Whole Exome Sequencing
Gene package Hereditary Congenital Defects, version 5.2, 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
Oesophagus atresia					
CHD7	608892	167	100	99	97
EFTUD2	603892	124	100	100	100
GLI3	165240	243	100	100	100
MID1	300552	112	100	100	97
MYCN	164840	112	100	100	97
SOX2	184429	257	100	100	100
Congenital Hernia Diaphragmatica					
GATA4	600576	164	100	100	100
GPC3	300037	86	100	98	92
LRP2	600073	118	100	99	97
MYRF	608329	219	100	100	99
NIPBL	608667	93	98	90	82
SMC3	606062	46	93	80	60
STRA6	610745	229	100	100	100
WT1	607102	175	100	100	100
ZFPM2	603693	127	100	100	99
Ano-rectal Marformation					
CASK	300172	60	96	86	70
CCNB1	123836	94	100	100	100
CCNQ	300708	126	81	81	81
FLNA	300017	206	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GLI3	165240	243	100	100	100
JAG1	601920	213	100	100	100
KDM6A	300128	57	95	86	73
KMT2D	602113	341	100	100	100
MED12	300188	99	100	100	98
MID1	300552	112	100	100	97
MNX1	142994	107	84	80	77
MYCN	164840	112	100	100	97
NOTCH2	600275	188	100	98	97
SALL1	602218	214	100	100	100
SALL4	607343	229	100	100	100
USP9X	300072	45	96	86	71
HSCR/CIPO/MMIHS					
AAAS	605378	175	100	100	100
ACTG2	102545	169	100	100	100
EDN3	131242	187	100	100	100
EDNRB	131244	135	100	99	94
FLNA	300017	206	100	100	100
KIFBP	609367	117	100	100	100
LMOD1	602715	134	100	100	100
MYH11	160745	171	100	100	99
MYL9	609905	264	100	100	99
MYLK	600922	195	100	100	100
NOS1	163731	201	100	100	100
PHOX2B	603851	140	100	95	89
RAD21	606462	63	100	98	93
RET	164761	217	100	100	100
SGO1	609168	55	96	90	82
SOX10	602229	381	100	100	100
VCL	193065	137	100	99	96
ZEB2	605802	289	100	100	100

- 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