Whole Exome Sequencing
Gene package Disorders of Sex Development (DSD), version 10, 26-2-2021

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy 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 filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). It is not excluded that pathogenic variants are being missed using this technology. 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	99.72	93.60	69.58
AMHR2	600956	100	100	99.36	92.55
ANOS1	300836	98.53	95	90.22	89.07
AR	313700	100	97.26	91.15	84.26
ARX	300382	85.61	80.04	71.89	58.82
ATRX	300032	100	98.81	94.83	79.42
BMP15	300247	100	100	100	98.60
BMP4	112262	100	100	99.31	95.95
CBX2	602770	96.35	82.96	78.12	77.91
CCNQ	300708	81.09	81.09	81.09	81.09
CDKN1C	600856	84.21	69.74	58.81	48.73
CHD7	608892	100	99.87	98.61	96.80
CREBBP	600140	99.64	98.59	96.38	88.55
CYB5A	613218	100	100	100	89.04
CYP11A1	118485	100	100	99.83	95.70
CYP11B1	610613	100	100	100	100
CYP17A1	609300	100	100	100	97.95
CYP19A1	107910	100	100	97.16	89.34

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CYP21A2	613815	100	98.01	87.09	68.69
DAZL	601486	88.41	85.88	80.94	69.14
DHCR7	602858	100	100	98.66	92.36
DHH	605423	100	100	100	98.56
DHX37	617362	100	98.44	92.84	73.83
DMRT1	602424	100	100	100	100
DMRT2	604935	100	90.51	86.52	39.57
DUSP6	602748	100	100	100	100
ESR1	133430	100	100	99.49	94.13
ESR2	601663	100	100	100	98.54
FEZF1	613301	100	100	100	98.97
FGF17	603725	100	100	100	97.13
FGF8	600483	93.90	88.15	87.21	62.09
FGFR1	136350	100	100	99.61	96.92
FGFR2	176943	100	100	99.54	97.18
FOXL2	605597	100	94.21	89.07	76.92
FSHB	136530	100	100	100	100
FSHR	136435	100	100	100	100
GATA4	600576	100	100	100	90.59
GNRH1	152760	100	100	100	95.98
GNRHR	138850	100	100	100	98.64
HDAC8	300269	100	100	97.79	84.00
HFM1	615684	96.63	93.89	90.14	77.14
HOXA13	142959	83.43	77.66	73.38	66.03
HS6ST1	604846	100	100	100	91.01
HSD17B3	605573	100	100	100	97.96
HSD3B2	613890	100	100	100	100
IL17RD	606807	100	100	99.92	95.62
INSL3	146738	100	100	98.31	78.34
KISS1	603286	100	100	93.50	66.08
LHB	152780	100	100	98.34	83.54
LHCGR	152790	100	99.61	98.25	88.95
MAMLD1	300120	100	100	100	97.35
MAP3K1	600982	99.72	96.77	93.15	86.89
MCM8	608187	100	99.07	94.57	83.25
MCM9	610098	100	100	100	99.47
MID1	300552	100	100	100	92.27
MYRF	608329	99.89	97.04	93.44	86.71
NR0B1	300473	100	100	99.41	95.90
NR2F2	107773	100	100	100	97.12
NR5A1	184757	100	100	100	95.38

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NSMF	608137	100	99.48	95.24	76.13
NUP107	607617	100	100	99.67	92.10
PBX1	176310	100	98.10	90.92	78.16
POLR3A	614258	100	100	99.87	98.36
POLR3H	No ID	100	100	99.86	89.34
POR	124015	100	100	100	98.25
PPP1R12A	602021	100	97.08	94.11	88.30
PPP2R3C	615902	100	100	99.35	93.45
PROK2	607002	100	91.97	79.01	75.16
PROKR2	607123	100	100	100	100
PSMC3IP	608665	100	100	99.08	91.49
RNF216	609948	100	100	99.84	97.23
RSPO1	609595	100	98.99	90.89	70.19
RXFP2	606655	100	99.92	98.61	92.93
SAMD9	610456	100	100	100	99.46
SEMA3A	603961	100	100	99.79	96.97
SOX3	313430	100	99.89	95.47	76.99
SOX8	605923	100	99.00	92.31	68.06
SOX9	608160	100	97.67	94.80	84.47
SPRY4	607984	100	100	100	99.13
SRD5A2	607306	100	100	92.83	63.94
SRY	480000	No coverage data			
STAG3	608489	100	100	99.04	92.01
STAR	600617	100	100	100	97.19
SYCE1	611486	100	100	100	88.92
TAC3	162330	100	100	100	99.14
TSPYL1	604714	100	100	100	98.05
WDR11	606417	100	100	100	97.65
WNT4	603490	97.70	91.59	90.29	85.86
WT1	607102	100	100	100	100
ZFPM2	603693	100	100	98.33	91.93
ZNRF3	612062	96.32	95.00	93.44	88.49

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 100 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