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

Add-on preliminary-evidence genes (5 genes)
CHMP2B HNRNPA2B1 MATR3 SIGMAR1 SQSTM1

Invitae Amyotrophic Lateral Sclerosis Panel (up to 19 genes)

Primary panel (15 genes)
ALS2 CHCHD10 DCTN1 FUS OPTN PFN1 SETX SOD1 SPG11 TARDBP TBK1 TFG
UBQLN2 VAPB VCP

Add-on preliminary-evidence genes (4 genes)
CHMP2B MATR3 SIGMAR1 SQSTM1

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 122 genes)

Primary panel (107 genes)
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 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 SLC5A7 SMN1, SMN2 SQSTM1 STAC3 STIM1 TAZ TCAP TIA1 TMEM5
TNNT1 TNPO3 TOR1AIP1 TPM2 TPM3 TRAPPC11 TRIM32 TTN VCP VMA21

Add-on preliminary-evidence genes (14 genes)
ALG14 HNRNPA2B1 HNRNPDL LAMB2 LIMS2 LRP4 MYF6 PREPL SNAP25 SUN1 SUN2 SYNE1
SYNE2 TMEM43

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

Invitae Congenital Myasthenic Syndrome Panel (up to 22 genes)

Primary panel (14 genes)
AGRN ALG2 CHAT CHRNA1 CHRNB1 CHRND CHRNE COLQ DOK7 DPAGT1 GFPT1 MUSK
RAPSN SLC5A7

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 NEUROLOGY GENE PANEL TESTS (continued)

NEUROMUSCULAR DISORDERS (continued)

Invitae Comprehensive Muscular Dystrophy Panel (up to 56 genes)	**Primary panel (48 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
	ISPD	ITGA7	LAMA2	LARGE1	LMNA	MYOT	PLEC	PNPLA2	POMGNT1	POMGNT2	POMK	POMT1
	POMT2	SGCA	SGCB	SGCD	SGCG	TCAP	TMEM5	TNPO3	TOR1AIP1	TRAPPC11	TRIM32	TTN
	Add-on preliminary-evidence genes (7 genes)											
	HNRNPDL	LIMS2	SUN1	SUN2	SYNE1	SYNE2	TMEM43					
	Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene (1 gene)											
	SMCHD1											
Invitae Congenital Muscular Dystrophy Panel (27 genes)	**Primary panel (27 genes)**											
	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	TCAP	TMEM5									
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 (31 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	TOR1AIP1	TRAPPC11	TRIM32	TTN					
	Add-on preliminary-evidence genes (2 genes)											
	HNRNPDL	LIMS2										
	Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene (1 gene)											
	SMCHD1											
Invitae Comprehensive Myopathy Panel (up to 52 genes)	**Primary panel (51 genes)**											
	ACTA1	ANO5	ATP2A1	BAG3	BIN1	CACNA1S	CAV3	CCDC78	CFL2	CNTN1	COL12A1	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 (1 gene)											
	MYF6											
Invitae Congenital Myopathy Panel (up to 28 genes)	**Primary panel (27 genes)**											
	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 genes (1 gene)											
	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 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 NEUROLOGY GENE PANEL TESTS (continued)

NEUROMUSCULAR DISORDERS (continued)

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 Periodic Paralysis Panel (3 genes)

Primary panel (3 genes)
CACNA1S KCNJ2 SCN4A

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 (72 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 SLC25A46 SLC52A2 SLC52A3 SLC5A7
SPG11 SPTLC1 SPTLC2 TFG TRIM2 TRPV4 TTR UBA1 VAPB VRK1 WNK1 YARS

Add-on preliminary-evidence genes (9 genes)
CCT5 FLRT1 HSPB3 LAS1L MARS PRDM12 SCN10A SETX SURF1

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

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

Primary panel (43 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 SLC25A46 SPG11 TFG TRIM2 TRPV4 YARS

Add-on preliminary-evidence genes (2 genes)
MARS 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 (21 genes)
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 gene)
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 Neuropathy Panel (up to 24 genes)

Primary panel (23 genes)
ATP7A BICD2 BSCL2 CHCHD10 DCTN1 DNAJB2 DYNC1H1 FBXO38 GARS HINT1 HSPB1 HSPB8
IGHMBP2 PLEKHG5 REEP1 SIGMAR SLC5A7 SMN1, SMN2 TRPV4 UBA1 VAPB VRK1

Add-on preliminary-evidence genes (1 gene)
HSPB3

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 (115 genes)

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	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	TOR1AIP1	TPM1	TPM2
TPM3	TRAPPC11	TRIM32	TTN	TTR	VCL	VCP					

Add-on preliminary-evidence genes (36 genes)

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 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 (133 genes)

ADSL	ALDH5A1	ALDH7A1	ALG13	ARHGEF9	ARX	ATP1A2	ATP1A3	ATRX	BRAT1	C12orf57	CACNA1A
CACNA2D2	CASK	CDKL5	CHD2	CHRNA2	CHRNA4	CHRNB2	CLCN4	CLN2 (TPP1)	CLN3	CLN5	CLN6
CLN8	CNTNAP2	CSTB	CTSD	DEPDC5	DNAJC5	DNM1	DOCK7	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	MTOR	NEDD4L	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	SLC12A5	SLC13A5	SLC19A3
SLC25A22	SLC2A1	SLC35A2	SLC6A1	SLC6A8	SLC9A6	SNX27	SPATA5	SPTAN1	ST3GAL5	STRADA	STX1B
STXBP1	SYN1	SYNGAP1	SYNJ1	SZT2	TBC1D24	TCF4	TSC1	TSC2	UBE3A	WWOX	ZDHHC9
ZEB2											

Add-on preliminary-evidence genes (50 genes)

ABAT	ARHGEF15	ATP6AP2	CACNA1H	CACNB4	CARS2	CASR	CBL	CERS1	CNTN2	COQ4	CPA6
DIAPH1	FARS2	FASN	GABBR2	GABRB2	GABRD	GAL	GPHN	JMJD1C	KCNA1	KCND2	KCNH5
KCNMA1	KPNA7	LMNB2	NECAP1	NPRL3	PIGG	PIGQ	PIK3AP1	PRDM8	PRICKLE2	PRIMA1	RBFOX1
RBFOX3	RELN	RYR3	SCN5A	SETD2	SIK1	SLC25A12	SLC35A3	SNAP25	SRPX2	ST3GAL3	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 65 genes)

Primary panel (53 genes)

ALDH7A1	ARHGEF9	ARX	BRAT1	CACNA2D2	CASK	CDKL5	CHD2	CLCN4	DNM1	DOCK7	EEF1A2
FOLR1	FRRS1L	GABRA1	GABRB3	GNAO1	GRIN1	GRIN2A	GRIN2B	HCN1	HNRNPU	IER3IP1	KCNA2
KCNB1	KCNQ2	KCNQ3	KCNT1	PCDH19	PIGA	PIGN	PIGO	PLCB1	PNKP	PNPO	PURA
SCN1A	SCN2A	SCN8A	SCN9A	SLC12A5	SLC13A5	SLC25A22	SLC2A1	SLC35A2	SLC6A1	SMC1A	SPTAN1
STXBP1	SYNGAP1	SZT2	TBC1D24	WWOX							

Add-on preliminary-evidence genes (12 genes)

ARHGEF15	ATP1A2	COQ4	GABBR2	GPHN	KCNH5	MTOR	NECAP1	NEDD4L	SCN1B	SIK1	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

