Whole Exome Sequencing
Gene package Craniosynostosis, version 3, 21-2-2020

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/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia 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).

Erasmus MC
University Medical Center Rotterdam

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	65	100	100	95
ADAMTSL4	610113	115	100	100	99
ALPL	171760	130	100	100	100
ALX3	606014	116	98	91	86
ALX4	605420	139	100	100	100
AXIN2	604025	108	100	100	98
BCL11B	606558	106	100	97	92
BPNT2	614010	146	100	100	97
CCBE1	612753	87	100	99	95
CDC45	603465	94	100	99	97
CHST3	603799	129	100	100	100
COLEC11	612502	168	100	100	100
CTSK	601105	65	100	100	98
CYP26B1	605207	159	100	100	100
EFNA4	601380	121	100	100	100
EFNB1	300035	68	100	100	100
ERF	611888	137	100	100	100
ESCO2	609353	58	100	100	95
FGF3	164950	122	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
FGF9	600921	80	100	100	100
FGFR1	136350	102	100	100	98
FGFR2	176943	74	100	100	95
FGFR3	134934	122	100	100	98
FLNA	300017	110	100	100	100
FLNB	603381	110	100	100	99
FREM1	608944	83	100	100	98
GLI3	165240	110	100	100	99
GNAS	139320	180	100	100	97
GNPTAB	607840	63	100	99	95
GPC3	300037	53	100	99	91
GPC4	300168	66	100	98	90
GUSB	611499	106	100	100	97
HRAS	190020	179	100	100	100
HUWE1	300697	53	100	96	83
IFT122	606045	111	100	100	99
IFT43	614068	75	100	100	100
IGF1R	147370	120	100	100	100
IHH	600726	133	100	100	100
IL11	147681	76	100	95	89
IL11RA	600939	103	100	100	100
IRX5	606195	99	100	100	100
JAG1	601920	96	100	100	96
KDM6A	300128	52	100	95	80
KMT2D	602113	114	100	100	99
KPTN	615620	150	100	100	100
KRAS	190070	83	100	100	87
LRP5	603506	146	100	99	98
MASP1	600521	112	100	100	99
MCPH1	607117	85	94	94	92
MEGF8	604267	134	100	99	98
MSX2	123101	82	100	100	98
NFIX	164005	154	100	100	100
NOTCH2	600275	98	100	99	98
OSTM1	607649	79	100	100	97
P4HB	176790	101	100	100	100
PHEX	300550	48	100	98	89

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
POLR1A	616404	100	100	100	98
POLR1C	610060	92	100	100	98
POLR1D	613715	81	100	100	100
POR	124015	170	100	100	100
RAB23	606144	104	100	100	100
RECQL4	603780	148	100	100	99
RUNX2	600211	107	100	100	100
SALL1	602218	120	100	100	99
SCARF2	613619	101	100	99	97
SEC24D	607186	75	100	100	95
SH3PXD2B	613293	122	100	100	99
SKI	164780	123	100	100	99
SMAD6	602931	165	100	97	87
SOX10	602229	72	100	97	89
SOX6	607257	78	100	100	98
STAT3	102582	90	100	100	99
TCF12	600480	72	100	100	98
TCOF1	606847	115	100	100	99
TLK2	608439	55	97	82	66
TWIST1	601622	132	100	100	95
WDR19	608151	71	100	100	97
WDR35	613602	62	100	100	94
ZIC1	600470	255	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