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

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

INVITAE PEDIATRIC GENE PANEL TESTS

Our testing menu contains a broad variety of panels curated by medical genetics experts for conditions that have pediatric onset, including epilepsies, brain malformations, ciliopathies, RASopathies, overgrowth syndromes, skeletal disorders, developmental disorders, and disorders of sex development.

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

CILIOPATHIES

Invitae Ciliopathies Panel (102 genes)

Primary panel (102 genes)

AHI1 ANKS6 ARL13B ARL6 ARMC4 B9D1 B9D2 BBS1 BBS10 BBS12 BBS2 BBS4
BBS5 BBS7 BBS9 C21orf59 C5orf42 CC2D2A CCDC103 CCDC114 CCDC151 CCDC39 CCDC40 CCDC65
CCNO CEP104 CEP120 CEP164 CEP290 CEP41 CEP83 CSPP1 DCDC2 DNAAF1 DNAAF2 DNAAF3
DNAAF5 DNAH1 DNAH11 DNAH5 DNAH8 DNAI1 DNAI2 DNAL1 DRC1 DYNC2H1 DYX1C1 EVC
EVC2 GAS8 GLIS2 IFT122 IFT140 IFT172 IFT80 INPP5E INVS IQCB1 KIAA0586 KIF7
LRRC6 MCIDAS MKKS MKS1 MRE11A NEK1 NEK8 NME8 NPHP1 NPHP3 NPHP4 OFD1
PDE6D PKD2 PKHD1 RPGR RPGRIP1L RSPH1 RSPH3 RSPH4A RSPH9 SDCCAG8 SPAG1 TCTN1
TCTN2 TCTN3 TMEM138 TMEM216 TMEM231 TMEM237 TMEM67 TRIM32 TTC21B TTC8 WDPCP WDR19
WDR34 WDR35 WDR60 XPNPEP3 ZMYND10 ZNF423

Invitae Skeletal Ciliopathies Panel (up to 18 genes)

Primary panel (17 genes)

CEP120 CSPP1 DYNC2H1 EVC EVC2 IFT80 IFT122 IFT140 IFT172 KIAA0586 NEK1 TCTN3
TTC21B WDR19 WDR34 WDR35 WDR60

Add-on FGFR3-related thanatophoric dysplasia gene (1 gene)

FGFR3

Invitae Primary Ciliary Dyskinesia Panel (up to 36 genes)

Primary panel (34 genes)

ARMC4 C21orf59 CCDC103 CCDC114 CCDC151 CCDC39 CCDC40 CCDC65 CCNO DNAAF1 DNAAF2 DNAAF3
DNAAF5 DNAH1 DNAH11 DNAH5 DNAH8 DNAI1 DNAI2 DNAL1 DRC1 DYX1C1 GAS8 LRRC6
MCIDAS NME8 OFD1 RPGR RSPH1 RSPH3 RSPH4A RSPH9 SPAG1 ZMYND10

Add-on preliminary-evidence gene (1 gene)

INVS

Add-on clinically overlapping gene (1 gene)

CFTR

Invitae Bardet-Biedl Syndrome Panel (16 genes)

Primary panel (16 genes)

ARL6 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 CEP290 MKKS MKS1
SDCCAG8 TRIM32 TTC8 WDPCP

Invitae Joubert and Meckel-Gruber Syndromes Panel (30 genes)

Primary panel (30 genes)

AHI1 ARL13B B9D1 B9D2 C5orf42 CC2D2A CEP104 CEP290 CEP41 CSPP1 INPP5E KIAA0586
KIF7 MKS1 MRE11A NPHP1 NPHP3 OFD1 PDE6D RPGRIP1L TCTN1 TCTN2 TCTN3 TMEM138
TMEM216 TMEM231 TMEM237 TMEM67 TTC21B ZNF423

Invitae Nephronophthisis Panel (27 genes)

Primary panel (27 genes)

AHI1 ANKS6 CC2D2A CEP164 CEP290 CEP83 DCDC2 GLIS2 IFT172 INVS IQCB1 NEK8
NPHP1 NPHP3 NPHP4 OFD1 PKHD1 RPGRIP1L SDCCAG8 TCTN1 TMEM216 TMEM237 TMEM67 TTC21B
WDR19 XPNPEP3 ZNF423

Invitae Oral-Facial-Digital Syndrome Type 1 Test (1 gene)

Primary panel (1 gene)

OFD1

Invitae Polycystic Kidney Disease Type 2 Panel (2 genes)

Primary panel (2 genes)

PKD2 PKHD1

Invitae Senior-Loken Syndrome Panel (8 genes)

Primary panel (8 genes)

CEP290 INVS IQCB1 NPHP1 NPHP3 NPHP4 SDCCAG8 WDR19

CONGENITAL HEART DEFECTS

Invitae Congenital Heart Defects and Heterotaxy Panel (up to 89 genes)

Primary panel (82 genes)

ACTC1 ACVR2B ALMS1 ANKS6 ARMC4 BBS10 BCOR BRAF C21orf59 CBL CCDC103 CCDC114
CCDC151 CCDC39 CCDC40 CCDC65 CCNO CEP290 CFAP53 CHD7 DNAAF1 DNAAF2 DNAAF3 DNAAF5
DNAH1 DNAH11 DNAH5 DNAH8 DNAI1 DNAI2 DNAL1 DRC1 DYX1C1 ELN FOXH1 GAS8
GATA4 GDF1 GJA1 GPC3 HRAS INVS JAG1 KRAS LEFTY2 LRRC6 MAP2K1 MAP2K2
MCIDAS MED13L MEIS2 MKS1 NEK8 NF1 NKX2-5 NKX2-6 NME8 NODAL NOTCH1 NOTCH2
NPHP3 NR2F2 NRAS NSD1 OFD1 PTPN11 RAF1 RIT1 RPGR RSPH1 RSPH3 RSPH4A
RSPH9 SHOC2 SOS1 SPAG1 TBX1 TBX5 TTC8 ZIC3 ZMYND10 ZNF423

Add-on preliminary-evidence genes (7 genes)

CFAP52 CRELD1 GATA6 HAND1 MYH6 SMAD6 ZFPM2

CYSTIC FIBROSIS AND CHRONIC PANCREATITIS

Invitae Cystic Fibrosis Test (up to 5 genes)

Primary panel (1 gene)

CFTR

Add-on chronic pancreatitis genes (4 genes)

CASR CTRC PRSS1 SPINK1

Invitae Chronic Pancreatitis Panel (5 genes)

Primary panel (5 genes)

CASR CFTR CTRC PRSS1 SPINK1

INVITAE

DEVELOPMENTAL DISORDERS

Invitae Alagille Syndrome Panel (2 genes)	Primary panel (2 genes) JAG1 NOTCH2
Invitae Alpha Thalassemia X-linked Intellectual Disability Test (1 gene)	Primary panel (1 gene) ATRX
Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel (2 genes)	Primary panel (2 genes) ACTB ACTG1
Invitae Branchiootorenal Spectrum Disorders Panel (up to 3 genes)	Primary panel (2 genes) EYA1 SIX1
	Add-on Townes-Brocks syndrome gene (1 gene) SALL1
Invitae Carpenter Syndrome Panel (2 genes)	Primary panel (2 genes) MEGF8 RAB23
Invitae CASR-Related Conditions Test (1 gene)	Primary panel (1 gene) CASR
Invitae CHARGE Syndrome Test (1 gene)	Primary panel (1 gene) CHD7
Invitae CHOPS Syndrome Test (1 gene)	Primary panel (1 gene) AFF4
Invitae Coffin-Lowry Syndrome Test (1 gene)	Primary panel (1 gene) RPS6KA3
Invitae Cohen Syndrome Test (1 gene)	Primary panel (1 gene) VPS13B
Invitae Cornelia de Lange Syndrome Panel (up to 6 genes)	Primary panel (5 genes) HDAC8 NIPBL RAD21 SMC1A SMC3
	Add-on preliminary-evidence genes (1 gene) EP300
Invitae Glass Syndrome Test (1 gene)	Primary panel (1 gene) SATB2
Invitae GLI3-Related Disorders Test (1 gene)	Primary panel (1 gene) GLI3
Invitae HPRT1-Related Disorders Test (1 gene)	Primary panel (1 gene) HPRT1
Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel (3 genes)	Primary panel (3 genes) ANOS1 CHD7 FGFR1
Invitae Kabuki Syndrome Panel (2 genes)	Primary panel (2 genes) KDM6A KMT2D
Invitae KAT6B-Related Disorders Test (1 gene)	Primary panel (1 gene) KAT6B
Invitae KBG Syndrome Test (1 gene)	Primary panel (1 gene) ANKRD11
Invitae MED12-Related Disorders Test (1 gene)	Primary panel (1 gene) MED12
Invitae Oculo-Facio-Cardio-Dental Syndrome Test (1 gene)	Primary panel (1 gene) BCOR
Invitae PTEN-Related Disorders Test (1 gene)	Primary panel (1 gene) PTEN
Invitae Renpenning Syndrome Test (1 gene)	Primary panel (1 gene) PQBP1
Invitae Rubinstein-Taybi Syndrome Panel (2 genes)	Primary panel (2 genes) CREBBP EP300
Invitae Simpson-Golabi-Behmel Syndrome Test (1 gene)	Primary panel (1 gene) GPC3
Invitae Smith-Lemli-Opitz Syndrome Test (1 gene)	Primary panel (1 gene) DHCR7
Invitae Sotos Syndrome Test (1 gene)	Primary panel (1 gene) NSD1
Invitae van der Woude Syndrome Panel (2 genes)	Primary panel (2 genes) GRHL3 IRF6
Invitae von Hippel-Lindau Syndrome Test (1 gene)	Primary panel (1 gene) VHL
Invitae WAGR Syndrome Panel (2 genes)	Primary panel (2 genes) PAX6 WT1
Invitae Weaver Syndrome Test (1 gene)	Primary panel (1 gene) EZH2

INVITAE PEDIATRIC GENE PANEL TESTS (continued)

DISORDERS OF SEX DEVELOPMENT/ENDOCRINOLOGY

Invitae Disorders of Male Sex Development Panel (up to 15 genes)

Primary panel (8 genes)
AR DHH MAP3K1 NR0B1 NR5A1 SRD5A2 SRY WT1

Add-on Kallman syndrome genes (4 genes)
ANOS1 CHD7 FGFR1 HESX1

Add-on alpha-thalassemia X-linked intellectual disability (1 gene)
ATRX

Add-on campomelic dysplasia gene (1 gene)
SOX9

Add-on Smith-Lemli-Opitz syndrome gene (1 gene)
DHCR7

Invitae Disorders of Female Sex Development Test (1 gene)

Primary panel (1 gene)
SRY

Invitae Androgen Insensitivity Panel (2 genes)

Primary panel (2 genes)
AR SRD5A2

Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel (3 genes)

Primary panel (3 genes)
ANOS1 CHD7 FGFR1

EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES

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 PEDIATRIC GENE PANEL TESTS (continued)

EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES (continued)

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

EYE DISORDERS

Invitae Aniridia Test (1 gene)

Primary panel (1 gene)
PAX6

Invitae Axenfeld-Rieger Panel (2 genes)

Primary panel (2 genes)
FOXC1 PITX2

Add-on aniridia gene (1 gene)
PAX6

Invitae Bardet-Biedl Syndrome Panel (16 genes)

Primary panel (16 genes)
ARL6 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 CEP290 MKKS MKS1
SDCCAG8 TRIM32 TTC8 WDPCP

Invitae CHARGE Syndrome Test (1 gene)

Primary panel (1 gene)
CHD7

Invitae Choroideremia Test (1 gene)

Primary panel (1 gene)
CHM

Invitae Congenital Cataracts Panel (up to 38 gene)

Primary panel (33 genes)
AGK BCOR BFSP1 BFSP2 CRYAA CRYAB CRYBA1 CRYBA4 CRYBB1 CRYBB2 CRYBB3 CRYGC
CRYGD CRYGS CTDP1 EPHA2 FAM126A FOXC1 FYCO1 GALK1 GCNT2 GJA3 GJA8 HSF4
MAF MIP NHS OCRL PAX6 PITX2 PITX3 SIL1 VSX2

Add-on preliminary-evidence genes (5 genes)
CHMP4B CRYGB LIM2 TDRD7 VIM

Invitae Duane-Radial Ray Syndrome Test (1 gene)

Primary panel (1 gene)
SALL4

Invitae Early-Onset Glaucoma Panel (3 genes)

Primary panel (3 genes)
CYP1B1 FOXC1 PITX2

Invitae Microphthalmia/Anophthalmia Disorders Panel (up to 20 genes)

Primary panel (17 genes)
ALDH1A3 BCOR BMP4 FOXE3 GDF6 MAB21L2 MFRP OTX2 PAX2 PRSS56 PXDN RARB
RAX SHH SOX2 STRA6 VSX2

Add-on preliminary-evidence genes (3 genes)
GDF3 HESX1 VAX1

Invitae Leber Congenital Amaurosis Panel (up to 21 genes)

Primary panel (19 genes)
AIPL1 CEP290 CRB1 CRX GDF6 GUCY2D IQCB1 KCNJ13 LCA5 LRAT NMNAT1 OTX2
PRPH2 RD3 RDH12 RPE65 RPGRIP1 SPATA7 TULP1

Add-on preliminary-evidence genes (2 genes)
BBS4 IMPDH1

Invitae Oculo-Facio-Cardio-Dental Syndrome Test (1 gene)

Primary panel (1 gene)
BCOR

Invitae Retinoblastoma Test (1 gene)

Primary panel (1 gene)
RB1

Invitae Senior-Loken Syndrome Panel (8 genes)

Primary panel (8 genes)
CEP290 INVS IQCB1 NPHP1 NPHP3 NPHP4 SDCCAG8 WDR19

OVERGROWTH SYNDROMES

Invitae Overgrowth and Macrocephaly Syndromes Panel (up to 25 genes)

Primary panel (20 genes)
AKT2 AKT3 CDKN1C CUL4B DIS3L2 DNMT3A EZH2 GLI3 GPC3 KPTN MED12 NF1
NFIX NPR2 NSD1 PHF6 PIK3R2 PTEN SETD2 SPRED1

Add-on preliminary-evidence genes (5 genes)
DICER1 EED PDGFRB RNF125 UPF3B

Invitae Perlman Syndrome Test (1 gene)

Primary panel (1 gene)
DIS3L2

Invitae Proteus Syndrome Test (1 gene)

Primary panel (1 gene)
AKT1

Invitae PTEN-Related Disorders Test (1 gene)

Primary panel (1 gene)
PTEN

INVITAE PEDIATRIC GENE PANEL TESTS (continued)

OVERGROWTH SYNDROMES (continued)

Invitae Simpson-Golabi-Behmel Syndrome Test (1 gene)	Primary panel (1 gene) GPC3
Invitae Sotos Syndrome Test (1 gene)	Primary panel (1 gene) NSD1
Invitae Weaver Syndrome Test (1 gene)	Primary panel (1 gene) EZH2

RASOPATHIES (NOONAN SPECTRUM DISORDERS)

Invitae RASopathies Comprehensive Panel (18 genes)	Primary panel (18 genes) A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RAF1 RASA1 RIT1 RRAS SHOC2 SOS1 SOS2 SPRED1
Invitae Cardio-Facio-Cutaneous Syndrome Panel (6 genes)	Primary panel (6 genes) BRAF KRAS MAP2K1 MAP2K2 SHOC2 SOS1
Invitae Costello Syndrome Test (1 gene)	Primary panel (1 gene) HRAS
Invitae Legius Syndrome Test (up to 2 genes)	Primary panel (1 gene) SPRED1 Add-on Legius syndrome gene (1 gene) NF1
Invitae Neurofibromatosis Type 1 Test (up to 2 genes)	Primary panel (1 gene) NF1 Add-on neurofibromatosis type 1 gene (1 gene) SPRED1
Invitae Noonan Syndrome Panel (up to 16 genes)	Primary panel (14 genes) A2ML1 BRAF CBL KRAS MAP2K1 MAP2K2 NRAS PTPN11 RAF1 RIT1 RRAS SHOC2 SOS1 SOS2 Add-on Baraitser-Winter cerebrofrontofacial syndrome genes (2 genes) ACTB ACTG1
Invitae Noonan Syndrome with Multiple Lentigines Pane (3 genes)	Primary panel (3 genes) BRAF PTPN11 RAF1

SKELETAL DISORDERS

Invitae Antley-Bixler Syndrome Test (up to 2 genes)	Primary panel (1 gene) POR Add-on craniosynostosis gene (1 gene) FGFR2
Invitae ARSE-Related Chondrodysplasia Punctata Test (up to 2 genes)	Primary panel (1 gene) ARSE Add-on NSDHL-related disorders gene (1 gene) NSDHL
Invitae Campomelic Dysplasia Test (1 gene)	Primary panel (1 gene) SOX9
Invitae Craniosynostosis Panel (up to 11 genes)	Primary panel (9 genes) ERF FGFR1 FGFR2 FGFR3 GLI3 MEGF8 MSX2 RAB23 TWIST1 Add-on 3MC and Treacher-Collins syndromes genes (2 genes) MASP1 TCOF1
Invitae Duane-Radial Ray Syndrome Test (1 gene)	Primary panel (1 gene) SALL4
Invitae Ellis van Creveld and Weyers Acrofacial Dysostosis Panel (2 genes)	Primary panel (2 genes) EVC EVC2
Invitae FGFR3-Related Disorders Test (1 gene)	Primary panel (1 gene) FGFR3
Invitae Hereditary Multiple Osteochondromas Panel (up to 3 genes)	Primary panel (2 genes) EXT1 EXT2 Add-on Langer-Giedion syndrome gene (1 gene) TRPS1
Invitae Holt-Oram Syndrome Test (1 gene)	Primary panel (1 gene) TBX5
Invitae NSDHL-Related Disorders Test (1 gene)	Primary panel (1 gene) NSDHL
Invitae Osteogenesis Imperfecta Panel (4 genes)	Primary panel (4 genes) COL1A1 COL1A2 CRTAP P3H1

SKELETAL DISORDERS (continued)

Invitae Skeletal Ciliopathies Panel (up to 18 genes)	Primary panel (17 genes) CEP120 CSPP1 DYNC2H1 EVC EVC2 IFT80 IFT122 IFT140 IFT172 KIAA0586 NEK1 TCTN3 TTC21B WDR19 WDR34 WDR35 WDR60 Add-on FGFR3-related thanatophoric dysplasia gene (1 gene) FGFR3
Invitae Thrombocytopenia Absent Radius Syndrome Test (1 gene)	Primary panel (1 gene) RBM8A
Invitae Townes-Brocks Syndrome Test (1 gene)	Primary panel (1 gene) SALL1
Invitae Treacher-Collins Syndrome Test (1 gene)	Primary panel (1 gene) TCOF1
Invitae Trichorhinophalangeal Syndrome Panel (2 genes)	Primary panel (2 genes) EXT1 TRPS1
Invitae Ulnar-Mammary Syndrome Test (1 gene)	Primary panel (1 gene) TBX3

SKIN DISORDERS

Invitae Cardio-Facio-Cutaneous Syndrome Panel (6 genes)	Primary panel (6 genes) BRAF KRAS MAP2K1 MAP2K2 SHOC2 SOS1
Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel (up to 10 genes)	Primary panel (8 genes) EDA EDAR EDARADD LTBP3 MSX1 NFKBIA PAX9 WNT10A Add-on Clouston syndrome and TP63-related disorder genes (2 genes) GJB6 TP63
Invitae Legius Syndrome Test (up to 2 genes)	Primary panel (1 gene) SPRED1 Add-on neurofibromatosis type 1 gene (1 gene) NF1
Invitae Noonan Syndrome with Multiple Lentigines Panel (3 genes)	Primary panel (3 genes) BRAF PTPN11 RAF1
Invitae PTEN-Related Disorders Test (1 gene)	Primary panel (1 gene) PTEN
Invitae TP63-Related Disorders Test (1 gene)	Primary panel (1 gene) TP63
Invitae van der Woude Syndrome Panel (2 genes)	Primary panel (2 genes) GRHL3 IRF6

CLINICAL AREA: HEREDITARY CANCER

PEDIATRIC ONCOLOGY

Invitae Pediatric Solid Tumors Panel (48 genes)	Primary panel (48 genes) ALK APC AXIN2 BAP1 BLM BMPR1A CDC73 CDKN1C DICER1 DIS3L2 EPCAM FH GPC3 HRAS MAX MEN1 MLH1 MSH2 MSH6 NBN NF1 NF2 PHOX2B PMS2 PRKAR1A PTCH1 PTEN RB1 RECQL4 RET SDHA SDHAF2 SDHB SDHC SDHD SMAD4 SMARCA4 SMARCB1 SMARCE1 STK11 SUFU TMEM127 TP53 TSC1 TSC2 VHL WRN WT1
Invitae Pediatric Hematologic Malignancies Panel (16 genes)	Primary panel (16 genes) ATM BLM CEBPA EPCAM GATA2 HRAS MLH1 MSH2 MSH6 NBN NF1 PMS2 RUNX1 TERC TERT TP53
Invitae Pediatric Nervous System/Brain Tumors Panel (up to 32 genes)	Primary panel (24 genes) ALK APC DICER1 EPCAM HRAS MEN1 MLH1 MSH2 MSH6 NF1 NF2 PHOX2B PMS2 PRKAR1A PTCH1 PTEN RB1 SMARCB1 SMARCE1 SUFU TP53 TSC1 TSC2 VHL Add-on hereditary paraganglioma pheochromocytoma genes (8 genes) MAX RET SDHA SDHAF2 SDHB SDHC SDHD TMEM127

INVITAE

HEADQUARTERS & LABORATORY | 1400 16th Street, San Francisco, CA 94103 | www.invitae.com | **IN THE US** | clientservices@invitae.com | p: 800-436-3037 | f: 415-276-4164
OUTSIDE THE US | globalsupport@invitae.com or visit www.invitae.com/contact | P/N F142-9

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS

A broad menu covering the vast majority of inherited metabolic disorders that are routinely tested as part of state newborn screening programs. Our curated panels are designed by medical genetics experts based on disorders and/or analyte results.

CLINICAL AREA: METABOLIC DISORDERS, NEWBORN SCREENING & IMMUNOLOGY

METABOLIC DISORDERS NEWBORN SCREENING CONFIRMATION

Invitae Metabolic Disorders Newborn Screening Confirmation Panel (up to 229 genes)

Primary panel (90 genes)

ABCD1	ABCD4	ACAD8	ACADM	ACADS	ACADSB	ACADVL	ACAT1	ACSF3	AHCY	ALDH4A1	ARG1
ASL	ASS1	AUH	BCKDHA	BCKDHB	BTD	CBS	CD320	CFTR	CPS1	CPT1A	CPT2
DBT	DNAJC19	ETFA	ETFB	ETFDH	ETHE1	FAH	FTCD	G6PD	GAA	GALE	GALK1
GALT	GCDH	GCH1	GLA	GNMT	GSS	HADH	HADHA	HADHB	HCFC1	HLCS	HMGCL
HPD	HSD17B10	IDUA	IVD	LMBRD1	MAT1A	MCCC1	MCCC2	MCEE	MLYCD	MMAA	MMAB
MMACHC	MMADHC	MTR	MTRR	MUT	NAGS	OAT	OPA3	OTC	PAH	PCBD1	PC
PCCA	PCCB	PPM1K	PRODH	PTS	QDPR	SERAC1	SLC22A5	SLC25A13	SLC25A15	SLC25A20	SMPD1
SPR	SUCLA2	SUCLG1	TAT	TAZ	TMEM70						

Add-on 2,4-dienoyl-CoA reductase deficiency genes (2 genes)

DECR1 NADK2

Add-on cerebral creatine deficiency genes (3 genes)

GAMT GATM SLC6A8

Add-on congenital disorders of glycosylation genes (102 genes)

ALG1	ALG11	ALG12	ALG13	ALG14	ALG2	ALG3	ALG6	ALG8	ALG9	ATP6V0A2	B3GALNT2
B3GALT6	B3GAT3	B3GLCT	B4GALNT1	B4GALT1	B4GALT7	B4GAT1	C1GALT1C1	CHST14	CHST3	CHST6	CHSY1
COG1	COG2	COG4	COG5	COG6	COG7	COG8	DDOST	DHDDS	DOLK	DPAGT1	DPM1
DPM2	DPM3	DSE	EOGT	EXT1	EXT2	FKRP	FKTN	G6PC3	GALNT3	GFPT1	GMPPA
GMPPB	GNE	ISPD	LARGE1	LFNG	MAGT1	MAN1B1	MGAT2	MOGS	MPDU1	MPI	NGLY1
NUS1	PAPSS2	PGM1	PGM3	PIGA	PIGL	PIGM	PIGN	PIGO	PIGQ	PIGT	PIGV
PIGW	PMM2	POFUT1	POGLUT1	POMGNT1	POMGNT2	POMK	POMT1	POMT2	RFT1	RPN2	SEC23A
SEC23B	SLC26A2	SLC35A1	SLC35A2	SLC35A3	SLC35C1	SLC35D1	SRD5A3	SSR4	ST3GAL3	ST3GAL5	STT3A
STT3B	TMEM165	TMEM5	TRIP11	TUSC3	XYLT1						

Add-on generalized leukodystrophies genes (6 genes)

ARSA ASPA GALC GM2A HEXA HEXB

Add-on glucose transporter type 1 deficiency gene (1 gene)

SLC2A1

Add-on glycine encephalopathy genes (6 genes)

AMT GCSH GLDC LIAS NFU1 SLC6A9

Add-on mucopolysaccharidosis type II gene (1 gene)

IDS

Add-on Niemann-Pick type C genes (2 gene)

NPC1 NPC2

Add-on pyridoxal 5'-phosphate-dependent epilepsy gene (1 gene)

PNPO

Add-on pyridoxine-responsive epilepsy gene (1 gene)

ALDH7A1

Add-on Smith-Lemli-Opitz syndrome gene (1 gene)

DHCR7

Add-on cerebrotendinous xanthomatosis gene (1 gene)

CYP27A1

Add-on 3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency gene (1 gene)

HMGCS2

Add-on neuronal ceroid lipofuscinosis genes (10 genes)

ATP13A2 CLN2 (TPP1) CLN3 CLN5 CLN6 CLN8 CTSD KCTD7 MFSD8 PPT1

Add-on succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency gene (1 gene)

OXCT1

Invitae Lysosomal Storage Disorders Newborn Screening Panel (6 genes)

Primary panel (6 genes)

GAA GALC GLA IDS IDUA SMPD1

Invitae X-Linked Adrenoleukodystrophy Newborn Screening Confirmation Test (1 gene)

Primary panel (1 gene)

ABCD1

PANELS BY ANALYTE

Invitae Low C0 Test (1 gene)

Primary panel (1 gene)

SLC22A5

Invitae Elevated C0/(C16+C18) Test (1 gene)

Primary panel (1 gene)

CPT1A

Invitae Elevated C3 Panel (up to 16 genes)

Primary panel (15 genes)

ABCD4	BTD	CD320	HCFC1	HLCS	LMBRD1	MCEE	MMAA	MMAB	MMACHC	MMADHC	MUT
PCCA	PCCB	TCN2									

Add-on ACSF3 gene (1 gene)

ACSF3

INVITAE

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

PANELS BY ANALYTE (continued)

Invitae Elevated C3-DC Test (1 gene)	**Primary panel (1 gene)** MLYCD
Invitae Elevated C4 Panel (3 genes)	**Primary panel (3 genes)** ACAD8 ACADS ETHE1
Invitae Elevated C4-OH Panel (2 genes)	**Primary panel (2 genes)** HADH HIBCH
Invitae Elevated C4 & C5 Panel (7 genes)	**Primary panel (7 genes)** ETFA ETFB ETFDH ETHE1 SLC52A1 SLC52A2 SLC52A3
Invitae Elevated C5 Panel (2 genes)	**Primary panel (2 genes)** ACADSB IVD
Invitae Elevated C5-DC Test (1 gene)	**Primary panel (1 gene)** GCDH
Invitae Elevated C5-OH Panel (13 genes)	**Primary panel (13 genes)** ACAT1 AUH BTD CLPB DNAJC19 HLCS HMGCL HSD17B10 MCCC1 MCCC2 OPA3 SERAC1 TAZ
Invitae Elevated C6, C8 & C10 Test (1 gene)	**Primary panel (1 gene)** ACADM
Invitae Elevated C14 & C14:1 Test (1 gene)	**Primary panel (1 gene)** ACADVL
Invitae Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel (2 genes)	**Primary panel (2 genes)** HADHA HADHB
Invitae Elevated C16, C16:1, C18, & C18:1 Panel (2 genes)	**Primary panel (2 genes)** CPT2 SLC25A20
Invitae Elevated Arginine Test (1 gene)	**Primary panel (1 gene)** ARG1
Invitae Elevated Citrulline Panel (up to 5 genes)	**Primary panel (4 genes)** ASL ASS1 PC SLC25A13 **Add-on dihydrolipoamide dehydrogenase deficiency gene (1 gene)** DLD
Invitae Low Citrulline Panel (3 genes)	**Primary panel (3 genes)** CPS1 NAGS OTC
Invitae Elevated Glycine Panel (up to 62 genes)	**Primary panel (6 genes)** AMT GLDC GCSH LIAS NFU1 SLC6A9 **Add-on organic acidemia genes (56 genes)** ACAD8 ACADSB ACAT1 ACSF3 ASPA AUH BCKDHA BCKDHB BTD D2HGDH DBT DHTKD1 DLD DNAJC19 ETFA ETFB ETFDH ETHE1 FBP1 FH FTCD GCDH GSS HIBCH HLCS HMGCL HSD17B10 IDH2 IVD L2HGDH MCCC1 MCCC2 MCEE MLYCD MMAA MMAB MMACHC MMADHC MUT NFU1 OGDH OPA3 OPLAH OXCT1 PCCA PCCB POLG PPM1K SERAC1 SLC13A5 SLC25A1 SLC25A19 SUCLA2 SUCLG1 TAZ TMEM70
Invitae Elevated Leucine Panel (5 genes)	**Primary panel (5 genes)** BCKDHA BCKDHB DBT DLD PPM1K
Invitae Elevated Methionine Panel (up to 6 genes)	**Primary panel (4 genes)** AHCY CBS GNMT MAT1A **Add-on additional causes of elevated methionine genes (2 genes)** FAH SLC25A13
Invitae Elevated Phenylalanine Panel (6 genes)	**Primary panel (6 genes)** GCH1 PAH PCBD1 PTS QDPR SPR
Invitae Elevated Proline Panel (2 genes)	**Primary panel (2 genes)** ALDH4A1 PRODH
Invitae Elevated Succinylacetone Test (1 gene)	**Primary panel (1 gene)** FAH
Invitae Elevated Tyrosine Panel (3 genes)	**Primary panel (3 genes)** FAH HPD TAT

AMINOACIDOPATHIES

Invitae Alkaptonuria Test (1 gene)	**Primary panel (1 gene)** HGD
Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel (11 genes)	**Primary panel (11 genes)** ABCD4 AMN CD320 CUBN GIF HCFC1 LMBRD1 MMACHC MMADHC TCN1 TCN2
Invitae Cystinuria Panel (3 genes)	**Primary panel (3 genes)** PREPL SLC3A1 SLC7A9
Invitae Disorders of Serine Biosynthesis Panel (3 genes)	**Primary panel (3 genes)** PHGDH PSAT1 PSPH
Invitae Glycine Encephalopathy Panel (6 genes)	**Primary panel (6 genes)** AMT GCSH GLDC LIAS NFU1 SLC6A9

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

AMINOACIDOPATHIES (continued)

Invitae Homocystinuria Panel (up to 19 genes)

Primary panel (4 genes)
CBS MTHFR MTR MTRR

Add-on combined methylmalonic acidemia and homocystinuria genes (11 genes)
ABCD4 AMN CD320 CUBN GIF HCFC1 LMBRD1 MMACHC MMADHC TCN1 TCN2

Add-on elevated methionine genes (4 genes)
AHCY CBS GNMT MAT1A

Invitae Hyperphenylalaninemia Panel (6 genes)

Primary panel (6 genes)
GCH1 PAH PCBD1 PTS QDPR SPR

Invitae Hyperprolinemia Panel (2 genes)

Primary panel (2 genes)
ALDH4A1 PRODH

Invitae Maple Syrup Urine Disease Panel (up to 5 genes)

Primary panel (4 genes)
BCKDHA BCKDHB DBT PPM1K

Add-on DLD gene (1 gene)
DLD

Invitae Tyrosinemia Panel (3 genes)

Primary panel (3 genes)
FAH HPD TAT

CARBOHYDRATE DISORDERS

Invitae Galactosemia Panel (3 genes)

Primary panel (3 genes)
GALE GALK1 GALT

Invitae Glucose-6-Phosphate Dehydrogenase Deficiency Test (1 gene)

Primary panel (1 gene)
G6PD

Invitae Glucose Transporter Type 1 Deficiency Syndrome Test (1 gene)

Primary panel (1 gene)
SLC2A1

Invitae Comprehensive Glycogen Storage Disease Panel (23 genes)

Primary panel (23 genes)
AGL ALDOA ENO3 FBP1 G6PC GAA GBE1 GYG1 GYS1 GYS2 LAMP2 LDHA
PFKM PGAM2 PHKA1 PHKA2 PHKB PHKG2 PYGL PYGM RBCK1 SLC2A2 SLC37A4

Invitae Liver Glycogen Storage Disease Panel (11 genes)

Primary panel (11 genes)
AGL FBP1 G6PC GBE1 GYS2 PHKA2 PHKB PHKG2 PYGL SLC2A2 SLC37A4

Invitae Muscle Glycogen Storage Disease Panel (up to 15 genes)

Primary panel (14 genes)
ALDOA ENO3 GAA GBE1 GYG1 GYS1 LAMP2 LDHA PFKM PGAM2 PHKA1 PHKB
PYGM RBCK1

Add-on PGM1 gene (1 gene)
PGM1

Invitae Hereditary Fructose Intolerance Test (1 gene)

Primary panel (1 gene)
ALDOB

Invitae Rare Carbohydrate Disorders Panel (2 genes)

Primary panel (2 genes)
FBP1 SLC5A1

CEREBROTENDINOUS XANTHOMATOSIS

Invitae Cerebrotendinous Xanthomatosis Test (up to 3 genes)

Primary panel (1 gene)
CYP27A1

Add-on sitosterolemia genes (2 genes)
ABCG5 ABCG8

CONGENITAL DISORDERS OF GLYCOSYLATION

Invitae Congenital Disorders of Glycosylation Panel (up to 103 genes)

Primary panel (50 genes)
ALG1 ALG11 ALG12 ALG13 ALG2 ALG3 ALG6 ALG8 ALG9 ATP6V0A2 B3GLCT CHST14
COG1 COG2 COG4 COG5 COG6 COG7 COG8 DHDDS DOLK DPAGT1 DPM1 DPM2
DPM3 G6PC3 GFPT1 GMPPA GMPPB MAGT1 MAN1B1 MGAT2 MOGS MPDU1 MPI NGLY1
PGM1 PGM3 PMM2 RFT1 SEC23B SLC35A1 SLC35A2 SLC35C1 SRD5A3 SSR4 ST3GAL5 TMEM165
TRIP11 TUSC3

Add-on preliminary evidence genes (11 genes)
ALG14 B4GALT1 DDOST NUS1 PIGM RPN2 SEC23A SLC35A3 ST3GAL3 STT3A STT3B

Add-on disorders of O-mannosylation genes (13 genes)
B3GALNT2 B4GAT1 FKRP FKTN GNE ISPD LARGE1 POMGNT1 POMGNT2 POMK POMT1 POMT2
TMEM5

Add-on glycosylation genes not involved in N-glycosylation genes (29 genes)
B3GALT6 B3GAT3 B4GALNT1 B4GALT7 C1GALT1C1 CHST3 CHST6 CHSY1 DSE EOGT EXT1 EXT2
GALNT3 LFNG PAPSS2 PIGA PIGL PIGM PIGN PIGO PIGQ PIGT PIGV PIGW
POFUT1 POGLUT1 SLC26A2 SLC35D1 XYLT1

CREATINE BIOSYNTHESIS AND TRANSPORT DISORDERS

Invitae Cerebral Creatine Deficiency Panel (3 genes)

Primary panel (3 genes)
GAMT GATM SLC6A8

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

CYSTIC FIBROSIS

Invitae Cystic Fibrosis Newborn Screening Confirmation Test (1 gene)	**Primary panel (1 gene)** CFTR

FATTY ACID OXIDATION DEFECTS

Invitae Fatty Acid Oxidation Defects Panel (up to 22 genes)	**Primary panel (18 genes)** ACADM ACADS ACADSB ACADVL CPT1A CPT2 ETFA ETFB ETFDH HADH HADHA HADHB HMGCL HMGCS2 MLYCD NADK2 SLC22A5 SLC25A20 **Add-on preliminary-evidence gene (1 gene)** DECR1 **Add-on riboflavin transporter deficiency genes (3 genes)** SLC52A1 SLC52A2 SLC52A3
Invitae Ketogenesis Disorders Panel (2 genes)	**Primary panel (2 genes)** HMGCL HMGCS2
Invitae Ketolysis Disorders Panel (2 genes)	**Primary panel (2 genes)** ACAT1 OXCT1
Invitae Medium Chain Acyl-CoA Dehydrogenase Deficiency Test (1 gene)	**Primary panel (1 gene)** ACADM
Invitae Multiple Acyl-CoA Dehydrogenase Deficiency Panel (up to 6 genes)	**Primary panel (3 genes)** ETFA ETFB ETFDH **Add-on riboflavin transporter deficiency genes (3 genes)** SLC52A1 SLC52A2 SLC52A3
Invitae Very Long Chain Acyl-CoA Dehydrogenase Deficiency Test (1 gene)	**Primary panel (1 gene)** ACADVL

LYSOSOMAL STORAGE DISORDERS

Invitae Comprehensive Lysosomal Storage Disorders Panel (up to 53 genes)	**Primary panel (52 genes)** AGA ARSA ARSB ASAH1 ATP13A2 CLN2 (TPP1) CLN3 CLN5 CLN6 CLN8 CTNS CTSA CTSD CTSF CTSK DNAJC5 FUCA1 GAA GALC GALNS GLA GLB1 GM2A GNPTAB GNPTG GNS GRN GUSB HEXA HEXB HGSNAT HYAL1 IDS IDUA KCTD7 LAMP2 LIPA MAN2B1 MANBA MCOLN1 MFSD8 NAGA NAGLU NEU1 NPC1 NPC2 PPT1 PSAP SGSH SLC17A5 SMPD1 SUMF1 **Add-on chitotriosidase deficiency gene (1 gene)** CHIT1
Invitae Cystinosis Test (1 gene)	**Primary panel (1 gene)** CTNS
Invitae Farber Lipogranulomatosis Test (1 gene)	**Primary panel (1 gene)** ASAH1
Invitae Fabry Disease Test (1 gene)	**Primary panel (1 gene)** GLA
Invitae GM2 Gangliosidosis Panel (3 genes)	**Primary panel (3 genes)** GM2A HEXA HEXB
Invitae Krabbe Disease Panel (up to 2 genes)	**Primary panel (1 gene)** GALC **Add-on prosaposin deficiency gene (1 gene)** PSAP
Invitae Lysosomal Acid Lipase Deficiency Test (1 gene)	**Primary panel (1 gene)** LIPA
Invitae Metachromatic Leukodystrophy Panel (up to 7 genes)	**Primary panel (3 genes)** ARSA PSAP SUMF1 **Add-on generalized leukodystrophies genes (4 genes)** ASPA GALC HEXA HEXB
Invitae Mucolipidosis Panel (4 genes)	**Primary panel (4 genes)** GNPTAB GNPTG MCOLN1 NEU1
Invitae Comprehensive Mucopolysaccharidoses (MPS) Panel (up to 23 genes)	**Primary panel (11 genes)** ARSB GALNS GLB1 GNS GUSB HGSNAT HYAL1 IDS IDUA NAGLU SGSH **Add-on mucolipidosis and oligosaccharidoses genes (12 genes)** AGA CTSA CTSK FUCA1 GNPTAB GNPTG MAN2B1 MANBA MCOLN1 NAGA NEU1 SLC17A5
Invitae Mucopolysaccharidosis Type I (MPS I) Test (up to 6 genes)	**Primary panel (1 gene)** IDUA **Add-on clinically overlapping lysosomal storage disorder genes (5 genes)** ARSB GNPTAB GUSB IDS SUMF1
Invitae Mucopolysaccharidosis Type II Test (up to 5 genes)	**Primary panel (1 gene)** IDS **Add-on clinically overlapping genes (4 gene)** GNPTAB GUSB IDUA SUMF1

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

LYSOSOMAL STORAGE DISORDERS (continued)

Invitae Mucopolysaccharidosis Type III (MPS III) Panel (4 genes)	**Primary panel (4 genes)** GNS HGSNAT NAGLU SGSH
Invitae Mucopolysaccharidosis Type IV (MPS IV) Panel (up to 3 genes)	**Primary panel (2 genes)** GALNS GLB1 **Add-on multiple sulfatase deficiency gene (1 gene)** SUMF1
Invitae Multiple Sulfatase Deficiency Test (up to 16 genes)	**Primary panel (1 gene)** SUMF1 **Add-on mucolipidosis and mucopolysaccharidosis genes (15 genes)** ARSB GALNS GLB1 GNPTAB GNPTG GNS GUSB HGSNAT HYAL1 IDS IDUA MCOLN1 NAGLU NEU1 SGSH
Invitae Comprehensive Neuronal Ceroid Lipofuscinoses Panel (up to 13 genes)	**Primary panel (9 genes)** CLN2 (TPP1) CLN3 CLN5 CLN6 CLN8 CTSD KCTD7 MFSD8 PPT1 **Add-on preliminary-evidence gene (1 gene)** ATP13A2 **Add-on adult-onset neuronal ceroid lipofuscinoses genes (3 gene)** CTSF DNAJC5 GRN
Invitae Niemann-Pick Disease Types A and B Test (up to 2 genes)	**Primary panel (1 gene)** SMPD1 **Add-on chitotriosidase deficiency gene (1 gene)** CHIT1
Invitae Niemann-Pick Disease Type C Panel (up to 4 genes)	**Primary panel (2 genes)** NPC1 NPC2 **Add-on lysosomal acid lipase deficiency gene (1 gene)** LIPA **Add-on chitotriosidase deficiency gene (1 gene)** CHIT1
Invitae Oligosaccharidoses Panel (up to 23 genes)	**Primary panel (8 genes)** AGA CTSA CTSK FUCA1 MAN2B1 MANBA NAGA SLC17A5 **Add-on mucolipidosis and mucopolysaccharidosis genes (15 genes)** ARSB GALNS GLB1 GNPTAB GNPTG GNS GUSB HGSNAT HYAL1 IDS IDUA MCOLN1 NAGLU NEU1 SGSH
Invitae Pompe Disease Test (up to 3 genes)	**Primary panel (1 gene)** GAA **Add-on Danon disease gene (1 gene)** LAMP2 **Add-on primary carnitine deficiency gene (1 gene)** SLC22A5
Invitae Prosaposin Deficiency Test (1 gene)	**Primary panel (1 gene)** PSAP
Invitae Sandhoff Disease Test (up to 2 genes)	**Primary panel (1 gene)** HEXB **Add-on Tay-Sachs disease gene (1 gene)** HEXA
Invitae Tay-Sachs Disease Test (up to 2 genes)	**Primary panel (1 gene)** HEXA **Add-on Sandhoff disease gene (1 gene)** HEXB

METAL TRANSPORT DISORDERS

Invitae ATP7A-Related Disorders (1 gene)	**Primary panel (1 gene)** ATP7A
Invitae Copper Metabolism Disorders Panel (5 genes)	**Primary panel (5 genes)** AP1S1 ATP7A ATP7B CP SLC33A1
Invitae Wilson Disease Test (1 gene)	**Primary panel (1 gene)** ATP7B

NEUROTRANSMITTER DISORDERS

Invitae Neurotransmitter Disorders Panel (up to 37 genes)	**Primary panel (27 genes)** ABAT ALDH5A1 ALDH7A1 AMT ARHGEF9 DBH DDC GAD1 GCH1 GCSH GLDC GLRA1 GLRB GPHN MAOA PCBD1 PHGDH PNPO PSAT1 PSPH PTS QDPR SLC25A22 SLC6A3 SLC6A5 SPR TH **Add-on neurodegeneration with brain iron accumulation genes (10 genes)** ATP13A2 C19orf12 COASY CP DCAF17 FA2H FTL PANK2 PLA2G6 WDR45
Invitae Hereditary Hyperekplexia Panel (6 genes)	**Primary panel (6 genes)** ARHGEF9 CLPB GLRA1 CLRB GPHN SLC6A5

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

ORGANIC ACIDEMIAS

Invitae Organic Acidemias Panel (up to 56 genes)

Primary panel (49 genes)
ACAD8 ACADSB ACAT1 ACSF3 ASPA AUH BCKDHA BCKDHB BTD D2HGDH DBT DNAJC19
ETFA ETFB ETFDH ETHE1 FBP1 FTCD GCDH GSS HIBCH HLCS HMGCL HSD17B10
IDH2 IVD L2HGDH MCCC1 MCCC2 MCEE MLYCD MMAA MMAB MMACHC MMADHC MUT
OPA3 OPLAH OXCT1 PCCA PCCB POLG PPM1K SERAC1 SLC25A1 SUCLA2 SUCLG1 TAZ
TMEM70

Add-on Kreb cycle defect genes (7 genes)
DHTKD1 DLD FH NFU1 OGDH SLC13A5 SLC25A19

Invitae 2-Hydroxyglutaric Aciduria Panel (4 genes)

Primary panel (4 genes)
D2HGDH IDH2 L2HGDH SLC25A1

Invitae 3-Methylcrotonyl CoA Carboxylase Panel (2 genes)

Primary panel (2 genes)
MCCC1 MCCC2

Invitae 3-Methylglutaconic Aciduria Panel (7 genes)

Primary panel (7genes)
AUH CLPB DNAJC19 OPA3 SERAC1 TAZ TMEM70

Invitae Barth Syndrome Test (1 gene)

Primary panel (1 gene)
TAZ

Invitae Biotinidase Deficiency Test (1 gene)

Primary panel (1 gene)
BTD

Invitae Canavan Disease Test (1 gene)

Primary panel (1 gene)
ASPA

Invitae Glutaric Acidemia Type I Test (1 gene)

Primary panel (1 gene)
GCDH

Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel (11 genes)

Primary panel (11 genes)
ABCD4 AMN CD320 CUBN GIF HCFC1 LMBRD1 MMADHC MMADHC TCN1 TCN2

Invitae Methylmalonic Acidemia Panel (up to 18 genes)

Primary panel (7 genes)
MMAA MMAB MMADHC MCEE MUT SUCLA2 SUCLG1

Add-on combined malonic and methylmalonic acidemia (1 gene)
ACSF3

Add-on combined methylmalonic acidemia and homocystinuria genes (10 genes)
ABCD4 AMN CD320 CUBN GIF HCFC1 LMBRD1 MMACHC TCN1 TCN2

Invitae Multiple Acyl-CoA Dehydrogenase Deficiency Panel (up to 6 genes)

Primary panel (3 genes)
ETFA ETFB ETFDH

Add-on riboflavin transporter deficiency genes (3 genes)
SLC52A1 SLC52A2 SLC52A3

Invitae Multiple Carboxylase Deficiency Panel (2 genes)

Primary panel (2 genes)
BTD HLCS

Invitae Propionic Acidemia Panel (up to 9 genes)

Primary panel (2 genes)
PCCA PCCB

Add-on methylmalonic acidemia genes (5 genes)
MMAA MMAB MMADHC MMACHC MUT

Add-on multiple carboxylase deficiency genes (2 genes)
BTD HLCS

PEROXISOMAL DISORDERS

Invitae Adult Refsum Disease Panel (2 genes)

Primary panel (2 genes)
PEX7 PHYH

Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Panel (3 genes)

Primary panel (3 genes)
AGPS GNPAT PEX7

Invitae X-linked Adrenoleukodystrophy Test (up to 15 genes)

Primary panel (1 gene)
ABCD1

Add-on peroxisomal acyl-CoA oxidase (ACOX1) deficiency gene (1 gene)
ACOX1

Add-on elevated very long chain fatty acids genes (13 genes)
HSD17B4 PEX1 PEX2 PEX3 PEX5 PEX6 PEX10 PEX12 PEX13 PEX14 PEX16 PEX19
PEX26

Invitae Zellweger Spectrum Disorder Panel (15 genes)

Primary panel (15 genes)
ACOX1 AMACR HSD17B4 PEX1 PEX10 PEX12 PEX13 PEX14 PEX16 PEX19 PEX2 PEX26
PEX3 PEX5 PEX6

PORPHYRIAS

Invitae Acute Hepatic Porphyrias Panel (4 genes)

Primary panel (4 genes)
ALAD CPOX HMBS PPOX

INVITAE METABOLIC DISORDERS AND NEWBORN SCREENING GENE PANEL TESTS (continued)

PURINE METABOLISM DISORDERS

Invitae Purine Metabolism Disorders Panel (up to 10 genes)

Primary panel (9 genes)

ADA	ADSL	AMPD1	HPRT1	GPHN	MOCOS	MOCS1	PNP	XDH

Add-on sulfite oxidase deficiency gene (1 gene)

SUOX

Invitae Lesch-Nyhan Syndrome Test (1 gene)

Primary panel (1 gene)

HPRT1

PYRUVATE METABOLISM AND TRICARBOXYLIC ACID CYCLE DEFECTS

Invitae 2-Ketoglutarate Dehydrogenase Deficiency Panel (up to 4 genes)

Primary panel (3 genes)

DLD	OGDH	SLC25A19

Add-on alpha-ketoadipic acid dehydrogenase deficiency gene (1 gene)

DHTKD1

Invitae Citrate Transporter Deficiency Test (1 gene)

Primary panel (1 gene)

SLC13A5

Invitae Dihydrolipoamide Dehydrogenase Deficiency Test (1 gene)

Primary panel (1 gene)

DLD

Invitae Fumarase Deficiency Test (1 gene)

Primary panel (1 gene)

FH

Invitae Pyruvate Carboxylase Deficiency Test (1 gene)

Primary panel (1 gene)

PC

Invitae Pyruvate Dehydrogenase Deficiency Panel (8 genes)

Primary panel (8 genes)

DLAT	DLD	LIAS	MPC1	PDHA1	PDHB	PDHX	PDP1

TREATABLE DISORDERS

Invitae Treatable Neurometabolic Disorders Panel (up to 133 genes)

Primary panel (92 genes)

ABCD1	ACAT1	AGA	ALDH5A1	ALDH7A1	AMN	AMT	ARG1	ARSA	ASL	ASS1	ATP7A
ATP7B	AUH	BCKDHA	BCKDHB	BTD	CBS	CLN2 (TPP1)	CP	CPS1	CUBN	CYP27A1	DBT
DHCR7	DLAT	DLD	ETFA	ETFB	ETFDH	ETHE1	GAMT	GATM	GCDH	GCH1	GCSH
GIF	GLA	GLDC	GLUD1	GUSB	HLCS	HMGCL	HMGCS2	HSD17B10	IDS	IDUA	IVD
LIPA	LMBRD1	MAN2B1	MCCC1	MCCC2	MMAA	MMAB	MMACHC	MMADHC	MOCS1	MTHFR	MTR
MTRR	MUT	NAGS	NPC1	NPC2	OTC	OXCT1	PAH	PANK2	PCBD1	PCCA	PCCB
PDHA1	PDHB	PDHX	PDP1	PHGDH	PNPO	PPM1K	PSAT1	PSPH	PTS	QDPR	SGSH
SLC19A3	SLC25A13	SLC25A15	SLC2A1	SLC6A8	SPR	TAT	TH				

Add-on neurometabolic conditions genes (41 genes)

ABAT	ADSL	AP1S1	ATP13A2	BCKDK	C19orf12	CLN3	CLN5	CLN6	CLN8	COASY	CTSD
D2HGDH	DBH	DCAF17	DDC	FA2H	FTL	GAD1	GNS	GPHN	HEXA	HEXB	HGSNAT
HPRT1	IDH2	KCTD7	L2HGDH	MAOA	MFSD8	MOCOS	NAGLU	PLA2G6	POLG	PPT1	SLC13A5
SLC33A1	SLC6A3	SUOX	WDR45	XDH							

Invitae Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD) Test (1 gene)

Primary panel (1 gene)

SLC19A3

UREA CYCLE DISORDERS

Invitae Urea Cycle Disorders Panel (up to 15 genes)

Primary panel (10 genes)

ALDH18A1	ARG1	ASL	ASS1	CPS1	NAGS	OAT	OTC	SLC25A13	SLC25A15

Add-on hyperammonemia genes (4 genes)

CA5A	GLUD1	GLUL	SLC7A7

Add-on hereditary orotic aciduria gene (1 gene)

UMPS

Invitae Arginase Deficiency Test (1 gene)

Primary panel (1 gene)

ARG1

Invitae Ornithine Transcarbamylase (OTC) Deficiency Test (up to 4 genes)

Primary panel (1 gene)

OTC

Add-on hereditary orotic aciduria gene (1 gene)

UMPS

Add-on low citrulline genes (2 genes)

CPS1	NAGS

INVITAE

HEADQUARTERS & LABORATORY | 1400 16th Street, San Francisco, CA 94103 | www.invitae.com | **IN THE US** | clientservices@invitae.com | p: 800-436-3037 | f: 415-276-4164
OUTSIDE THE US | globalsupport@invitae.com or visit www.invitae.com/contact | P/N F152-7

INVITAE CARDIOLOGY GENETIC TESTS

A comprehensive menu for heritable heart and vascular conditions curated by medical genetics experts for specific indications and clinical scenarios.

CLINICAL AREA: CARDIOLOGY

ARRHYTHMIA AND CARDIOMYOPATHY

Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel (up to 149 genes)

Primary panel (67 genes)

ABCC9	ACTC1	ACTN2	AGL	ANK2	BAG3	CACNA1C	CACNB2	CALM1	CALM2	CALM3	CASQ2
CAV3	CRYAB	CSRP3	DES	DMD	DOLK	DSC2	DSG2	DSP	EMD	EYA4	FHL1
FKRP	FKTN	FLNC	GAA	GLA	GPD1L	HCN4	JUP	KCNA5	KCNE1	KCNE2	KCNH2
KCNJ2	KCNQ1	LAMP2	LMNA	MYBPC3	MYH7	MYL2	MYL3	MYL4	NKX2-5	PKP2	PLN
PRKAG2	RAF1	RBM20	RYR2	SCN5A	SGCD	SLC22A5	TAZ	TCAP	TGFB3	TMEM43	TNNC1
TNNI3	TNNT2	TPM1	TRDN	TTN	TTR	VCL					

Add-on preliminary-evidence genes (47 genes)

AKAP9	ANKRD1	CACNA2D1	CALR3	CHRM2	CTF1	CTNNA3	DTNA	FHL2	GATA4	GATA6	GATAD1
GJA5	ILK	JPH2	KCND3	KCNE3	KCNE5	KCNJ5	KCNJ8	KCNK3	LAMA4	LDB3	LRRC10
MED12	MYH6	MYLK2	MYOM1	MYOZ2	MYPN	NEBL	NEXN	NPPA	PDLIM3	PLEKHM2	PRDM16
RANGRF	SCN10A	SCN1B	SCN2B	SCN3B	SCN4B	SLMAP	SNTA1	TMPO	TRPM4	TXNRD2	

Add-on RASopathy genes not included in panel (17 genes)

A2ML1	BRAF	CBL	HRAS	KRAS	MAP2K1	MAP2K2	NF1	NRAS	PTPN11	RASA1	RIT1
RRAS	SHOC2	SOS1	SOS2	SPRED1							

Add-on autosomal recessive syndromic pediatric cardiomyopathy genes (8 genes)

ACADVL	ALMS1	CPT2	DNAJC19	ELAC2	MTO1	SDHA	TMEM70

Add-on sudden unexpected death in epilepsy (SUDEP) genes for arrhythmia and cardiomyopathy (10 genes)

DEPDC5	KCNQ2	KCNQ3	KCNT1	PCDH19	PRRT2	SCN1A	SCN8A	SCN9A	SLC2A1

ARRHYTHMIA

Invitae Arrhythmia Comprehensive Panel (up to 73 genes)

Primary panel (39 genes)

ABCC9	ACTN2	ANK2	CACNA1C	CACNB2	CALM1	CALM2	CALM3	CASQ2	CAV3	DES	DSC2
DSG2	DSP	EMD	FLNC	GPD1L	HCN4	JUP	KCNA5	KCNE1	KCNE2	KCNH2	KCNJ2
KCNQ1	LMNA	MYL4	NKX2-5	PKP2	PLN	PRKAG2	RBM20	RYR2	SCN5A	TMEM43	TNNI3
TNNT2	TRDN	TTN									

Add-on preliminary-evidence genes (24 genes)

AKAP9	ANKRD1	CACNA2D1	CTNNA3	GJA5	KCND3	KCNE3	KCNE5	KCNJ5	KCNJ8	KCNK3	LDB3
NPPA	PDLIM3	RANGRF	SCN10A	SCN1B	SCN2B	SCN3B	SCN4B	SLMAP	SNTA1	TGFB3	TRPM4

Add-on sudden unexpected death in epilepsy (SUDEP) genes for arrhythmia (10 genes)

DEPDC5	KCNQ2	KCNQ3	KCNT1	PCDH19	PRRT2	SCN1A	SCN8A	SCN9A	SLC2A1

Invitae Arrhythmogenic Cardiomyopathy Panel (up to 24 genes)

Primary panel (19 genes)

ACTN2	DES	DSC2	DSG2	DSP	EMD	FLNC	JUP	LMNA	PKP2	PLN	PRKAG2
RBM20	RYR2	SCN5A	TMEM43	TNNI3	TNNT2	TTN					

Add-on preliminary-evidence genes (5 genes)

ANKRD1	CTNNA3	LDB3	PDLIM3	TGFB3

Invitae Brugada Syndrome Panel (up to 20 genes)

Primary panel (8 genes)

ABCC9	CACNA1C	CACNB2	GPD1L	HCN4	KCNH2	PKP2	SCN5A

Add-on preliminary-evidence genes (12 genes)

CACNA2D1	KCND3	KCNE3	KCNE5	KCNJ8	RANGRF	SCN10A	SCN1B	SCN2B	SCN3B	SLMAP	TRPM4

Invitae Catecholaminergic Polymorphic Ventricular Tachycardia Panel (8 genes)

Primary panel (8 genes)

ANK2	CALM1	CALM2	CALM3	CASQ2	KCNJ2	RYR2	TRDN

Invitae Long QT Syndrome Panel (up to 17 genes)

Primary panel (13 genes)

ANK2	CACNA1C	CALM1	CALM2	CALM3	CAV3	KCNE1	KCNE2	KCNH2	KCNJ2	KCNQ1	SCN5A
TRDN											

Add-on preliminary-evidence genes (4 genes)

AKAP9	KCNJ5	SCN4B	SNTA1

Invitae Short QT Syndrome Panel (up to 6 genes)

Primary panel (5 genes)

CACNA1C	CACNB2	KCNH2	KCNJ2	KCNQ1

Add-on preliminary-evidence gene (1 gene)

CACNA2D1

INVITAE CARDIOLOGY GENETIC TESTS (continued)

CARDIOMYOPATHY

Invitae Cardiomyopathy Comprehensive Panel (up to 106 genes)

Primary panel (50 genes)

ABCC9 ACTC1 ACTN2 AGL BAG3 CACNA1C CAV3 CRYAB CSRP3 DES DMD DOLK
DSC2 DSG2 DSP EMD EYA4 FHL1 FKRP FKTN FLNC GAA GLA HCN4
JUP LAMP2 LMNA MYBPC3 MYH7 MYL2 MYL3 PKP2 PLN PRKAG2 RAF1 RBM20
RYR2 SCN5A SGCD SLC22A5 TAZ TCAP TMEM43 TNNC1 TNNI3 TNNT2 TPM1 TTN
TTR VCL

Add-on preliminary-evidence genes (31 genes)

ANKRD1 CALR3 CHRM2 CTF1 CTNNA3 DTNA FHL2 GATA4 GATA6 GATAD1 ILK JPH2
LAMA4 LDB3 LRRC10 MED12 MYH6 MYLK2 MYOM1 MYOZ2 MYPN NEBL NEXN NKX2-5
NPPA PDLIM3 PLEKHM2 PRDM16 TGFB3 TMPO TXNRD2

Add-on RASopathy genes not included in panel (17 genes)

A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RASA1 RIT1
RRAS SHOC2 SOS1 SOS2 SPRED1

Add-on genes associated with autosomal recessive syndromic pediatric cardiomyopathy (8 genes)

ACADVL ALMS1 CPT2 DNAJC19 ELAC2 MTO1 SDHA TMEM70

Invitae Arrhythmogenic Cardiomyopathy Panel (up to 23 genes)

Primary panel (19 genes)

ACTN2 DES DSC2 DSG2 DSP EMD JUP LMNA PKP2 PLN PRKAG2 RBM20
RYR2 SCN5A TGFB3 TMEM43 TNNI3 TNNT2 TTN

Add-on preliminary-evidence genes (4 genes)

ANKRD1 CTNNA3 LDB3 PDLIM3

Invitae Dilated Cardiomyopathy Panel (up to 70 genes)

Primary panel (41 genes)

ABCC9 ACTC1 ACTN2 BAG3 CAV3 CRYAB CSRP3 DES DMD DOLK DSC2 DSG2
DSP EMD EYA4 FKRP FKTN FLNC JUP LAMP2 LMNA MYBPC3 MYH7 PKP2
PLN RAF1 RBM20 RYR2 SCN5A SGCD SLC22A5 TAZ TCAP TMEM43 TNNC1 TNNI3
TNNT2 TPM1 TTN TTR VCL

Add-on preliminary-evidence genes (23 genes)

ANKRD1 CHRM2 CTF1 FHL2 GATA4 GATA6 GATAD1 ILK LAMA4 LDB3 LRRC10 MED12
MYH6 MYPN NEBL NEXN NKX2-5 NPPA PDLIM3 PLEKHM2 PRDM16 TMPO TXNRD2

Add-on genes associated with autosomal recessive syndromic pediatric cardiomyopathy (6 genes)

ACADVL ALMS1 CPT2 DNAJC19 SDHA TMEM70

Invitae Hypertrophic Cardiomyopathy Panel (up to 58 genes)

Primary panel (26 genes)

ACTC1 ACTN2 AGL BAG3 CACNA1C CAV3 CSRP3 DES FHL1 FLNC GAA GLA
LAMP2 MYBPC3 MYH7 MYL2 MYL3 PLN PRKAG2 TCAP TNNC1 TNNI3 TNNT2 TPM1
TTR VCL

Add-on preliminary-evidence genes (12 genes)

ANKRD1 CALR3 GATA4 JPH2 LDB3 MYH6 MYLK2 MYOM1 MYOZ2 MYPN NEXN PDLIM3

Add-on RASopathy genes (18 genes)

A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RAF1 RASA1
RIT1 RRAS SHOC2 SOS1 SOS2 SPRED1

Add-on genes associated with autosomal recessive syndromic pediatric cardiomyopathy (4 genes)

ACADVL CPT2 ELAC2 MTO1

Invitae Left Ventricular Noncompaction Panel (up to 19 genes)

Primary panel (15 genes)

ACTC1 DSP HCN4 LAMP2 LMNA MYBPC3 MYH7 PLN RYR2 SCN5A TAZ TNNI3
TNNT2 TPM1 VCL

Add-on preliminary-evidence genes (4 genes)

DTNA LDB3 PLEKHM2 PRDM16

Invitae Transthyretin Amyloidosis Test (1 gene)

Primary panel (1 gene)

TTR

Invitae Hereditary Hemochromatosis Panel (5 genes)

Primary panel (5 genes)

HAMP HFE HFE2 SLC40A1 TFR2

Invitae RASopathies Comprehensive Panel (18 genes)

Primary panel (18 genes)

A2ML1 BRAF CBL HRAS KRAS MAP2K1 MAP2K2 NF1 NRAS PTPN11 RAF1 RASA1
RIT1 RRAS SHOC2 SOS1 SOS2 SPRED1

INVITAE CARDIOLOGY GENETIC TESTS (continued)

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	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	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

AORTOPATHY AND CONNECTIVE TISSUE DISORDERS

Invitae Aortopathy Comprehensive Panel (up to 25 genes)

Primary panel (23 genes)

ACTA2	CBS	COL3A1	COL5A1	COL5A2	EFEMP2	FBN1	FBN2	FLNA	MED12	MYH11	MYLK
NOTCH1	PLOD1	PRKG1	SKI	SLC2A10	SMAD3	SMAD4	TGFB2	TGFB3	TGFBR1	TGFBR2	

Add-on preliminary-evidence genes (2 genes)

MAT2A	SMAD6

Invitae Ehlers-Danlos Syndrome Panel (15 genes)

Primary panel (15 genes)

ADAMTS2	ATP7A	CHST14	COL1A1	COL12A1	COL1A2	COL3A1	COL5A1	COL5A2	CRTAP	FLNA	FKBP14
P3H1	PLOD1	SLC39A13									

Invitae Loeys-Dietz Syndrome Panel (up to 6 genes)

Primary panel (4 genes)

SMAD3	TGFB2	TGFBR1	TGFBR2

Add-on clinically overlapping genes (2 genes)

FBN1	TGFB3

Invitae Marfan Syndrome Test (1 gene)

Primary panel (1 gene)

FBN1

FAMILIAL HYPERCHOLESTEROLEMIA

Invitae Familial Hypercholesterolemia Panel (4 genes)

Primary panel (4 genes)

APOB	LDLR	LDLRAP1	PCSK9

PULMONARY HYPERTENSION

Invitae Pulmonary Arterial Hypertension Panel (up to 9 genes)

Primary panel (4 genes)

ACVRL1	BMPR2	CAV1	ENG

Add-on preliminary-evidence genes (5 genes)

BMPR1B	GDF2	KCNA5	KCNK3	SMAD9

Invitae Hereditary Hemorrhagic Telangiectasia Panel (5 genes)

Primary panel (5 genes)

ACVRL1	ENG	GDF2	RASA1	SMAD4

Invitae Capillary Malformation-Arteriovenous Malformation Syndrome Test (up to 4 genes)

Primary panel (1 gene)

RASA1

Add-on hereditary hemorrhagic telangiectasia genes (3 genes)

ACVRL1	ENG	SMAD4

CONGENITAL HEART DISEASE

Invitae Congenital Heart Disease Panel (42 genes)

Primary panel (42 genes)

ACTC1	ACVR2B	ALMS1	BCOR	BRAF	CBL	CHD7	CRELD1	ELN	FOXH1	GATA4	GATA6
GDF1	GJA1	GPC3	HAND1	HRAS	JAG1	KRAS	LEFTY2	MAP2K1	MAP2K2	MED13L	MEIS2
MYH6	NKX2-5	NKX2-6	NODAL	NOTCH1	NR2F2	NRAS	NSD1	PTPN11	RAF1	RIT1	SHOC2
SMAD6	SOS1	TBX1	TBX5	ZFPM2	ZIC3						

Invitae CHARGE Syndrome Test (1 gene)

Primary panel (1 gene)

CHD7

Invitae Holt-Oram Syndrome Test (1 gene)

Primary panel (1 gene)

TBX5

Invitae RASopathies Comprehensive Panel (18 genes)

Primary panel (18 genes)

A2ML1	BRAF	CBL	HRAS	KRAS	MAP2K1	MAP2K2	NF1	NRAS	PTPN11	RAF1	RASA1
RIT1	RRAS	SHOC2	SOS1	SOS2	SPRED1						

Invitae Sotos Syndrome Test (1 gene)

Primary panel (1 gene)

NSD1

ABOUT INVITAE

Invitae's mission is to bring comprehensive genetic information into mainstream medical practice to improve the quality of healthcare for everyone. Our goal is to aggregate most of the world's genetic tests into a single service with higher quality, faster turnaround time, and lower price than many single-gene tests today.

For more information about Invitae's genetic tests, please visit www.invitae.com.

INVITAE

HEADQUARTERS & LABORATORY | 1400 16th Street, San Francisco, CA 94103 | www.invitae.com | **IN THE US** | clientservices@invitae.com | p: 800-436-3037 | f: 415-276-4164
OUTSIDE THE US | globalsupport@invitae.com or visit www.invitae.com/contact | Updated January 5, 2018