Invitae is pleased to announce 25 new and updated neurological and pediatric-related panels within the following disease areas:

- Deafness
- Developmental Disorders
- Epilepsy Seizures and Developmental Brain Abnormalities
- Eye Disorders
- Kidney Disorders and Progressive Renal Disease
- Neurodevelopmental Disorders
- Neuromuscular Disorders
- Pulmonary Disorders
- Skeletal Disorders
- Skin Disorders

Deafness		
Invitae Comprehensive Deafness Panel *(new genes in **teal**)*	224 genes	ABHD12, ABHD5, ACOX1, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANLN, **ARSB,** ARSG, **ATP1A3,** ATP2B2, ATP6V1B1, **BCAP31,** BCS1L, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, **DBH, DCAF17,** DCDC2, DFNA5, DFNB59, DIABLO, DIAPH1, DIAPH3, DMXL2, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FGFR3, FOXC1, FOXI1, **GALNS,** GATA3, GDF6, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB6, **GLB1, GNS,** GPSM2, GRHL2, GRXCR1, GRXCR2, **GUSB,** HARS, HARS2, HGF, **HGSNAT,** HOMER2, HSD17B4, **HYAL1, IDS, IDUA,** ILDR1, JAG1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LHX3, LOXHD1, LOXL3, LRP2, LRTOMT, MAN2B1, MARVELD2, MCM2, MEOX1, MET, MIR96, **MITF, MPZ,** MSRB3, MYH14, MYH7B, MYH9, MYO15A, MYO18B, MYO3A, MYO6, MYO7A, **NAGLU,** NARS2, NDRG1, NF2, NLRP3, **NOG,** OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PCGF2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RAI1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, **SCP2,** SERAC1, SERPINB6, **SGSH,** SH3TC2, SIX1, SIX5, SLC12A2, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC44A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, **TMEM126A,** TMEM132E, TMIE, TMPRSS3, TPRN, TRIOBP, **TRRAP,** TSPEAR, TUBB4B, TWNK, UBR1, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN, ZNF469
Developmental disorders		
Invitae Facial Dysostosis and Frontonasal Dysplasia Panel **(new panel)**	28 genes	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
Invitae Cornelia de Lange Syndrome and Related Disorders Panel *(new genes in **teal**)*	31 genes	**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**

Epilepsy seizures and developmental brain abnormalities

Invitae Epilepsy Panel *(new genes in **teal**)*	302 genes	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
Invitae Brain Malformations Panel *(new genes in **teal**)*	163 genes	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**

Eye disorders

Invitae Congenital Stationary Night Blindness Panel **(new panel)**	22 genes	CABP4, CACNA1F, CACNA2D4, CHM, CYP4V2, FRMD7, GNAT1, GNB3, GPR179, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RBP4, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1
Invitae Corneal Dystrophies Panel **(new panel)**	33 genes	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
Invitae Achromatopsia Panel **(new panel)**	8 genes	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, RGS9, RGS9BP

Eye disorders *(continued)*		
Invitae Microphthalmia, Anophthalmia, Coloboma (MAC) and Anterior Segment Dysgenesis Panel *(new genes in **teal**)*	81 genes	**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**
Kidney disorders and progressive renal disease		
Invitae Renal Tubular Disorders Panel **(new panel)**	39 genes	ACE, AGT, AGTR1, AQP2, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CA2, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CUL3, EGF, FOXI1, FXYD2, GATM, GNA11, HSD11B2, KCNJ1, KCNJ10, KLHL3, MAGED2, NR3C2, OCRL, REN, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC34A1, SLC4A1, SLC4A4, TRPM6, WNK1, WNK4
Invitae Progressive Renal Disease Panel *(new genes in **teal**)*	195 genes	**ACE, ACTB,** ACTN4, **ADAMTS13, AGT, AGTR1, AHI1, ALG8, ALG9, ALMS1, AMN, ANKS6,** ANLN, APOL1, **APRT, AQP2, ARHGAP24, ARHGDIA, ARL6, ATP6V0A4, ATP6V1B1, AVP, AVPR2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BSND, CA2, CACNA1D, CACNA1H, CASR, CD151,** CD2AP, **CDKN1C, CEP164, CEP19, CEP290, CEP83, CEP89, CFHR5, CHRM3, CLCN5, CLCNKB, CLDN16, CLDN19, COL4A1,** COL4A3, COL4A4, COL4A5, **COL4A6, COQ2, COQ6, COQ8B, CPLANE1,** CRB2, **CTNS, CUBN, CYP24A1, DCDC2, DGKE, DHCR7, DICER1, DLC1, DNAJB11, DZIP1L, EGF, EMP2, EYA1, FAM20A, FAN1, FAT1, FN1, FOXI1, FOXP3, FREM1, GANAB, GATA3, GATM, GLA, GLIS2, GRHPR, GSN,** HNF1A, **HNF1B, HPRT1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43,** INF2, **INVS, IQCB1, ITGA3, ITGA6, ITGB4, JAG1, KANK2, KANK4, KAT6B, KCNJ1, KCNJ10, LAMA5, LAMB2, LCAT, LDHA,** LMX1B, **LPIN1, LRP4, LRP5, LYZ, LZTFL1, MAGED2, MAGI2, MAPKBP1, MEFV, MKKS, MYH9,** MYO1E, **NEK8, NOTCH2, NPHP1, NPHP3, NPHP4,** NPHS1, NPHS2, **NSDHL, NUP107, NUP133, wNUP160, NUP205, NUP85, NUP93, OFD1,** PAX2, **PBX1, PDSS2, PHEX,** PKD2, PKHD1, **PLCE1, PODXL, PREPL, PTPRO, REN, RMND1, RPGRIP1L, SALL1, SALL4, SARS2, SDCCAG8, SEC61A1, SEMA3E, SGPL1, SIX1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC37A4, SLC41A1, SLC4A1, SLC4A4, SLC7A7, SMARCAL1, SYNPO, TBC1D8B, TMEM67, TNS2, TP63, TRIM32,** TRPC6, **TSC1, TSC2, TTC21B, TTC8, UMOD, WDPCP, WDR19, WDR73, WT1, XPNPEP3, XPO5, ZNF423**
Neurodevelopmental disorders		
Invitae Neurodevelopmental Disorders Panel **(new panel)**	241 genes	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, 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, GBRG2, 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,S MARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SON, SOS1, SOS2, SOX11, SPAST, SPATA5, SPTAN1, STAG1, STXBP1,S URF1, SYNGAP1, TAF1, TBCK, TBL1XR1, TCF20, TCF4, TELO2, TPP1, TRAPPC9, TRRAP, TSC1,T SC2, TUBA1A, UBE3A, UNC80, USP9X, VPS13B, WDR45, WWOX, ZBTB18, ZBTB20, ZC4H2, ZDHHC9, ZEB2, ZIC2, ZMIZ1, ZMYND11

Neuromuscular disorders

Invitae Comprehensive Neuromuscular Disorders Panel *(new genes in **teal**)*	210 genes	**ABHD5, ACAD9, ACADM, ACADVL,** ACTA1, ADSSL1, **AGK, AGL,** AGRN, **AHCY, ALDOA,** ALG14, ALG2, **AMACR,** AMPD1, ANO5, ATP2A1, **ATP7B,** B3GALNT2, B4GAT1, BAG3, BIN1, **C1QBP,** CACNA1S, CAPN3, CASQ1, CAV3, CCDC78, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, **COQ2, COQ4, COQ7, COQ8A, COQ9, COX15, COX20, COX6B1, CPT1A,** CPT2, CRYAB, **CTDP1,** DAG1, DDC, DES, **DGUOK,** DMD, **DNA2,** DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYSF, EMD, **ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2,** FHL1, FKBP14, FKRP, FKTN, **FLAD1,** FLNC, GAA, **GATM, GBE1, GFER,** GFPT1, GMPPB, GNE, GOSR2, GYG1, GYS1, HACD1, **HADH, HADHA, HADHB, HMBS,** HNRNPA2B1, HNRNPDL, ISCU, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LMNA, LMOD3, **LPIN1, MAN2B1,** MAP3K20, MATR3, MEGF10, **MGME1,** MICU1, **MPV17,** MTM1, MUSK, MYH2, **MYH3,** MYH7, MYL2, MYO18B, MYOT, MYPN, NEB, **OPA1, OPA3,** ORAI1, **PDSS1, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB,** PLEC, PNPLA2, **PNPLA8, POGLUT1, POLG, POLG2,** POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, **PUS1, PYGM,** PYROXD1, RAPSN, **RBCK1, RNASEH1, RRM2B,** RXYLT1, RYR1, SCN4A, **SDHA,** SELENON, SGCA, SGCB, SGCD, SGCG, **SIL1, SLC16A1,** SLC18A3, **SLC22A5, SLC25A20, SLC25A3, SLC25A4, SLC25A42,** SLC5A7, SMCHD1, SMN1, SMN2, **SMPX,** SPEG, SQSTM1, STAC3, STIM1, **SUCLA2, SUCLG1,** SYT2, **TANGO2,** TAZ, TCAP, TIA1, TK2, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, **TRMT5, TSFM,** TTN, **TWNK, TYMP,** VAMP1, VCP, VMA21, **YARS2**
Add-on Preliminary-evidence Genes for Neuromuscular Disorders *(new genes in **teal**)*	19 genes	**ATP1A2, ATP5D, ATP5E, CHCHD10,** KLHL9, LAMB2, LIMS2, LRP4, **MCM3AP,** MTMR14, **SLC25A32,** SNAP25, SUN1, SUN2, SYNE1, SYNE2, TMEM43, TNNT3, **TOP3A**
Invitae Comprehensive Muscular Dystrophy Panel *(new genes in **teal**)*	53 genes	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
Invitae Limb-Girdle Muscular Dystrophy Panel *(new genes in **teal**)*	38 genes	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
Invitae Comprehensive Myopathy Panel *(new genes in **teal**)*	72 genes	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
Invitae Rhabdomyolysis and Metabolic Myopathy Panel *(new genes in **teal**)*	129 genes	**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** **REMOVED: SLC25A32 *available through add-on panel**
Add-on Preliminary-evidence Genes for Rhabdomyolysis and Metabolic Myopathy **(new panel)**	5 genes	ATP5D, ATP5E, CHCHD10, SLC25A32, TOP3A

Pulmonary disorders		
Invitae Surfactant Metabolism Panel **(new panel)**	21 genes	ABCA3, COPA, CSF2RA, CSF2RB, DKC1, FLNA, FOXF1, GATA2, ITGA3, MARS, NKX2-1, NOTCH2, PARN, RTEL1, SFTPB, SFTPC, SLC7A7, TERC, TERT, TINF2, TMEM173
Skeletal disorders		
Invitae Microcephalic Primordial Dwarfism and Seckel Syndrome Panel **(new panel)**	38 genes	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
Invitae Osteogenesis Imperfecta and Bone Fragility Panel *(new genes in **teal**)*	67 genes	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
Invitae Skeletal Disorders Panel *(new genes in **teal**)*	358 genes	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**

Skin disorders		
Invitae Hypopigmentation Panel **(new panel)**	47 genes	ACD, AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10orf11, CLCN7, CTC1, DKC1, DTNBP1, EDN3, EDNRB, EPG5, FRMD7, GNAI3, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LYST, MITF, MLPH, MYO5A, NHP2, NOP10, OCA2, PARN, PAX3, RAB27A, RET, RTEL1, SLC24A5, SLC38A8, SLC45A2, SNAI2, SOX10, TERC, TERT, TINF2, TYR, TYRP1, USB1, WRAP53
Invitae Ectodermal Dysplasia and Related Disorders Panel *(new genes in **teal**)*	73 genes	**ANTXR1, APCDD1, ATP7A, AXIN2, BCS1L, BMP4, CDH3, CDSN, CLDN1, CTSC, DSG4, DSP,** EDA, EDAR, EDARADD, **EGFR, ERCC2, ERCC3, ERCC8, GJA1, GJB2, GJB6, GRHL2, GTF2E2, GTF2H5, HOXC13, HR, IRF6, JUP, KANK2, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT25, KRT6A, KRT6B, KRT6C, KRT71, KRT74, KRT83, KRT85, LIPH, LPAR6, LRP6, LSS,** LTBP3, **MBTPS2, MPLKIP,** MSX1, **NECTIN1, NECTIN4,** NFKBIA, PAX9, **PKP1, POC1A, PORCN, PTH1R, RIN2, RNF113A, RPL21, SMOC2, SNRPE, SOX18, SPINK5, TFAP2B, TP63, TRPS1, TRPV3, TSPEAR,** WNT10A, **WNT10B**

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