Whole Exome Sequencing
Gene package Disorders of Sex Development (DSD), version 9, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + 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/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. It is not excluded that pathogenic mutations 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AKR1C2	600450	131	91	91	91
AKR1C4	600451	127	100	100	100
AMH	600957	179	100	100	100
AMHR2	600956	209	100	100	100
ANOS1	300836	85	100	98	94
AR	313700	134	100	99	96
ARX	300382	100	92	89	84
ATRX	300032	37	92	74	52
BMP15	300247	111	100	100	100
BMP4	112262	224	100	100	100
CBX2	602770	220	100	99	97
CCNQ	300708	126	81	81	81
CDKN1C	600856	156	92	87	83
CHD7	608892	167	100	99	97
CREBBP	600140	275	99	99	98
CYB5A	613218	109	100	95	89
CYP11A1	118485	205	100	100	100
CYP11B1	610613	301	100	100	100
CYP17A1	609300	201	100	97	94
CYP19A1	107910	101	100	100	98
CYP21A2	613815	193	100	100	100
DAZL	601486	38	82	65	52
DHCR7	602858	243	100	100	100
DHH	605423	335	100	100	100
DHX37	617362	201	100	100	99
DMRT1	602424	169	100	100	100
DMRT2	604935	137	100	99	98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
DUSP6	602748	271	100	100	100
ESR1	133430	181	100	100	100
FEZF1	613301	262	100	100	100
FGF17	603725	222	100	100	100
FGF8	600483	142	100	97	94
FGFR1	136350	185	100	100	100
FGFR2	176943	145	100	100	99
FOXL2	605597	274	100	100	100
FSHB	136530	111	100	100	98
FSHR	136435	148	100	100	100
GATA4	600576	164	100	100	100
GNRH1	152760	107	100	100	100
GNRHR	138850	96	100	100	96
HDAC8	300269	73	100	100	95
HFM1	615684	34	79	58	43
HOXA13	142959	221	91	85	82
HS6ST1	604846	330	100	100	100
HSD17B3	605573	117	100	98	89
HSD3B2	613890	170	100	100	100
IL17RD	606807	160	100	100	99
INSL3	146738	195	100	100	100
KISS1	603286	162	100	100	100
LHB	152780	161	100	100	100
LHCGR	152790	93	99	94	87
MAMLD1	300120	125	100	100	100
MAP3K1	600982	97	99	98	95
MCM8	608187	54	97	86	72
MCM9	610098	121	100	100	99
MID1	300552	112	100	100	97
NR0B1	300473	193	100	100	100
NR2F2	107773	270	100	100	98
NR5A1	184757	293	100	100	100
NSMF	608137	215	100	100	100
NUP107	607617	55	97	91	79
POLR3A	614258	143	100	100	99
POLR3H	No ID	183	100	100	100
POR	124015	296	100	100	100
PPP2R3C	615902	58	97	83	64
PROK2	607002	84	100	100	100
PROKR2	607123	340	100	100	100
PSMC3IP	608665	107	100	99	95
RNF216	609948	149	100	98	97
RSPO1	609595	204	100	100	100
RXFP2	606655	72	93	86	78
SEMA3A	603961	102	100	99	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
SOX3	313430	155	100	100	100
SOX8	605923	169	100	100	100
SOX9	608160	207	100	100	99
SPRY4	607984	277	100	100	100
SRD5A2	607306	114	100	100	100
SRY	480000	382	100	100	100
STAG3	608489	143	100	100	99
STAR	600617	186	100	100	100
SYCE1	611486	159	100	100	100
TAC3	162330	133	100	100	100
TSPYL1	604714	185	100	100	100
WDR11	606417	87	99	96	93
WNT4	603490	289	100	92	92
WT1	607102	175	100	100	100
ZFPM2	603693	127	100	100	99
ZNRF3	612062	198	97	96	96

- OMIM release used: 8-9-2019
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x