Клінічна область Порушення скелету

	Панелі:	Гени:
1	Синдроми надмірного росту / Invitae Overgrowth Syndromes Panel - 53 гена	ABCC9, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD4, CHD8, CUL4B, DICER1, DIS3L2, DNMT3A, EED, EZH2, GFAP, GLI3, GNAS, GPC3, HEPACAM, HERC1, KPTN, L1CAM, MED12, MLC1, MPDZ, MTOR, NFIA, NFIX, NONO, NPR2, NSD1, OFD1, PDGFRB, PHF21A, PIK3R2, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RASA1, RIN2, RNF125, SETD2, STRADA, SYN1, TBC1D7, TCF20, UPF3B, ZBTB20
2	Синдроми надмірного росту та макроцефалії / Invitae Overgrowth and Macrocephaly Syndromes Panel - 26 генів	AKT2, AKT3, CDKN1C, CUL4B, DIS3L2, DNMT3A, EZH2, GLI3, GPC3, KPTN, MED12, MTOR, NF1, NFIX, NPR2, NSD1, PHF6, PIK3R2, PTEN, SETD2, SPRED1, DICER1, EED, PDGFRB, RNF125, UPF3B
3	Синдром Сімпсона-Голабі-Бехмеля / Invitae Simpson-Golabi-Behmel Syndrome Test - 1 ген	GPC3
4	Синдром Вівера / Invitae Weaver Syndrome Test - 1 ген	EZH2
5	Мікроцефальна примордіальна карликовість і синдром Секеля / Invitae Microcephalic Primordial Dwarfism and Seckel Syndrome Panel - 38 генів	ATR, ATRIP, CDC45, CDC6, CDK5RAP2, CDT1, CENPJ, CEP135, CEP152, CEP63, CEP97, CRIPT, DNA2, DNMT3A, DONSON, GMNN, LARP7, LIG4, MCM5, MCPH1, NIN, NSMCE2, ORC1, ORC4, ORC6, PCNT, PLK4, POC1A, RBBP8, RNU4ATAC, RTTN, SRCAP, TRAIP, TUBGCP4, TUBGCP6, UBE3B, WDR4, XRCC4
6	Синдром Адамса-Олівера / Invitae Invitae Adams-Oliver Syndrome Panel - 8 генів	ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1
7	Спондилокостальний дизостоз / Invitae Spondylocostal Dysostosis Panel - 8 генів	DLL1, DLL3, DMRT2, HES7, LFNG, MESP2, RIPPLY2, TBX6
8	Вади розвитку кінцівок і пальців / Invitae Limb and Digital Malformations Panel - 177 генів	ACVR1, ADAMTS10, ADAMTS17, AFF4, AHI1, ANKRD11, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, B3GLCT, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP2, BMP4, BMPR1B, BTRC, C2CD3, CACNA1C, CC2D2A, CCNQ, CDH3, CEP104, CEP120, CEP290, CEP41, CHSY1, CHUK, CKAP2L, CPLANE1, CREBBP, CSPP1, DDX59, DHCR7, DHODH, DLL4, DLX5, DLX6, DOCK6, DPF2, DVL1, DVL3, DYNC1I1, EOGT, EP300, ESCO2, EVC, EVC2, FAT1, FBLN1, FBN1, FBXW4, FGF10, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FMN1, FRAS1, FREM2, FZD2, GDF5, GDF6, GJA1, GLI2, GLI3, GNAS, GSC, HDAC4, HDAC8, HOXA13, HOXD13, IFT57, IHH, INPP5E, KDM6A, KIAA0586, KIF7, KMT2A, KMT2D, LMBR1, LRP4, LTBP2, LTBP3, MAP3K20, MEGF8, MGP, MKKS, MKS1, MRE11, MYCN, NECTIN1, NECTIN4, NIPBL, NOG, NOTCH1, NPHP1, NPHP3, NSDHL, NXN, OFD1, PDE3A, PDE4D, PDE6D, PGM3, PHF6, PIGV, PITX1, POLR1A, PORCN, PRKAR1A, PRMT7, PTDSS1, PTHLH, RAB23, RAD21, RBM8A, RBPJ, RECQL4, ROR2, RPGRIP1L, SALL1, SALL4, SC5D, SDCCAG8, SF3B4, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SOST, SOX11, SOX9, TBX15, TBX3, TBX5, TCTN1, TCTN2, TCTN3, TGDS, THPO, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TP63, TRIM32, TRPS1, TRPV4, TTC21B, TTC8, VAC14, WDPCP, WNT10B, WNT3, WNT5A, WNT7A, ZNF423, ZSWIM6
9	Синдром Стіклера / Invitae Stickler Syndrome Panel - 9 генів	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, LRP2, VCAN
10	Х-зчеплена гіпофосфатемія / Invitae X-Linked Hypophosphatemia Test - 1 ген	PHEX
11	Гіпофосфатемія / Invitae Hypophosphatemia Panel - 17 генів	ALPL, CLCN5, CTNS, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, GNAS, OCRL, PHEX, SLC34A1, SLC34A3, VDR

12	Розлади скелета / Invitae Skeletal Disorders Panel - 358 генів	ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, AFF4, AGA, AGPS, AIFM1, ALPL, AMER1, ANKH, ANO5, ARCN1, ARSB, ARSE, ASCC1, ASPM, ATR, ATRIP, B3GALT6, B3GAT3, B4GALT7, BGN, BMP1, BMP2, BMPER, BMPR1B, C2CD3, CA2, CANT1, CASR, CCDC8, CDC45, CDC6, CDK5RAP2, CDKN1C, CDT1, CENPJ, CEP120, CEP135, CEP152, CEP63, CEP97, CFAP410, CHST14, CHST3, CHUK, CLCN5, CLCN7, COG1, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CRIPT, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTNS, CTSA, CTSK, CUL7, CWC27, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DIP2C, DLL1, DLL3, DLX3, DMP1, DMRT2, DNA2, DNMT3A, DONSON, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, EXOC6B, EXOSC2, EXT1, EXT2, EXTL3, FAH, FAM111A, FAM20C, FAM46A, FAR1, FAT4, FBN1, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FTO, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GHRHR, GHSR, GJA1, GLB1, GMNN, GNAS, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GSC, GUSB, GZF1, HES7, HGSNAT, HPGD, HSPG2, HYAL1, IARS2, ICK, IDS, IDUA, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IGF1, IGF2, IHH, IMPAD1, INPPL1, INTU, JAG1, KAT6B, KIAA0586, KIAA0753, KIF22, KL, KMT2A, LARP7, LBR, LEMD3, LFNG, LIFR, LIG4, LMNA, LMX1B, LONP1, LOXL3, LRP4, LRP5, LRRK1, LTBP2, LTBP3, MAFB, MAN2B1, MANBA, MAP3K7, MATN3, MBTPS1, MBTPS2, MCM5, MCPH1, MEOX1, MESDC2, MESP2, MGP, MMP13, MMP14, MMP2, MMP9, MNX1, MSX2, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NIN, NKX3-2, NOG, NOTCH2, NPPC, NPR2, NPR3, NSDHL, NSMCE2, NTRK1, NXN, OBSL1, OCRL, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PCGF2, PCNT, PCYT1A, PDE4D, PEX5, PEX7, PGM3, PHEX, PIK3C2A, PISD, PKDCC, PLK4, PLOD2, PLS3, POC1A, POLR1A, POP1, POR, PPIB, PPP3CA, PRKAR1A, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB33B, RBBP8, RECQL4, RIPPLY2, RMRP, RNU4ATAC, ROR2, RSPO2, RSPRY1, RTTN, RUNX2, SC5D, SEC24D, SERPINF1, SERPINH1, SETBP1, SFRP4, SGMS2, SGSH, SH3PXD2B, SLC17A5, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SLCO5A1, SMAD4, SMARCAL1, SNRPB, SNX10, SOX9, SP7, SPARC, SQSTM1, SRCAP, SUCO, SULF1, SUMF1, TAB2, TAPT1, TBCE, TBX15, TBX3, TBX5, TBX6, TBXAS1, TCIRG1, TCTEX1D2, TCTN3, TGFB1, TMEM165, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TRAF3IP1, TRAIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRIP4, TRMT10A, TRPS1, TRPV4, TTC21B, TUBGCP4, TUBGCP6, TYROBP, UBE3B, VAC14, VDR, VPS33A, WDR19, WDR34, WDR35, WDR4, WDR60, WISP3, WNT1, WNT3, WNT3A, WNT5A, XRCC4, XYLT1, XYLT2, ZMPSTE24, ZNF687
13	Синдром Антлі-Бікслера / Invitae Antley-Bixler Syndrome Test - 2 гена	POR, FGFR2
14	Точкова хондродисплазія, пов’язана з ARSE / Invitae ARSE-Related Chondrodysplasia Punctata Test - 2 гена	ARSE, NSDHL
15	Кампомелічна дисплазія / Invitae Campomelic Dysplasia Test - 1 ген	SOX9
16	Краніосиностоз / Invitae Craniosynostosis Panel - 67 генів	ALPL, ASXL1, B3GAT3, CD96, CDC45, CDT1, COLEC11, CYP26B1, EFNA4, EFNB1, ERF, ESCO2, FBN1, FGF9, FGFR1, FGFR2, FGFR3, FREM1, GLI3, GPC3, IFT122, IFT140, IFT43, IGF1R, IL11RA, KAT6A, KAT6B, MASP1, MEGF8, MSX2, NFIA, ORC1, ORC4, ORC6, P4HB, PHEX, POR, PPP3CA, RAB23, RECQL4, RSPRY1, RUNX2, SCARF2, SEC24D, SIX2, SKI, SLC25A24, SMAD2, SMAD3, SMAD6, SOX6, SPECC1L, STAT3, TCF12, TCOF1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMCO1, TWIST1, WDR19, WDR35, ZEB2, ZIC1, MASP1, TCOF1
17	Акрофаціальний дизостоз Елліса-ван Кревельда та Вейєрса / Invitae Ellis-van Creveld and Weyers Acrofacial Dysostosis Panel - 2 гена	EVC, EVC2
18	Спадкові множинні остеохондроми / Invitae Hereditary Multiple Osteochondromas Panel - 3 гена	EXT1, EXT2, TRPS1
19	Недосконалий остеогенез і крихкість кісток / Invitae Osteogenesis Imperfecta and Bone Fragility Panel - 67 генів	ALPL, ANO5, ASCC1, B3GAT3, B4GALT7, BMP1, CA2, CLCN5, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CTNS, CTSK, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FAM46A, FGF23, FGFR1, FKBP10, GNAS, GORAB, IFITM5, LRP5, LRRK1, MBTPS2, MESDC2, NBAS, NOTCH2, NTRK1, OCRL, OSTM1, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SFRP4, SGMS2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SNX10, SP7, SPARC, SUCO, TAPT1, TCIRG1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TRIP4, VDR, WNT1, WNT3A, XYLT2
20	Перинатальна летальна скелетна дисплазія та скелетні ціліопатії / Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel - 72 генів	AGPS, ALPL, ASCC1, CEP120, CFAP410, CHUK, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CREB3L1, CRTAP, CSPP1, DLL1, DLL3, DMRT2, DYNC2H1, DYNC2LI1, EBP, EVC, EVC2, FAM111A, FGFR2, FGFR3, GNPAT, GPX4, HES7, HSPG2, ICK, IFT122, IFT140, IFT172, IFT43, IFT52, IFT74, IFT80, IFT81, IMPAD1, INPPL1, INTU, KIAA0586, KIAA0753, LBR, LFNG, LRP5, MBTPS2, MESP2, NEK1, P3H1, PAM16, PEX5, PEX7, PPIB, RIPPLY2, SLC26A2, SLC35D1, SOX9, TBX6, TCTEX1D2, TMEM38B, TRAF3IP1, TRIP11, TRIP4, TRPV4, TTC21B, WDR19, WDR34, WDR35, WDR60, WNT1, WNT3
21	Трихорінофаланговий синдром / Invitae Trichorhinophalangeal Syndrome Panel - 2 гена	EXT1, TRPS1
22	Розлади сполучної тканини / Invitae Connective Tissue Disorders Panel - 92 гена	ABCC6, ABL1, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ARIH1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B4GALT7, BGN, C1S, CBS, CHST14, CHST3, COG7, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, CRTAP, DCHS1, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, FLNB, FOXE3, GGCX, GORAB, HCN4, LEMD3, LOX, LOXL3, LTBP2, LTBP3, LTBP4, LZTS1, MAT2A, MED12, MFAP5, MYH11, MYLK, NOG, NOTCH1, P3H1, PKD2, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC26A2, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SPARC, TALDO1, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, ZNF469