Whole Exome Sequencing
Gene package Collagenopathy, version 1, 30-9-2021

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/). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy 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 filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). It is not excluded that pathogenic variants 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ABCC6	603234	99.56	98.03	96.73	84.24
ABL1	189980	100	100	100	99.21
ACVR1	102576	100	100	100	99.36
ADAMTS10	608990	100	100	99.74	94.21
ADAMTS17	607511	95.41	90.13	86.31	82.79
ADAMTS2	604539	98.59	96.10	95.14	89.77
ADAMTSL2	612277	45.35	42.70	40.34	33.50
ADAMTSL4	610113	100	99.10	98.06	92.03
AEBP1	602981	100	100	99.56	94.39
ALDH18A1	138250	100	100	100	98.26
ALPL	171760	100	100	100	99.94
ATP7A	300011	100	100	99.49	95.63
B3GALT6	615291	79.89	76.12	72.74	70.01
B3GAT3	606374	100	100	100	95.97
B4GALT7	604327	98.64	93.64	93.64	85.79
BMP1	112264	100	100	99.82	95.41
BPNT2	614010	100	100	100	99.11
C1R	613785	100	100	99.79	96.81
C1S	120580	100	100	100	96.76

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CBS	613381	100	100	100	100
CHST14	608429	99.09	95.47	92.86	89.81
CHST3	603799	100	100	100	100
COL10A1	120110	100	100	99.18	97.24
COL11A1	120280	100	99.17	97.37	87.82
COL11A2	120290	100	100	98.35	87.06
COL12A1	120320	100	99.75	98.55	92.77
COL1A1	120150	100	100	99.00	92.15
COL1A2	120160	100	99.09	96.75	86.41
COL2A1	120140	100	100	99.72	94.32
COL3A1	120180	99.82	98.39	96.02	89.41
COL5A1	120215	100	99.17	97.94	93.25
COL5A2	120190	100	99.89	98.64	90.01
COL6A1	120220	100	98.94	96.91	92.16
COL6A2	120240	100	100	99.49	95.69
COL6A3	120250	100	100	99.77	98.78
COL9A1	120210	100	100	97.67	84.62
COL9A2	120260	100	96.12	88.64	64.54
COL9A3	120270	100	99.22	95.30	79.74
CREB3L1	616215	99.94	97.66	93.07	78.85
CRTAP	605497	100	100	100	98.66
DSE	605942	100	100	100	100
EFEMP1	601548	100	100	100	99.82
EFEMP2	604633	100	99.97	97.06	84.43
ELN	130160	100	99.13	95.08	75.43
FBLN5	604580	91.62	91.62	91.62	90.51
FBN1	134797	100	100	100	99.50
FBN2	612570	100	100	100	99.22
FKBP10	607063	100	99.59	98.23	92.39
FKBP14	614505	100	100	100	100
FLNA	300017	99.98	98.83	97.47	92.13
FLNB	603381	100	100	99.93	98.27
GNAS	139320	100	100	100	98.21
IFITM5	614757	100	100	100	100
IPO8	605600	100	100	99.54	96.46
LEMD3	607844	100	100	99.50	94.34
LRP5	603506	97.91	97.91	97.46	94.32
LTBP1	150390	99.95	99.01	97.95	92.28
LTBP2	602091	100	98.58	95.78	88.07
LTBP3	602090	99.90	98.83	94.23	80.91

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LTBP4	604710	99.23	96.89	94.33	85.75
MAP3K7	602614	100	100	99.17	97.17
MESD	607783	100	100	100	99.41
P3H1	610339	100	100	100	100
PLOD1	153454	100	99.54	96.86	87.82
PLOD2	601865	96.30	91.47	91.02	86.20
PLOD3	603066	100	96.13	94.04	80.47
PLS3	300131	100	100	100	97.90
PPIB	123841	100	100	100	100
PRDM5	614161	100	100	98.66	91.35
PYCR1	179035	100	100	100	96.80
RIN2	610222	100	100	100	97.40
SERPINF1	172860	100	100	100	99.22
SERPINH1	600943	100	100	100	100
SKI	164780	97.53	92.04	86.04	74.94
SLC26A2	606718	100	100	100	100
SLC2A10	606145	100	100	100	98.61
SLC39A13	608735	100	100	100	97.99
SMAD3	603109	100	100	100	99.75
SP7	606633	100	100	100	94.45
TAB2	605101	100	100	100	99.55
TAPT1	612758	95.71	88.94	83.17	77.21
TGFB2	190220	100	100	100	95.93
TGFB3	190230	100	100	100	97.89
TGFBR1	190181	93.12	93.12	93.12	93.12
TGFBR2	190182	100	100	100	97.70
TMEM38B	611236	100	100	100	100
TNXB	600985	62.44	48.29	42.12	35.83
VAV3	605541	100	100	99.58	89.87
VCAN	118661	100	100	100	100
WNT1	164820	100	100	100	95.59
WNT11	603699	95.18	88.47	83.22	74.35
XYLT1	608124	98.57	94.72	91.00	86.04
XYLT2	608125	94.49	94.49	94.49	92.50
ZNF469	612078	100	99.63	98.38	93.54

- OMIM release used: 23-9-2021
- The statistics above are based on a set of 150 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

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