Whole Exome Sequencing
Gene package Craniosynostosis, version 6, 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ABCC9	601439	100	100	99.94	98.26
ADAMTSL4	610113	100	99.01	98.00	91.41
ALPL	171760	100	100	99.83	98.30
ALX3	606014	92.43	80.12	73.22	73.13
ALX4	605420	100	97.18	93.11	77.84
ASXL1	612990	98.42	98.42	98.42	97.19
AXIN2	604025	100	100	99.63	97.62
BCL11B	606558	100	99.02	96.99	90.41
BPNT2	614010	100	100	100	98.64
CCBE1	612753	100	100	100	98.23
CDC45	603465	100	100	100	98.02
CHST3	603799	100	100	100	100
COLEC11	612502	100	100	97.89	89.70
CTSK	601105	100	100	100	98.94
CYP26B1	605207	100	100	99.88	94.75
EFNA4	601380	100	100	97.86	89.23
EFNB1	300035	100	100	99.87	88.27
ERF	611888	100	100	98.23	86.34
ESCO2	609353	100	100	100	100
FGF3	164950	100	98.33	94.59	84.43
FGF9	600921	100	100	100	96.35

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
FGFR1	136350	100	100	100	97.93
FGFR2	176943	100	100	99.68	98.15
FGFR3	134934	100	99.60	98.55	90.61
FLNA	300017	99.95	99.05	97.51	95.12
FREM1	608944	100	100	100	98.80
GLI3	165240	100	100	100	99.89
GNAS	139320	100	100	100	96.95
GPC3	300037	100	100	99.55	95.55
GPC4	300168	100	100	100	99.51
GUSB	611499	100	100	100	98.65
HRAS	190020	100	100	100	100
HUWE1	300697	100	99.65	98.90	93.92
IFT122	606045	100	100	99.77	95.61
IFT43	614068	100	100	100	98.69
IGF1R	147370	100	100	99.30	96.66
IHH	600726	100	99.38	96.83	92.88
IL11RA	600939	100	99.37	95.58	79.68
IRX5	606195	100	100	97.45	82.94
JAG1	601920	100	98.67	97.58	97.41
KAT6A	601408	100	100	99.50	97.02
KDM6A	300128	100	100	99.30	92.95
KMT2D	602113	100	100	99.75	97.30
KPTN	615620	100	100	97.45	84.30
KRAS	190070	100	100	100	100
LRP5	603506	97.91	97.91	97.26	93.39
MASP1	600521	100	100	100	98.59
MCPH1	607117	99.21	97.13	95.33	89.69
MEGF8	604267	99.98	98.45	95.05	82.55
MSX2	123101	100	100	97.68	90.43
NOTCH2	600275	99.86	98.85	98.02	95.09
OSTM1	607649	100	97.73	86.41	69.34
P4HB	176790	100	99.66	97.08	83.57
PHEX	300550	100	100	100	99.44
POLR1A	616404	100	100	99.83	95.51
POLR1B	602000	95.91	95.91	95.91	95.55
POLR1C	610060	100	100	100	94.29
POLR1D	613715	100	100	100	90.09
POR	124015	96.93	96.93	96.93	96.64
PRRX1	167420	100	97.91	91.90	76.93
RAB23	606144	100	100	100	100
RECQL4	603780	100	99.95	99.35	94.89

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
RUNX2	600211	100	100	100	100
SALL1	602218	100	100	100	99.07
SCARF2	613619	98.01	91.54	80.70	63.30
SEC24D	607186	100	100	99.79	96.68
SH3PXD2B	613293	100	98.73	96.69	90.06
SIX1	601205	100	100	100	96.52
SKI	164780	97.72	91.07	85.13	73.30
SLC25A24	608744	100	100	100	96.17
SMAD3	603109	100	100	100	99.63
SMAD6	602931	100	87.44	66.07	48.41
SMO	601500	100	97.31	95.12	89.20
SOX10	602229	100	99.66	97.70	90.57
SOX6	607257	100	100	99.91	96.62
SP7	606633	100	100	100	93.02
SPECC1L	614140	100	100	100	100
STAT3	102582	100	100	100	95.99
TCF12	600480	100	100	100	99.58
TCOF1	606847	100	99.96	96.81	86.51
TFAP2B	601601	100	99.21	97.47	90.92
TGFBR1	190181	93.12	93.12	93.12	93.12
TGFBR2	190182	100	100	100	98.71
TLK2	608439	100	100	100	96.23
TWIST1	601622	99.84	88.74	82.75	71.01
WDR19	608151	100	100	99.75	97.05
WDR35	613602	100	99.78	99.23	96.28
ZIC1	600470	100	100	98.79	94.43
ZNF462	617371	100	99.92	99.52	98.00

- OMIM release used: 23-9-2021
- 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