

Whole Exome Sequencing

Gene package Craniosynostosis, version 3.1, 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



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ABCC9	601439	77	100	99	94
ADAMTSL4	610113	244	100	100	100
ALPL	171760	323	100	100	100
ALX3	606014	202	100	98	96
ALX4	605420	206	100	99	97
AXIN2	604025	193	100	100	100
BCL11B	606558	242	100	100	99
BPNT2	614010	192	100	100	100
CCBE1	612753	163	95	95	95
CDC45	603465	172	100	100	100
CHST3	603799	345	100	100	100
COLEC11	612502	234	100	100	100
CTSK	601105	108	100	100	100
CYP26B1	605207	414	100	100	100
EFNA4	601380	253	100	100	100
EFNB1	300035	143	100	100	100
ERF	611888	220	100	100	100
ESCO2	609353	88	97	89	81
FGF3	164950	219	100	100	100
FGF9	600921	122	100	100	95
FGFR1	136350	185	100	100	100
FGFR2	176943	145	100	100	99
FGFR3	134934	241	100	100	100
FLNA	300017	206	100	100	100
FLNB	603381	193	100	100	99
FREM1	608944	123	100	99	96
GLI3	165240	243	100	100	100
GNAS	139320	269	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GNPTAB	607840	87	100	99	95
GPC3	300037	86	100	98	92
GPC4	300168	93	100	99	97
GUSB	611499	222	100	100	100
HRAS	190020	426	100	100	100
HUWE1	300697	90	100	98	93
IFT122	606045	141	100	100	98
IFT43	614068	133	100	100	100
IGF1R	147370	176	100	100	100
IHH	600726	276	100	100	100
IL11	147681	155	100	99	95
IL11RA	600939	193	100	100	100
IRX5	606195	211	100	100	100
JAG1	601920	213	100	100	100
KDM6A	300128	57	95	86	73
KMT2D	602113	341	100	100	100
KPTN	615620	242	100	100	100
KRAS	190070	59	90	78	77
LRP5	603506	325	98	98	98
MASP1	600521	195	100	100	100
MCPH1	607117	114	87	84	81
MEGF8	604267	242	100	100	100
MSX2	123101	170	100	100	100
NFIX	164005	273	100	100	100
NOTCH2	600275	188	100	98	97
OSTM1	607649	118	93	70	64
P4HB	176790	174	100	100	100
PHEX	300550	69	100	98	87
POLR1A	616404	161	100	100	100
POLR1C	610060	136	100	100	100
POLR1D	613715	123	100	100	100
POR	124015	296	100	100	100
RAB23	606144	64	100	88	85
RECQL4	603780	312	100	100	100
RUNX2	600211	160	100	100	100
SALL1	602218	214	100	100	100
SCARF2	613619	234	100	99	97
SEC24D	607186	96	100	99	96
SH3PXD2B	613293	196	100	100	100
SKI	164780	208	100	100	98
SMAD6	602931	188	100	100	100
SOX10	602229	381	100	100	100
SOX6	607257	118	96	96	95
STAT3	102582	135	100	100	99
TCF12	600480	94	100	99	96
TCOF1	606847	201	100	100	98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TLK2	608439	83	98	90	80
TWIST1	601622	187	100	100	98
WDR19	608151	75	100	96	92
WDR35	613602	75	97	93	87
ZIC1	600470	322	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 (± 10 bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (± 10 bp flanking introns) that is covered at least 10x, 20x or 30x