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Клінічна область Неврологія

	Панелі:	Гени:
1	Комплексні нейропатії / Invitae Comprehensive Neuropathies Panel - 111 генів	AARS, AIFM1, APOA1, ASAH1, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CHCHD10, COX6A1, CYP27A1, CYP7B1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EXOSC9, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GLA, GNB4, GSN, HARS, HEXA, HINT1, HMBS, HSPB1, HSPB8, IGHMBP2, INF2, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, POLG, POLG2, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN11A, SCN9A, SEPT9, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SMN2, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, VRK1, WNK1, YARS, ARHGEF10, CCT5, HSPB3, LAS1L, MICAL1, SCN10A, SGPL1, SLC25A21, SLC52A1
2	Хвороба Шарко-Марі-Тута / Invitae Charcot-Marie-Tooth Disease Comprehensive Panel - 60 генів	AARS, AIFM1, ATP1A1, BAG3, BSCL2, COX6A1, DHTKD1, DNAJB2, DNM2, DRP2, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KIF5A, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NDRG1, NEFH, NEFL, PDK3, PLEKHG5, PMP2, PMP22, PRPS1, PRX, RAB7A, SBF1, SBF2, SH3TC2, SLC25A46, SPG11, SURF1, TFG, TRIM2, TRPV4, YARS, ARHGEF10, MICAL1, SGPL1
3	Нейропатія дрібних волокон / Invitae Small Fiber Neuropathy Test - 2 гена	SCN9A, SCN10A
4	Спадкова сенсорна та вегетативна нейропатія / Invitae Hereditary Sensory and Autonomic Neuropathy Panel - 16 генів	ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN11A, SCN9A, SPTLC1, SPTLC2, WNK1, CCT5
5	Спадкова моторна нейропатія / Invitae Hereditary Motor Neuropathy Panel - 30 генів	ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC9, FBXO38, GARS, HEXA, HINT1, HSPB1, HSPB8, IGHMBP2, MORC2, PLEKHG5, REEP1, SIGMAR1, SLC5A7, SMN1, SMN2, TRPV4, UBA1, VAPB, VRK1, HSPB3, LAS1L, SLC25A21
6	Спінальна м'язова атрофія / Invitae Spinal Muscular Atrophy Panel - 2 гена	SMN1, SMN2
7	Спадкова спастична параплегія / Invitae Hereditary Spastic Paraplegia Comprehensive Panel - 78 генів	ABCD1, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12orf65, CAPN1, CPT1C, CYP27A1, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HACE1, HEXA, HSPD1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, NIPA1, NKX6-2, NT5C2, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, UCHL1, VAMP1, WASHC5, ZFYVE26, ADGRB2, AMPD2, ARSI, ATP2B4, C19orf12, CCT5, DSTYK, EXOSC3, IBA57, KLC2, PGAP1, SLC33A1, USP8, VPS37A, ZFR, ZFYVE27
8	Сімейна дизавтономія / Invitae Familial Dysautonomia Test - 1 ген	ELP1
9	Комплексна дистонія / Invitae Dystonia Comprehensive Panel - 41 ген	ACTB, ADCY5, ANO3, ATP1A3, ATP7B, BCAP31, CIZ1, COL6A3, CYP27A1, GCH1, GNAL, GNAO1, HEXA, HPCA, KCNMA1, KCTD17, KMT2B, MECR, PANK2, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, SGCE, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SPR, TH, THAP1, TOR1A, TUBB4A, VAC14, VPS13A, VPS13D, XPR1, DRD2, MED20, TOR1AIP1
10	Спадкова хвороба Паркінсона та паркінсонізм / Invitae Hereditary Parkinson Disease and Parkinsonism Panel - 29 ген	ATP13A2, ATP7B, CHCHD2, CSF1R, DCTN1, DNAJC6, FBXO7, GBA, GCH1, LRRK2, PARK7, PDE8B, PINK1, PLA2G6, PRKN, PRKRA, RAB39B, SLC6A3, SNCA, SPR, SYNJ1, TH, TMEM230, VPS13C, VPS35, XPR1, MAPT, PODXL, UCHL1

11	Спадковий бічний аміотрофічний склероз, лобно-скронева деменція та хвороба Альцгеймера / Invitae Hereditary Amyotrophic Lateral Sclerosis, Frontotemporal Dementia and Alzheimer Disease Panel - 42 гена	ALS2, ANG, ANXA11, APP, CHCHD10, CHMP2B, DCTN1, ERBB4, FUS, GRN, HEXA, HNRNPA2B1, ITM2B, KIF5A, MAPT, OPTN, PFN1, PRNP, PSEN1, PSEN2, SETX, SNCA, SOD1, SORL1, SPG11, SQSTM1, TARDBP, TBK1, TFG, TREM2, UBQLN2, VAPB, VCP, ATP13A2, DDHD1, ERLIN1, FIG4, LRRK2, MATR3, NEFH, SIGMAR1, TIA1
12	Бічний аміотрофічний склероз / Invitae Amyotrophic Lateral Sclerosis Panel - 30 генів	ALS2, ANG, ANXA11, CHCHD10, DCTN1, ERBB4, FUS, HEXA, KIF5A, OPTN, PFN1, SETX, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TFG, UBQLN2, VAPB, VCP, ATP13A2, CHMP2B, DDHD1, ERLIN1, FIG4, MATR3, NEFH, SIGMAR1, TIA1
13	Лобно-скронева деменція / Invitae Frontotemporal Dementia Panel - 15 генів	CHCHD10, CHMP2B, DCTN1, FUS, GRN, HNRNPA2B1, MAPT, SQSTM1, TARDBP, TBK1, TREM2, UBQLN2, VCP, LRRK2, PSEN1
14	Спадкова пріонова хвороба / Invitae Hereditary Prion Disease Test - 1 ген	PRNP
15	Сімейна геміплегічна мігрень / Invitae Familial Hemiplegic Migraine Panel - 8 генів	ATP1A2, ATP1A3, CACNA1A, PRRT2, SCN1A, SLC1A3, SLC2A1, KCNK18
16	Спадкова хвороба дрібних судин головного мозку / Invitae Hereditary Cerebral Small Vessel Disease Panel - 10 генів	APP, CBS, COL4A1, COL4A2, CST3, FOXC1, GLA, HTRA1, NOTCH3, TREX1
17	Септооптична дисплазія / Invitae Septo-optic Dysplasia Panel - 8 генів	GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3

18	Лейкодистрофія та генетична лейкоенцефалопатія / Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel - 792 гена	AARS, AARS2, ABAT, ABCA1, ABCD1, ABCD4, ABHD5, ACADS, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACSF3, ACTA2, ACTB, ACY1, ADAR, ADGRG1, ADK, ADNP, ADSL, AGA, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALG12, ALG13, ALG2, ALG6, ALG9, AMACR, AMPD2, AMT, ANK3, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOPT1, ARCN1, ARFGEF2, ARHGAP31, ARHGEF9, ARNT2, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL2, ATP13A2, ATP5A1, ATP6AP2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, ATRX, AUH, B3GALNT2, B4GALNT1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCL11B, BCS1L, BICD2, BMP4, BOLA3, BPTF, BRAT1, BTD, C12orf57, C12orf65, C19orf12, C2CD3, CACNA1A, CACNA1E, CARS2, CBS, CC2D2A, CCDC88A, CDC42, CDKL5, CEP290, CHMP1A, CLCN2, CLCN4, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNNM2, CNOT1, CNTNAP1, CNTNAP2, COA7, COASY, COG7, COL18A1, COL3A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CPLANE1, CPLX1, CPS1, CRAT, CREBBP, CRIPT, CRLF1, CSF1R, CSPP1, CTBP1, CTC1, CTDP1, CTNS, CTSA, CTSD, CUL4B, CYB5R3, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DGUOK, DHCR24, DHFR, DHX37, DLD, DLL4, DMXL2, DNM1L, DNM2, DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DPYS, DYRK1A, EARS2, EDNRB, EHMT1, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL1, ELOVL4, ENTPD1, EOGT, EPG5, EPRS, ERCC1, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, EXOSC2, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGFRL1, FH, FIG4, FKRP, FKTN, FLVCR2, FOLR1, FOXC1, FOXG1, FOXRED1, FUCA1, FUK, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GALC, GALT, GAN, GATAD2B, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GM2A, GMNN, GNAO1, GNS, GOT2, GPHN, GRIN1, GRM7, GTF2H5, GTPBP2, HACE1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HLCS, HMGCL, HNRNPU, HSD17B10, HSD17B4, HSPD1, IARS, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, INPP5E, IREB2, ISCA1, ISCA2, ITPA, ITPR1, IVD, JAM3, KARS, KAT6B, KATNB1, KCNJ10, KCNJ2, KCNMA1, KCNT1, KCTD7, KDM1A, KIAA0556, KIAA0586, KIAA0753, KIDINS220, KIF1A, KIF1C, KLHL7, KMT2E, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LARGE1, LARS2, LETM1, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM7, MAG, MAGEL2, MAN2B1, MANBA, MARS2, MAST1, MAT1A, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MED17, MED25, MEF2C, MFSD2A, MFSD8, MGP, MICU1, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MORC2, MPLKIP, MPV17, MPZ, MRPL12, MRPL44, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, NACC1, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NFE2L2, NFU1, NGLY1, NKX6-2, NOTCH1, NPC1, NPC2, NPHP1, NRXN1, NSUN3, NT5C2, NTRK2, NUBPL, NUP62, OAT, OCRL, OPA1, OPA3, OSGEP, OTC, PACS1, PAFAH1B1, PAH, PANK2, PARS2, PAX1, PC, PCCA, PCCB, PCDH12, PCGF2, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHGDH, PHYH, PIGA, PIGB, PIGG, PIGL, PIGM, PIGN, PIGP, PIGQ, PIGT, PIGU, PIGV, PIK3C2A, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLP1, PMM2, PMP22, PNKP, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPP1R15B, PPP2R1A, PPP3CA, PPT1, PRF1, PRKDC, PRODH, PROSC, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, PUS3, PYCR2, QARS, QRSL1, RAB11B, RAB3GAP1, RAB3GAP2, RARS, RARS2, RBPJ, REPS1, RERE, RHOBTB2, RIN2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIP1L, RPIA, RPS6KC1, RRM2B, RTTN, RXYLT1, SAMD9L, SAMHD1, SCN3A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SETBP1, SGSH, SH3TC2, SHOC2, SHPK, SLC12A2, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC35A2, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A8, SLC6A9, SLC9A1, SLC9A6, SMC1A, SNAP29, SNIP1, SNORD118, SNRPB, SNX14, SOD1, SON, SOX10, SOX2, SPART, SPAST, SPATA5, SPG11, SPG7, SPTAN1, SQSTM1, SRD5A3, SSR4, ST3GAL5, STAG2, STAMBP, STAT1, STAT2, STN1, STRADA, STX11, STXBP1, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYNE1, SYNJ1, TACO1, TAF2, TANGO2, TARS2, TBC1D24, TBCD, TBCE, TBCK, TBX1, TCF4, TCTN2, TIMM50,

TIMMDC1, TM4SF20, TMEM106B, TMEM126B, TMEM165, TMEM216, TMEM67, TMEM70, TMTC3, TOE1, TP53RK, TPI1, TPK1, TRAPPC11, TRAPPC9, TREX1, TRMT10A, TRMT5, TSC1, TSC2, TSEN54, TTC19, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUFM, TWNK, TXN2, TYMP, UBA5, UBE2A, UBE3A, UFM1, UNC13D, UPB1, USP7, VARS2, VPS11, VPS13D, VPS33A, WARS2, WDR45, WDR73, WHSC1, WWOX, YARS, YME1L1, ZEB2, ZFYVE26, ZIC1, ZNF335, ALDH7A1, APP, CHMP2B, CP, CTSF, DNAJC5, DNMT1, FTL, GBE1, GRN, HTRA1, KIAA1161, KIF5A, LMNB1, MAPT, NOTCH3, PDGFB, PDGFRB, PDYN, PHAX, PINK1, PRNP, PSEN1, RNF216, SCP2, SLC20A2, TREM2, TYROBP, VCP, VPS13A, XK, XPR1

19	Спектр розладів дитячого церебрального паралічу / Invitae Cerebral Palsy Spectrum Disorders Panel - 424 гена	ABAT, ABCD1, ACADM, ACADVL, ACAT1, ACBD5, ACOX1, ACTB, ADAR, ADCY5, ADD3, ADNP, ADSL, AFG3L2, AGAP1, AHDC1, AHI1, AKT3, ALDH18A1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, ALG3, ALS2, AMACR, AMPD2, AMT, ANO3, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARHGEF9, ARL6IP1, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ATAD1, ATL1, ATM, ATP13A2, ATP1A3, ATP7A, ATP7B, ATP8A2, ATRX, AUH, AUTS2, B4GALNT1, BCAP31, BCKDHA, BCKDHB, BICD2, BSCL2, BTD, C12orf65, C19orf12, CACNA1A, CACNA1G, CAMTA1, CAPN1, CASK, CBS, CCDC88C, CCT5, CDKL5, CEP290, CHD8, CHRNA1, CIZ1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLTC, COASY, COL4A1, COL4A2, COL6A3, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ9, CPS1, CPT1C, CREBBP, CTBP1, CTNNB1, CTSD, CYP27A1, CYP2U1, CYP7B1, DARS, DARS2, DBH, DBT, DCAF17, DDC, DDHD1, DDHD2, DDX3X, DGKZ, DHDDS, DHFR, DLAT, DLD, DMD, DNAJC12, DNM2, DPAGT1, DYNC1H1, DYRK1A, EEF2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, ENTPD1, EPHA4, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, FA2H, FAM126A, FARS2, FAT2, FGF12, FGF14, FH, FOLR1, FOXG1, FRRS1L, FTL, FUCA1, GABRA2, GAD1, GALC, GAMT, GATM, GBA, GBA2, GCDH, GCH1, GFAP, GJC2, GLB1, GLDC, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GNS, GPHN, GPR88, GRID2, GRIN1, GRIN2B, GRM1, HACE1, HESX1, HEXA, HEXB, HGSNAT, HLCS, HMGCL, HPCA, HPRT1, HSD17B10, HSD17B4, HSPD1, IBA57, IFIH1, IQSEC2, IREB2, ITPA, ITPR1, KANK1, KAT6A, KCNA2, KCNC3, KCNJ6, KCNMA1, KCNQ2, KCNT1, KCTD17, KCTD7, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KMT2A, KMT2B, KMT2C, L1CAM, L2HGDH, LAMA2, LIAS, LMBRD1, MAG, MAOA, MAP2K1, MARS2, MAST1, MCCC1, MCCC2, MCEE, MECP2, MECR, MFSD8, MICU1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOCS3, MPC1, MTHFR, MTOR, MTPAP, MTR, MTRR, MTTP, MUT, NAA10, NAA35, NAGLU, NAGS, NBAS, NGLY1, NIPA1, NKX2-1, NPC1, NPC2, NPHP1, NT5C2, NUS1, OTC, PAFAH1B1, PAH, PAK3, PALM, PANK2, PCBD1, PCCA, PCCB, PCDH12, PDE10A, PDE2A, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PHIP, PIGN, PIGT, PLA2G6, PLD3, PLP1, PLXNA2, PMM2, PNKD, PNP, PNPLA6, PNPO, POLG, POLR3A, PPT1, PRKRA, PROSC, PRRT2, PRUNE1, PSAT1, PSPH, PTPN11, PTS, PURA, QDPR, RAB3GAP1, RAB3GAP2, RANBP2, REEP1, REEP2, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RTN2, SACS, SAMHD1, SATB2, SCN1A, SCN2A, SCN3A, SCN8A, SETD5, SGCE, SGSH, SHH, SIL1, SIX3, SLC16A2, SLC17A5, SLC18A2, SLC19A3, SLC1A4, SLC25A15, SLC25A22, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SON, SPART, SPAST, SPATA5, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, ST3GAL5, STAMBP, STUB1, STXBP1, SUCLA2, SUCLG1, SUOX, SURF1, SYNGAP1, TAF1, TANGO2, TBC1D24, TBCK, TBL1XR1, TCF4, TECPR2, TFG, TGIF1, TGM6, TH, THAP1, TMEM240, TMEM67, TOR1A, TREX1, TRPC3, TSEN54, TTBK2, TTPA, TUBA1A, TUBB2A, TUBB2B, TUBB3, TUBB4A, UBE3A, UCHL1, VAC14, VAMP1, VPS13A, VPS13D, VPS37A, WARS2, WASHC5, WDR45, WDR62, ZBTB18, ZC4H2, ZEB2, ZFR, ZFYVE26, ZIC1, ZIC2, ZIC4
20	Вади розвитку головного мозку / Invitae Brain Malformations Panel - 163 гена	ACTB, ACTG1, ADGRG1, ADNP, AHDC1, AKT3, AMPD2, APC2, ARFGEF2, ARID1A, ARID1B, ARX, ASNS, ASPM, ATP6V0A2, B3GALNT2, B4GAT1, BMP4, C19orf12, CASK, CCM2, CCND2, CDK13, CDK5, CDON, CHMP1A, CIT, CNOT1, CNOT3, COASY, COL18A1, COL3A1, COL4A1, COL4A2, CP, CRADD, CUL4B, DAG1, DCHS1, DCX, DIAPH1, DISP1, DLL1, DMXL2, DPF2, DYNC1H1, EIF2AK2, EMC1, EML1, ERMARD, EXOSC3, FA2H, FAT4, FGFR1, FIG4, FKRP, FKTN, FLNA, FOXA2, FTL, GAS1, GLI2, GMPPB, GPSM2, GRIN2B, HIVEP2, IER3IP1, IQSEC2, ISPD, KATNB1, KCNMA1, KIF11, KIF1BP, KIF2A, KIF7, KMT2E, KRIT1, L1CAM, LAMA1, LAMB1, LAMC3, LARGE1, LRP2, MACF1, MED12, MED17, MFSD2A, MRE11, NDE1, NEDD4L, NPRL3, OCLN, OPHN1, PAFAH1B1, PANK2, PDCD10, PHGDH, PIK3R2, PLA2G6, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R12A, PTCH1, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RARS2, RBM10, RELN, RERE, RTTN, RXYLT1, SEPSECS, SHH, SIN3A, SIX3, SLC25A19, SMARCA4, SMARCB1, SMARCE1, SMC1A, SNAP29, SON, SOX2, SRD5A3, STAG2, STAMBP, STIL, TBC1D20, TGIF1, TMTC3, TOE1, TRRAP, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, UBE3B, USP7, USP9X, VLDLR, VPS13A, VRK1, WDR45, WDR62, YWHAE, ZBTB18, ZBTB20, ZIC2, ZMIZ1

21	Церебрально-лицевий синдром Барайцера-Вінтера / Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel - 2 гена	ACTB, ACTG1
22	Церебральні кавернозні мальформації / Invitae Cerebral Cavernous Malformations Panel - 3 гена	CCM2, C39KRIT1, PDCD10
23	Церебральні кавернозні мальформації / Invitae Holoprosencephaly Panel - 10 генів	FGFR1, GLI2, SHH, SIX3, TGIF1, ZIC2, CDON, FOXH1, NODAL, PTCH1
24	Синдроми Ретта і Ангельмана та пов'язані з ними розлади / Invitae Rett and Angelman Syndromes and Related Disorders Panel - 40 генів	ADSL, ALDH5A1, ARX, ATRX, CAMK2B, CDKL5, CLTC, CNTNAP2, CTNNB1, DDX3X, DYRK1A, EHMT1, FOLR1, FOXG1, GABBR2, GRIA3, GRIN2A, GRIN2B, HDAC8, IQSEC2, KANSL1, KCNA2, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN2A, SCN8A, SLC6A1, SLC9A6, SMC1A, STXBP1, SYNGAP1, TBL1XR1, TCF4, UBE3A, WDR45, ZEB2
25	Комплекс туберозного склерозу / Invitae Tuberous Sclerosis Complex Panel - 2 гена	TSC1, TSC2
26	Епілепсія / Invitae Epilepsy Panel - 320 генів	AARS, ABAT, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG1, ALG12, ALG13, ALG6, AMACR, AMT, AP2M1, AP3B2, ARG1, ARHGEF9, ARSA, ARX, ASAH1, ASNS, ATAD1, ATP1A1, ATP1A2, ATP1A3, ATP6AP2, ATP7A, ATRX, BRAT1, C12orf57, CACNA1A, CACNA1B, CACNA1E, CACNA2D2, CAD, CAMK2B, CARS2, CASK, CCDC88A, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CLCN6, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLTC, CNTN2, CNTNAP2, COG5, COL18A1, CSTB, CTNNB1, CTSD, CYFIP2, CYP27A1, DDC, DDX3X, DEAF1, DENND5A, DEPDC5, DHDDS, DHFR, DIAPH1, DMXL2, DNAJC5, DNM1, DNM1L, DOCK7, DYNC1H1, DYRK1A, ECHS1, EEF1A2, EHMT1, EMC1, EPM2A, FAR1, FARS2, FBXO11, FGF12, FLNA, FOLR1, FOXG1, FOXP1, FRRS1L, GABBR2, GABRA1, GABRA2, GABRB1, GABRB2, GABRB3, GABRG2, GAMT, GATAD2B, GATM, GCH1, GLDC, GLRA1, GLRB, GNAO1, GNB1, GOSR2, GPAA1, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, GRIN2D, GTPBP3, HCN1, HDAC8, HEXA, HNRNPU, IER3IP1, IFIH1, IQSEC2, ITPA, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCND2, KCNH1, KCNH2, KCNH5, KCNJ10, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD7, KIF1A, KIF2A, KIF5A, KPNA7, LAMC3, LGI1, LIAS, MBD5, MDH2, MECP2, MEF2C, MFSD8, MICAL1, MOCS1, MOCS2, MTOR, NACC1, NAGLU, NECAP1, NEDD4L, NEXMIF, NGLY1, NHLRC1, NPC1, NPC2, NPRL3, NRXN1, NTRK2, NUS1, PACS1, PACS2, PAFAH1B1, PCDH19, PCLO, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PIGA, PIGB, PIGG, PIGN, PIGO, PIGP, PIGQ, PIGV, PIGW, PLAA, PLCB1, PNKD, PNKP, PNPO, PNPT1, POLG, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PRICKLE1, PRIMA1, PROSC, PRRT2, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, QARS, QDPR, RAB11A, RAB11B, RAI1, RALA, RANBP2, RELN, RFT1, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGDI, RORB, RUSC2, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCP2, SERPINI1, SETBP1, SGCE, SGSH, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMC1A, SNAP25, SNX27, SPATA5, SPTAN1, ST3GAL3, ST3GAL5, STAG2, STRADA, STX1B, STXBP1, STXBP2, SUMF1, SUOX, SURF1, SYN1, SYNGAP1, SYNJ1, SZT2, TANGO2, TBC1D24, TBCK, TBL1XR1, TCF4, TH, TK2, TPK1, TREX1, TRIM8, TSC1, TSC2, TSFM, TUBB2A, UBA5, UBE3A, UNC80, WDR45, WWOX, YWHAG, ZDHHC9, ZEB2, ZSWIM6, ARHGEF15, CACNA1H, CERS1, FASN, GABRD, GUF1, IDH3A, JMJD1C, LMNB2, MOCS3, PIK3AP1, PRDM8, PRICKLE2, RBFOX1, RBFOX3, SCN5A, SNIP1, TUBA8

27	Порушення розвитку нервової системи / Invitae Neurodevelopmental Disorders (NDD) Panel - 241 ген	ACTB, ACTG1, ADNP, ADSL, AGA, AHDC1, ALDH5A1, ALDH7A1, AMER1, ANKRD11, AP1S2, ARG1, ARID1A, ARID1B, ARSA, ARX, ASNS, ASXL1, ATP1A3, ATP7A, ATRX, AUTS2, BCAP31, BRAF, BRAT1, BRD4, BRWD3, CACNA1A, CACNA1E, CAMK2B, CASK, CBL, CC2D2A, CDK13, CDKL5, CHD2, CHD7, CHD8, CLCN4, CLN2 (TPP1), CLN3, CLN5, CLN6, CLTC, CNTNAP2, COL4A1, CREBBP, CTNNB1, CUL3, DDC, DDX3X, DEAF1, DHCR7, DNM1L, DNMT3A, DOCK6, DPF2, DYNC1H1, DYRK1A, EEF1A2, EFTUD2, EHMT1, EP300, EZH2, FGD1, FOLR1, FOXG1, FOXP1, GABBR2, GABRB3, GABRG2, GALC, GAMT, GATAD2B, GATM, GLB1, GM2A, GNAO1, GNAS, GNS, GPC3, GRIA3, GRIN1, GRIN2A, GRIN2B, HDAC8, HEXA, HEXB, HGSNAT, HIVEP2, HNRNPK, HNRNPU, HRAS, HUWE1, IDS, IDUA, IGF1R, IL1RAPL1, IQSEC2, ITPR1, KANSL1, KAT6A, KAT6B, KCNA2, KCNB1, KCNH1, KCNQ2, KCNT1, KDM5C, KDM6A, KIF1A, KMT2A, KMT2B, KMT2D, KMT2E, KRAS, L1CAM, LZTR1, MAGEL2, MAN1B1, MAP2K1, MAP2K2, MBD5, MECP2, MED12, MED13L, MEF2C, MFSD8, MID1, MTOR, NAA10, NAA15, NAGLU, NALCN, NEXMIF, NF1, NFIA, NFIX, NGLY1, NHS, NIPBL, NONO, NPC1, NR2F1, NRAS, NRXN1, NSD1, NSUN2, OCRL, OPHN1, OTC, PACS1, PACS2, PAH, PCBD1, PCDH19, PDHA1, PGAP3, PHF21A, PHF6, PHIP, PLA2G6, PMM2, POLG, PPM1D, PPP1CB, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PTEN, PTPN11, PTS, PURA, QDPR, RAD21, RAF1, RAI1, RBM10, RIT1, RPS6KA3, SATB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SETBP1, SETD5, SGSH, SHOC2, SIN3A, SLC13A5, SLC16A2, SLC2A1, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SON, SOS1, SOS2, SOX11, SPAST, SPATA5, SPTAN1, STAG1, STXBP1, SURF1, SYNGAP1, TAF1, TBCK, TBL1XR1, TCF20, TCF4, TELO2, TRAPPC9, TRRAP, TSC1, TSC2, TUBA1A, UBE3A, UNC80, USP9X, VPS13B, WDR45, WWOX, ZBTB18, ZBTB20, ZC4H2, ZDHHC9, ZEB2, ZIC2, ZMIZ1, ZMYND11
28	Вроджений міастенічний синдром / Invitae Congenital Myasthenic Syndrome Panel - 25 генів	AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, MUSK, PREPL, RAPSN, SLC18A3, SLC5A7, SYT2, VAMP1
29	Схильність до злоякісної гіпертермії / Invitae Malignant Hyperthermia Susceptibility Panel - 3 гена	CACNA1S, RYR1, STAC3
30	Комплексна м'язова дистрофія / Invitae Comprehensive Muscular Dystrophy Panel - 53 гена	ANO5, B3GALNT2, B4GAT1, CAPN3, CAV3, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, GOSR2, HNRNPDL, ISPD, ITGA7, LAMA2, LARGE1, LMNA, MYOT, PLEC, PNPLA2, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TK2, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
31	Дистрофінопатії / Invitae Dystrophinopathies Test - 1 ген	DMD
32	М'язова дистрофія пояса кінцівок / Invitae Limb-Girdle Muscular Dystrophy Panel - 38 генів	ANO5, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DYSF, FKRP, FKTN, GAA, GMPPB, GOSR2, HNRNPDL, ISPD, LAMA2, LMNA, MYOT, PLEC, PNPLA2, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TK2, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
33	Комплексна міопатія / Invitae Comprehensive Myopathy Panel - 74 гена	ACTA1, ADSSL1, AMPD1, ANO5, ATP2A1, BAG3, BIN1, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FKBP14, FLNC, GNE, GYG1, GYS1, HACD1, HNRNPA2B1, ISCU, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMP2, LDB3, LMNA, LMOD3, MAP3K20, MATR3, MEGF10, MICU1, MTM1, MYH2, MYH7, MYL2, MYO18B, MYOT, MYPN, NEB, ORAI1, PYROXD1, RYR1, SCN4A, SELENON, SMPX, SPEG, SQSTM1, STAC3, STIM1, TAZ, TIA1, TK2, TNNT1, TPM2, TPM3, TTN, VCP, VMA21
34	Періодичний параліч / Invitae Periodic Paralysis Panel - 6 генів	ATP1A2, CACNA1S, KCNJ2, MCM3AP, RYR1, SCN4A
35	Міотонія та вроджена параміотонія / Invitae Myotonia and Paramyotonia Congenita Panel - 2 гена	CLCN1, SCN4A

36	Рабдоміоліз і метаболічна міопатія / Invitae Rhabdomyolysis and Metabolic Myopathy Panel - 134 гена	ABHD5, ACAD9, ACADM, ACADVL, AGK, AGL, AHCY, ALDOA, AMACR, AMPD1, ANO5, ATP2A1, ATP7B, B3GALNT2, B4GAT1, C1QBP, CACNA1S, CAPN3, CASQ1, CAV3, CHAT, CHKB, COQ2, COQ4, COQ7, COQ8A, COQ9, COX15, COX20, COX6B1, CPT1A, CPT2, CTDP1, DAG1, DGUOK, DMD, DNA2, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FHL1, FKRP, FKTN, FLAD1, GAA, GATM, GBE1, GFER, GMPPB, GYG1, GYS1, HADH, HADHA, HADHB, HMBS, ISCU, ISPD, ITGA7, LAMA2, LAMP2, LARGE1, LDHA, LPIN1, MAN2B1, MGME1, MICU1, MPV17, MYH3, OPA1, OPA3, PDSS1, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, PNPLA8, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PUS1, PYGM, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, SCN4A, SDHA, SGCA, SGCB, SGCD, SGCG, SIL1, SLC16A1, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SLC25A42, STAC3, SUCLA2, SUCLG1, TANGO2, TCAP, TK2, TNPO3, TRIM32, TRMT5, TSFM, TWNK, TYMP, YARS2, ATP5D, ATP5E, CHCHD10, SLC25A32, TOP3A
37	Дизостоз обличчя та лобно-назальна дисплазія / Invitae Facial Dysostosis and Frontonasal Dysplasia Panel - 28 генів	ABHD5, ACAD9, ACADM, ACADVL, AGK, AGL, AHCY, ALDOA, AMACR, AMPD1, ANO5, ATP2A1, ATP7B, B3GALNT2, B4GAT1, C1QBP, CACNA1S, CAPN3, CASQ1, CAV3, CHAT, CHKB, COQ2, COQ4, COQ7, COQ8A, COQ9, COX15, COX20, COX6B1, CPT1A, CPT2, CTDP1, DAG1, DGUOK, DMD, DNA2, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FHL1, FKRP, FKTN, FLAD1, GAA, GATM, GBE1, GFER, GMPPB, GYG1, GYS1, HADH, HADHA, HADHB, HMBS, ISCU, ISPD, ITGA7, LAMA2, LAMP2, LARGE1, LDHA, LPIN1, MAN2B1, MGME1, MICU1, MPV17, MYH3, OPA1, OPA3, PDSS1, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, PNPLA8, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PUS1, PYGM, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, SCN4A, SDHA, SGCA, SGCB, SGCD, SGCG, SIL1, SLC16A1, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SLC25A42, STAC3, SUCLA2, SUCLG1, TANGO2, TCAP, TK2, TNPO3, TRIM32, TRMT5, TSFM, TWNK, TYMP, YARS2, ATP5D, ATP5E, CHCHD10, SLC25A32, TOP3A
38	Дизостоз обличчя та лобно-назальна дисплазія / Invitae Facial Dysostosis and Frontonasal Dysplasia Panel - 28 генів	ALX1, ALX3, ALX4, CHD7, DHODH, EDN1, EDNRA, EFNB1, EFTUD2, EVC, EVC2, GATA1, GNAI3, IRX5, PDE4D, PLCB4, POLR1A, POLR1C, POLR1D, PRKAR1A, RPL11, RPL5, RPS28, SF3B4, TCOF1, TSR2, TWIST1, ZSWIM6
39	Синдром Алажілля / Invitae Alagille Syndrome Panel - 2 гена	JAG1, NOTCH2
40	Альфа-таласемія Х-зчеплена інтелектуальна недостатність / Invitae Alpha Thalassemia X-linked Intellectual Disability Test - 1 ген	ATRX
41	Синдром Карпентера / Invitae Carpenter Syndrome Panel - 2 гена	MEGF8, RAB23
42	Синдром Кофіна-Лоурі / Invitae Coffin-Lowry Syndrome Test - 1 ген	RPS6KA3
43	Синдром Коена / Invitae Cohen Syndrome Test - 1 ген	VPS13B
44	Синдром Корнелії де Ланге та пов'язані з ним розлади / Invitae Cornelia de Lange Syndrome and Related Disorders Panel - 31 ген	ADNP, AFF4, ANKRD11, ARID1A, ARID1B, BRD4, CREBBP, DPF2, EP300, ESCO2, HDAC8, KMT2A, MED13L, NIPBL, PHF6, PHIP, RAD21, SETD5, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SOX11, SRCAP, STAG1, STAG2, TAF1, TAF6, ZMYND11
45	Гіпогонадотропний гіпогонадизм / Invitae Hypogonadotropic Hypogonadism Panel - 46 генів	ANOS1, AXL, CCDC141, CHD4, CHD7, CYP19A1, DHCR7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IGSF10, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, LMNA, NR0B1, NSMF, PCSK1, PLXNA1, POLR3B, PROK2, PROKR2, PROP1, RELN, RNF216, SEMA3A, SOX10, SOX2, SPRY4, SRA1, SRY, TAC3, TACR3, WDR11
46	Синдром Кабукі / Invitae Kabuki Syndrome Panel - 2 гена	KDM6A, KMT2D
47	Синдром Рубінштейна-Тайбі / Invitae Rubinstein-Taybi Syndrome Panel - 2 гена	CREBBP, EP300

48	Синдром Сімпсона-Голабі-Бехмеля / Invitae Simpson-Golabi-Behmel Syndrome Test - 1 ген	GPC3
49	Синдром Ван дер Вуда / Invitae van der Woude Syndrome Panel - 2 гена	GRHL3, IRF6
50	Синдром Вівера / Invitae Weaver Syndrome Test - 1 ген	EZH2
51	Порушення статевого розвитку / Invitae Disorders of Sex Development Panel - 53 гена	AMH, AMHR2, ANOS1, AR, ARX, ATRX, B3GLCT, CBX2, CCNQ, CHD7, CKAP2L, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHH, DMRT1, ESR2, FRAS1, FREM2, H6PD, HHAT, HOXA13, HSD17B3, HSD3B2, KL, LHCGR, LHX3, LHX4, MAMLD1, MAP3K1, NR0B1, NR5A1, POR, PROP1, PSMC3IP, RSPO1, SOX9, SPECC1L, SRD5A2, SRY, STAR, TNK2, TOE1, TSPYL1, TWIST2, UBR1, WNT4, WNT9B, WT1, WWOX, ZFPM2