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

MOVEMENT DISORDERS

Invitae Dystonia Panel (up to 21 genes)

Primary panel (16 genes)
ANO3 ATP1A3 GCH1 GNAL PARK2 PNKD PRKRA PRRT2 SGCE SLC2A1 SLC6A3 SPR
TH THAP1 TOR1A TUBB4A

Add-on preliminary-evidence genes (5 genes)
CIZ1 DRD2 HPCA KCTD17 TOR1AIP1

Invitae Hereditary Parkinson's Disease & Parkinsonism Panel (up to 17 genes)

Primary panel (15 genes)
ATP13A2 DCTN1 DNAJC6 FBXO7 GCH1 LRRK2 PARK2 PARK7 PINK1 PRKRA SLC6A3 SNCA
SPR TH VPS35

Add-on preliminary-evidence genes (2 genes)
CHCHD2 MAPT

NEURODEGENERATIVE DISORDERS

Invitae Combined Hereditary Dementia and Amyotrophic Lateral Sclerosis Panel (up to 27 genes)

Primary panel (21 genes)
ALS2 APP CHCHD10 DCTN1 FUS GRN MAPT OPTN PFN1 PRNP PSEN1 PSEN2
SNCA SOD1 SPG11 TARDBP TBK1 TFG UBQLN2 VAPB VCP

Add-on preliminary-evidence genes (6 genes)
CHMP2B HNRNPA2B1 MATR3 SETX SIGMAR1 SQSTM1

Invitae Amyotrophic Lateral Sclerosis Panel (up to 19 genes)

Primary panel (13 genes)
ALS2 CHCHD10 DCTN1 FUS OPTN PFN1 SOD1 SPG11 TARDBP TBK1 TFG UBQLN2
VAPB

Add-on preliminary-evidence genes (6 genes)
CHMP2B MATR3 SETX SIGMAR1 SQSTM1 VCP

Invitae Frontotemporal Dementia Panel (up to 13 genes)

Primary panel (9 genes)
CHCHD10 DCTN1 FUS GRN MAPT TARDBP TBK1 UBQLN2 VCP

Add-on preliminary-evidence genes (4 genes)
CHMP2B HNRNPA2B1 PSEN1 SQSTM1

Invitae Hereditary Alzheimer's Disease Panel (3 genes)

Primary panel (3 genes)
APP PSEN1 PSEN2

Invitae Hereditary Parkinson's Disease & Parkinsonism Panel (up to 17 genes)

Primary panel (15 genes)
ATP13A2 DCTN1 DNAJC6 FBXO7 GCH1 LRRK2 PARK2 PARK7 PINK1 PRKRA SLC6A3 SNCA
SPR TH VPS35

Add-on preliminary-evidence genes (2 genes)
CHCHD2 MAPT

Invitae Hereditary Prion Disease Test (1 gene)

Primary panel (1 gene)
PRNP

NEUROMUSCULAR DISORDERS

Invitae Comprehensive Neuromuscular Disorders Panel (up to 121 genes)

Primary panel (104 genes)
ACTA1 AGRN ALG2 ANO5 ATP2A1 B3GALNT2 B4GAT1 BAG3 BIN1 CACNA1S CAPN3 CAV3
CCDC78 CFL2 CHAT CHKB CHRNA1 CHRNB1 CHRND CHRNE CLCN1 CNTN1 COL6A1 COL6A2
COL6A3 COLQ CPT2 CRYAB DAG1 DES DMD DNAJB6 DNM2 DOK7 DPAGT1 DPM1
DPM2 DPM3 DYSF EMD FHL1 FKBP14 FKRP FKTN FLNC GAA GFPT1 GMPPB
GNE 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 RAPSN RYR1 SCN4A SELENON SGCA SGCB
SGCD SGCG SMN1, SMN2 SQSTM1 STAC3 STIM1 TAZ TCAP TIA1 TMEM5 TNNT1 TNPO3
TPM2 TPM3 TRAPPC11 TRIM32 TTN VCP VMA21

Add-on preliminary-evidence genes (16 genes)
ALG14 COL12A1 HNRNPA2B1 HNRNPDL LAMB2 LIMS2 LRP4 MYF6 PREPL SNAP25 SUN1 SUN2
SYNE1 SYNE2 TMEM43 TOR1AIP1

Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene (1 gene)
SMCHD1

Invitae Congenital Myasthenic Syndrome Panel (up to 21 genes)

Primary panel (13 genes)
AGRN ALG2 CHAT CHRNA1 CHRNB1 CHRND CHRNE COLQ DOK7 DPAGT1 GFPT1 MUSK
RAPSN

Add-on preliminary-evidence genes (8 genes)
ALG14 GMPPB LAMB2 LRP4 PLEC PREPL SCN4A SNAP25

Invitae Malignant Hyperthermia Susceptibility Panel (2 genes)

Primary panel (2 genes)
CACNA1S RYR1

INVITAE

INVITAE NEUROLOGY GENE PANEL TESTS (continued)

NEUROMUSCULAR DISORDERS (continued)

Invitae Comprehensive Muscular Dystrophy Panel (up to 56 genes)	**Primary panel (46 genes)** ANO5 B3GALNT2 B4GAT1 CAPN3 CAV3 CHKB 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 SGCA SGCB SGCD SGCG TCAP TMEM5 TNPO3 TRAPPC11 TRIM32 TTN **Add-on preliminary-evidence genes (9 genes)** COL12A1 HNRNPDL LIMS2 SUN1 SUN2 SYNE1 SYNE2 TMEM43 TOR1AIP1 **Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene (1 gene)** SMCHD1
Invitae Congenital Muscular Dystrophy Panel (up to 27 genes)	**Primary panel (26 genes)** B3GALNT2 B4GAT1 CHKB COL6A1 COL6A2 COL6A3 DAG1 DMD DPM1 DPM2 DPM3 FKRP FKTN GMPPB ISPD ITGA7 LAMA2 LARGE1 LMNA POMGNT1 POMGNT2 POMK POMT1 POMT2 TCAP TMEM5 **Add-on preliminary-evidence gene (1 gene)** COL12A1
Invitae Dystroglycanopathy Panel (17 genes)	**Primary panel (17 genes)** B3GALNT2 B4GAT1 DAG1 DPM1 DPM2 DPM3 FKRP FKTN GMPPB ISPD LARGE1 POMGNT1 POMGNT2 POMK POMT1 POMT2 TMEM5
Invitae Dystrophinopathies Test (1 gene)	**Primary panel (1 gene)** DMD
Invitae Emery-Dreifuss Muscular Dystrophy Panel (up to 8 genes)	**Primary panel (3 genes)** EMD FHL1 LMNA **Add-on preliminary-evidence genes (5 genes)** SUN1 SUN2 SYNE1 SYNE2 TMEM43
Invitae Limb-Girdle Muscular Dystrophy Panel (up to 34 genes)	**Primary panel (30 genes)** ANO5 CAPN3 CAV3 DAG1 DES DMD DNAJB6 DYSF FKRP FKTN GAA GMPPB ISPD LMNA MYOT PLEC PNPLA2 POMGNT1 POMK POMT1 POMT2 SGCA SGCB SGCD SGCG TCAP TNPO3 TRAPPC11 TRIM32 TTN **Add-on preliminary-evidence genes (3 genes)** HNRNPDL LIMS2 TOR1AIP1 **Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene (1 gene)** SMCHD1
Invitae Comprehensive Myopathy Panel (up to 52 genes)	**Primary panel (50 genes)** ACTA1 ANO5 ATP2A1 BAG3 BIN1 CACNA1S CAV3 CCDC78 CFL2 CNTN1 COL6A1 COL6A2 COL6A3 CPT2 CRYAB DES DNAJB6 DNM2 DYSF FHL1 FKBP14 FLNC GNE 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 genes (2 genes)** COL12A1 MYF6
Invitae Congenital Myopathy Panel (up to 28 genes)	**Primary panel (26 genes)** ACTA1 BIN1 CCDC78 CFL2 CNTN1 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 genes (2 genes)** COL12A1 MYF6
Invitae Autophagic Vacuolar Myopathy Panel (3 genes)	**Primary panel (3 genes)** DES LAMP2 VMA21
Invitae Central Core Disease Test (1 gene)	**Primary panel (1 gene)** RYR1
Invitae Centronuclear Myopathy Panel (up to 7 genes)	**Primary panel (6 genes)** BIN1 CCDC78 DNM2 MTM1 RYR1 TTN **Add-on preliminary-evidence gene (1 gene)** MYF6
Invitae Congenital Fiber-Type Disproportion Panel (7 genes)	**Primary panel (7 genes)** ACTA1 LMNA MYH7 RYR1 SELENON TPM2 TPM3
Invitae Distal Myopathy Panel (18 genes)	**Primary panel (18 genes)** ANO5 BAG3 CAV3 CRYAB DES DNAJB6 DYSF FHL1 FLNC GNE LDB3 MATR3 MYH7 MYOT SQSTM1 TIA1 TTN VCP
Invitae Hyperkalemic Periodic Paralysis Test (1 gene)	**Primary panel (1 gene)** SCN4A
Invitae Hypokalemic Periodic Paralysis Panel (3 genes)	**Primary panel (3 genes)** CACNA1S KCNJ2 SCN4A

INVITAE NEUROLOGY GENE PANEL TESTS (continued)

NEUROMUSCULAR DISORDERS (continued)

Invitae Inclusion Body Myopathy Panel (up to 5 genes)

Primary panel (4 genes)
GNE MYH2 TTN VCP

Add-on preliminary-evidence gene (1 gene)
HNRNPA2B1

Invitae Multiminicore Disease Panel (2 genes)

Primary panel (2 genes)
RYR1 SELENON

Invitae Myofibrillar Myopathy Panel (8 genes)

Primary panel (8 genes)
BAG3 CRYAB DES DNAJB6 FHL1 FLNC LDB3 MYOT

Invitae Nemaline Myopathy Panel (11 genes)

Primary panel (11 genes)
ACTA1 CFL2 KBTBD13 KLHL40 KLHL41 LMOD3 MYPN NEB TNNT1 TPM2 TPM3

Invitae Type VI Collagenopathy Panel (up to 4 genes)

Primary panel (3 genes)
COL6A1 COL6A2 COL6A3

Add-on preliminary-evidence gene (1 gene)
COL12A1

Invitae Myotonia and Paramyotonia Congenita Panel (2 genes)

Primary panel (2 genes)
CLCN1 SCN4A

Invitae Spinal Muscular Atrophy Panel (2 genes)

Primary panel (2 genes)
SMN1, SMN2

NEUROPATHIES AND RELATED DISORDERS

Invitae Comprehensive Neuropathies Panel (up to 83 genes)

Primary panel (70 genes)
AARS AIFM1 ATL1 ATL3 ATP7A BICD2 BSCL2 CHCHD10 DCTN1 DNAJB2 DNM2 DNMT1
DST DYNC1H1 EGR2 FAM134B FBXO38 FGD4 FIG4 GAN GARS GDAP1 GJB1 GNB4
HARS HINT1 HSPB1 HSPB8 IGHMBP2 IKBKAP INF2 KIF1A LITAF LMNA LRSAM1 MED25
MFN2 MORC2 MPZ MTMR2 NDRG1 NEFL NGF NTRK1 PDK3 PLEKHG5 PMP22 PRPS1
PRX RAB7A REEP1 SBF2 SCN11A SCN9A SH3TC2 SIGMAR1 SLC52A2 SLC52A3 SLC5A7 SPG11
SPTLC1 SPTLC2 TFG TRIM2 TRPV4 TTR UBA1 VAPB WNK1 YARS

Add-on preliminary-evidence genes (11 genes)
CCT5 FLRT1 HSPB3 LAS1L MARS PRDM12 SCN10A SETX SLC25A46 SURF1 VRK1

Add-on spinal muscular atrophy genes (2 genes)
SMN1, SMN2

Invitae Charcot-Marie-Tooth Disease Comprehensive Panel (up to 45 genes)

Primary panel (42 genes)
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 SPG11 TFG TRIM2 TRPV4 YARS

Add-on preliminary-evidence genes (3 genes)
MARS SLC25A46 SURF1

Invitae Charcot-Marie-Tooth Disease Autosomal Dominant Panel (up to 25 genes)

Primary panel (24 genes)
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 gene)
MARS

Invitae Charcot-Marie-Tooth Disease Autosomal Recessive Panel (up to 22 genes)

Primary panel (20 genes)
DNAJB2 EGR2 FGD4 FIG4 GDAP1 HINT1 IGHMBP2 LMNA LRSAM1 MED25 MFN2 MTMR2
NDRG1 NEFL PLEKHG5 PRX SBF2 SH3TC2 SPG11 TRIM2

Add-on preliminary-evidence genes (2 genes)
SLC25A46 SURF1

Invitae Charcot-Marie-Tooth Disease X-linked Panel (4 genes)

Primary panel (4 genes)
AIFM1 GJB1 PDK3 PRPS1

Invitae Hereditary Sensory and Autonomic Neuropathy Panel (up to 17 genes)

Primary panel (15 genes)
ATL1 ATL3 DNMT1 DST FAM134B IKBKAP KIF1A NGF NTRK1 RAB7A SCN11A SCN9A
SPTLC1 SPTLC2 WNK1

Add-on preliminary-evidence genes (2 genes)
CCT5 PRDM12

Invitae Familial Dysautonomia Test (1 gene)

Primary panel (1 gene)
IKBKAP

Invitae Hereditary Motor Neuropathies Panel (up to 24 genes)

Primary panel (22 genes)
ATP7A BICD2 BSCL2 CHCHD10 DCTN1 DNAJB2 DYNC1H1 FBXO38 GARS HINT1 HSPB1 HSPB8
IGHMBP2 PLEKHG5 REEP1 SIGMAR SLC5A7 SMN1, SMN2 TRPV4 UBA1 VAPB

Add-on preliminary-evidence genes (2 genes)
HSPB3 VRK1

Invitae Spinal Muscular Atrophy Panel (2 genes)

Primary panel (2 genes)
SMN1, SMN2

INVITAE NEUROLOGY GENE PANEL TESTS (continued)

NEUROPATHIES AND RELATED DISORDERS (continued)

Invitae Small Fiber Neuropathy Test (up to 2 genes)

Primary panel (1 gene)

SCN9A

Add-on preliminary-evidence gene (1 gene)

SCN10A

Invitae Riboflavin Transporter Deficiency Neuronopathy Panel (2 genes)

Primary panel (2 genes)

SLC52A2 SLC52A3

Invitae Hereditary Spastic Paraplegia Comprehensive Panel (up to 64 genes)

Primary panel (43 genes)

ABCD1	ALDH18A1	ALS2	AP4B1	AP4E1	AP4M1	AP4S1	AP5Z1	ATL1	B4GALNT1	BSCL2	C12orf65
CYP2U1	CYP7B1	DDHD1	DDHD2	ERLIN2	FA2H	GBA2	GJC2	HSPD1	KDM5C	KIF1A	KIF1C
KIF5A	L1CAM	NIPA1	NT5C2	PLP1	PNPLA6	REEP1	RTN2	SACS	SLC16A2	SPAST	SPG11
SPG20	SPG21	SPG7	TECPR2	VAMP1	WASHC5	ZFYVE26					

Add-on preliminary-evidence genes (21 genes)

AMPD2	ARL6IP1	ARSI	ATP13A2	C19orf12	CCT5	CPT1C	ENTPD1	ERLIN1	EXOSC3	IBA57	MAG
PGAP1	RAB3GAP2	REEP2	SLC33A1	TFG	USP8	VPS37A	ZFR	ZFYVE27			

Invitae Hereditary Spastic Paraplegia Autosomal Dominant Panel (up to 16 genes)

Primary panel (12 genes)

ALDH18A1	ATL1	BSCL2	HSPD1	KIF1A	KIF5A	NIPA1	REEP1	RTN2	SPAST	VAMP1	WASHC5

Add-on preliminary-evidence genes (4 genes)

CPT1C REEP2 SLC33A1 ZFYVE27

Invitae Hereditary Spastic Paraplegia Autosomal Recessive Panel (up to 46 genes)

Primary panel (28 genes)

ALDH18A1	ALS2	AP4B1	AP4E1	AP4M1	AP4S1	AP5Z1	B4GALNT1	C12orf65	CYP2U1	CYP7B1	DDHD1
DDHD2	ERLIN2	FA2H	GBA2	GJC2	KIF1A	KIF1C	NT5C2	PNPLA6	SACS	SPG11	SPG20
SPG21	SPG7	TECPR2	ZFYVE26								

Add-on preliminary-evidence genes (18 genes)

AMPD2	ARL6IP1	ARSI	ATP13A2	C19orf12	CCT5	ENTPD1	ERLIN1	EXOSC3	IBA57	MAG	PGAP1
RAB3GAP2	REEP2	TFG	USP8	VPS37A	ZFR						

Invitae Hereditary Spastic Paraplegia X-linked Panel (5 genes)

Primary panel (5 genes)

ABCD1 KDM5C L1CAM PLP1 SLC16A2

CARDIOMYOPATHY AND SKELETAL MUSCLE DISEASE

Invitae Cardiomyopathy and Skeletal Muscle Disease Panel (up to 158 genes)

Primary panel (113 genes)

ABCC9	ACTA1	ACTC1	ACTN2	AGL	ANO5	ATP2A1	B3GALNT2	B4GAT1	BAG3	BIN1	CACNA1C
CAPN3	CAV3	CCDC78	CFL2	CHKB	CNTN1	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
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	RYR1	RYR2
SCN5A	SELENON	SGCA	SGCB	SGCD	SGCG	SLC22A5	SQSTM1	STAC3	STIM1	TAZ	TCAP
TIA1	TMEM43	TMEM5	TNNC1	TNNI3	TNNT1	TNNT2	TNPO3	TPM1	TPM2	TPM3	TRAPPC11
TRIM32	TTN	TTR	VCL	VCP							

Add-on preliminary-evidence genes (38 genes)

ANKRD1	CALR3	CHRM2	COL12A1	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
TOR1AIP1	TXNRD2										

Add-on autosomal recessive syndromic pediatric cardiomyopathy genes (7 genes)

ACADVL	ALMS1	DNAJC19	ELAC2	MTO1	SDHA	TMEM70

INVITAE NEUROLOGY GENE PANEL TESTS (continued)

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

EPILEPSY

Invitae Epilepsy Panel (up to 189 genes)

Primary panel (125 genes)

ADSL	ALDH5A1	ALDH7A1	ALG13	ARHGEF9	ARX	ATP1A2	ATP1A3	ATRX	BRAT1	C12orf57	CACNA2D2
CASK	CDKL5	CHD2	CHRNA2	CHRNA4	CHRNB2	CLN2 (TPP1)	CLN3	CLN5	CLN6	CLN8	CNTNAP2
CSTB	CTSD	DEPDC5	DNAJC5	DNM1	DYRK1A	EEF1A2	EFHC1	EHMT1	EPM2A	FOLR1	FOXG1
FRRS1L	GABRA1	GABRB3	GABRG2	GAMT	GATM	GLDC	GLRA1	GNAO1	GOSR2	GRIN1	GRIN2A
GRIN2B	HCN1	HNRNPU	IER3IP1	IQSEC2	ITPA	KANSL1	KCNA2	KCNB1	KCNC1	KCNH2	KCNJ10
KCNQ2	KCNQ3	KCNT1	KCTD7	KIAA2022	LGI1	LIAS	MBD5	MECP2	MEF2C	MFSD8	NGLY1
NHLRC1	NRXN1	PACS1	PCDH19	PIGA	PIGN	PIGO	PLCB1	PNKD	PNKP	PNPO	POLG
PPT1	PRICKLE1	PRRT2	PURA	QARS	ROGDI	SATB2	SCARB2	SCN1A	SCN1B	SCN2A	SCN3A
SCN8A	SCN9A	SERPINI1	SGCE	SLC13A5	SLC19A3	SLC25A22	SLC2A1	SLC35A2	SLC6A1	SLC6A8	SLC9A6
SNX27	SPATA5	SPTAN1	STX1B	STXBP1	SYN1	SYNGAP1	SYNJ1	SZT2	TBC1D24	TCF4	TSC1
TSC2	UBE3A	WWOX	ZDHHC9	ZEB2							

Add-on preliminary-evidence genes (58 genes)

ABAT	ARHGEF15	ATP6AP2	CACNA1A	CACNA1H	CACNB4	CARS2	CASR	CBL	CERS1	CLCN4	CNTN2
COQ4	CPA6	DIAPH1	DOCK7	FARS2	FASN	GABBR2	GABRB2	GABRD	GAL	GPHN	JMJD1C
KCNA1	KCND2	KCNH5	KCNMA1	KPNA7	LMNB2	MTOR	NECAP1	NEDD4L	NPRL3	PIGG	PIGQ
PIK3AP1	PRDM8	PRICKLE2	PRIMA1	RBFOX1	RBFOX3	RELN	RYR3	SCN5A	SETD2	SIK1	SLC12A5
SLC25A12	SLC35A3	SNAP25	SRPX2	ST3GAL3	ST3GAL5	STRADA	TBL1XR1	TPK1	WDR45		

Add-on genes for glycine encephalopathy (3 genes)

AMT GCSH GLDC

Add-on FLNA gene (1 gene)

FLNA

Add-on PTEN gene (1 gene)

PTEN

Add-on RANBP2 gene (1 gene)

RANBP2

Invitae Alternating Hemiplegia of Childhood Panel (up to 5 genes)

Primary panel (2 genes)

ATP1A2 ATP1A3

Add-on clinically overlapping genes (3 genes)

CACNA1A SCN1A SLC2A1

Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel (2 genes)

Primary panel (2 genes)

ACTB ACTG1

Invitae Cerebral Cavernous Malformations Panel (3 genes)

Primary panel (3 genes)

CCM2 KRIT1 PDCD10

Invitae CHARGE Syndrome Test (1 gene)

Primary panel (1 gene)

CHD7

Invitae Early Infantile Epileptic Encephalopathy Panel (up to 63 genes)

Primary panel (49 genes)

ALDH7A1	ARHGEF9	ARX	BRAT1	CACNA2D2	CASK	CDKL5	CHD2	DNM1	EEF1A2	FOLR1	FRRS1L
GABRA1	GABRB3	GNAO1	GRIN1	GRIN2B	HCN1	HNRNPU	IER3IP1	KCNA2	KCNB1	KCNQ2	KCNQ3
KCNT1	PCDH19	PIGA	PIGN	PIGO	PLCB1	PNKP	PNPO	PURA	SCN1A	SCN2A	SCN8A
SCN9A	SLC2A1	SLC13A5	SLC25A22	SLC35A2	SLC6A1	SMC1A	SPTAN1	STXBP1	SYNGAP1	SZT2	TBC1D24
WWOX											

Add-on preliminary-evidence genes (14 genes)

ARHGEF15	CLCN4	COQ4	DOCK7	GABBR2	GPHN	KCNH5	MTOR	NECAP1	NEDD4L	SCN1B	SIK1
SLC12A5	ST3GAL3										

Invitae Holoprosencephaly Panel (up to 9 genes)

Primary panel (5 genes)

GLI2 SHH SIX3 TGIF1 ZIC2

Add-on preliminary-evidence genes (4 genes)

CDON FOXH1 NODAL PTCH1

Invitae Neurodegeneration with Brain Iron Accumulation Panel (up to 14 genes)

Primary panel (11 genes)

ATP13A2 C19orf12 COASY CP DCAF17 FTL FUCA1 PANK2 PLA2G6 SQSTM1 WDR45

Add-on preliminary-evidence genes (3 genes)

FA2H KIF1A TRIM32

Invitae Rett/Angelman and Related Disorders Panel (up to 26 genes)

Primary panel (22 genes)

ADSL	ALDH5A1	ATRX	CDKL5	CNTNAP2	DYRK1A	EHMT1	FOXG1	IQSEC2	KANSL1	MBD5	MECP2
MEF2C	NGLY1	NRXN1	SATB2	SCN8A	SLC9A6	STXBP1	TCF4	UBE3A	ZEB2		

Add-on preliminary-evidence genes (4 genes)

GABRD JMJD1C TBL1XR1 WDR45

Invitae Tuberous Sclerosis Complex Panel (2 genes)

Primary panel (2 genes)

TSC1 TSC2

