INVITAE

ORDER ONLINE at www.invitae.com/signin.

ORDER ID
For Invitae internal use only

Requisition Form
NEUROLOGY TRF905-11

PATIENT INFORMATION

First name | MI | Last name

Date of birth (MM/DD/YYYY) | Sex ◯ M ◯ F | MRN (medical record number)

Ancestry ◯ Asian ◯ Black/African American ◯ White/Caucasian ◯ Ashkenazi Jewish ◯ Hispanic ◯ Native American ◯ Pacific Islander ◯ Other:

Email address (for report access after release by medical professional)

Phone | Is this patient deceased? ◯ Yes ◯ No Deceased date:

Address | City

State | ZIP code | Country

SPECIMEN INFORMATION

Label each tube with the patient's full name, date of birth, and specimen collection date.
A requisition form MUST accompany each specimen. www.invitae.com/specimen-requirements

Specimen type : ◯ Blood ◯ Saliva ◯ Assisted saliva ◯ DNA - source:

DNA must be extracted in a CLIA or other suitably certified laboratory

We are unable to accept blood/saliva from patients with:

- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Collection date (MM/DD/YYYY)

Special cases
◯ History of/current hematologic malignancy
◯ Resubmission

REASON FOR TESTING

Primary indication:

ICD-10 codes

Previous results

Testing for a personal history of disease? ◯ **Yes** ◯ **No** If yes, describe below.
Age at diagnosis: _____

ORGANIZATION INFORMATION

Organization name and address

Organization name

Phone | Fax

Address | City

State | ZIP code | Country

Primary clinical contact

Name | Role/title

Phone | NPI

Email address (for report access)

Ordering physician

◯ **Same as primary clinical contact**

Name | NPI

Email address (for report access)

Additional clinical or laboratory contact (optional)

Name | Email address (for report access)

Letter of Medical Necessity (LMN)

◯ I have attached an LMN and/or other documentation for insurance billing purposes.

◯ I agree to allow Invitae to transfer the information from this requisition to an LMN and/or other documentation using the ordering physician's name as the signature for insurance billing.

Family history? ◯ **Yes** ◯ **No** If yes, describe in detail below or attach pedigree. If there is a known familial variant, indicate here.

◯ INSURANCE BILLING (U.S. ONLY)

◯ **I have attached a copy of the patient's card**

Insurance company name | Member ID#

Patient relation to policy holder: ◯ Self ◯ Child ◯ Spouse ◯ Other

Policy holder name | Prior-authorization #

◯ PATIENT PAY BILLING

Invitae will send an electronic invoice to the patient email listed above

◯ INSTITUTIONAL BILLING

◯ **Send invoice to organization address above**

Billing contact name | Phone | Fax

Billing email address

Billing address | City

State | ZIP code | Country

◯ OTHER BILLING

Invitae partner code:

By signing this form, the medical professional acknowledges that the individual/family member authorized to make decisions for the individual (collectively, the "Patient") has been supplied information regarding and consented to undergo genetic testing, substantially as set forth in Invitae's Informed Consent for Genetic Testing (www.invitae.com/patient-consent), and has been informed that Invitae may notify them of clinical updates related to genetic test results (in consultation with the ordering medical professional as indicated). The Patient has further been informed and hereby authorizes Invitae Corporation ("Invitae") and its designees to release information concerning testing to their insurer in order to process and/or appeal claims on behalf of the Patient. For amounts received directly, the Patient agrees to remit payment to Invitae for testing services rendered. I acknowledge that I offered pre-test Genetic Counseling to the Patient, if required by their insurer. In addition to the above, I attest that I am the ordering physician, or I am authorized by the ordering physician to order this test, or I am authorized under applicable state law to order this test.

Medical professional signature | **Date**

ORDER INSTRUCTIONS

Select a pre-curated test, combine multiple tests, or customize your own test for each patient. Invitae's pricing is per clinical area for initial order and re-requisition. All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send in two sample tubes and your order will represent two billable events. Your test results will be delivered as two reports. Please contact Client Services with any questions. For Invitae's full test menu, please visit www.invitae.com.

RE-REQUISITION

Invitae offers one re-requisition at no additional charge within 90 days for genes within the original clinical area. For more information and to request online, please visit www.invitae.com/re-requisition.

FAMILY VARIANT TESTING

Invitae offers Family Variant Testing at no additional charge within 90 days for the genes in which the original family member's variant was identified. In such cases, please use the Family Variant Testing/VUS Resolution requisition form (TRF920), available at www.invitae.com/forms.

To request a complimentary specimen collection kit visit www.invitae.com/request-a-kit

SHIPPING INSTRUCTIONS

Please ship specimen overnight in insulated containers:

Attn: Invitae Client Services
1400 16th Street
San Francisco, CA 94103
USA

PRELIMINARY-EVIDENCE GENES

Invitae's primary panels contain genes for which there is definitive evidence that variants in these genes cause specific diseases. Preliminary-evidence genes are genes for which there is only early evidence of a relationship between variants in these genes and specific diseases. All preliminary-evidence genes are indicated as such on the requisition form below.

ASSAY

Invitae is a CAP-accredited and CLIA-certified clinical diagnostic laboratory performing full-gene sequencing and deletion/duplication analysis using next-generation sequencing technology (NGS). Search for details on the analysis of any gene in our test catalog at www.invitae.com/physician/search.

Invitae continually updates its panels based on the most recent evidence. Please note that if an order is placed using an older version of this form, Invitae reserves the right to upgrade any ordered panel(s) to the current version(s). To avoid confusion, please consider placing your order using our online test catalog.

CLINICAL AREA: NEUROLOGY

Test code	Test name	# gene(s)	Gene list
Movement Disorders			
○ 03351	Invitae Dystonia Panel	16	ANO3, ATP1A3, GCH1, GNAL, PARK2, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SLC6A3, SPR, TH, THAP1, TOR1A, TUBB4A
	○ 03351.1 Add-on preliminary-evidence genes	5	CIZ1, DRD2, HPCA, KCTD17, TOR1AIP1
○ 03352	Invitae Hereditary Parkinson's Disease & Parkinsonism Panel	15	ATP13A2, DCTN1, DNAJC6, FBXO7, GCH1, LRRK2, PARK2, PARK7, PINK1, PRKRA, SLC6A3, SNCA, SPR, TH, VPS35
	○ 03352.1 Add-on preliminary-evidence genes	2	CHCHD2, MAPT
Neurodegenerative Disorders			
○ 03502	Invitae Combined Hereditary Dementia and Amyotrophic Lateral Sclerosis Panel	22	ALS2, APP, CHCHD10, DCTN1, FUS, GRN, MAPT, OPTN, PFN1, PRNP, PSEN1, PSEN2, SETX, SNCA, SOD1, SPG11, TARDBP, TBK1, TFG, UBQLN2, VAPB, VCP
	○ 03502.1 Add-on preliminary-evidence genes	5	CHMP2B, HNRNPA2B1, MATR3, SIGMAR1, SQSTM1
○ 03503	Invitae Amyotrophic Lateral Sclerosis Panel	15	ALS2, CHCHD10, DCTN1, FUS, OPTN, PFN1, SETX, SOD1, SPG11, TARDBP, TBK1, TFG, UBQLN2, VAPB, VCP
	○ 03503.1 Add-on preliminary-evidence genes	4	CHMP2B, MATR3, SIGMAR1, SQSTM1
○ 03505	Invitae Frontotemporal Dementia Panel	9	CHCHD10, DCTN1, FUS, GRN, MAPT, TARDBP, TBK1, UBQLN2, VCP
	○ 03505.1 Add-on preliminary-evidence genes	4	CHMP2B, HNRNPA2B1, PSEN1, SQSTM1
○ 03504	Invitae Hereditary Alzheimer's Disease Panel	3	APP, PSEN1, PSEN2
○ 03352	Invitae Hereditary Parkinson's Disease & Parkinsonism Panel	15	ATP13A2, DCTN1, DNAJC6, FBXO7, GCH1, LRRK2, PARK2, PARK7, PINK1, PRKRA, SLC6A3, SNCA, SPR, TH, VPS35
	○ 03352.1 Add-on preliminary-evidence genes	2	CHCHD2, MAPT
○ 03506	Invitae Hereditary Prion Disease Test	1	PRNP

CLINICAL AREA: NEUROLOGY (continued)

Test code	Test name		# gene(s)	Gene list
Neuromuscular Disorders				
○ 03280	Invitae Comprehensive Neuromuscular Disorders Panel		107	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
	○ 03280.1	Add-on preliminary-evidence genes	14	ALG14, HNRNPA2B1, HNRNPDL, LAMB2, LIMS2, LRP4, MYF6, PREPL, SNAP25, SUN1, SUN2, SYNE1, SYNE2, TMEM43
	○ 03280.2	Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene	1	SMCHD1
○ 03281	Invitae Congenital Myasthenic Syndrome Panel		14	AGRN, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, MUSK, RAPSN, SLC5A7
	○ 03281.1	Add-on preliminary-evidence genes	8	ALG14, GMPPB, LAMB2, LRP4, PLEC, PREPL, SCN4A, SNAP25
○ 03285	Invitae Malignant Hyperthermia Susceptibility Panel		2	CACNA1S, RYR1
○ 03291	Invitae Comprehensive Muscular Dystrophy Panel		48	ANO5, B3GALNT2, B4GAT1, CAPN3, CAV3, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, ISPD, ITGA7, LAMA2, LARGE1, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TMEM5, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
	○ 03291.1	Add-on preliminary-evidence genes	7	HNRNPDL, LIMS2, SUN1, SUN2, SYNE1, SYNE2, TMEM43
	○ 03291.2	Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene	1	SMCHD1
○ 03292	Invitae Congenital Muscular Dystrophy Panel		27	B3GALNT2, B4GAT1, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DMD, DPM1, DPM2, DPM3, FKRP, FKTN, GMPPB, ISPD, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, TCAP, TMEM5
○ 03302	Invitae Dystroglycanopathy Panel		17	B3GALNT2, B4GAT1, DAG1, DPM1, DPM2, DPM3, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, TMEM5
○ 03301	Invitae Dystrophinopathies Test		1	DMD
○ 03303	Invitae Emery-Dreifuss Muscular Dystrophy Panel		3	EMD, FHL1, LMNA
	○ 03303.1	Add-on preliminary-evidence genes	5	SUN1, SUN2, SYNE1, SYNE2, TMEM43
○ 03304	Invitae Limb-Girdle Muscular Dystrophy Panel		31	ANO5, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DYSF, FKRP, FKTN, GAA, GMPPB, ISPD, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
	○ 03304.1	Add-on preliminary-evidence genes	2	HNRNPDL, LIMS2
	○ 03304.2	Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene	1	SMCHD1
○ 03361	Invitae Comprehensive Myopathy Panel		51	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
	○ 03361.1	Add-on preliminary-evidence gene	1	MYF6

CLINICAL AREA: NEUROLOGY (continued)

Test code	Test name	# gene(s)	Gene list
Neuromuscular Disorders (continued)			
○ 03362	Invitae Congenital Myopathy Panel	27	ACTA1, BIN1, CCDC78, CFL2, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, DNM2, FKBP14, KBTBD13, KLHL40, KLHL41, LMOD3, MEGF10, MTM1, MYH7, MYPN, NEB, RYR1, SELENON, STAC3, TNNT1, TPM2, TPM3, TTN
	○ 03362.1 Add-on preliminary-evidence gene	1	MYF6
○ 03371	Invitae Autophagic Vacuolar Myopathy Panel	3	DES, LAMP2, VMA21
○ 03363	Invitae Central Core Disease Test	1	RYR1
○ 03364	Invitae Centronuclear Myopathy Panel	6	BIN1, CCDC78, DNM2, MTM1, RYR1, TTN
	○ 03364.1 Add-on preliminary-evidence gene	1	MYF6
○ 03365	Invitae Congenital Fiber-Type Disproportion Panel	7	ACTA1, LMNA, MYH7, RYR1, SELENON, TPM2, TPM3
○ 03366	Invitae Distal Myopathy Panel	18	ANO5, BAG3, CAV3, CRYAB, DES, DNAJB6, DYSF, FHL1, FLNC, GNE, LDB3, MATR3, MYH7, MYOT, SQSTM1, TIA1, TTN, VCP
○ 03373	Invitae Periodic Paralysis Panel	3	CACNA1S, KCNJ2, SCN4A
○ 03374	Invitae Inclusion Body Myopathy Panel	4	GNE, MYH2, TTN, VCP
	○ 03374.1 Add-on preliminary-evidence gene	1	HNRNPA2B1
○ 03367	Invitae Multiminicore Disease Panel	2	RYR1, SELENON
○ 03368	Invitae Myofibrillar Myopathy Panel	8	BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, LDB3, MYOT
○ 03369	Invitae Nemaline Myopathy Panel	11	ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3
○ 03370	Invitae Type VI Collagenopathy Panel	3	COL6A1, COL6A2, COL6A3
	○ 03370.1 Add-on preliminary-evidence gene	1	COL12A1
○ 03375	Invitae Myotonia and Paramyotonia Congenita Panel	2	CLCN1, SCN4A
○ 03245	Invitae Spinal Muscular Atrophy Panel	2	SMN1, SMN2
Neuropathies and Related Disorders			
○ 03200	Invitae Comprehensive Neuropathies Panel	72	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
	○ 03200.1 Add-on preliminary-evidence genes	9	CCT5, FLRT1, HSPB3, LAS1L, MARS, PRDM12, SCN10A, SETX, SURF1
	○ 03200.2 Add-on spinal muscular atrophy genes	2	SMN1, SMN2
○ 03201	Invitae Charcot-Marie-Tooth Disease Comprehensive Panel	43	AARS, AIFM1, BSCL2, DNAJB2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, LITAF, LMNA, LRSAM1, MED25, MFN2, MORC2, MPZ, MTMR2, NDRG1, NEFL, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, SBF2, SH3TC2, SLC25A46, SPG11, TFG, TRIM2, TRPV4, YARS
	○ 03201.1 Add-on preliminary-evidence genes	2	MARS, SURF1
○ 03211	Invitae Charcot-Marie-Tooth Disease Autosomal Dominant Panel	24	AARS, BSCL2, DNM2, DYNC1H1, EGR2, GARS, GDAP1, GNB4, HARS, HSPB1, HSPB8, INF2, LITAF, LMNA, LRSAM1, MFN2, MORC2, MPZ, NEFL, PMP22, RAB7A, TFG, TRPV4, YARS
	○ 03211.1 Add-on preliminary-evidence gene	1	MARS

CLINICAL AREA: NEUROLOGY (continued)

Test code	Test name	# gene(s)	Gene list
Neuropathies and Related Disorders (continued)			
○ 03212	Invitae Charcot-Marie-Tooth Disease Autosomal Recessive Panel	21	DNAJB2, EGR2, FGD4, FIG4, GDAP1, HINT1, IGHMBP2, LMNA, LRSAM1, MED25, MFN2, MTMR2, NDRG1, NEFL, PLEKHG5, PRX, SBF2, SH3TC2, SLC25A46, SPG11, TRIM2
	○ 03212.1 Add-on preliminary-evidence gene	1	SURF1
○ 03213	Invitae Charcot-Marie-Tooth Disease X-linked Panel	4	AIFM1, GJB1, PDK3, PRPS1
○ 03230	Invitae Hereditary Sensory and Autonomic Neuropathy Panel	15	ATL1, ATL3, DNMT1, DST, FAM134B, IKBKAP, KIF1A, NGF, NTRK1, RAB7A, SCN11A, SCN9A, SPTLC1, SPTLC2, WNK1
	○ 03230.1 Add-on preliminary-evidence genes	2	CCT5, PRDM12
○ 03461	Invitae Familial Dysautonomia Test	1	IKBKAP
○ 03240	Invitae Hereditary Motor Neuropathies Panel	23	ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, FBXO38, GARS, HINT1, HSPB1, HSPB8, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SLC5A7, SMN1, SMN2, TRPV4, UBA1, VAPB, VRK1
	○ 03240.1 Add-on preliminary-evidence gene	1	HSPB3
○ 03245	Invitae Spinal Muscular Atrophy Panel	2	SMN1, SMN2
○ 03220	Invitae Small Fiber Neuropathy Test	1	SCN9A
	○ 03220.1 Add-on preliminary-evidence gene	1	SCN10A
○ 03270	Invitae Riboflavin Transporter Deficiency Neuronopathy Panel	2	SLC52A2, SLC52A3
○ 03251	Invitae Hereditary Spastic Paraplegia Comprehensive Panel	43	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
	○ 03251.1 Add-on preliminary-evidence genes	21	AMPD2, ARL6IP1, ARSI, ATP13A2, C19orf12, CCT5, CPT1C, ENTPD1, ERLIN1, EXOSC3, IBA57, MAG, PGAP1, RAB3GAP2, REEP2, SLC33A1, TFG, USP8, VPS37A, ZFR, ZFYVE27
○ 03261	Invitae Hereditary Spastic Paraplegia Autosomal Dominant Panel	12	ALDH18A1, ATL1, BSCL2, HSPD1, KIF1A, KIF5A, NIPA1, REEP1, RTN2, SPAST, VAMP1, WASHC5
	○ 03261.1 Add-on preliminary-evidence genes	4	CPT1C, REEP2, SLC33A1, ZFYVE27
○ 03262	Invitae Hereditary Spastic Paraplegia Autosomal Recessive Panel	28	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
	○ 03262.1 Add-on preliminary-evidence genes	18	AMPD2, ARL6IP1, ARSI, ATP13A2, C19orf12, CCT5, ENTPD1, ERLIN1, EXOSC3, IBA57, MAG, PGAP1, RAB3GAP2, REEP2, TFG, USP8, VPS37A, ZFR
○ 03263	Invitae Hereditary Spastic Paraplegia X-linked Panel	5	ABCD1, KDM5C, L1CAM, PLP1, SLC16A2

CLINICAL AREA: NEUROLOGY (continued)

Test code	Test name		# gene(s)	Gene list
Cardiomyopathy and Skeletal Muscle Disease				
○ 02252	Invitae Cardiomyopathy and Skeletal Muscle Disease Panel		115	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
	○ 02252.1	Add-on preliminary-evidence genes	36	ANKRD1, CALR3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, HNRNPDL, ILK, JPH2, LAMA4, LDB3, LIMS2, LRRC10, MYF6, MYH6, MYLK2, MYOM1, MYOZ2, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, SUN1, SUN2, SYNE1, SYNE2, TGFB3, TMPO, TXNRD2
	○ 02252.2	Add-on autosomal recessive syndromic pediatric cardiomyopathy genes	7	ACADVL, ALMS1, DNAJC19, ELAC2, MTO1, SDHA, TMEM70

NEUROLOGY INDIVIDUAL GENES

○ A2ML1	○ ANO3	○ BMPR2	○ CHD7	○ CRTAP	○ DPM3	○ FBXO7	○ GJA1
○ AARS	○ ANO5	○ BRAF	○ CHKB	○ CRYAB	○ DRD2	○ FGD4	○ GJA5
○ ABCC9	○ AP4B1	○ BSCL2	○ CHMP2B	○ CSRP3	○ DSC2	○ FHL1	○ GJB1
○ ABCD1	○ AP4E1	○ C12orf65	○ CHRM2	○ CTF1	○ DSG2	○ FHL2	○ GJC2
○ ACADVL	○ AP4M1	○ C19orf12	○ CHRNA1	○ CTNNA3	○ DSP	○ FIG4	○ GLA
○ ACTA1	○ AP4S1	○ CACNA1C	○ CHRNB1	○ CYP2U1	○ DST	○ FKBP14	○ GMPPB
○ ACTA2	○ AP5Z1	○ CACNA1S	○ CHRND	○ CYP7B1	○ DTNA	○ FKRP	○ GNAL
○ ACTB	○ APOB	○ CACNA2D1	○ CHRNE	○ DAG1	○ DYNC1H1	○ FKTN	○ GNB4
○ ACTC1	○ APP	○ CACNB2	○ CHST14	○ DCTN1	○ DYSF	○ FLNA	○ GNE
○ ACTN2	○ ARL6IP1	○ CALM1	○ CIZ1	○ DDHD1	○ EFEMP2	○ FLNC	○ GPC3
○ ACVR2B	○ ARSI	○ CALM2	○ CLCN1	○ DDHD2	○ EGR2	○ FLRT1	○ GPD1L
○ ACVRL1	○ ATL1	○ CALM3	○ CNTN1	○ DEPDC5	○ ELAC2	○ FOXH1	○ GRN
○ ADAMTS2	○ ATL3	○ CALR3	○ COL12A1	○ DES	○ ELN	○ FUS	○ HAMP
○ AGL	○ ATP13A2	○ CAPN3	○ COL1A1	○ DMD	○ EMD	○ GAA	○ HAND1
○ AGRN	○ ATP1A3	○ CASQ2	○ COL1A2	○ DNAJB2	○ ENG	○ GAN	○ HARS
○ AIFM1	○ ATP2A1	○ CAV1	○ COL3A1	○ DNAJB6	○ ENTPD1	○ GARS	○ HCN4
○ AKAP9	○ ATP7A	○ CAV3	○ COL5A1	○ DNAJC19	○ ERLIN1	○ GATA4	○ HFE
○ ALDH18A1	○ B3GALNT2	○ CBL	○ COL5A2	○ DNAJC6	○ ERLIN2	○ GATA6	○ HFE2
○ ALG14	○ B4GALNT1	○ CBS	○ COL6A1	○ DNM2	○ EXOSC3	○ GATAD1	○ HINT1
○ ALG2	○ B4GAT1	○ CCDC78	○ COL6A2	○ DNMT1	○ EYA4	○ GBA2	○ HNRNPA2B1
○ ALMS1	○ BAG3	○ CCT5	○ COL6A3	○ DOK7	○ FA2H	○ GCH1	○ HNRNPDL
○ ALS2	○ BCOR	○ CFL2	○ COLQ	○ DOLK	○ FAM134B	○ GDAP1	○ HPCA
○ AMPD2	○ BICD2	○ CHAT	○ CPT1C	○ DPAGT1	○ FBN1	○ GDF1	○ HRAS
○ ANK2	○ BIN1	○ CHCHD10	○ CPT2	○ DPM1	○ FBN2	○ GDF2	○ HSPB1
○ ANKRD1	○ BMPR1B	○ CHCHD2	○ CRELD1	○ DPM2	○ FBXO38	○ GFPT1	○ HSPB3

NEUROLOGY INDIVIDUAL GENES (continued)

○ HSPB8	○ LAMB2	○ MYH7	○ PKP2	○ RTN2	○ SLC6A3	○ TGFB2	○ VPS35
○ HSPD1	○ LAMP2	○ MYL2	○ PLEC	○ RYR1	○ SLMAP	○ TGFB3	○ VPS37A
○ IBA57	○ LARGE1	○ MYL3	○ PLEKHG5	○ RYR2	○ SMAD3	○ TGFBR1	○ VRK1
○ IGHMBP2	○ LAS1L	○ MYL4	○ PLEKHM2	○ SACS	○ SMAD4	○ TGFBR2	○ WASHC5
○ IKBKAP	○ LDB3	○ MYLK	○ PLN	○ SBF2	○ SMAD6	○ TH	○ WNK1
○ ILK	○ LDLR	○ MYLK2	○ PLOD1	○ SCN10A	○ SMAD9	○ THAP1	○ YARS
○ INF2	○ LDLRAP1	○ MYOM1	○ PLP1	○ SCN11A	○ SMCHD1	○ TIA1	○ ZFPM2
○ ISPD	○ LEFTY2	○ MYOT	○ PMP22	○ SCN1A	○ SMN1, SMN2	○ TMEM43	○ ZFR
○ ITGA7	○ LIMS2	○ MYOZ2	○ PNKD	○ SCN1B	○ SNAP25	○ TMEM5	○ ZFYVE26
○ JAG1	○ LITAF	○ MYPN	○ PNPLA2	○ SCN2B	○ SNCA	○ TMEM70	○ ZFYVE27
○ JPH2	○ LMNA	○ NDRG1	○ PNPLA6	○ SCN3B	○ SNTA1	○ TMPO	○ ZIC3
○ JUP	○ LMOD3	○ NEB	○ POMGNT1	○ SCN4A	○ SOD1	○ TNNC1	
○ KBTBD13	○ LRP4	○ NEBL	○ POMGNT2	○ SCN4B	○ SOS1	○ TNNI3	
○ KCNA5	○ LRRC10	○ NEFL	○ POMK	○ SCN5A	○ SOS2	○ TNNT1	
○ KCND3	○ LRRK2	○ NEXN	○ POMT1	○ SCN8A	○ SPAST	○ TNNT2	
○ KCNE1	○ LRSAM1	○ NF1	○ POMT2	○ SCN9A	○ SPG11	○ TNPO3	
○ KCNE2	○ MAG	○ NGF	○ PRDM12	○ SDHA	○ SPG20	○ TOR1A	
○ KCNE3	○ MAP2K1	○ NIPA1	○ PRDM16	○ SELENON	○ SPG21	○ TOR1AIP1	
○ KCNE5	○ MAP2K2	○ NKX2-5	○ PREPL	○ SETX	○ SPG7	○ TPM1	
○ KCNH2	○ MAPT	○ NKX2-6	○ PRKAG2	○ SGCA	○ SPR	○ TPM2	
○ KCNJ2	○ MARS	○ NODAL	○ PRKG1	○ SGCB	○ SPRED1	○ TPM3	
○ KCNJ5	○ MAT2A	○ NOTCH1	○ PRKRA	○ SGCD	○ SPTLC1	○ TRAPPC11	
○ KCNJ8	○ MATR3	○ NPPA	○ PRNP	○ SGCE	○ SPTLC2	○ TRDN	
○ KCNK3	○ MED12	○ NR2F2	○ PRPS1	○ SGCG	○ SQSTM1	○ TRIM2	
○ KCNQ1	○ MED13L	○ NRAS	○ PRRT2	○ SH3TC2	○ STAC3	○ TRIM32	
○ KCNQ2	○ MED25	○ NSD1	○ PRX	○ SHOC2	○ STIM1	○ TRPM4	
○ KCNQ3	○ MEGF10	○ NT5C2	○ PSEN1	○ SIGMAR1	○ SUN1	○ TRPV4	
○ KCNT1	○ MFN2	○ NTRK1	○ PSEN2	○ SKI	○ SUN2	○ TTN	
○ KCTD17	○ MORC2	○ OPTN	○ PTPN11	○ SLC16A2	○ SURF1	○ TTR	
○ KDM5C	○ MPZ	○ P3H1	○ RAB3GAP2	○ SLC22A5	○ SYNE1	○ TUBB4A	
○ KIF1A	○ MTM1	○ PARK2	○ RAB7A	○ SLC25A46	○ SYNE2	○ TXNRD2	
○ KIF1C	○ MTMR2	○ PARK7	○ RAF1	○ SLC2A1	○ TARDBP	○ UBA1	
○ KIF5A	○ MTO1	○ PCDH19	○ RANGRF	○ SLC2A10	○ TAZ	○ UBQLN2	
○ KLHL40	○ MUSK	○ PCSK9	○ RAPSN	○ SLC33A1	○ TBK1	○ USP8	
○ KLHL41	○ MYBPC3	○ PDK3	○ RASA1	○ SLC39A13	○ TBX5	○ VAMP1	
○ KRAS	○ MYF6	○ PDLIM3	○ RBM20	○ SLC40A1	○ TCAP	○ VAPB	
○ L1CAM	○ MYH11	○ PFN1	○ REEP1	○ SLC52A2	○ TECPR2	○ VCL	
○ LAMA2	○ MYH2	○ PGAP1	○ REEP2	○ SLC52A3	○ TFG	○ VCP	
○ LAMA4	○ MYH6	○ PINK1	○ RIT1	○ SLC5A7	○ TFR2	○ VMA21	

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Epilepsy, Seizures, and Developmental Brain Abnormalities			
○ 03401	Invitae Epilepsy Panel	133	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, 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, SMC1A, SNX27, SPATA5, SPTAN1, ST3GAL5, STRADA, STX1B, STXBP1, SYN1, SYNJ1, SYNGAP1, SZT2, TBC1D24, TCF4, TSC1, TSC2, UBE3A, WWOX, ZDHHC9, ZEB2
	○ 03401.1 Add-on preliminary-evidence genes	50	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
	○ 03401.2 Add-on genes for glycine encephalopathy	3	AMT, GCSH, GLDC
	○ 03401.3 Add-on FLNA gene	1	FLNA
	○ 03401.4 Add-on PTEN gene	1	PTEN
	○ 03401.5 Add-on RANBP2 gene	1	RANBP2
○ 03407	Invitae Alternating Hemiplegia of Childhood Panel	2	ATP1A2, ATP1A3
	○ 03407.1 Add-on clinically overlapping Genes	3	CACNA1A, SCN1A, SLC2A1
○ 04741	Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel	2	ACTB, ACTG1
○ 04422	Invitae Cerebral Cavernous Malformations Panel	3	CCM2, KRIT1, PDCD10
○ 04211	Invitae CHARGE Syndrome Test	1	CHD7
○ 03402	Invitae Early Infantile Epileptic Encephalopathy Panel	53	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
	○ 03402.1 Add-on preliminary-evidence genes	12	ARHGEF15, ATP1A2, COQ4, GABBR2, GPHN, KCNH5, MTOR, NECAP1, NEDD4L, SCN1B, SIK1, ST3GAL3
○ 04424	Invitae Holoprosencephaly Panel	5	GLI2, SHH, SIX3, TGIF1, ZIC2
	○ 04424.1 Add-on preliminary-evidence genes	4	CDON, FOXH1, NODAL, PTCH1
○ 03414	Invitae Neurodegeneration with Brain Iron Accumulation Panel	11	ATP13A2, C19orf12, COASY, CP, DCAF17, FTL, FUCA1, PANK2, PLA2G6, SQSTM1, WDR45
	○ 03414.1 Add-on preliminary-evidence genes	3	FA2H, KIF1A, TRIM32
○ 03404	Invitae Rett/Angelman and Related Disorders Panel	22	ADSL, ALDH5A1, ATRX, CDKL5, CNTNAP2, DYRK1A, EHMT1, FOXG1, IQSEC2, KANSL1, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN8A, SLC9A6, STXBP1, TCF4, UBE3A, ZEB2
	○ 03404.1 Add-on preliminary-evidence genes	4	GABRD, TBL1XR1, JMJD1C, WDR45
○ 01721	Invitae Tuberous Sclerosis Complex Panel	2	TSC1, TSC2

EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES INDIVIDUAL GENES

○ ABAT	○ CDKL5	○ DYNC1H1	○ GLRA1	○ KIF1A	○ PLA2G6	○ SCN5A	○ ST3GAL5
○ ACTB	○ CDON	○ DYRK1A	○ GNAO1	○ KPNA7	○ PLCB1	○ SCN8A	○ STRADA
○ ACTG1	○ CERS1	○ EEF1A2	○ GOSR2	○ KRIT1	○ PNKD	○ SCN9A	○ STX1B
○ ADSL	○ CHD2	○ EFHC1	○ GPHN	○ LGI1	○ PNKP	○ SERPINI1	○ STXBP1
○ ALDH5A1	○ CHD7	○ EHMT1	○ GRIN1	○ LIAS	○ PNPO	○ SETD2	○ SYN1
○ ALDH7A1	○ CHRNA2	○ EPM2A	○ GRIN2A	○ LMNB2	○ POLG	○ SGCE	○ SYNGAP1
○ ALG13	○ CHRNA4	○ FA2H	○ GRIN2B	○ MBD5	○ PPT1	○ SHH	○ SYNJ1
○ AMT	○ CHRNB2	○ FARS2	○ HCN1	○ MECP2	○ PRDM8	○ SIK1	○ SZT2
○ ARHGEF15	○ CLCN4	○ FASN	○ HNRNPU	○ MEF2C	○ PRICKLE1	○ SIX3	○ TBC1D24
○ ARHGEF9	○ CLN2 (TPP1)	○ FOLR1	○ IER3IP1	○ MFSD8	○ PRICKLE2	○ SLC12A5	○ TBL1XR1
○ ARX	○ CLN3	○ FOXG1	○ IQSEC2	○ MTOR	○ PRIMA1	○ SLC13A5	○ TCF4
○ ATP13A2	○ CLN5	○ FOXH1	○ ITPA	○ NECAP1	○ PRRT2	○ SLC19A3	○ TGIF1
○ ATP1A2	○ CLN6	○ FRRS1L	○ JMJD1C	○ NEDD4L	○ PTCH1	○ SLC25A12	○ TPK1
○ ATP1A3	○ CLN8	○ FTL	○ KANSL1	○ NGLY1	○ PTEN	○ SLC25A22	○ TRIM32
○ ATP6AP2	○ CNTN2	○ FUCA1	○ KCNA1	○ NHLRC1	○ PURA	○ SLC2A1	○ TSC1
○ ATRX	○ CNTNAP2	○ GABBR2	○ KCNA2	○ NODAL	○ QARS	○ SLC35A2	○ TSC2
○ BRAT1	○ COASY	○ GABRA1	○ KCNB1	○ NPRL3	○ RANBP2	○ SLC35A3	○ UBE3A
○ C12orf57	○ COQ4	○ GABRA6	○ KCNC1	○ NRXN1	○ RBFOX1	○ SLC6A1	○ WDR45
○ C19orf12	○ CP	○ GABRB2	○ KCND2	○ PACS1	○ RBFOX3	○ SLC6A8	○ WWOX
○ CACNA1A	○ CPA6	○ GABRB3	○ KCNH2	○ PANK2	○ RELN	○ SLC9A6	○ ZDHHC9
○ CACNA1H	○ CSTB	○ GABRD	○ KCNH5	○ PCDH19	○ ROGDI	○ SMC1A	○ ZEB2
○ CACNA2D2	○ CTSD	○ GABRG2	○ KCNJ10	○ PDCD10	○ RYR3	○ SNAP25	○ ZIC2
○ CACNB4	○ DCAF17	○ GAL	○ KCNMA1	○ PIGA	○ SATB2	○ SNX27	
○ CARS2	○ DEPDC5	○ GAMT	○ KCNQ2	○ PIGG	○ SCARB2	○ SPATA5	
○ CASK	○ DIAPH1	○ GATM	○ KCNQ3	○ PIGN	○ SCN1A	○ SPTAN1	
○ CASR	○ DNAJC5	○ GCSH	○ KCNT1	○ PIGO	○ SCN1B	○ SQSTM1	
○ CBL	○ DNM1	○ GLDC	○ KCTD7	○ PIGQ	○ SCN2A	○ SRPX2	
○ CCM2	○ DOCK7	○ GLI2	○ KIAA2022	○ PIK3AP1	○ SCN3A	○ ST3GAL3	

