100
BMP4	112262	100	100	99.10	95.74
CBX2	602770	94.88	80.58	78.12	78.12
CCNQ	300708	81.09	81.09	81.09	81.09
CDKN1C	600856	84.56	72.52	60.78	51.01
CHD7	608892	100	100	99.89	98.69
CREBBP	600140	100	99.21	97.19	91.73
CYB5A	613218	100	100	100	98.61
CYP11A1	118485	100	100	100	97.30
CYP11B1	610613	100	100	100	100
CYP17A1	609300	100	100	100	100
CYP19A1	107910	100	100	97.87	90.17

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
CYP21A2	613815	100	98.84	88.37	70.89
DAZL	601486	94.98	86.01	84.33	72.27
DHCR7	602858	100	100	99.42	94.57
DHH	605423	100	100	100	98.16
DHX37	617362	100	98.74	94.47	79.88
DMRT1	602424	100	100	100	100
DMRT2	604935	100	100	94.44	59.82
DUSP6	602748	100	100	100	100
ESR1	133430	100	100	99.87	96.57
ESR2	601663	100	100	100	99.80
FEZF1	613301	100	100	100	100
FGF17	603725	100	100	100	96.12
FGF8	600483	93.90	92.43	87.21	66.49
FGFR1	136350	100	100	100	97.93
FGFR2	176943	100	100	99.68	98.15
FOXL2	605597	100	94.43	90.64	80.31
FSHB	136530	100	100	100	100
FSHR	136435	100	100	100	100
GATA4	600576	100	100	100	94.74
GNRH1	152760	100	100	100	98.85
GNRHR	138850	100	100	100	100
HDAC8	300269	100	100	99.68	90.30
HFM1	615684	98.14	95.14	93.68	82.66
HOXA13	142959	83.26	77.66	74.25	68.15
HS6ST1	604846	100	100	100	97.53
HSD17B3	605573	100	100	100	100
HSD3B2	613890	100	100	100	100
IL17RD	606807	100	100	100	99.43
INSL3	146738	100	100	98.49	78.34
KISS1	603286	100	100	99.78	68.06
LHB	152780	100	100	100	87.37
LHCGR	152790	100	100	99.44	96.83
MAMLD1	300120	100	100	100	99.67
MAP3K1	600982	100	98.34	94.70	89.36
MCM8	608187	100	100	98.80	88.87
MCM9	610098	88.54	88.54	88.54	88.54
MID1	300552	100	100	100	97.46
MYRF	608329	99.79	97.35	93.93	87.17
NR0B1	300473	100	100	100	95.72
NR2F2	107773	100	100	100	98.23
NR5A1	184757	100	100	100	98.37

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
NSMF	608137	100	99.21	95.34	83.30
NUP107	607617	100	100	100	95.86
PBX1	176310	100	99.08	92.76	80.44
POLR3A	614258	100	100	100	99.12
POLR3H	No ID	100	100	100	93.37
POR	124015	96.93	96.93	96.93	96.64
PPP1R12A	602021	100	99.11	96.27	90.75
PPP2R3C	615902	100	100	100	97.22
PROK2	607002	100	94.11	82.98	75.16
PROKR2	607123	100	100	100	100
PSMC3IP	608665	100	100	100	97.10
RNF216	609948	100	100	100	99.64
RSPO1	609595	100	99.27	91.68	71.32
RXFP2	606655	100	100	100	97.39
SAMD9	610456	100	100	100	99.87
SEMA3A	603961	100	100	100	99.28
SOX3	313430	100	100	94.29	73.05
SOX8	605923	100	99.57	95.35	71.96
SOX9	608160	100	97.10	94.90	86.77
SPRY4	607984	100	100	100	100
SRD5A2	607306	100	98.86	93.23	67.01
SRY	480000	0	0	0	0
STAG3	608489	100	100	99.59	94.65
STAR	600617	100	100	100	98.64
SYCE1	611486	100	100	100	90.40
TAC3	162330	100	100	100	100
TSPYL1	604714	100	100	100	99.21
WDR11	606417	100	100	100	99.32
WNT4	603490	97.40	91.59	90.72	86.60
WT1	607102	100	100	100	100
ZFPM2	603693	100	100	99.70	95.45
ZNRF3	612062	96.49	95.21	93.99	90.09

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 104 samples
- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x