

Whole Exome Sequencing

Gene package Skeletal Dysplasia, version 5, 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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
ABCC9	601439	100	100	99.94	98.26
ACAN	155760	93.76	92.96	92.06	89.17
ACP5	171640	100	100	100	100
ACTB	102630	100	100	100	99.18
ACVR1	102576	100	100	100	98.54
ADAMTS10	608990	100	100	99.84	93.42
ADAMTS17	607511	95.41	90.63	86.38	82.36
ADAMTSL2	612277	44.37	42.14	39.23	32.87
AGA	613228	100	100	100	97.41
AGPS	603051	99.54	95.91	95.16	92.04
ALG12	607144	100	100	100	100
ALG3	608750	100	100	98.86	94.65
ALG9	606941	99.91	99.91	98.98	91.73
ALMS1	606844	99.89	99.40	98.78	96.05
ALPL	171760	100	100	99.83	98.30
AMER1	300647	100	100	100	100
ANKH	605145	100	100	100	99.13
ANKRD11	611192	100	99.03	97.64	94.15
ANO5	608662	100	99.81	98.88	93.14
ARSB	611542	100	100	99.12	89.26
ARSL	300180	100	100	100	92.65
B3GALT6	615291	77.76	74.28	72.00	69.31
B3GAT3	606374	100	100	100	94.34
B4GALT7	604327	100	93.64	93.32	84.20

HGNC approved gene symbol	OMIM gene ID	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
BMP1	112264	100	100	99.73	94.82
BMPER	608699	100	99.34	98.17	93.50
BMPR1B	603248	100	100	100	96.53
BPNT2	614010	100	100	100	98.64
BRAF	164757	95.04	94.58	93.81	88.27
BTK	300300	100	100	100	99.57
CA2	611492	100	100	100	100
CANT1	613165	100	100	100	98.50
CBL	165360	100	100	100	99.03
CCDC134	618788	100	100	100	92.50
CCDC8	614145	100	100	99.20	95.23
CCN6	603400	100	100	99.14	93.80
CDC45	603465	100	100	100	98.02
CDC6	602627	100	100	100	99.32
CDKN1C	600856	84.56	72.52	60.78	51.01
CDT1	605525	99.78	93.23	88.41	76.52
CEP120	613446	100	100	100	97.66
CFAP410	603191	100	100	99.67	86.95
CHST3	603799	100	100	100	100
CLCN5	300008	100	100	99.65	95.35
CLCN7	602727	100	100	99.97	94.84
COG1	606973	100	100	100	97.42
COL10A1	120110	100	100	98.89	95.77
COL11A1	120280	100	99.21	97.82	88.98
COL11A2	120290	100	99.90	95.62	84.23
COL1A1	120150	100	100	98.54	89.55
COL1A2	120160	100	99.02	95.78	84.61
COL2A1	120140	100	100	99.42	93.30
COL9A1	120210	100	100	98.08	86.28
COL9A2	120260	100	96.25	87.33	63.80
COL9A3	120270	100	100	96.30	79.94
COLEC11	612502	100	100	97.89	89.70
COMP	600310	98.23	92.72	92.72	85.65
CREB3L1	616215	99.94	97.86	93.04	79.44
CREBBP	600140	100	99.21	97.19	91.73
CRTAP	605497	100	100	100	99.78
CSGALNACT1	616615	100	100	100	100
CTSA	613111	100	99.85	99.59	92.57
CTSK	601105	100	100	100	98.94
CUL7	609577	100	100	99.75	99.20
CYP26B1	605207	100	100	99.88	94.75
CYP27B1	609506	100	100	100	94.45
DDR2	191311	100	100	100	99.86
DHCR24	606418	100	100	100	97.77

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DLL3	602768	100	99.21	92.03	65.12
DLX3	600525	100	100	99.95	91.75
DMP1	600980	100	100	100	99.85
DONSON	611428	99.24	95.31	91.49	77.13
DPM1	603503	100	100	100	99.38
DVL1	601365	99.96	94.38	90.21	77.51
DYM	607461	100	100	100	98.47
DYNC2H1	603297	99.09	98.87	97.49	91.46
DYNC2I1	615462	99.92	96.05	87.67	76.33
DYNC2I2	613363	100	100	98.01	84.76
DYNLT2B	617353	100	100	100	100
EBP	300205	100	100	99.55	91.56
EIF2AK3	604032	100	99.38	96.83	92.39
ENPP1	173335	93.66	91.98	89.22	86.14
EVC	604831	94.29	94.26	93.73	87.77
EVC2	607261	100	99.86	99.03	95.16
EXT1	608177	100	100	100	99.43
EXT2	608210	100	100	100	99.64
EXTL3	605744	100	100	100	100
FAM111A	615292	100	100	100	98.56
FAM20C	611061	100	96.80	92.44	77.92
FBN1	134797	100	100	100	99.38
FERMT3	607901	100	99.08	94.02	88.34
FGD1	300546	100	98.24	94.73	84.21
FGF23	605380	100	100	100	100
FGF8	600483	93.90	92.43	87.21	66.49
FGF9	600921	100	100	100	96.35
FGFR1	136350	100	100	100	97.93
FGFR2	176943	100	100	99.68	98.15
FGFR3	134934	100	99.60	98.55	90.61
FIG4	609390	100	100	100	99.50
FKBP10	607063	100	99.79	98.23	89.65
FLNA	300017	99.95	99.05	97.51	95.12
FLNB	603381	100	100	99.77	97.55
FN1	135600	100	100	99.99	98.23
FUCA1	612280	100	100	100	99.42
FZD2	600667	99.13	97.17	95.63	92.24
GALNS	612222	100	100	99.41	87.98
GALNT3	601756	100	100	100	99.74
GDF3	606522	100	100	100	97.75
GDF5	601146	100	100	100	100
GDF6	601147	100	100	98.68	75.80
GH1	139250	100	100	100	100
GHR	600946	100	100	100	98.74

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GHRH	139190	100	100	100	77.10
GHRHR	139191	100	97.78	93.59	80.90
GHSR	601898	100	100	100	100
GJA1	121014	100	100	100	100
GLB1	611458	100	100	99.74	96.88
GLI2	165230	99.88	98.56	96.02	88.28
GLI3	165240	100	100	100	99.89
GMNN	602842	100	97.86	89.02	73.90
GNAS	139320	100	100	100	96.95
GNPAT	602744	100	100	100	96.02
GNPTAB	607840	100	100	99.79	98.44
GNPTG	607838	100	97.58	94.41	93.66
GNS	607664	100	97.83	96.15	93.60
GORAB	607983	100	100	100	96.50
GPC6	604404	100	100	100	97.91
GPX4	138322	100	96.02	92.64	88.95
GUSB	611499	100	100	100	98.65
HDAC4	605314	99.52	96.20	94.64	82.86
HES7	608059	92.21	75.71	65.45	40.78
HESX1	601802	100	100	96.38	88.11
HGSNAT	610453	94.18	93.42	91.23	83.05
HMGA2	600698	66.22	59.31	59.31	40.40
HOXA13	142959	83.26	77.66	74.25	68.15
HPGD	601688	100	100	100	97.44
HRAS	190020	100	100	100	100
HSPA9	600548	100	100	99.79	98.19
HSPG2	142461	99.45	99.11	97.49	90.45
HYLS1	610693	100	100	100	97.16
IDH2	147650	100	98.22	91.50	90.70
IDS	300823	100	100	98.17	89.78
IDUA	252800	99.80	90.46	84.30	72.67
IFITM5	614757	100	100	100	100
IFT122	606045	100	100	99.77	95.61
IFT140	614620	100	100	99.07	94.18
IFT172	607386	100	100	99.58	97.91
IFT43	614068	100	100	100	98.69
IFT80	611177	100	100	99.70	91.35
IGF1	147440	100	100	100	93.53
IGF1R	147370	100	100	99.30	96.66
IGF2	147470	100	100	100	96.83
IGFALS	601489	100	100	100	99.42
IGFBP3	146732	100	100	100	83.93
IGSF1	300137	100	100	100	98.88
IHH	600726	100	99.38	96.83	92.88

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IKBKB	603258	100	100	98.85	93.75
IKBKG	300248	36.54	25.95	25.35	20.68
IL2RG	308380	100	100	100	97.91
INPPL1	600829	98.63	95.71	94.66	87.53
KDELR2	609024	100	100	99.05	79.28
KIAA0753	617112	100	100	99.77	98.11
KIF22	603213	100	100	99.38	96.04
KIF7	611254	99.38	95.78	90.74	79.67
KMT2A	159555	99.87	99.34	98.67	96.60
KRAS	190070	100	100	100	100
LBR	600024	100	100	100	94.80
LEMD3	607844	100	100	99.60	95.17
LFNG	602576	100	100	100	90.22
LHX3	600577	100	99.59	96.19	84.31
LHX4	602146	100	100	98.49	91.16
LIFR	151443	100	99.80	97.37	88.03
LMX1B	602575	100	98.62	91.29	86.54
LONP1	605490	100	99.29	97.44	91.37
LRP4	604270	100	100	99.92	97.75
LRP5	603506	97.91	97.91	97.26	93.39
LRRK1	610986	100	99.46	98.14	92.98
LTBP2	602091	99.94	98.73	95.67	86.85
LTBP3	602090	99.59	98.30	93.77	80.79
LZTR1	600574	100	100	100	98.04
MAN2B1	609458	100	99.96	97.78	88.49
MANBA	609489	100	100	99.97	99.23
MAP2K1	176872	100	100	100	97.96
MAP2K2	601263	100	100	98.27	87.68
MAP3K7	602614	100	100	99.30	97.80
MATN3	602109	91.90	86.65	84.98	84.98
MBTPS2	300294	100	100	98.23	93.11
MEOX1	600147	100	100	100	85.77
MESD	607783	100	100	100	99.28
MESP2	605195	100	97.48	94.84	83.92
MGP	154870	100	99.59	90.39	89.05
MMP13	600108	100	100	100	99.81
MMP14	600754	100	100	99.79	94.45
MMP2	120360	100	100	98.68	90.60
MMP9	120361	100	100	99.85	92.50
MTAP	156540	100	100	100	100
MYH3	160720	100	99.58	98.72	94.95
MYO18B	607295	100	99.86	98.77	92.52
NAGLU	609701	100	94.32	91.15	85.27
NANS	605202	100	100	100	98.37

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NBAS	608025	100	100	100	98.26
NEK1	604588	100	99.49	98.10	94.33
NEK9	609798	100	100	99.41	96.32
NEU1	608272	100	100	100	99.34
NIN	608684	100	100	99.77	97.46
NIPBL	608667	100	100	99.65	97.00
NKX3-2	602183	100	100	98.56	89.99
NOTCH2	600275	99.86	98.85	98.02	95.09
NPPA	108780	100	100	100	100
NPPB	600295	100	100	97.94	69.48
NPPC	600296	100	100	93.54	67.46
NPR1	108960	100	99.61	96.41	85.70
NPR2	108961	100	100	100	99.03
NPR3	108962	100	100	100	100
NRAS	164790	100	100	100	99.54
OBSL1	610991	100	100	99.58	95.21
OFD1	300170	100	99.97	99.28	94.92
ORC1	601902	100	100	100	98.62
ORC4	603056	100	100	100	94.48
ORC6	607213	100	100	100	95.93
OSTM1	607649	100	97.73	86.41	69.34
OTX2	600037	100	100	100	100
P3H1	610339	100	100	100	100
P4HA1	176710	100	100	100	96.76
P4HB	176790	100	99.66	97.08	83.57
PAM16	614336	100	100	97.68	94.74
PAPPA	176385	95.63	94.15	92.03	89.68
PAPPA2	619485	100	100	99.79	97.63
PAPSS2	603005	100	100	100	95.75
PCNT	605925	100	100.00	99.21	93.69
PCYT1A	123695	100	100	100	99.80
PDE4D	600129	100	100	100	100
PEX5	600414	100	100	99.73	93.66
PEX7	601757	100	100	99.06	87.85
PHEX	300550	100	100	100	99.44
PHGDH	606879	100	100	99.78	94.15
PHYH	602026	100	89.33	89.33	83.03
PIK3CA	171834	100	100	100	99.11
PIK3R1	171833	100	100	100	98.75
PIK3R3	606076	100	100	100	95.77
PITX1	602149	99.90	94.06	88.92	74.90
PITX2	601542	100	96.62	93.01	82.88
PLEKHM1	611466	98.79	97.89	96.21	85.01
PLK4	605031	100	100	100	98.37

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PLOD2	601865	96.78	95.59	91.29	86.31
PLOD3	603066	100	97.38	93.62	79.16
PLS3	300131	100	100	100	99.06
POC1A	614783	100	100	100	97.36
POP1	602486	100	100	100	97.75
POR	124015	96.93	96.93	96.93	96.64
POU1F1	173110	100	96.73	91.84	88.02
PPIB	123841	100	100	100	100
PRKAR1A	188830	100	100	100	100
PROKR2	607123	100	100	100	100
PROP1	601538	100	100	95.93	71.75
PSAT1	610936	100	100	100	96.43
PTDSS1	612792	100	100	100	99.46
PTH1R	168468	100	100	100	95.75
PTPN11	176876	98.37	98.37	98.37	98.37
RAB33B	605950	100	100	100	100
RAF1	164760	100	100	100	96.29
RASGRP2	605577	100	99.98	98.54	89.89
RBM8A	605313	100	100	100	94.55
RBPJ	147183	100	99.91	98.49	96.34
RIPPLY2	609891	100	100	100	95.37
RIT1	609591	100	100	100	100
ROR2	602337	100	100	99.42	95.71
RPGRIP1L	610937	99.73	98.44	96.86	92.30
RPL10	312173	100	100	100	100
RSPRY1	616585	100	100	100	100
RUNX2	600211	100	100	100	100
SBDS	607444	100	100	100	100
SCARF2	613619	98.01	91.54	80.70	63.30
SCUBE3	614708	100	99.97	98.68	96.29
SEC24D	607186	100	100	99.79	96.68
SERPINF1	172860	100	100	100	98.71
SERPINH1	600943	100	100	100	100
SGMS2	611574	100	100	100	100
SGSH	605270	93.52	93.52	93.52	93.25
SH3PXD2B	613293	100	98.73	96.69	90.06
SHOC2	602775	100	100	100	98.53
SHOX	312865	100	100	97.34	77.77
SHOX2	602504	100	99.92	98.57	86.16
SLC10A7	611459	100	100	100	83.19
SLC17A5	604322	93.58	93.58	93.58	92.55
SLC25A24	608744	100	100	100	96.17
SLC26A2	606718	100	100	100	100
SLC29A3	612373	100	97.60	97.60	97.48

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SLC34A3	609826	100	99.46	98.48	91.09
SLC35D1	610804	100	100	99.00	91.53
SLC39A13	608735	100	100	100	96.56
SLCO2A1	601460	100	100	98.73	87.75
SLCO5A1	613543	100	100	99.30	96.27
SMAD4	600993	100	100	100	99.47
SMARCAL1	606622	100	100	100	97.64
SNRPB	182282	100	100	97.97	84.94
SNX10	614780	100	100	100	100
SOS1	182530	100	99.96	97.71	96.03
SOS2	601247	99.61	98.32	97.02	91.85
SOST	605740	100	100	100	100
SOX2	184429	100	100	99.49	93.56
SOX3	313430	100	100	94.29	73.05
SOX9	608160	100	97.10	94.90	86.77
SP7	606633	100	100	100	93.02
SPARC	182120	100	100	100	95.22
SPINK5	605010	100	99.84	97.49	87.43
SPR	182125	100	100	99.05	88.20
SRCAP	611421	100	99.95	99.28	95.12
STAT3	102582	100	100	100	95.99
STAT5B	604260	100	99.03	96.66	89.84
SULF1	610012	100	100	99.13	95.62
SUMF1	607939	100	100	100	100
TAPT1	612758	96.16	88.94	84.83	80.69
TBCE	604934	100	100	99.25	96.43
TBX15	604127	100	100	99.39	97.43
TBX4	601719	100	98.25	94.05	87.49
TBX6	602427	100	100	100	90.67
TBXAS1	274180	100	100	100	100
TCIRG1	604592	100	98.97	94.86	86.76
TCTN2	613846	100	100	99.43	95.27
TCTN3	613847	100	100	100	98.55
TENT5A	611357	100	100	100	100
TGFB1	190180	100	100	100	99.69
TMEM165	614726	100	100	100	96.75
TMEM216	613277	100	100	100	99.45
TMEM231	614949	100	100	100	95.56
TMEM38B	611236	100	100	100	100
TNFRSF11A	603499	90.20	90.20	87.10	81.42
TNFRSF11B	602643	100	100	100	100
TNFSF11	602642	100	100	100	100
TRAPPC2	300202	100	100	100	93.81
TRIP11	604505	100	99.69	98.38	92.09

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TRPS1	604386	100	100	100	100
TRPV4	605427	100	100	100	96.69
TTC21B	612014	100	100	100	99.65
VDR	601769	100	96.80	91.53	77.28
WDR19	608151	100	100	99.75	97.05
WDR35	613602	100	99.78	99.23	96.28
WNT1	164820	100	100	100	95.13
WNT5A	164975	100	100	98.71	96.17
XRCC4	194363	100	100	100	95.91
XYLT1	608124	98.78	95.46	92.27	85.10
XYLT2	608125	94.49	94.49	94.49	90.39
ZBTB16	176797	100	100	100	100
ZMPSTE24	606480	100	100	100	100

- 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 ($\pm 10bp$ flanking introns) that is covered at least 10x, 20x, 30x or 50x