

INVITAE NEUROLOGY GENE PANEL TESTS

A broad menu covering the vast majority of inherited neurological conditions with both large comprehensive and smaller condition-specific panels curated by medical genetics experts.

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

| Test name | # gene(s) | Gene list |
|--|-----------|---|
| Movement Disorders | | |
| Invitae Dystonia Panel | 18 | ANO3, ATP1A3, ATP7B, GCH1, GNAL, HEXA, PNKD, PRKN, PRKRA, PRRT2, SGCE, SLC2A1, SLC6A3, SPR, TH, THAP1, TOR1A, TUBB4A |
| Add-on preliminary-evidence genes | 5 | CIZ1, DRD2, HPCA, KCTD17, TOR1AIP1 |
| Invitae Hereditary Parkinson's Disease & Parkinsonism Panel | 16 | ATP7B, ATP13A2, DCTN1, DNAJC6, FBXO7, GCH1, LRRK2, PARK7, PINK1, PRKN, PRKRA, SLC6A3, SNCA, SPR, TH, VPS35 |
| Add-on preliminary-evidence genes | 2 | CHCHD2, MAPT |
| Neurodegenerative Disorders | | |
| Invitae Combined Hereditary Dementia and Amyotrophic Lateral Sclerosis Panel | 23 | ALS2, APP, CHCHD10, DCTN1, FUS, GRN, KIF5A, MAPT, OPTN, PFN1, PRNP, PSEN1, PSEN2, SETX, SNCA, SOD1, SPG11, TARDBP, TBK1, TFG, UBQLN2, VAPB, VCP |
| Add-on preliminary-evidence genes | 5 | CHMP2B, HNRNPA2B1, MATR3, SIGMAR1, SQSTM1 |
| Invitae Amyotrophic Lateral Sclerosis Panel | 16 | ALS2, CHCHD10, DCTN1, FUS, KIF5A, OPTN, PFN1, SETX, SOD1, SPG11, TARDBP, TBK1, TFG, UBQLN2, VAPB, VCP |
| Add-on preliminary-evidence genes | 4 | CHMP2B, MATR3, SIGMAR1, SQSTM1 |
| Invitae Frontotemporal Dementia Panel | 9 | CHCHD10, DCTN1, FUS, GRN, MAPT, TARDBP, TBK1, UBQLN2, VCP |
| Add-on preliminary-evidence genes | 4 | CHMP2B, HNRNPA2B1, PSEN1, SQSTM1 |
| Invitae Hereditary Alzheimer's Disease Panel | 3 | APP, PSEN1, PSEN2 |
| Invitae Hereditary Parkinson's Disease & Parkinsonism Panel | 16 | ATP7B, ATP13A2, DCTN1, DNAJC6, FBXO7, GCH1, LRRK2, PARK7, PINK1, PRKN, PRKRA, SLC6A3, SNCA, SPR, TH, VPS35 |
| Add-on preliminary-evidence genes | 2 | CHCHD2, MAPT |
| Invitae Hereditary Prion Disease Test | 1 | PRNP |
| Neuromuscular Disorders | | |
| Invitae Comprehensive Neuromuscular Disorders Panel | 109 | ACTA1, AGRN, ALG2, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1S, CAPN3, CAV3, CCDC78, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRYAB, DAG1, DES, DMD, DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYF, EMD, FHL1, FKBP14, FKRP, FKTN, FLNC, GAA, GFPT1, GMPPB, GNE, GYS1, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LDB3, LMNA, LMOD3, MATR3, MEGF10, MTM1, MUSK, MYH2, MYH7, MYL2, MYOT, MYPN, NEB, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, RAPSN, RXYLT1, RYR1, SCN4A, SELENON, SGCA, SLC6A3, SLC6A7, SLC5A7, SMN1, SMN2, SQSTM1, STAC3, STIM1, TAZ, TCAP, TIA1, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, VCP, VMA21 |
| Add-on preliminary-evidence genes | 13 | ALG14, HNRNPA2B1, HNRNPDL, LAMB2, LIMS2, LRP4, MYF6, SNAP25, SUN1, SUN2, SYNE1, SYNE2, TMEM43 |
| Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene | 1 | SMCHD1 |
| Invitae Congenital Myasthenic Syndrome Panel | 16 | AGRN, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, MUSK, PREPL, RAPSN, SLC5A7 |
| Add-on preliminary-evidence genes | 6 | ALG14, LAMB2, LRP4, PLEC, SCN4A, SNAP25 |
| Invitae Malignant Hyperthermia Susceptibility Panel | 2 | CACNA1S, RYR1 |

INVITAE NEUROLOGY GENE PANEL TESTS

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

| Test name | # gene(s) | Gene list |
|---|-----------|---|
| Neuromuscular Disorders (continued) | | |
| Invitae Comprehensive Muscular Dystrophy Panel | 48 | ANO5, B3GALNT2, B4GAT1, CAPN3, CAV3, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, ISPD, ITGA7, LAMA2, LARGE1, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SGCA, SGCB, SGCD, SGCC, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN |
| Add-on preliminary-evidence genes | 7 | HNRNPDL, LIMS2, SUN1, SUN2, SYNE1, SYNE2, TMEM43 |
| Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene | 1 | SMCHD1 |
| Invitae Congenital Muscular Dystrophy Panel | 27 | B3GALNT2, B4GAT1, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DMD, DPM1, DPM2, DPM3, FKRP, FKTN, GMPPB, ISPD, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, TCAP |
| Invitae Dystroglycanopathy Panel | 17 | B3GALNT2, B4GAT1, DAG1, DPM1, DPM2, DPM3, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 |
| Invitae Dystrophinopathies Test | 1 | DMD |
| Invitae Emery-Dreifuss Muscular Dystrophy Panel | 3 | EMD, FHL1, LMNA |
| Add-on preliminary-evidence genes | 5 | SUN1, SUN2, SYNE1, SYNE2, TMEM43 |
| Invitae Limb-Girdle Muscular Dystrophy Panel | 31 | ANO5, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DYSF, FKRP, FKTN, GAA, GMPPB, ISPD, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCC, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN |
| Add-on preliminary-evidence genes | 2 | HNRNPDL, LIMS2 |
| Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene | 1 | SMCHD1 |
| Invitae Comprehensive Myopathy Panel | 52 | ACTA1, ANO5, ATP2A1, BAG3, BIN1, CACNA1S, CAV3, CCDC78, CFL2, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FKBP14, FLNC, GNE, GYS1, KBTBD13, KCNJ2, KLHL40, KLHL41, LDB3, LMNA, LMOD3, MATR3, MEGF10, MTM1, MYH7, MYL2, MYOT, MYPN, NEB, RYR1, SCN4A, SELENON, SQSTM1, STAC3, STIM1, TIA1, TNNT1, TPM2, TPM3, TTN, VCP |
| Add-on preliminary-evidence gene | 1 | MYF6 |
| Invitae Congenital Myopathy Panel | 27 | ACTA1, BIN1, CCDC78, CFL2, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, DNM2, FKBP14, KBTBD13, KLHL40, KLHL41, LMOD3, MEGF10, MTM1, MYH7, MYPN, NEB, RYR1, SELENON, STAC3, TNNT1, TPM2, TPM3, TTN |
| Add-on preliminary-evidence gene | 1 | MYF6 |
| Invitae Autophagic Vacuolar Myopathy Panel | 3 | DES, LAMP2, VMA21 |
| Invitae Central Core Disease Test | 1 | RYR1 |
| Invitae Centronuclear Myopathy Panel | 6 | BIN1, CCDC78, DNM2, MTM1, RYR1, TTN |
| Add-on preliminary-evidence gene | 1 | MYF6 |
| Invitae Congenital Fiber-Type Disproportion Panel | 7 | ACTA1, LMNA, MYH7, RYR1, SELENON, TPM2, TPM3 |
| Invitae Distal Myopathy Panel | 18 | ANO5, BAG3, CAV3, CRYAB, DES, DNAJB6, DYSF, FHL1, FLNC, GNE, LDB3, MATR3, MYH7, MYOT, SQSTM1, TIA1, TTN, VCP |
| Invitae Inclusion Body Myopathy Panel | 4 | GNE, MYH2, TTN, VCP |
| Add-on preliminary-evidence gene | 1 | HNRNPA2B1 |

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CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

| Test name | # gene(s) | Gene list |
|---|-----------|--|
| Neuromuscular Disorders (continued) | | |
| Invitae Multiminicore Disease Panel | 2 | RYR1, SELENON |
| Invitae Myofibrillar Myopathy Panel | 8 | BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, LDB3, MYOT |
| Invitae Nemaline Myopathy Panel | 11 | ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3 |
| Invitae Periodic Paralysis Panel | 4 | CACNA1S, KCNJ2, RYR1, SCN4A |
| Invitae Type VI Collagenopathy Panel | 3 | COL6A1, COL6A2, COL6A3 |
| Add-on preliminary-evidence gene | 1 | COL12A1 |
| Invitae Myotonia and Paramyotonia Congenita Panel | 2 | CLCN1, SCN4A |
| Invitae Spinal Muscular Atrophy Panel | 2 | SMN1, SMN2 |
| Neuropathies and Related Disorders | | |
| Invitae Comprehensive Neuropathies Panel | 72 | AARS, AIFM1, ATL1, ATL3, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, ELP1, FBXO38, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KIF1A, LITAF, LMNA, LRSAM1, MED25, MFN2, MORC2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF2, SCN11A, SCN9A, SH3TC2, SIGMAR1, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SPG11, SPTLC1, SPTLC2, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, WNK1, YARS, VRK1 |
| Add-on preliminary-evidence genes | 9 | CCT5, FLRT1, HSPB3, LAS1L, MARS, PRDM12, SCN10A, SETX, SURF1 |
| Add-on spinal muscular atrophy genes | 2 | SMN1, SMN2 |
| Invitae Charcot-Marie-Tooth Disease Comprehensive Panel | 43 | AARS, AIFM1, BSCL2, DNAJB2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, LITAF, LMNA, LRSAM1, MED25, MFN2, MORC2, MPZ, MTMR2, NDRG1, NEFL, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, SBF2, SH3TC2, SLC25A46, SPG11, TFG, TRIM2, TRPV4, YARS |
| Add-on preliminary-evidence genes | 2 | MARS, SURF1 |
| Invitae Charcot-Marie-Tooth Disease Autosomal Dominant Panel | 24 | AARS, BSCL2, DNM2, DYNC1H1, EGR2, GARS, GDAP1, GNB4, HARS, HSPB1, HSPB8, INF2, LITAF, LMNA, LRSAM1, MFN2, MORC2, MPZ, NEFL, PMP22, RAB7A, TFG, TRPV4, YARS |
| Add-on preliminary-evidence gene | 1 | MARS |
| Invitae Charcot-Marie-Tooth Disease Autosomal Recessive Panel | 21 | DNAJB2, EGR2, FGD4, FIG4, GDAP1, HINT1, IGHMBP2, LMNA, LRSAM1, MED25, MFN2, MTMR2, NDRG1, NEFL, PLEKHG5, PRX, SBF2, SH3TC2, SLC25A46, SPG11, TRIM2 |
| Add-on preliminary-evidence gene | 1 | SURF1 |
| Invitae Charcot-Marie-Tooth Disease X-linked Panel | 4 | AIFM1, GJB1, PDK3, PRPS1 |
| Invitae Hereditary Sensory and Autonomic Neuropathy Panel | 15 | ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, RAB7A, RETREG1, SCN11A, SCN9A, SPTLC1, SPTLC2, WNK1 |
| Add-on preliminary-evidence genes | 2 | CCT5, PRDM12 |
| Invitae Familial Dysautonomia Test | 1 | ELP1 |
| Invitae Hereditary Motor Neuropathies Panel | 23 | ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, FBXO38, GARS, HINT1, HSPB1, HSPB8, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SLC5A7, SMN1, SMN2, TRPV4, UBA1, VAPB, VRK1 |
| Add-on preliminary-evidence gene | 1 | HSPB3 |
| Invitae Spinal Muscular Atrophy Panel | 2 | SMN1, SMN2 |

INVITAE NEUROLOGY GENE PANEL TESTS

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

| Test name | # gene(s) | Gene list |
|---|-----------|--|
| Neuropathies and Related Disorders (continued) | | |
| Invitae Small Fiber Neuropathy Test | 1 | SCN9A |
| Add-on preliminary-evidence gene | 1 | SCN10A |
| Invitae Riboflavin Transporter Deficiency Neuronopathy Panel | 2 | SLC52A2, SLC52A3 |
| Invitae Hereditary Spastic Paraplegia Comprehensive Panel | 45 | ABCD1, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ATL1, B4GALNT1, BSCL2, C12orf65, CYP2U1, CYP7B1, DDHD1, DDHD2, ERLIN2, FA2H, GBA2, GJC2, HEXA, HSPD1, KDM5C, KIF1A, KIF1C, KIF5A, L1CAM, NIPA1, NT5C2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SACS, SLC16A2, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, VAMP1, WASHC5, ZFYVE26 |
| Add-on preliminary-evidence genes | 20 | AMPD2, ARL6IP1, ARSI, ATP13A2, C19orf12, CCT5, CPT1C, ENTPD1, ERLIN1, EXOSC3, IBA57, MAG, PGAP1, RAB3GAP2, SLC33A1, TFG, USP8, VPS37A, ZFR, ZFYVE27 |
| Invitae Hereditary Spastic Paraplegia Autosomal Dominant Panel | 13 | ALDH18A1, ATL1, BSCL2, HSPD1, KIF1A, KIF5A, NIPA1, REEP1, REEP2, RTN2, SPAST, VAMP1, WASHC5 |
| Add-on preliminary-evidence genes | 3 | CPT1C, SLC33A1, ZFYVE27 |
| Invitae Hereditary Spastic Paraplegia Autosomal Recessive Panel | 30 | ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, B4GALNT1, C12orf65, CYP2U1, CYP7B1, DDHD1, DDHD2, ERLIN2, FA2H, GBA2, GJC2, HEXA, KIF1A, KIF1C, NT5C2, PNPLA6, REEP2, SACS, SPART, SPG11, SPG21, SPG7, TECPR2, ZFYVE26 |
| Add-on preliminary-evidence genes | 17 | AMPD2, ARL6IP1, ARSI, ATP13A2, C19orf12, CCT5, ENTPD1, ERLIN1, EXOSC3, IBA57, MAG, PGAP1, RAB3GAP2, TFG, USP8, VPS37A, ZFR |
| Invitae Hereditary Spastic Paraplegia X-linked Panel | 5 | ABCD1, KDM5C, L1CAM, PLP1, SLC16A2 |
| Cardiomyopathy and Skeletal Muscle Disease | | |
| Invitae Cardiomyopathy and Skeletal Muscle Disease Panel | 116 | ABCC9, ACTA1, ACTC1, ACTN2, AGL, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1C, CAPN3, CAV3, CCDC78, CFL2, CHKB, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, CSRP3, DAG1, DES, DMD, DNAJB6, DNM2, DOLK, DPM1, DPM2, DPM3, DSC2, DSG2, DSP, DYSF, EMD, EYA4, FHL1, FKBP14, FKRP, FKTN, FLNC, GAA, GLA, GMPPB, GNE, GYS1, HCN4, ISPD, ITGA7, JUP, KBTBD13, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LMNA, LMOD3, MATR3, MEGF10, MTM1, MYBPC3, MYH7, MYL2, MYL3, MYOT, MYPN, NEB, PKP2, PLEC, PLN, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRKAG2, RAF1, RBM20, RXYLT1, RYR1, RYR2, SCN5A, SELENON, SGCA, SGCB, SGCD, SGGC, SLC22A5, SQSTM1, STAC3, STIM1, TAZ, TCAP, TIA1, TMEM43, TNNC1, TNNI3, TNNT1, TNNT2, TNPO3, TOR1AIP1, TPM1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, TTR, VCL, VCP |
| Add-on preliminary-evidence genes | 36 | ANKRD1, CALR3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, HNRNPDL, ILK, JPH2, LAMA4, LDB3, LIMS2, LRRC10, MYF6, MYH6, MYLK2, MYOM1, MYOZ2, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, SUN1, SUN2, SYNE1, SYNE2, TGFB3, TMPO, TXNRD2 |
| Add-on autosomal recessive syndromic pediatric cardiomyopathy genes | 7 | ACADVL, ALMS1, DNAJC19, ELAC2, MTO1, SDHA, TMEM70 |

INVITAE NEUROLOGY GENE PANEL TESTS

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

| Test name | # gene(s) | Gene list |
|--|-----------|--|
| Epilepsy, Seizures, and Developmental Brain Abnormalities | | |
| Invitae Epilepsy Panel | 146 | ADSL, ALDH5A1, ALDH7A1, ALG13, ARHGEF9, ARX, ATP1A2, ATP1A3, ATRX, BRAT1, C12orf57, CACNA1A, CACNA2D2, CARS2, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRN2, CLCN4, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DEPDC5, DNAJC5, DNM1, DOCK7, DYRK1A, EEF1A2, EFHC1, EHMT1, EPM2A, FARS2, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GLRA1, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, IER3IP1, IQSEC2, ITPA, JMJD1C, KANSL1, KCNA2, KCNB1, KCNC1, KCNH2, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, LGI1, LIAS, MBD5, MECP2, MEF2C, MFS2D, MTOR, NEDD4L, NEXMIF, NGLY1, NHLRC1, NPRL3, NRXN1, PACS1, PCDH19, PIGA, PIGN, PIGO, PLCB1, PNKD, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRIMA1, PRRT2, PURA, QARS, RELN, ROGDI, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SERPINI1, SGCE, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SNX27, SPATA5, SPTAN1, ST3GAL5, STRADA, STX1B, STXB1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TPK1, TSC1, TSC2, UBE3A, WDR45, WWOX, ZDHHC9, ZEB2 |
| Add-on preliminary-evidence genes | 35 | ABAT, ARHGEF15, ATP6AP2, CACNA1H, CACNB4, CASR, CERS1, CNTN2, CPA6, DIAPH1, FASN, GABRD, GAL, GPHN, KCNA1, KCND2, KCNH5, KPNA7, LMNB2, NECAP1, PIGG, PIQ3, PIK3AP1, PRDM8, PRICKLE2, RBFOX1, RBFOX3, RYR3, SCN5A, SETD2, SLC35A3, SNAP25, SRPX2, ST3GAL3, TBL1XR1 |
| Add-on genes for glycine encephalopathy | 3 | AMT, GCSH, GLDC |
| Add-on FLNA gene | 1 | FLNA |
| Add-on PTEN gene | 1 | PTEN |
| Add-on RANBP2 gene | 1 | RANBP2 |
| Invitae Alternating Hemiplegia of Childhood Panel | 2 | ATP1A2, ATP1A3 |
| Add-on clinically overlapping Genes | 3 | CACNA1A, SCN1A, SLC2A1 |
| Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel | 2 | ACTB, ACTG1 |
| Invitae Cerebral Cavernous Malformations Panel | 3 | CCM2, KRIT1, PDCD10 |
| Invitae CHARGE Syndrome Test | 1 | CHD7 |
| Invitae Early Infantile Epileptic Encephalopathy Panel | 59 | ALDH7A1, ARHGEF9, ARX, BRAT1, CACNA2D2, CASK, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, FARS2, FOLR1, FRRS1L, GABBR2, GABRA1, GABRB3, GNAO1, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, IER3IP1, KCNA2, KCNB1, KCNMA1, KCNQ2, KCNQ3, KCNT1, PCDH19, PIGA, PIGN, PIGO, PLCB1, PNKP, PNPO, PURA, SCN1A, SCN2A, SCN8A, SCN9A, SIK1, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SMC1A, SPTAN1, STXB1, SYNGAP1, SZT2, TBC1D24, WDR45, WWOX |
| Add-on preliminary-evidence genes | 10 | ARHGEF15, ATP1A2, COQ4, GPHN, KCNH5, MTOR, NECAP1, NEDD4L, SCN1B, ST3GAL3 |
| Invitae Holoprosencephaly Panel | 6 | FGFR1, GLI2, SHH, SIX3, TGIF1, ZIC2 |
| Add-on preliminary-evidence genes | 4 | CDON, FOXH1, NODAL, PTCH1 |
| Invitae Neurodegeneration with Brain Iron Accumulation Panel | 11 | ATP13A2, C19orf12, COASY, CP, DCAF17, FTL, FUCA1, PANK2, PLA2G6, SQSTM1, WDR45 |
| Add-on preliminary-evidence genes | 3 | FA2H, KIF1A, TRIM32 |
| Invitae Rett/Angelman and Related Disorders Panel | 24 | ADSL, ALDH5A1, ATRX, CDKL5, CNTNAP2, DYRK1A, EHMT1, FOXG1, GABBR2, IQSEC2, KANSL1, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN8A, SLC9A6, STXB1, TCF4, UBE3A, WDR45, ZEB2 |
| Add-on preliminary-evidence genes | 4 | GABRD, HDAC8, TBL1XR1, JMJD1C |
| Invitae Tuberous Sclerosis Complex Panel | 2 | TSC1, TSC2 |