

## **Kötőszöveti betegségek (Connective tissue disorders) génpanel v:2022.01.26.**

Panel tartalma 115 gén:

ACTA2, ABCC6, ABL1, 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, COL9A1, COL9A2, COL9A3, CRTAP, CSGALNACT1, DCHS1, DSE, EFEMP2, ELN, ERBIN, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, FLNB, FOXE3, GGCX, GORAB, HCN4, KIF22, LEMD3, LOX, LOXL3, LRP2, LTBP2, LTBP3, LTBP4, LZTS1, MAT2A, MED12, MFAP5, MYH11, MYLK, NEK9, NEPRO, NOG, NOTCH1, NT5E, P3H1, P4HA1, PKD2, PLOD1, PLOD3, PLP1, PRDM5, PRKG1, PYCR1, RIN2, ROCK2, SERPING1, SGMS2, SHOC2, SKI, SLC26A2, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SMS, SPARC, TALDO1, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TNFRSF1A, TNXB, UPF3B, VCAN, ZNF469.

Forrás:

Invitae Connective Tissue Disorders Panel

<https://www.invitae.com/pt/physician/tests/434340/> (Megtekintve: 2022.01.26.)

Connective Tissue Disorders Panel

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

(Megtekintve: 2022.01.26.)

Connective Tissue NGS Panel

<https://www.fulgentgenetics.com/Connective-Tissue> (Megtekintve: 2022.01.26.)