Whole Exome Sequencing
Gene package Skeletal Dysplasia, Version 2.2, 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).

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	77	100	99	94
ACAN	155760	234	93	92	92
ACP5	171640	280	100	100	100
ACTB	102630	253	100	100	100
ACVR1	102576	104	100	100	99
ADAMTS10	608990	267	100	100	100
ADAMTS17	607511	177	98	96	94
ADAMTSL2	612277	97	47	45	44
AGA	613228	83	100	98	90
AGPS	603051	64	96	94	85
ALG12	607144	224	100	100	100
ALG3	608750	213	100	100	100
ALG9	606941	78	97	95	92
ALMS1	606844	116	100	99	96
ALPL	171760	323	100	100	100
AMER1	300647	240	100	100	100
ANKH	605145	171	100	100	100
ANKRD11	611192	247	100	99	97
ANO5	608662	61	94	87	75
ARSB	611542	130	100	100	98
ARSL	300180	101	100	100	100
B3GALT6	615291	283	83	80	78
B3GAT3	606374	243	100	100	100
B4GALT7	604327	254	100	100	100
BMP1	112264	301	100	100	100
BMPER	608699	116	100	100	99

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BMPR1B	603248	79	100	98	92
BRAF	164757	84	100	99	94
BTK	300300	77	100	99	95
CA2	611492	116	100	100	100
CANT1	613165	318	100	100	100
CBL	165360	130	100	100	99
CCDC8	614145	247	100	100	100
CCN6	603400	110	100	99	97
CDC45	603465	172	100	100	100
CDC6	602627	186	100	100	98
CDKN1C	600856	156	92	87	83
CDT1	605525	222	100	100	98
CEP120	613446	84	99	94	89
CFAP410	603191	250	100	100	100
CHST3	603799	345	100	100	100
CLCN5	300008	118	100	96	91
CLCN7	602727	244	100	100	100
COG1	606973	164	100	100	99
COL10A1	120110	266	100	100	100
COL11A1	120280	86	99	95	89
COL11A2	120290	186	100	100	99
COL1A1	120150	246	100	100	100
COL1A2	120160	131	98	97	96
COL2A1	120140	203	100	100	99
COL9A1	120210	99	100	98	96
COL9A2	120260	153	100	100	100
COL9A3	120270	185	100	100	100
COLEC11	612502	234	100	100	100
COMP	600310	286	100	100	100
CREB3L1	616215	178	100	100	100
CREBBP	600140	275	99	99	98
CRTAP	605497	155	100	100	100
CSGALNACT1	616615	159	100	100	98
CTSA	613111	181	100	100	100
CTSK	601105	108	100	100	100
CUL7	609577	281	100	100	100
CYP26B1	605207	414	100	100	100
CYP27B1	609506	256	100	100	100
DDR2	191311	131	100	100	100
DHCR24	606418	207	100	100	100
DLL3	602768	150	100	100	100
DLX3	600525	271	100	100	100
DMP1	600980	92	100	96	96
DONSON	611428	78	95	93	89

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DPM1	603503	79	98	89	73
DVL1	601365	251	100	100	100
DYM	607461	82	100	99	93
DYNC2H1	603297	57	94	84	73
EBP	300205	110	100	100	100
EIF2AK3	604032	95	97	93	90
ENPP1	173335	67	93	85	78
EVC	604831	189	94	92	91
EVC2	607261	181	100	100	99
EXT1	608177	298	100	100	100
EXT2	608210	142	100	100	100
EXTL3	605744	223	100	100	100
FAM111A	615292	108	100	99	97
FAM20C	611061	200	100	100	100
FBN1	134797	141	100	100	99
FERMT3	607901	243	100	100	100
FGD1	300546	126	100	100	100
FGF23	605380	272	100	100	100
FGF8	600483	142	100	97	94
FGF9	600921	122	100	100	95
FGFR1	136350	185	100	100	100
FGFR2	176943	145	100	100	99
FGFR3	134934	241	100	100	100
FIG4	609390	88	100	96	91
FKBP10	607063	195	100	100	100
FLNA	300017	206	100	100	100
FLNB	603381	193	100	100	99
FN1	135600	150	100	100	100
FUCA1	612280	172	100	100	100
FZD2	600667	394	100	100	99
GALNS	612222	220	100	100	100
GALNT3	601756	74	99	89	82
GDF3	606522	173	100	100	100
GDF5	601146	246	100	100	100
GDF6	601147	147	100	100	100
GH1	139250	200	100	100	100
GHR	600946	105	100	95	88
GHRHR	139191	163	100	100	100
GHSR	601898	463	100	100	99
GJA1	121014	143	100	100	100
GLB1	611458	149	100	100	100
GLI2	165230	260	100	100	100
GLI3	165240	243	100	100	100
GMNN	602842	45	91	69	59

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GNAS	139320	269	100	100	100
GNPAT	602744	80	97	94	86
GNPTAB	607840	87	100	99	95
GNPTG	607838	302	100	100	100
GNS	607664	105	100	100	99
GORAB	607983	77	100	93	83
GPC6	604404	135	100	100	100
GPX4	138322	285	100	100	99
GUSB	611499	222	100	100	100
HDAC4	605314	227	100	99	97
HES7	608059	144	100	100	98
HESX1	601802	47	79	62	57
HGSNAT	610453	93	99	94	92
HOXA13	142959	221	91	85	82
HPGD	601688	79	96	77	60
HRAS	190020	426	100	100	100
HSPA9	600548	114	100	99	97
HSPG2	142461	283	99	99	99
HYLS1	610693	114	100	100	100
IDH2	147650	236	100	100	100
IDS	300823	91	100	98	94
IDUA	252800	224	100	100	98
IFITM5	614757	322	100	100	100
IFT122	606045	141	100	100	98
IFT140	614620	199	100	100	100
IFT172	607386	143	100	100	98
IFT43	614068	133	100	100	100
IFT80	611177	49	92	82	69
IGF1	147440	117	100	100	100
IGF1R	147370	176	100	100	100
IGFALS	601489	372	100	100	100
IGSF1	300137	91	100	100	98
IHH	600726	276	100	100	100
IKBKB	603258	159	100	100	100
IKBKG	300248	35	36	26	26
IL2RG	308380	88	100	100	97
IMPAD1	614010	192	100	100	100
INPPL1	600829	195	100	100	99
KIAA0753	617112	101	100	97	95
KIF22	603213	243	100	100	100
KIF7	611254	224	100	100	98
KMT2A	159555	118	100	100	98
KRAS	190070	59	90	78	77
LBR	600024	72	96	94	86

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LEMD3	607844	108	96	91	83
LFNG	602576	221	88	86	85
LHX3	600577	232	100	100	100
LHX4	602146	206	100	100	100
LIFR	151443	66	87	80	72
LMX1B	602575	257	100	100	100
LONP1	605490	264	100	100	100
LRP4	604270	228	100	100	99
LRP5	603506	325	98	98	98
LRRK1	610986	225	99	98	97
LTBP2	602091	241	100	100	100
LTBP3	602090	223	100	99	99
LZTR1	600574	242	100	100	100
MAN2B1	609458	213	100	100	100
MANBA	609489	93	100	99	94
MAP2K1	176872	148	100	100	99
MAP2K2	601263	276	100	100	100
MAP3K7	602614	83	99	96	90
MATN3	602109	144	100	95	91
MBTPS2	300294	52	99	94	81
MEOX1	600147	212	100	100	100
MESP2	605195	184	100	100	100
MGP	154870	68	89	89	89
MMP13	600108	94	100	100	98
MMP14	600754	213	100	100	100
MMP2	120360	178	100	100	100
MMP9	120361	228	100	100	100
MTAP	156540	109	100	100	97
MYH3	160720	137	100	100	99
MYO18B	607295	248	100	100	98
NAGLU	609701	182	100	100	100
NANS	605202	140	100	100	99
NBAS	608025	103	100	98	94
NEK1	604588	53	92	81	70
NEK9	609798	100	100	100	97
NEU1	608272	238	100	100	100
NIN	608684	97	98	94	91
NKX3-2	602183	185	100	100	100
NOTCH2	600275	188	100	98	97
NPPC	600296	156	100	100	100
NPR2	108961	199	100	100	100
NRAS	164790	107	100	100	99
OBSL1	610991	283	100	100	100
OFD1	300170	51	97	80	64

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ORC1	601902	148	100	100	100
ORC4	603056	43	96	83	61
ORC6	607213	104	100	98	89
OSTM1	607649	118	93	70	64
OTX2	600037	208	100	100	100
P3H1	610339	199	100	100	100
P4HB	176790	174	100	100	100
PAM16	614336	190	100	100	100
PAPPA2	No ID	199	100	100	100
PAPSS2	603005	124	100	100	100
PCNT	605925	234	100	96	94
PCYT1A	123695	126	100	100	99
PDE4D	600129	88	100	96	90
PEX5	600414	172	100	100	100
PEX7	601757	94	100	100	94
PHEX	300550	69	100	98	87
PHGDH	606879	199	100	100	100
PHYH	602026	99	100	97	87
PIK3R1	171833	65	100	93	82
PITX1	602149	285	100	100	100
PITX2	601542	197	100	100	97
PLEKHM1	611466	191	99	98	98
PLK4	605031	77	98	90	83
PLOD2	601865	68	91	89	83
PLS3	300131	50	100	92	73
POC1A	614783	216	100	100	100
POP1	602486	129	100	100	99
POR	124015	296	100	100	100
POU1F1	173110	65	92	91	85
PPIB	123841	189	100	100	100
PRKAR1A	188830	98	100	100	99
PROKR2	607123	340	100	100	100
PROP1	601538	186	100	100	100
PSAT1	610936	103	100	98	90
PTDSS1	612792	101	100	100	100
PTH1R	168468	266	100	100	100
PTPN11	176876	80	98	94	89
RAB33B	605950	201	100	100	98
RAF1	164760	124	100	98	97
RASGRP2	605577	310	100	100	100
RBM8A	605313	82	100	100	95
RBPJ	147183	79	99	91	81
RIPPLY2	609891	129	90	64	64
RIT1	609591	123	100	100	100

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RMRP	157660	No coverage data			
RNU4ATAC	601428	No coverage data			
ROR2	602337	278	100	100	98
RPGRIP1L	610937	80	95	91	83
RPL10	312173	136	100	100	100
RSPRY1	616585	93	100	96	94
RUNX2	600211	160	100	100	100
SBDS	607444	131	100	100	99
SCARF2	613619	234	100	99	97
SEC24D	607186	96	100	99	96
SERPINF1	172860	117	100	100	98
SERPINH1	600943	369	100	100	100
SGSH	605270	272	99	94	94
SH3PXD2B	613293	196	100	100	100
SHOC2	602775	74	100	100	96
SHOX	312865	45	50	50	48
SLC10A7	611459	67	94	81	68
SLC17A5	604322	82	100	94	85
SLC25A24	608744	95	97	91	89
SLC26A2	606718	138	100	100	100
SLC29A3	612373	233	100	100	100
SLC34A3	609826	248	100	100	100
SLC35D1	610804	89	92	88	76
SLC39A13	608735	246	100	100	100
SLCO2A1	601460	152	100	100	99
SLCO5A1	613543	148	91	91	89
SMAD4	600993	78	100	97	95
SMARCAL1	606622	146	100	100	98
SNRPB	182282	124	100	100	100
SNX10	614780	71	100	100	98
SOS1	182530	74	94	89	79
SOS2	601247	64	95	80	67
SOST	605740	330	100	100	100
SOX2	184429	257	100	100	100
SOX3	313430	155	100	100	100
SOX9	608160	207	100	100	99
SP7	606633	258	100	100	100
SPARC	182120	141	100	100	100
SPINK5	605010	65	96	87	77
SPR	182125	142	100	100	100
SRCAP	611421	186	100	100	100
STAT3	102582	135	100	100	99
STAT5B	604260	157	100	100	100
SULF1	610012	99	100	98	92

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SUMF1	607939	174	100	100	100
TAPT1	612758	58	94	80	66
TBCE	604934	69	100	96	87
TBX15	604127	193	100	100	99
TBX4	601719	204	100	100	100
TBX6	602427	280	100	100	100
TBXAS1	274180	174	100	100	96
TCIRG1	604592	242	100	100	100
TCTEX1D2	617353	146	100	100	88
TCTN2	613846	108	100	95	93
TCTN3	613847	126	100	100	98
TGFB1	190180	274	100	100	100
TMEM165	614726	92	100	99	93
TMEM216	613277	142	100	100	100
TMEM231	614949	271	100	100	98
TMEM38B	611236	89	100	100	93
TNFRSF11A	603499	178	95	95	95
TNFRSF11B	602643	122	100	100	97
TNFSF11	602642	119	100	100	100
TRAPPC2	300202	40	88	60	43
TRIP11	604505	44	90	73	58
TRPS1	604386	210	100	100	100
TRPV4	605427	299	100	100	100
TTC21B	612014	63	99	95	83
VDR	601769	176	100	100	100
WDR19	608151	75	100	96	92
WDR34	613363	177	100	100	100
WDR35	613602	75	97	93	87
WDR60	615462	85	99	95	88
WNT1	164820	280	100	100	100
WNT5A	164975	316	100	100	100
XRCC4	194363	39	95	77	54
XYLT1	608124	191	100	98	95
XYLT2	608125	298	100	100	99
ZBTB16	176797	309	100	100	100
ZMPSTE24	606480	73	98	87	77

- 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

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