

Whole Exome Sequencing

Gene package Aneurysm, version 3.1, 22-11-2017



Technical information

DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate 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).



Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ABL1	Leukemia, Philadelphia chromosome-positive, resistant to imatinib	189980	84	100	100	97
ACTA2	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834	102620	97	100	100	100
BGN	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106	301870	52	100	98	89
COL1A1	{Bone mineral density variation QTL, osteoporosis}, 166710 Caffey disease, 114000 Ehlers-Danlos syndrome, classic, 130000 Ehlers-Danlos syndrome, type VIIA, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220	120150	87	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
COL1A2	Ehlers-Danlos syndrome, cardiac valvular form, 225320 Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710	120160	52	100	98	90
COL3A1	Ehlers-Danlos syndrome, type IV, 130050	120180	89	100	100	99
COL5A1	Ehlers-Danlos syndrome, classic type, 130000	120215	85	100	98	97
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	94	100	100	100
EFEMP2	Cutis laxa, autosomal recessive, type IB, 614437	604633	76	100	100	100
ELN	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500	130160	70	100	100	98
FBN1	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328	134797	111	100	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	51	100	98	89
FLNA	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244	300017	79	100	100	100
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 Cataract 34, multiple types, 612968	601094	32	82	68	54
GATA5	No OMIM phenotype	611496	66	100	100	99
LOX	Aortic aneurysm, familial thoracic 10, 617168	153455	107	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MAT2A	No OMIM phenotype	601468	70	100	100	91
MFAP5	Aortic aneurysm, familial thoracic 9, 616166	601103	35	100	96	65
MYH11	Aortic aneurysm, familial thoracic 4, 132900	160745	76	100	99	91
MYLK	Aortic aneurysm, familial thoracic 7, 613780	600922	79	100	100	96
NOTCH1	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730	190198	87	100	99	98
PLOD1	Ehlers-Danlos syndrome, type VI, 225400	153454	57	100	100	97
PRKG1	Aortic aneurysm, familial thoracic 8, 615436	176894	45	100	99	88
ROBO4	No OMIM phenotype	607528	70	100	100	97
SKI	Shprintzen-Goldberg syndrome, 182212	164780	76	100	100	99
SLC2A10	Arterial tortuosity syndrome, 208050	606145	94	100	100	96
SMAD2	No OMIM phenotype	601366	47	100	97	88
SMAD3	Loeys-Dietz syndrome 3, 613795	603109	99	100	100	100
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	59	100	100	96
SMAD6	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439	602931	90	100	100	99
TGFB2	Loeys-Dietz syndrome 4, 614816	190220	54	100	100	94
TGFB3	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582	190230	69	100	100	100
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	96	95	93	92
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	190182	124	100	100	100

- Gene symbols according HGNC
- OMIM release used: 2-6-2017
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 96 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (± 10 bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (± 10 bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (± 10 bp flanking introns) that is covered at least 20x