G.LAB

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Клінічна область Офтальмологія - 518 генів*

	Панелі:	Гени:
1	Вроджена стаціонарна нічна сліпота / Invitae Congenital Stationary Night Blindness Panel - 22 гена	CABP4, CACNA1F, CACNA2D4, CHM, CYP4V2, FRMD7, GNAT1, GNB3, GPR179, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RBP4, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1
2	Дистрофії рогівки / Invitae Corneal Dystrophies Panel - 33 гена	CHRDL1, CHST6, COL17A1, COL5A1, COL8A2, CYP4V2, DCN, FOXE3, GJA8, GRHL2, GSN, KERA, KRT12, KRT3, LCAT, LOXHD1, MAF, MIR184, NLRP1, OVOL2, PAX6, PIKFYVE, PITX2, PRDM5, PXDN, SLC4A11, TACSTD2, TGFBI, UBIAD1, VSX1, ZEB1, ZNF143, ZNF469
3	Ахроматопсія / Invitae Achromatopsia Panel - 8 генів	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, RGS9, RGS9BP
4	Макулярна дистрофія та макулярна дегенерація / Invitae Macular Dystrophy Panel - 36 генів	ABCA4, BEST1, C1QTNF5, CDH3, CERKL, CFI, CHST6, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, FSCN2, GUCA1B, HMCN1, IMPG1, IMPG2, KCNV2, MFSD8, NMNAT1, PRDM13, PROM1, PRPH2, RAX2, RBP3, RBP4, RDH12, RDH5, RLBP1, RP1L1, RPGRIP1, RS1, SIX6, TIMP3
5	Альбінізм / Invitae Oculocutaneous Albinism Panel - 23 гена	AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10orf11, DTNBP1, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MLPH, MYO5A, OCA2, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1
6	Септо-оптична дисплазія / Invitae Septo-optic Dysplasia Panel - 8 генів	GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3
7	Синдром Альпорта / Invitae Alport Syndrome Panel - 6 генів	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9
8	Синдром Стіклера / Invitae Stickler Syndrome Panel - 9 генів	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, LRP2, VCAN
9	Глаукома / Invitae Glaucoma Panel - 27 генів	ASB10, ATOH7, BMP4, COL4A1, COL8A2, CYP1B1, EXO5, FOXC1, FOXE3, LMX1B, LTBP2, MAF, MFRP, MYOC, OPTN, PAX6, PIK3R1, PITX2, PITX3, PRPF8, PRSS56, PXDN, SH3PXD2B, SIX6, SLC4A4, TEK, WDR36
10	Мікрофтальмія, анофтальмія, колобома (MAC) і дисгенезія переднього сегмента / Invitae Microphthalmia, Anophthalmia, Coloboma (MAC) and Anterior Segment Dysgenesis Panel - 81 ген	ABCB6, ADAMTS18, ALDH1A3, ALX1, ASPH, BCOR, BMP4, BMP7, C12orf57, CDON, CHD7, COL4A1, CPAMD8, CRYAA, CRYBA4, CYP1B1, DCDC1, ELP4, ERCC2, ERCC5, ERCC6, FAT1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FZD5, GDF6, GJA1, GRIP1, HCCS, HESX1, HMGB3, HMX1, ITPR1, KERA, MAB21L2, MFRP, MIR204, MITF, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, PITX3, PORCN, PRDM5, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, RERE, SALL2, SHH, SIX3, SIX6, SLC38A8, SMCHD1, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, UBE3B, VAX1, VSX1, VSX2, WNT2B, YAP1, ZDBF2, ZIC2

11	Спадкові захворювання сітківки / Invitae Inherited Retinal Disorders Panel - 330 генів	ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADAMTSL4, ADGRA3, ADGRV1, ADIPOR1, AGBL5, AHI1, AHR, AIPL1, ALMS1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ASRGL1, ATF6, ATOH7, B9D1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C10orf11, C12orf65, C1QTNF5, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CHM, CIB2, CISD2, CLCC1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPLANE1, CRB1, CRX, CSPP1, CTNNA1, CTSD, CWC27, CYP4V2, DHDDS, DHX32, DHX38, DNAJC17, DRAM2, DSCAML1, DTHD1, EFEMP1, ELOVL4, EMC1, ERCC6, EXOSC2, EYS, FAM161A, FBLN5, FLVCR1, FRMD7, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GNS, GPR143, GPR179, GPR45, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS, HGSNAT, HK1, HMCN1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IFT80, IFT81, IFT88, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIAA0586, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, LYST, LZTFL1, MAK, MAPKAPK3, MERTK, MFN2, MFRP, MFSD8, MIR204, MKKS, MKS1, MPDZ, MTPAP, MTTP, MYO7A, NAGLU, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OR2W3, OTX2, P3H2, PAX2, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POC5, POMGNT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP1, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGR (ORF15), RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCLT1, SDCCAG8, SEMA4A, SGSH, SIX6, SLC24A1, SLC24A5, SLC45A2, SLC7A14, SNRNP200, SPATA7, SPP2, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMED7, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WDR34, WFS1, WHRN, ZNF408, ZNF423, ZNF513
12	Синдром Барде-Бідля / Invitae Bardet-Biedl Syndrome Panel - 28 генів	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CEP164, CEP19, CEP290, FBN3, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, SCLT1, SDCCAG8, TRAPPC3, TRIM32, TTC8, WDPCP
13	Катаракта / Invitae Cataracts Panel - 107 гена	ABCA3, ABCB6, ABHD12, ADAMTS18, ADAMTSL4, AGK, ALDH18A1, BCOR, BEST1, BFSP1, BFSP2, CHMP4B, CLN3, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXC1, FOXE3, FTL, FYCO1, FZD4, GALK1, GALT, GCNT2, GFER, GJA1, GJA3, GJA8, HMX1, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, MAF, MIP, MIR184, MYH9, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PEX10, PEX11B, PEX16, PEX2, PEX7, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RDH11, RECQL4, RGS6, RNLS, RRAGA, SC5D, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TFAP2A, TMEM70, UNC45B, VIM, VSX2, WDR87, WFS1, WRN, XYLT2, CHMP4B, CRYGB, LIM2, VIM
14	Мікрофтальмія, анофтальмія, колобома (MAC) і дисгенезія переднього сегмента / Invitae Microphthalmia, Anophthalmia, Coloboma (MAC) and Anterior Segment Dysgenesis Panel - 81 ген	ABCB6, ADAMTS18, ALDH1A3, ALX1, ASPH, BCOR, BMP4, BMP7, C12orf57, CDON, CHD7, COL4A1, CPAMD8, CRYAA, CRYBA4, CYP1B1, DCDC1, ELP4, ERCC2, ERCC5, ERCC6, FAT1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FZD5, GDF6, GJA1, GRIP1, HCCS, HESX1, HMGB3, HMX1, ITPR1, KERA, MAB21L2, MFRP, MIR204, MITF, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, PITX3, PORCN, PRDM5, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, RERE, SALL2, SHH, SIX3, SIX6, SLC38A8, SMCHD1, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, UBE3B, VAX1, VSX1, VSX2, WNT2B, YAP1, ZDBF2, ZIC2