CGAT – Connective Tissue Disorder Panel v.1.0 (11/2022)

Panel coverage statistics

This panel contains 114 RefSeq genes. The names of the genes are in HGNC format this is current as of the versioning date of this panel. There is on average 97.1% coverage of all coding exons at 20x and 98.8% coverage at 10x in analyzable target regions from the panel-based exome backbone (v8 SureSelect). This also includes 10 base pairs of flanking intronic DNA in all available transcripts. Asterisk(*) represent genes in which <20x coverage spanning at least 50% of a given exon is calculated. Please note that sequence variants in certain exons of genes can occur in repetitive DNA or highly homologous genomic architecture inherent in the human genome, and therefore will not be analyzed.

Panel description

Individuals with inherited connective tissue disorders including but not limited to Marfan, Loeys-Dietz, Thoracic Aortic Aneurysmal Dissections, Ehlers-Danlos Syndrome, Stickler, Cutis Laxa, Marfan-like and related disorders have rare yet recurrent aberrations in a gene involved in structure or function of connective tissue. Such disorders can include a constellation of features including but not limited to joints, eyes, skin, and cardiovascular system that can be affected. This panel is designed to detect sequence-based variants in the form of single nucleotide, insertion/deletion, and copy number alterations.

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If ordering a custom panel, please provide gene name in the designated orderable field in Epic.

*ABCC6**	*COL11A2*	*GGCX*	*SKI*	*KIF22*
ABL1	*COL12A1*	*GORAB*	*SLC26A2*	*LRP2*
ACTA2	*COL1A1*	*HCN4*	*SLC2A10*	*NEK9*
ACVR1	*COL1A2*	*LEMD3*	*SLC39A13*	*NEPRO*
ADAMTS10	*COL2A1*	*LOX*	*SMAD2*	*NT5E*
ADAMTS17	*COL3A1*	*LOXL3*	*SMAD3*	*P4HA1*
ADAMTS2	*COL4A1*	*LTBP2*	*SMAD4*	*PLP1*
ADAMTSL4	*COL5A1*	*LTBP3*	*SMAD6*	*ROCK2*
AEBP1	*COL5A2*	*LTBP4*	*SPARC*	*SERPING1*
ALDH18A1	*COL9A1*	*LZTS1*	*TALDO1*	*SHOC2*
ARIH1	*COL9A2*	*MAT2A*	*TGFB1*	*SMS*
ATP6V0A2	*COL9A3*	*MED12*	*TGFB2*	*TNFRSF1A*
ATP6V1A	*CRTAP*	*MFAP5*	*TGFB3*	*TNXB**
ATP6V1E1	*DCHS1*	*MYH11*	*TGFBR1*	*VCAN*
ATP7A	*DSE*	*MYLK*	*TGFBR2*	
B3GALT6	*EFEMP2*	*NOG*	*UPF3B*	
B3GAT3	*ELN*	*NOTCH1*	*ZNF469*	
B4GALT7	*FBLN5*	*P3H1*	*ADAMTSL2**	
BGN	*FBN1*	*PKD2*	*ALPL*	
C1S	*FBN2*	*PLOD1*	*ATP6AP1*	
CBS	*FKBP14*	*PLOD3*	*BPNT2*	
CHST14	*FLCN*	*PRDM5*	*C1R*	
CHST3	*FLNA*	*PRKG1*	*CHD4*	
COG7	*FLNB*	*PYCR1*	*CSGALNACT1*	
COL11A1	*FOXE3*	*RIN2*	*ERBIN*	

ABCC6 exons 1-10, *ADAMTSL2* exons 10-19 *& TNXB* exons 32-44 are not analyzed due to multiple homologous sequences in the genome