

Kötőszöveti betegségek (Connective tissue disorders) génpanel v:2025-05-21

Panel tartalma 126 gén:

ABCC6, ABL1, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, ADAMTSL4, AEBP1, ALDH18A1, ALPL, ARIH1, ATP6AP1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B4GALT7, BGN, BPNT2, C1R, C1S, CBS, CHD4, CHST14, CHST3, COG7, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, CRTAP, CSGALNACT1, DCC, DCHS1, DSE, EFEMP1, EFEMP2, ELN, ERBIN, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, FLNB, FOXE3, GGCX, GORAB, HCN4, IPO8, KIF22, LEMD3, LOX, LOXL3, LRP2, LTBP1, LTBP2, LTBP3, LTBP4, LZTS1, MAT2A, MED12, MFAP5, MOCS1, MYH11, MYLK, NEK9, NEPRO, NOG, NOTCH1, NT5E, P3H1, P4HA1, PIEZO2, PKD2, PLOD1, PLOD3, PLP1, PRDM5, PRKG1, PYCR1, RIN2, ROBO3, ROCK2, SERPING1, SGMS2, SHOC2, SKI, SLC26A2, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SMS, SPARC, TALDO1, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, THBS2, TNFRSF1A, TNXB, UPF3B, VCAN, ZNF469

Forrás:

Invitae Connective Tissue Disorders Panel

<https://www.invitae.com/pt/physician/tests/434340/>

Connective Tissue Disorders Panel

<https://www.preventiongenetics.com/testInfo?val=Connective+Tissue+Disorders+Panel>

Connective Tissue NGS Panel

<https://www.fulgentgenetics.com/Connective-Tissue>

Ehlers Danlos syndrome with a likely monogenic cause (Version 4.1)

<https://panelapp.genomicsengland.co.uk/panels/53/>

(Megtekintve: 2025-05-21)