

## Whole Exome Sequencing

### Gene package Aneurysm, version 3, 1-7-2017



#### Technical information

After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: <http://bio-bwa.sourceforge.net/>). Variant detection is performed by Genome Analysis Toolkit (reference: <http://www.broadinstitute.org/gatk/>). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. 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
ABL1	Leukemia, Philadelphia chromosome-positive, resistant to imatinib	189980	97	100	100
ACTA2	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834	102620	96	100	100
BGN	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106	301870	63	100	100
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	76	99	94
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	71	100	96
COL3A1	Ehlers-Danlos syndrome, type IV, 130050	120180	63	99	93

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
COL5A1	Ehlers-Danlos syndrome, classic type, 130000	120215	77	99	97
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	70	99	97
EFEMP2	Cutis laxa, autosomal recessive, type IB, 614437	604633	82	100	100
ELN	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500	130160	63	100	99
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	86	100	100
FBN2	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118	612570	90	100	98
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	55	100	99
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 Cataract 34, multiple types, 612968	601094	40	73	58
GATA5	No OMIM phenotype	611496	54	100	100
LOX	Aortic aneurysm, familial thoracic 10, 617168	153455	87	100	100
MAT2A	No OMIM phenotype	601468	37	93	74
MFAP5	Aortic aneurysm, familial thoracic 9, 616166	601103	38	95	80
MYH11	Aortic aneurysm, familial thoracic 4, 132900	160745	90	100	100
MYLK	Aortic aneurysm, familial thoracic 7, 613780	600922	102	100	100
NOTCH1	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730	190198	86	100	99
PLOD1	Ehlers-Danlos syndrome, type VI, 225400	153454	88	100	98
PRKG1	Aortic aneurysm, familial thoracic 8, 615436	176894	46	97	82

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ROBO4	No OMIM phenotype	607528	47	99	90
SKI	Shprintzen-Goldberg syndrome, 182212	164780	55	99	97
SLC2A10	Arterial tortuosity syndrome, 208050	606145	95	100	100
SMAD2	No OMIM phenotype	601366	94	100	96
SMAD3	Loeys-Dietz syndrome 3, 613795	603109	93	100	100
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	76	100	98
SMAD6	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439	602931	71	94	84
TGFB2	Loeys-Dietz syndrome 4, 614816	190220	107	100	100
TGFB3	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582	190230	96	100	100
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	111	94	94
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	190182	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 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x