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 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 (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

