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Alnylam Act®

HEREDITARY ATTR AMYLOIDOSIS

Alnylam is sponsoring no-charge genetic testing for individuals who may carry a gene mutation known to be associated with hereditary ATTR (hATTR) amyloidosis. The Alnylam Act® program was developed to reduce barriers to genetic testing and counseling to help people make more informed decisions about their health.

ABOUT hATTR AMYLOIDOSIS

hATTR amyloidosis is an inherited, rapidly progressive, debilitating, and fatal disease. It is caused by variants in the transthyretin (TTR) gene that results in misfolded TTR proteins accumulating as amyloid fibrils in multiple tissues including the nerves, heart, and gastrointestinal tract. hATTR amyloidosis can lead to significant morbidity, disability, and mortality, with a median survival of 4.7 years following a diagnosis. The benefits of genetic testing may include the ability to:

- Help patients consider clinical trials
- Shorten the time to diagnosis and prevent misdiagnoses
- Identify risk of disease for patients and their family members

Visit www.invitae.com/en/alnylam-act-ttr to learn more and order a test for your patient.

Brought to you by:

Alnylam® PHARMACEUTICALS

INVITAE

GENETIC TESTING WITH INVITAE

The Alnylam Act® program offers testing with one of the following three options:

Invitae Cardiomyopathy Comprehensive Panel

Testing for ~ 80 genes associated with inherited cardiomyopathy conditions, including hATTR amyloidosis

Invitae Comprehensive Neuropathies Panel

Testing for ~ 100 genes that cause dominant, recessive, and X-linked hereditary neuropathies, including hATTR amyloidosis

Invitae Hereditary Transthyretin-mediated Amyloidosis (hATTR Amyloidosis) Test

Single-gene genetic testing for the TTR gene, which is associated with hATTR amyloidosis

PATIENT ELIGIBILITY

Patients 18 years or older with a suspected diagnosis or a confirmed family history of hATTR amyloidosis are invited to take part in the Alnylam Act® program. Signs and symptoms of hATTR amyloidosis can include:

- Sensory and/or motor neuropathy (e.g., neuropathic pain, alternation sensation [sensitivity to pain and temperature], numbness and tingling, muscle weakness, impaired balance, difficulty walking)
- Autonomic dysfunction (e.g., nausea and vomiting, changes in GI motility [diarrhea, constipation, gastroparesis, early satiety], orthostatic hypotension [fainting and dizziness upon standing], sexual dysfunction, bladder dysfunction)
- Heart disease (e.g., shortness of breath, edema, palpitations, arrhythmias, conduction abnormalities, heart failure, abnormal cardiac imaging [echo, MRI, or technetium])
- Bilateral carpal tunnel syndrome
- Spinal stenosis or spinal radiculopathy
- Ocular changes (e.g., blurred vision, blindness, dry eyes, glaucoma, visual field abnormalities, retinal detachment)
- Biopsy positive for amyloid

For more information or to order a test, please visit www.invitae.com/en/alnylam-act-ttr or contact Invitae Client Services at clientservices@invitae.com or 800-436-3037.

The Alnylam Act® program was created to provide access to genetic testing and counseling to patients as a way to help people make more informed decisions about their health.

- *While Alnylam provides financial support for this program, tests and services are performed by independent third parties.*
- *Healthcare professionals must confirm that patients meet certain criteria to use the program.*
- *Alnylam receives de-identified patient data from this program, but at no time does Alnylam receive patient-identifiable information. Alnylam uses healthcare professional contact information for research and commercial purposes.*
- *Genetic testing is available in the US and certain other countries. Genetic counseling is available in the US.*
- *Healthcare professionals or patients who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use, or support any Alnylam product.*
- *No patients, healthcare professionals, or payers, including government payers, are billed for this program.*

HEADQUARTERS | 1400 16th Street, San Francisco, CA 94103 | **ONLINE** | www.invitae.com | **CONTACT** | www.invitae.com/contact | p: 800-436-3037 | f: 415-276-4164

INVITAE AlnylamAct®

ORDER ID
For Invitae internal use only

Requisition Form
ALNYLAM ACT® (hATTR AMYLOIDOSIS) TRF921-9

This requisition form can be used to submit an order for the **Alnylam Act®**, a sponsored testing program for genetic disorders brought to you by **Alnylam Pharmaceuticals** and **Invitae Corporation**.

INSTRUCTIONS: Review the ordering options and then complete all sections of this form. Your ordering option will be indicated in the test selection section.

ORDERING OPTIONS

1. ALNYLAM ACT® (hATTR AMYLOIDOSIS) PROGRAM

For individuals that meet the eligibility criteria below and wish to receive the program specific genetic testing panels.

REQUIRED: You must select below the appropriate eligibility criteria for this patient.

Patient must be age 18 or older AND have a family history or suspected diagnosis of hATTR amyloidosis with one or more of the following signs and symptoms. Please check all that apply:

- ◯ Family history of hATTR amyloidosis
- ◯ Sensory and/or motor neuropathy (e.g., neuropathic pain, alternation sensation [sensitivity to pain and temperature], numbness and tingling, muscle weakness, impaired balance, difficulty walking)
- ◯ Autonomic dysfunction (e.g., nausea and vomiting, changes in GI motility [diarrhea, constipation, gastroparesis, early satiety], orthostatic hypotension [fainting and dizziness upon standing], sexual dysfunction, bladder dysfunction)
- ◯ Bilateral carpal tunnel syndrome
- ◯ Spinal stenosis or spinal radiculopathy
- ◯ Heart disease (e.g., shortness of breath, edema, palpitations, arrhythmias, conduction abnormalities, heart failure, abnormal cardiac imaging [echo, MRI, or technetium])
- ◯ Renal abnormalities (e.g., renal insufficiency and/or proteinuria)
- ◯ Ocular changes (e.g., blurred vision, blindness, dry eyes, glaucoma, visual field abnormalities, retinal detachment)
- ◯ Biopsy positive for amyloid
- ◯ Other (must be completed if checked): _____

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING

For relatives of program participants who received a Pathogenic/Likely Pathogenic result or approved VUS who want to receive gene specific family follow-up testing at no additional charge. Relatives do not need to meet the eligibility criteria listed above, but must be age 18 or older. Learn more at www.invitae.com/family.

PATIENT INFORMATION

First name	MI	Last name

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

Ancestry ◯ Asian ◯ Black/African American ◯ White/Caucasian ◯ Ashkenazi Jewish ◯ Hispanic ◯ Native American ◯ Pacific Islander ◯ French Canadian ◯ Sephardic Jewish ◯ Mediterranean ◯ Other: _____

Phone	Email address (report access after clinician releases)

Address	City

State/Prov	ZIP/Postal code	Country

Ship a kit to this patient (optional) by faxing or emailing this completed form to Invitae

Kit type: ◯ Buccal swab kit ◯ Saliva kit

Ship to: ◯ Address above ◯ Alternate address: _____

SPECIMEN INFORMATION

Specimen type: Blood (3-mL purple EDTA) -OR- Buccal Swabs (OCD-100, 2 devices) -OR- Saliva (Oragene™) -OR- DNA source: _____

We are unable to accept blood/buccal/saliva from patients with:
- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Specimen collection date (MM/DD/YYYY): *If not provided, the day before specimen receipt will be used*

Special cases: ◯ History of/current hematologic malignancy in patient

INVITAE PARTNER CODE

TTR [ ] *(write in country code, two characters)*

CLINICIAN INFORMATION

Organization name	

Phone	Fax

Address	City

State/Prov	ZIP/Postal code	Country

Primary clinical contact name (if different from ordering provider)	NPI

Primary clinical contact email address (for report access)

Ordering provider (select one ordering provider by marking the checkbox before the name)

	Name	NPI	Email address (for report access)
◯	jkaslf jslfj		
◯			
◯	s		
◯			
◯			
◯			

Additional clinical or laboratory contacts (optional, to share access to order online)

◯ **Share this order with the primary clinical contact's default clinical team, manage at** invitae.com

Name	Email address (for report access)

Name	Email address (for report access)

detect

Cardiomyopathy and Arrhythmia

INVITAE

Sponsored, no-charge genetic testing and counseling for individuals suspected of having a familial arrhythmia or cardiomyopathy

IMPROVE DIAGNOSIS, RISK STRATIFICATION, AND MANAGEMENT

IDENTIFY AT-RISK FAMILY MEMBERS—BEFORE A LIFE-THREATENING EVENT

POST-TEST GENETIC COUNSELING FOR US AND CANADA PATIENTS OFFERED AT NO CHARGE

Detect Cardiomyopathy and Arrhythmia offers the Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel to test up to 150 genes associated with inherited arrhythmia and cardiomyopathy conditions, including:

- Hypertrophic cardiomyopathy
- Dilated cardiomyopathy
- Arrhythmogenic cardiomyopathy
- Left ventricular noncompaction
- Long QT syndrome
- Brugada syndrome
- Catecholaminergic polymorphic ventricular tachycardia

All major cardiology professional societies, including the American College of Cardiology, American Heart Association, Heart Rhythm Society, and Heart Failure Society of America, recommend genetic testing.[1–6]

www.invitae.com/DetectCardio

While third parties and commercial organizations may provide financial support for this program, tests and services are performed by Invitae. Healthcare professionals must confirm that patients meet certain criteria to use the program. Third parties and commercial organizations may receive de-identified patient data from this program, but at no time would they receive patient identifiable information. Third parties and commercial organizations may receive contact information for healthcare professionals who use this program. Genetic testing is available in the US, Canada, Argentina, Australia, Brazil, Chile, Colombia, and Mexico only. Healthcare professionals and patients who participate in this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any other products or services from Invitae or from third parties or commercial organizations.

1. Ackerman MJ et al. Heart Rhythm. 2011;8;1308-39.
2. Gersh BJ et al. J Am. Coll Cardiol. 2011;58:2703-38.
3. Priori SG et al. Heart Rhythm. 2013;10:1932-63.
4. Al-Khatib SM et al. J Am Coll Cardiol. 2018;72:e91-e220.
5. Hershberger RE et al. J. Card. Fail. 2018;24,281-302.
6. Towbin JA et al. Heart Rhythm. 2019. doi:10.1016/j.hrthm.2019.05.007.

HEADQUARTERS | 1400 16th Street, San Francisco, CA 94103 | **ONLINE** | www.invitae.com | **CONTACT** | www.invitae.com/contact

INVITAE

detect
Cardiomyopathy and Arrhythmia

ORDER ID
For Invitae internal use only

Requisition Form
Detect Cardiomyopathy and Arrhythmia TRF955-6

This requisition form can be used to submit an order for the **Invitae Detect Cardiomyopathy and Arrhythmia program**, a sponsored testing program for familial cardiomyopathies or arrhythmias.

INSTRUCTIONS: Review the ordering options and then complete all sections of this form. Your ordering option will be indicated in the test selection section.

ORDERING OPTIONS

1. DETECT CARDIOMYOPATHY AND ARRHYTHMIA PROGRAM

For individuals that meet the eligibility criteria below and wish to receive the program specific genetic testing panels.

REQUIRED: You must select below the appropriate eligibility criteria for this patient.

This program is available to patients suspected of having a familial cardiomyopathy or arrhythmia (select one or more):**

◯ Suspicion or known diagnosis of a familial cardiomyopathy or arrhythmia
Diagnosis: ◯HCM ◯DCM ◯ARVC ◯LVNC ◯LQTS ◯CPVT ◯BrS ◯Other: _____
Age at diagnosis: _____
Index of clinical suspicion: ◯High ◯Moderate ◯Low

◯ Family history of a primary cardiomyopathy or arrhythmia
Diagnosis: ◯HCM ◯DCM ◯ARVC ◯LVNC ◯LQTS ◯CPVT ◯BrS ◯Other: _____

◯ Family history of unexplained sudden cardiac death
Age(s): _____

◯ Patient is deceased* ◯Yes ◯No

**If the patient is deceased please also complete the postmortem consent form located at www.invitae.com/postmortem-consent*

***Please note that this program is available only in select countries; for a complete list please visit https://www.invitae.com/en/detect-cardiomyopathy-arrhythmia/*

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING

For relatives of program participants who received a Pathogenic/Likely Pathogenic result or approved VUS who want to receive gene specific family follow-up testing at no additional charge. Relatives do not need to meet the eligibility criteria listed above. Learn more at www.invitae.com/family.

PATIENT INFORMATION

First name	MI	Last name
Date of birth (MM/DD/YYYY)	Biological sex ◯M ◯F	MRN (medical record number)

Ancestry ◯Asian ◯Black/African American ◯White/Caucasian ◯Ashkenazi Jewish ◯Hispanic ◯Native American ◯Pacific Islander ◯French Canadian ◯Sephardic Jewish ◯Mediterranean ◯Other: _____

Phone	Email address (report access after clinician releases)	
Address		City
State/Prov	ZIP/Postal code	Country

Ship a kit to this patient (optional) by faxing or emailing this completed form to Invitae

Kit type: ◯Buccal swab kit ◯Saliva kit

Ship to: ◯Address above ◯Alternate address: _____

SPECIMEN INFORMATION

Specimen type: Blood (3-mL purple EDTA) -OR- Buccal Swabs (OCD-100, 2 devices) -OR- Saliva (Oragene™) -OR- DNA source: _____

We are unable to accept blood/buccal/saliva from patients with:
- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Specimen collection date (MM/DD/YYYY):
If not provided, the day before specimen receipt will be used

Special cases: ◯History of/current hematologic malignancy in patient

INVITAE PARTNER CODE CARDIO

CLINICIAN INFORMATION

Organization name		
Phone	Fax	
Address	City	
State/Prov	ZIP/Postal code	Country

Primary clinical contact name (if different from ordering provider)	NPI (US only)
Primary clinical contact email address (for report access)	

Ordering provider (select one ordering provider by marking the checkbox before the name)

	Name	NPI (US only)	Email address (for report access)
◯			
◯			
◯			
◯			
◯			
◯			

Additional clinical or laboratory contacts (optional, to share access to order online)

◯ **Share this order with the primary clinical contact's default clinical team, manage at invitae.com**

Name	Email address (for report access)
Name	Email address (for report access)

Transparent pricing

MAKING GENETIC TESTING MORE ACCESSIBLE THAN EVER

Genetic testing should be affordable and accessible. At Invitae, we believe in ethical and transparent billing. We offer multiple billing options and have an exceptional Client Services team ready to work with you.

For a flat price, your healthcare provider can order testing on any number of genes within a single clinical area. (For example, hereditary cancer is a clinical area, as are cardiology and pediatric genetics.)

$100
OUT OF POCKET

Typically patients pay no more than $100 out of pocket for one of our tests

$250
PATIENT PAY

If preferred, patients have the option to pay $250 per clinical area

INSURANCE BILLING

Invitae can bill insurance directly for panel tests; please visit www.invitae.com/in-network-partners for a list of insurance companies that have brought Invitae in-network. We also accept Medicare and Medicaid. You won't need to contact your insurance company; Invitae will work directly with them to coordinate coverage and payment.

Out-of-pocket expenses

Regardless of whether our laboratory is in-network or out-of-network with your insurance provider, Invitae is committed to making genetic testing affordable. For testing related to a personal or family history of breast, ovarian, colorectal, or uterine cancer (also referred to as HBOC and Lynch syndrome), Invitae offers an out-of-pocket cost estimator, accessible at www.invitae.com/patient-billing. Typically patients pay no more than $100 out of pocket for one of our tests. If you receive a bill for more than $100, please call our billing experts at 800-436-3037 for access to patient programs.

PATIENT PAY

If preferred, you have the option to pay $250 per clinical area for panel testing. To take advantage of this pricing, you must submit payment upfront and in full before test results are released. In addition, your clinician must place the order online and provide your e-mail address. The patient-pay option is available as a prepaid option only and does not allow Invitae to submit claims to your insurance company. It also does not allow Invitae to apply financial assistance programs.

PATIENT ASSISTANCE PROGRAM

Invitae is committed to making genetic testing affordable and accessible by removing financial and logistical barriers. Our Patient Assistance Program (PAP) is available to patients in the US who undergo testing with Invitae and meet income criteria. Please contact Client Services to learn more about our interest-free payment plans and financial assistance program.

continued on next page

Genetic counseling resources at Invitae

Our experienced, board-certified genetic counselors are available by telephone to answer your questions about genetic testing, including:

- On-demand conversations with genetic counselors who can answer brief questions about testing and test results
- Comprehensive genetic counseling session, after you receive your test results from your healthcare provider, to discuss the results and what they mean for you and your family members*

To ask brief questions or schedule a comprehensive genetic counseling session, please call Invitae at **1-800-436-3037** Monday through Friday, 5 am to 5 pm Pacific and ask to speak with a genetic counselor. These services are included in the cost of the testing.

Scan to schedule an appointment with one of our genetics experts.

** Comprehensive genetic counseling sessions available for hereditary cancer, cardiology, ophthalmology, reproductive health, and proactive health screens only at this time.*

INVITAE

ORDER ONLINE at www.invitae.com/signin.

ORDER ID
For Invitae internal use only

INVITAE REQUISITION FORM

PATIENT INFORMATION

First name | MI | Last name | Date of birth (MM/DD/YYYY)

Biological sex: ○ Male ○ Female

MRN (medical record number)

Ancestry: ○ Asian ○ Black/African American ○ White/Caucasian ○ Ashkenazi Jewish ○ Hispanic ○ Native American ○ Pacific Islander ○ French Canadian ○ Sephardic Jewish ○ Mediterranean ○ Other: _____

Email address (for billing contact and report access after clinician releases)

Mobile phone (for billing contact)

Address

City | State/Prov | Zip/Postal code | Country

Ship a saliva kit to this patient (to submit this request, fax this completed requisition form to Invitae Client Services at 415-276-4164)

○ Ship kit to address above ○ Ship kit to alternate address: _____

CLINICAL INFORMATION

Organization name	Phone	Fax

Address	City	State/Prov	ZIP/Postal Code	Country

CLINICAL TEAM

Primary clinical contact (contact for general inquires)

Name	NPI	Email address (for report access)

Ordering provider ○ Same as primary clinical contact

For your convenience, we have provided multiple fields below to pre-populate your organization's provider list. For each order, indicate one ordering provider.

	Name	NPI	Email address (for report access)
○			
○			
○			
○			
○			

Additional clinical or laboratory contacts (optional; share online access to this order with the contacts below)

○ Share this order with the primary clinical contact's default clinical team (establish and manage team online at www.invitae.com/signin)

Name	Email address (for report access)	Name	Email address (for report access)
Name	Email address (for report access)	Name	Email address (for report access)

INSURANCE BILLING (attach front and back of insurance card)

Attach clinical notes, medical records, and/or letter of medical necessity (LMN) to prevent delays. We do not accept insurance for certain tests *or* patients outside the US. www.invitae.com/billing

Policyholder name	Patient relationship to policyholder: ○ Self ○ Spouse ○ Child ○ Other: _____			Medicare insurance billing only (select one): ○ Patient was treated as a hospital inpatient (more than a 24 hour stay) in the last 14 days ○ Not a hospital patient
Primary insurance company name	Primary member ID#	Primary insurance phone	Prior-authorization #	
Secondary insurance company name	Secondary member ID#	Secondary insurance phone	Prior-authorization #	

PATIENT PAY BILLING

Invitae will send an electronic invoice to the patient email listed above. Insurance will not be billed.

INSTITUTIONAL BILLING

Invitae will send an invoice to the organization address above. Please contact Invitae if this order should be billed to a different location.

PARTNERSHIP PROGRAMS

Invitae partner code:

INVITAE

Patient's first name | Patient's last name

INVITAE DIAGNOSTIC REQUISITION FORM

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

Collection date (MM/DD/YYYY)

If not provided, date will be 1 day prior to our receipt of specimen. For DNA, provide date retrieved from archive.

Specimen type

○ Blood ○ Saliva ○ DNA - source: _____

DNA must be extracted in a CLIA or other suitably certified laboratory. We are unable to accept blood or saliva from patients with allogeneic bone marrow transplants or a blood transfusion <2 weeks prior to specimen collection.

Specimen ID (IB # on tube):

Is this patient deceased? ○ Yes ○ No

Deceased date (MM/DD/YYYY)

REASON FOR TESTING

Primary indication:

ONCOLOGY

○ Hereditary breast and ovarian cancer (HBOC) syndrome
○ Lynch syndrome
○ Polyposis (FAP)
○ Other: _____

CARDIOLOGY

○ Aortopathy ○ Cardiomyopathy
○ Arrhythmia ○ Other: _____

OTHER

○ Neurology
○ Other: _____

ICD-10 codes (required for insurance billing)

PERSONAL HISTORY

Is/was this patient affected or symptomatic†? ○ Yes ○ No If yes, describe below and attach clinical notes.

Age at diagnosis: _____

†Symptomatic means the patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

Is there a hematological malignancy in this patient (current or history of)? ○ Yes ○ No

Has this patient had genetic testing before? ○ Yes ○ No If yes, write test results and attach the report.

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ○ Yes ○ No
If yes, describe below and attach pedigree and/or clinical notes.

Relationship to patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

TEST SELECTION

OPTION 1: SELECT AN INVITAE PANEL FROM OUR TEST CATALOG

Select your desired test(s) from the attached test catalog and discard any pages without a selection.

OPTION 2: INVITAE TEST CODE

Indicate test IDs here (reference www.invitae.com/tests or our test catalog). Test IDs containing add-on codes will include the original panel as well as the add-on.

Test code	Add-on code (optional)	Test code	Add-on code (optional)

OR

Invitae supports customization of your test. To create a custom panel, log in to your Invitae portal account or contact Client Services. Then indicate the ID associated with that panel here.

Custom panel ID

OPTION 3: FAMILY FOLLOW-UP TESTING

Invitae family follow-up testing is available at no additional charge for blood relatives of patients who receive pathogenic or likely pathogenic results (or approved VUS). Learn more at www.invitae.com/family.

Invitae proband RQ# _____

Relationship to proband _____

Gene(s) _____

Variant(s) _____

Invitae's family follow-up testing analyzes the variant(s) indicated above. If you would like this report to include any variants of uncertain significance and be eligible for re-requisition, please include billing information on this requisition form and check here: ☐

AUTOMATIC REFLEX: Invitae offers one re-requisition at no additional charge for tests within the same clinical area (www.invitae.com/re-requisition). Preschedule it here or in your Invitae portal.

Conditions for reflex: ○ Regardless of initial results ○ Only if negative (no pathogenic/likely pathogenic results)

Reflex test: **Test code** _____ . _____ **Add-on code (optional)**

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/forms). For orders originating outside the US, the Patient has been informed their personal information and specimen will be transferred to and processed in the US. The Patient has been informed that Invitae may notify them of clinical updates related to genetic test results (in consultation with the ordering medical professional). If insurance billing is selected, the Patient has been informed and authorizes Invitae Corporation ("Invitae") and its designees to release information concerning testing to their insurer. The medical professional agrees to allow Invitae (1) to transfer the information from this TRF to a letter of medical necessity and/or other documentation using the medical professional's name as the signature as well as (2) assist the patient in obtaining pre-test genetic counseling from a third-party service, as required by the patient's insurance provider. I acknowledge that the Patient has agreed that if the Patient's insurer does not reimburse Invitae in full for any reason then Invitae may bill the Patient for the services and the Patient will remit payment to Invitae. For amounts the Patient receives from the insurer, the Patient has agreed to remit payment to Invitae for services rendered. I acknowledge that I offered pre-test genetic counseling to the Patient, if required by their insurer. I attest that I am authorized under applicable law to order this test.

Medical professional signature (required) | **Date (MM/DD/YYYY)**

WITHIN THE US | p: 800-436-3037 | f: 415-276-4164 | e: clientservices@invitae.com | OUTSIDE THE US | p: +1 415-930-4018 | www.invitae.com/contact | e: globalsupport@invitae.com
INVITAE | 1400 16th Street, San Francisco, CA 94103, USA | www.invitae.com |

CARDIOLOGY TEST CATALOG

All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send a separate specimen for each clinical area. Each clinical area represents an individual billable event and report. Contact Client Services with any questions. For Invitae's full test menu, please visit www.invitae.com.

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

Test code	Test name	# gene(s)	Gene list
Hereditary Hemorrhagic Telangiectasia and Vascular Malformations			
○ 02352	Invitae Hereditary Hemorrhagic Telangiectasia and Vascular Malformations Panel	6	ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4
Arrhythmia and Cardiomyopathy			
○ 02101	Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel	100	ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BRAF, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CBL, CDH2, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GATA4, GATA5, GJA5, GLA, HCN4, HRAS, JUP, KCNE1, KCNH2, KCNJ2, KCNQ1, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYL4, MYLK3, NF1, NKX2-5, NRAS, PCCA, PCCB, PKP2, PLN, PPA2, PPCS, PPP1CB, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RYR2, SCN5A, SDHA, SGCD, SHOC2, SLC22A5, SOS1, SOS2, SPRED1, TAZ, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL
○ 02101.1	Add-on preliminary-evidence genes	57	A2ML1, AKAP9, ANK2, ANKRD1, CACNA2D1, CACNB2, CALR3, CAV3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA6, GATAD1, GPD1L, HAND1, ILK, JPH2, KCNA5, KCND3, KCNE2, KCNE3, KCNE5, KCNJ5, KCNJ8, KCNK3, KIF20A, KLF10, LAMA4, LDB3, LRRC10, MAP3K8, MED12, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NPPA, PDLIM3, PLEKHM2, PRDM16, RANGRF, RASA2, RRAS, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SLMAP, SNTA1, TMPO, TXNRD2
○ 02101.4	Add-on sudden unexpected death in epilepsy (SUDEP) genes for arrhythmia and cardiomyopathy	11	DEPDC5, KCNA1, KCNQ2, KCNQ3, KCNT1, PCDH19, PRRT2, SCN1A, SCN8A, SCN9A, SLC2A1
Arrhythmia			
○ 02211	Invitae Long QT Syndrome Panel	10	CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN
○ 02211.1	Add-on preliminary-evidence genes	7	AKAP9, ANK2, CAV3, KCNE2, KCNJ5, SCN4B, SNTA1
○ 02213	Invitae Catecholaminergic Polymorphic Ventricular Tachycardia Panel	7	CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN
○ 02214	Invitae Short QT Syndrome Panel	3	KCNH2, KCNJ2, KCNQ1
○ 02214.1	Add-on preliminary-evidence gene	3	CACNA1C, CACNA2D1, CACNB2
○ 02201	Invitae Arrhythmia Comprehensive Panel	41	ABCC9, ACTN2, BAG3, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CDH2, DES, DSC2, DSG2, DSP, EMD, FLNC, GATA4, GATA5, GJA5, HCN4, JUP, KCNE1, KCNH2, KCNJ2, KCNQ1, LMNA, MYL4, NKX2-5, PKP2, PLN, PPA2, PRKAG2, RBM20, RYR2, SCN5A, TMEM43, TNNI3, TNNT2, TRDN, TRPM4, TTN
○ 02201.1	Add-on preliminary-evidence genes	28	AKAP9, ANK2, ANKRD1, CACNA2D1, CACNB2, CAV3, CTNNA3, GATA6, GPD1L, KCNA5, KCND3, KCNE2, KCNE3, KCNE5, KCNJ5, KCNJ8, KCNK3, LDB3, NPPA, PDLIM3, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SLMAP, SNTA1
○ 02201.2	Add-on sudden unexpected death in epilepsy (SUDEP) genes for arrhythmia	11	DEPDC5, KCNA1, KCNQ2, KCNQ3, KCNT1, PCDH19, PRRT2, SCN1A, SCN8A, SCN9A, SLC2A1
○ 02212	Invitae Brugada Syndrome Test	1	SCN5A
○ 02212.1	Add-on preliminary-evidence genes	19	ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNH2, KCNE3, KCNE5, KCNJ8, PKP2, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SLMAP, TRPM4
Cardiomyopathy			
○ 02261	Invitae Hypertrophic Cardiomyopathy Panel	30	ACADVL, ACTC1, ACTN2, AGL, ALPK3, BAG3, CACNA1C, CPT2, CSRP3, DES, ELAC2, FHL1, FLNC, GAA, GLA, LAMP2, MTO1, MYBPC3, MYH7, MYL2, MYL3, PLN, PRKAG2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL
○ 02261.1	Add-on preliminary-evidence genes	14	ANKRD1, CALR3, CAV3, GATA4, JPH2, KLF10, LDB3, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, PDLIM3
○ 02265	Invitae Hereditary Transthyretin-mediated amyloidosis (hATTR amyloidosis) Test	1	TTR

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CARDIOLOGY TEST CATALOG

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

Test code	Test name	# gene(s)	Gene list
Cardiomyopathy (continued)			
○ 04151	Invitae RASopathies Comprehensive Panel	18	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1
○ 02262	Invitae Dilated Cardiomyopathy and Left Ventricular Noncompaction Panel	54	ABCC9, ACADVL, ACTC1, ACTN2, ALMS1, ALPK3, BAG3, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FKRP, FKTN, FLNC, HCN4, JUP, LAMP2, LMNA, MYBPC3, MYH7, MYLK3, PCCA, PCCB, PKP2, PLN, PPCS, RAF1, RBM20, RYR2, SCN5A, SDHA, SGCD, SLC22A5, TAZ, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL
	○ 02262.1 Add-on preliminary-evidence genes	26	ANKRD1, CAV3, CHRM2, CTF1, DTNA, FHL2, GATA4, GATA6, GATAD1, HAND1, ILK, LAMA4, LDB3, LRRC10, MED12, MYH6, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, TMPO, TXNRD2
○ 02251	Invitae Cardiomyopathy Comprehensive Panel	82	ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BRAF, CACNA1C, CBL, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GLA, HCN4, HRAS, JUP, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYLK3, NF1, NRAS, PCCA, PCCB, PKP2, PLN, PPCS, PPP1CB, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RYR2, SCN5A, SDHA, SGCD, SHOC2, SLC22A5, SOS1, SOS2, SPRED1, TAZ, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL
	○ 02251.1 Add-on preliminary-evidence genes	39	A2ML1, ANKRD1, CALR3, CAV3, CDH2, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, HAND1, ILK, JPH2, KIF20A, KLF10, LAMA4, LDB3, LRRC10, MAP3K8, MED12, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, RASA2, RRAS, TMPO, TXNRD2
○ 02263	Invitae Arrhythmogenic Cardiomyopathy Panel	22	ACTN2, BAG3, CDH2, DES, DSC2, DSG2, DSP, EMD, FLNC, JUP, LMNA, PKP2, PLN, PPA2, PRKAG2, RBM20, RYR2, SCN5A, TMEM43, TNNI3, TNNT2, TTN
	○ 02263.1 Add-on preliminary-evidence genes	5	ANKRD1, CTNNA3, LDB3, NKX2-5, PDLIM3
○ 05201	Invitae Hereditary Hemochromatosis Panel	5	HAMP, HFE, HJV, SLC40A1, TFR2
Aortopathy and Connective Tissue Disorders			
○ 02301	Invitae Aortopathy Comprehensive Panel	29	ACTA2, ADAMTS10, BGN, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, FLNA, FOXE3, LOX, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2
	○ 02301.1 Add-on preliminary-evidence genes	6	ARIH1, HCN4, LTBP3, MAT2A, PLOD3, SMAD6
○ 02311	Invitae Loeys-Dietz Syndrome Panel	7	FBN1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2
○ 02313	Invitae Ehlers-Danlos Syndrome Panel	17	ADAMTS2, ATP7A, B3GALT6, B4GALT7, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CRTAP, FKBP14, FLNA, P3H1, PLOD1, SLC39A13
○ 02312	Invitae Marfan Syndrome Test	1	FBN1
Lipid Disorders			
○ 02401	Invitae Familial Hypercholesterolemia Panel	4	APOB, LDLR, LDLRAP1, PCSK9
○ 53698	Invitae Comprehensive Lipidemia Panel	25	ABCA1, ABCG5, ABCG8, ANGPTL3, APOA1, APOA5, APOB, APOC2, CETP, CREB3L3, CYP27A1, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPA, LIPG, LMF1, LPL, LRP6, MTTP, PCSK9, PNPLA2, SAR1B
	○ 53698.1 Add-on preliminary-evidence genes	11	APOA4, APOC3, CYP7A1, GALNT2, GCKR, LIPC, LIPI, MYLIP, PLTP, SCARB1, ZHX3
Pulmonary Hypertension			
○ 02351	Invitae Pulmonary Arterial Hypertension Panel	12	ACVRL1, AQP1, ATP13A3, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNK3, SMAD9, SOX17, TBX4
	○ 02351.1 Add-on preliminary-evidence genes	2	BMPR1B, KCNA5
Congenital Heart Disease			
○ 04151	Invitae RASopathies Comprehensive Panel	18	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1

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CARDIOLOGY TEST CATALOG

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

Test code	Test name	# gene(s)	Gene list
Congenital Heart Disease (continued)			
○ 04204	Invitae Congenital Heart Disease Panel	55	ACTC1, ACVR2B, ALMS1, BCOR, BRAF, CASZ1, CBL, CHD7, CRELD1, ELN, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HAND1, HAND2, HRAS, JAG1, KDM6A, KMT2D, KRAS, LEFTY2, MAP2K1, MAP2K2, MED13L, MEIS2, MESP1, MYH6, NFATC1, NKX2-5, NKX2-6, NODAL, NOTCH1, NR2F2, NRAS, NSD1, PLD1, PTPN11, RAF1, RBFOX2, RIT1, ROBO1, SHOC2, SMAD6, SOS1, TAB2, TBX1, TBX20, TBX5, TFAP2B, ZFPM2, ZIC3

CARDIOLOGY AND NEUROLOGY INDIVIDUAL GENES

○ A2ML1	○ BGN	○ CSRP3	○ GALNT2	○ KCNJ2	○ MAP3K8	○ NRAS	○ SAR1B	○ TBX20
○ ABCA1	○ BMPR1B	○ CTF1	○ GATA4	○ KCNJ5	○ MAT2A	○ NSD1	○ SCARB1	○ TBX4
○ ABCC9	○ BMPR2	○ CTNNA3	○ GATA5	○ KCNJ8	○ MED12	○ P3H1	○ SCN10A	○ TBX5
○ ABCG5	○ BRAF	○ CYP27A1	○ GATA6	○ KCNK3	○ MED13L	○ PCCA	○ SCN1A	○ TCAP
○ ABCG8	○ CACNA1C	○ CYP7A1	○ GATAD1	○ KCNQ1	○ MEIS2	○ PCCB	○ SCN1B	○ TFAP2B
○ ACADVL	○ CACNA1D	○ DEPDC5	○ GCKR	○ KCNQ2	○ MESP1	○ PCDH19	○ SCN2B	○ TFR2
○ ACTA2	○ CACNA2D1	○ DES	○ GDF1	○ KCNQ3	○ MFAP5	○ PCSK9	○ SCN3B	○ TGFB2
○ ACTC1	○ CACNB2	○ DMD	○ GDF2	○ KCNT1	○ MRAS	○ PDLIM3	○ SCN4B	○ TGFB3
○ ACTN2	○ CALM1	○ DNAJC19	○ GJA1	○ KDM6A	○ MTO1	○ PKP2	○ SCN5A	○ TGFBR1
○ ACVR2B	○ CALM2	○ DOLK	○ GJA5	○ KIF20A	○ MTTP	○ PLD1	○ SCN8A	○ TGFBR2
○ ACVRL1	○ CALM3	○ DSC2	○ GLA	○ KLF10	○ MYBPC3	○ PLEKHM2	○ SCN9A	○ TMEM43
○ ADAMTS10	○ CALR3	○ DSG2	○ GPC3	○ KMT2D	○ MYH11	○ PLN	○ SDHA	○ TMEM70
○ ADAMTS2	○ CASQ2	○ DSP	○ GPD1	○ KRAS	○ MYH6	○ PLOD1	○ SGCD	○ TMPO
○ AGL	○ CASZ1	○ DTNA	○ GPD1L	○ LAMA4	○ MYH7	○ PLOD3	○ SHOC2	○ TNNC1
○ AKAP9	○ CAV1	○ EFEMP2	○ GPIHBP1	○ LAMP2	○ MYL2	○ PLTP	○ SKI	○ TNNI3
○ ALMS1	○ CAV3	○ EIF2AK4	○ HAMP	○ LCAT	○ MYL3	○ PNPLA2	○ SLC22A5	○ TNNI3K
○ ALPK3	○ CBL	○ ELAC2	○ HAND1	○ LDB3	○ MYL4	○ PPA2	○ SLC2A1	○ TNNT2
○ ANGPTL3	○ CBS	○ ELN	○ HAND2	○ LDLR	○ MYLIP	○ PPCS	○ SLC2A10	○ TPM1
○ ANK2	○ CDH2	○ EMD	○ HCN4	○ LDLRAP1	○ MYLK	○ PPP1CB	○ SLC39A13	○ TRDN
○ ANKRD1	○ CETP	○ ENG	○ HFE	○ LEFTY2	○ MYLK2	○ PRDM16	○ SLC40A1	○ TRPM4
○ APOA1	○ CHD7	○ EPHB4	○ HJV	○ LIPA	○ MYLK3	○ PRKAG2	○ SLMAP	○ TTN
○ APOA4	○ CHRM2	○ EYA4	○ HRAS	○ LIPC	○ MYOM1	○ PRKG1	○ SMAD2	○ TTR
○ APOA5	○ CHST14	○ FBN1	○ ILK	○ LIPG	○ MYOZ2	○ PRRT2	○ SMAD3	○ TXNRD2
○ APOB	○ COL12A1	○ FBN2	○ JAG1	○ LIPI	○ MYPN	○ PTPN11	○ SMAD4	○ VCL
○ APOC2	○ COL1A1	○ FHL1	○ JPH2	○ LMF1	○ NEBL	○ RAF1	○ SMAD6	○ ZFPM2
○ APOC3	○ COL1A2	○ FHL2	○ JUP	○ LMNA	○ NEXN	○ RANGRF	○ SMAD9	○ ZHX3
○ AQP1	○ COL3A1	○ FKBP14	○ KCNA1	○ LOX	○ NF1	○ RASA1	○ SNTA1	○ ZIC3
○ ARIH1	○ COL5A1	○ FKRP	○ KCNA5	○ LPL	○ NFATC1	○ RASA2	○ SOS1	
○ ATP13A3	○ COL5A2	○ FKTN	○ KCND3	○ LRP6	○ NKX2-5	○ RBFOX2	○ SOS2	
○ ATP7A	○ CPT2	○ FLNA	○ KCNE1	○ LRRC10	○ NKX2-6	○ RBM20	○ SOX17	
○ B3GALT6	○ CREB3L3	○ FLNC	○ KCNE2	○ LTBP3	○ NODAL	○ RIT1	○ SPRED1	
○ B4GALT7	○ CRELD1	○ FOXE3	○ KCNE3	○ LZTR1	○ NOTCH1	○ ROBO1	○ TAB2	
○ BAG3	○ CRTAP	○ FOXH1	○ KCNE5	○ MAP2K1	○ NPPA	○ RRAS	○ TAZ	
○ BCOR	○ CRYAB	○ GAA	○ KCNH2	○ MAP2K2	○ NR2F2	○ RYR2	○ TBX1	

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INVITAE

Patient's first name

Patient's last name

NEUROLOGY TEST CATALOG

All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send a separate specimen for each clinical area. Each clinical area represents an individual billable event and report. Contact Client Services with any questions. For Invitae's full test menu, please visit www.invitae.com.

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

Test code	Test name	# gene(s)	Gene list
Movement Disorders			
○ 03351	Invitae Dystonia Comprehensive Panel	38	ACTB, ADCY5, ANO3, ATP1A3, ATP7B, BCAP31, CIZ1, COL6A3, CYP27A1, GCH1, GNAL, GNAO1, HEXA, HPCA, KCNMA1, KCTD17, KMT2B, MECR, PANK2, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, SGCE, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SPR, TH, THAP1, TOR1A, TUBB4A, VAC14, VPS13A, VPS13D, XPR1
	○ 03351.1 Add-on preliminary-evidence genes	3	DRD2, MED20, TOR1AIP1
○ 03352	Invitae Hereditary Parkinson's Disease & Parkinsonism Panel	26	ATP13A2, ATP7B, CHCHD2, CSF1R, DCTN1, DNAJC6, FBXO7, GBA, GCH1, LRRK2, PARK7, PDE8B, PINK1, PLA2G6, PRKN, PRKRA, RAB39B, SLC6A3, SNCA, SPR, SYNJ1, TH, TMEM230, VPS13C, VPS35, XPR1
	○ 03352.1 Add-on preliminary-evidence genes	3	MAPT, PODXL, UCHL1
Neurovascular Disorders			
○ 53700	Invitae Familial Hemiplegic Migraine Panel	7	ATP1A2, ATP1A3, CACNA1A, PRRT2, SCN1A, SLC1A3, SLC2A1
	○ 53700.1 Add-on preliminary-evidence gene	1	KCNK18
○ 53701	Invitae Hereditary Cerebral Small Vessel Disease Panel	10	APP, CBS, COL4A1, COL4A2, CST3, FOXC1, GLA, HTRA1, NOTCH3, TREX1
○ 53702	Invitae Hereditary Moyamoya Disease Panel	2	GUCY1A1, RNF213
	○ 53702.1 Add-on preliminary-evidence gene	1	CCER2
Neurodegenerative Disorders			
○ 03352	Invitae Hereditary Parkinson's Disease & Parkinsonism Panel	26	ATP13A2, ATP7B, CHCHD2, CSF1R, DCTN1, DNAJC6, FBXO7, GBA, GCH1, LRRK2, PARK7, PDE8B, PINK1, PLA2G6, PRKN, PRKRA, RAB39B, SLC6A3, SNCA, SPR, SYNJ1, TH, TMEM230, VPS13C, VPS35, XPR1
	○ 03352.1 Add-on preliminary-evidence genes	3	MAPT, PODXL, UCHL1
○ 03502	Invitae Hereditary Amyotrophic Lateral Sclerosis, Frontotemporal Dementia and Alzheimer Disease Panel	33	ALS2, ANG, ANXA11, APP, CHCHD10, CHMP2B, DCTN1, ERBB4, FUS, GRN, HEXA, HNRNPA2B1, ITM2B, KIF5A, MAPT, OPTN, PFN1, PRNP, PSEN1, PSEN2, SETX, SNCA, SOD1, SORL1, SPG11, SQSTM1, TARDBP, TBK1, TFG, TREM2, UBQLN2, VAPB, VCP
	○ 03502.1 Add-on preliminary-evidence genes	9	ATP13A2, DDHD1, ERLIN1, FIG4, LRRK2, MATR3, NEFH, SIGMAR1, TIA1
○ 03503	Invitae Amyotrophic Lateral Sclerosis Panel	21	ALS2, ANG, ANXA11, CHCHD10, DCTN1, ERBB4, FUS, HEXA, KIF5A, OPTN, PFN1, SETX, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TFG, UBQLN2, VAPB, VCP
	○ 03503.1 Add-on preliminary-evidence genes	9	ATP13A2, CHMP2B, DDHD1, ERLIN1, FIG4, MATR3, NEFH, SIGMAR1, TIA1
○ 03505	Invitae Frontotemporal Dementia Panel	13	CHCHD10, CHMP2B, DCTN1, FUS, GRN, HNRNPA2B1, MAPT, SQSTM1, TARDBP, TBK1, TREM2, UBQLN2, VCP
	○ 03505.1 Add-on preliminary-evidence genes	2	LRRK2, PSEN1
○ 03504	Invitae Hereditary Alzheimer's Disease Panel	3	APP, PSEN1, PSEN2
○ 03506	Invitae Hereditary Prion Disease Test	1	PRNP
Neuromuscular Disorders			
○ 03281	Invitae Congenital Myasthenic Syndrome Panel	21	AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, MUSK, PREPL, RAPSN, SLC18A3, SLC5A7, SYT2, VAMP1
	○ 03281.1 Add-on preliminary-evidence genes	4	LAMB2, LRP4, SCN4A, SNAP25
○ 03285	Invitae Malignant Hyperthermia Susceptibility Panel	3	CACNA1S, RYR1, STAC3
○ 03292	Invitae Congenital Muscular Dystrophy Panel	29	B3GALNT2, B4GAT1, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DMD, DPM1, DPM2, DPM3, FKRP, FKTN, GMPPB, GOSR2, ISPD, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, TCAP, TK2
○ 03373	Invitae Periodic Paralysis Panel	6	ATP1A2, CACNA1S, KCNJ2, MCM3AP, RYR1, SCN4A

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NEUROLOGY TEST CATALOG

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

Test code	Test name	# gene(s)	Gene list
Neuromuscular Disorders (continued)			
○ 03280	Invitae Comprehensive Neuromuscular Disorders Panel	131	ACTA1, ADSSL1, AGRN, ALG14, ALG2, AMPD1, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1S, CAPN3, CASQ1, CAV3, CCDC78, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL13A1, 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, GOSR2, GYG1, GYS1, HACD1, HNRNPA2B1, HNRNPDL, ISCU, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LDB3, LMNA, LMOD3, MAP3K20, MATR3, MEGF10, MICU1, MTM1, MUSK, MYH2, MYH7, MYL2, MYO18B, MYOT, MYPN, NEB, ORAI1, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, PYROXD1, RAPSN, RXYLT1, RYR1, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SLC18A3, SLC5A7, SMCHD1, SMN1, SMN2, SPEG, SQSTM1, STAC3, STIM1, SYT2, TAZ, TCAP, TIA1, TK2, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, VAMP1, VCP, VMA21
	○ 03280.1 Add-on preliminary-evidence genes	12	KLHL9, LAMB2, LIMS2, LRP4, MTMR14, SNAP25, SUN1, SUN2, SYNE1, SYNE2, TMEM43, TNNT3
○ 03291	Invitae Comprehensive Muscular Dystrophy Panel	52	ANO5, B3GALNT2, B4GAT1, CAPN3, CAV3, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, GOSR2, HNRNPDL, ISPD, ITGA7, LAMA2, LARGE1, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TK2, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
	○ 03291.1 Add-on preliminary-evidence genes	7	KBTBD13, LIMS2, SUN1, SUN2, SYNE1, SYNE2, TMEM43
○ 03304	Invitae Limb-Girdle Muscular Dystrophy Panel	37	ANO5, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DYSF, FKRP, FKTN, GAA, GMPPB, GOSR2, HNRNPDL, ISPD, LAMA2, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TK2, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
	○ 03304.1 Add-on preliminary-evidence genes	2	KBTBD13, LIMS2
○ 03301	Invitae Dystrophinopathies Test	1	DMD
○ 03361	Invitae Comprehensive Myopathy Panel	71	ACTA1, ADSSL1, AMPD1, ANO5, ATP2A1, BAG3, BIN1, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FKBP14, FLNC, GNE, GYG1, GYS1, HACD1, HNRNPA2B1, ISCU, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMP2, LDB3, LMNA, LMOD3, MAP3K20, MATR3, MEGF10, MICU1, MTM1, MYH2, MYH7, MYL2, MYO18B, MYOT, MYPN, NEB, ORAI1, PYROXD1, RYR1, SCN4A, SELENON, SPEG, SQSTM1, STAC3, STIM1, TAZ, TIA1, TK2, TNNT1, TPM2, TPM3, TTN, VCP, VMA21
	○ 03361.1 Add-on preliminary-evidence genes	2	KLHL9, MTMR14
○ 03362	Invitae Congenital Myopathy Panel	36	ACTA1, BIN1, CACNA1S, CCDC78, CFL2, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, DNM2, FKBP14, HACD1, KBTBD13, KLHL40, KLHL41, LMOD3, MAP3K20, MEGF10, MICU1, MTM1, MYH7, MYL2, MYO18B, MYPN, NEB, PYROXD1, RYR1, SELENON, SPEG, STAC3, TK2, TNNT1, TPM2, TPM3, TTN
	○ 03362.1 Add-on preliminary-evidence gene	1	MTMR14
○ 03375	Invitae Myotonia and Paramyotonia Congenita Panel	2	CLCN1, SCN4A
○ 03245	Invitae Spinal Muscular Atrophy Panel	2	SMN1, SMN2
○ 53699	Invitae Hereditary Rhabdomyolysis Panel	101	ACAD9, ACADM, ACADVL, AGL, ALDOA, AMACR, AMPD1, ANO5, ATP2A1, ATP7B, B3GALNT2, B4GAT1, CACNA1S, CAPN3, CASQ1, CAV3, CHKB, COQ8A, COQ9, CPT1A, CPT2, CTDP1, DAG1, DGUOK, DMD, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, ENO3, ETFA, ETFB, ETFDH, FDX2, FHL1, FKRP, FKTN, FLAD1, GAA, GBE1, GMPPB, GYG1, GYS1, HADH, HADHA, HADHB, HMBS, ISCU, ISPD, ITGA7, LAMA2, LAMP2, LARGE1, LDHA, LPIN1, OPA1, OPA3, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PYGM, RBCK1, RRM2B, RXYLT1, RYR1, SCN4A, SDHA, SGCA, SGCB, SGCD, SGCG, SIL1, SLC16A1, SLC22A5, SLC25A20, SLC25A32, STAC3, SUCLA2, SUCLG1, TANGO2, TCAP, TK2, TNPO3, TRIM32, TSFM, TWNK, TYMP

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NEUROLOGY TEST CATALOG

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

Test code	Test name	# gene(s)	Gene list
Neuropathies and Related Disorders			
○ 03200	Invitae Comprehensive Neuropathies Panel	101	AARS, AIFM1, APOA1, ASAH1, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CHCHD10, COX6A1, CYP27A1, CYP7B1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EXOSC9, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GLA, GNB4, GSN, HARS, HEXA, HINT1, HMBS, HSPB1, HSPB8, IGHMBP2, INF2, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, POLG, POLG2, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN11A, SCN9A, SEPT9, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SMN2, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, VRK1, WNK1, YARS
	○ 03200.1 Add-on preliminary-evidence genes	9	ARHGEF10, CCT5, HSPB3, LAS1L, MICAL1, SCN10A, SGPL1, SLC25A21, SLC52A1
○ 03201	Invitae Charcot-Marie-Tooth Disease Comprehensive Panel	57	AARS, AIFM1, ATP1A1, BAG3, BSCL2, COX6A1, DHTKD1, DNAJB2, DNM2, DRP2, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KIF5A, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NDRG1, NEFH, NEFL, PDK3, PLEKHG5, PMP2, PMP22, PRPS1, PRX, RAB7A, SBF1, SBF2, SH3TC2, SLC25A46, SPG11, SURF1, TFG, TRIM2, TRPV4, YARS
	○ 03201.1 Add-on preliminary-evidence genes	3	ARHGEF10, MICAL1, SGPL1
○ 03230	Invitae Hereditary Sensory and Autonomic Neuropathy Panel	15	ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN11A, SCN9A, SPTLC1, SPTLC2, WNK1
	○ 03230.1 Add-on preliminary-evidence genes	1	CCT5
○ 03461	Invitae Familial Dysautonomia Test	1	ELP1
○ 03240	Invitae Hereditary Motor Neuropathy Panel	27	ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC9, FBXO38, GARS, HEXA, HINT1, HSPB1, HSPB8, IGHMBP2, MORC2, PLEKHG5, REEP1, SIGMAR1, SLC5A7, SMN1, SMN2, TRPV4, UBA1, VAPB, VRK1
	○ 03240.1 Add-on preliminary-evidence gene	3	HSPB3, LAS1L, SLC25A21
○ 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
○ 03251	Invitae Hereditary Spastic Paraplegia Comprehensive Panel	62	ABCD1, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12orf65, CAPN1, CPT1C, CYP27A1, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HACE1, HEXA, HSPD1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, NIPA1, NKX6-2, NT5C2, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, UCHL1, VAMP1, WASHC5, ZFYVE26
	○ 03251.1 Add-on preliminary-evidence genes	16	ADGRB2, AMPD2, ARSI, ATP2B4, C19orf12, CCT5, DSTYK, EXOSC3, IBA57, KLC2, PGAP1, SLC33A1, USP8, VPS37A, ZFR, ZFYVE27

CARDIOLOGY AND NEUROLOGY INDIVIDUAL GENES

○ AARS	○ ADSSL1	○ AMACR	○ AP4M1	○ ASAH1	○ ATP7A	○ BSCL2	○ CBS	○ CHMP2B
○ ABCD1	○ AGL	○ AMPD1	○ AP4S1	○ ATL1	○ ATP7B	○ C12orf65	○ CCDC78	○ CHRNA1
○ ACAD9	○ AGRN	○ AMPD2	○ AP5Z1	○ ATL3	○ B3GALNT2	○ C19orf12	○ CCER2	○ CHRNB1
○ ACADM	○ AIFM1	○ ANG	○ APOA1	○ ATP13A2	○ B4GALNT1	○ CACNA1A	○ CCT5	○ CHRND
○ ACADVL	○ ALDH18A1	○ ANO3	○ APP	○ ATP1A1	○ B4GAT1	○ CACNA1S	○ CFL2	○ CHRNE
○ ACTA1	○ ALDOA	○ ANO5	○ ARG1	○ ATP1A2	○ BAG3	○ CAPN1	○ CHAT	○ CIZ1
○ ACTB	○ ALG14	○ ANXA11	○ ARHGEF10	○ ATP1A3	○ BCAP31	○ CAPN3	○ CHCHD10	○ CLCN1
○ ADCY5	○ ALG2	○ AP4B1	○ ARL6IP1	○ ATP2A1	○ BICD2	○ CASQ1	○ CHCHD2	○ CNTN1
○ ADGRB2	○ ALS2	○ AP4E1	○ ARSI	○ ATP2B4	○ BIN1	○ CAV3	○ CHKB	○ COL12A1

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NEUROLOGY TEST CATALOG

CLINICAL AREA: CARDIOLOGY AND NEUROLOGY

CARDIOLOGY AND NEUROLOGY INDIVIDUAL GENES (continued)

○ COL13A1	○ DYNC1H1	○ GLA	○ KDM5C	○ MTM1	○ PMP2	○ SCN11A	○ SORL1	○ TRAPPC11
○ COL4A1	○ DYSF	○ GMPPB	○ KIDINS220	○ MTMR14	○ PMP22	○ SCN1A	○ SPART	○ TREM2
○ COL4A2	○ EGR2	○ GNAL	○ KIF1A	○ MTMR2	○ PNKD	○ SCN4A	○ SPAST	○ TREX1
○ COL6A1	○ ELP1	○ GNAO1	○ KIF1C	○ MUSK	○ PNPLA2	○ SCN9A	○ SPEG	○ TRIM2
○ COL6A2	○ EMD	○ GNB4	○ KIF5A	○ MYH2	○ PNPLA6	○ SDHA	○ SPG11	○ TRIM32
○ COL6A3	○ ENO3	○ GNE	○ KLC2	○ MYH7	○ PODXL	○ SELENON	○ SPG21	○ TRPV4
○ COLQ	○ ENTPD1	○ GOSR2	○ KLHL40	○ MYL2	○ POLG	○ SEPT9	○ SPG7	○ TSFM
○ COQ8A	○ ERBB4	○ GRN	○ KLHL41	○ MYO18B	○ POLG2	○ SETX	○ SPR	○ TTN
○ COQ9	○ ERLIN1	○ GSN	○ KLHL9	○ MYOT	○ POMGNT1	○ SGCA	○ SPTLC1	○ TTR
○ COX6A1	○ ERLIN2	○ GUCY1A1	○ KMT2B	○ MYPN	○ POMGNT2	○ SGCB	○ SPTLC2	○ TUBB4A
○ CPT1A	○ ETFA	○ GYG1	○ L1CAM	○ NDRG1	○ POMK	○ SGCD	○ SQSTM1	○ TWNK
○ CPT1C	○ ETFB	○ GYS1	○ LAMA2	○ NEB	○ POMT1	○ SGCE	○ STAC3	○ TYMP
○ CPT2	○ ETFDH	○ HACD1	○ LAMB2	○ NEFH	○ POMT2	○ SGCG	○ STIM1	○ UBA1
○ CRYAB	○ EXOSC3	○ HACE1	○ LAMP2	○ NEFL	○ PRDM12	○ SGPL1	○ SUCLA2	○ UBQLN2
○ CSF1R	○ EXOSC9	○ HADH	○ LARGE1	○ NGF	○ PREPL	○ SH3TC2	○ SUCLG1	○ UCHL1
○ CST3	○ FA2H	○ HADHA	○ LAS1L	○ NIPA1	○ PRKN	○ SIGMAR1	○ SUN1	○ USP8
○ CTDP1	○ FARS2	○ HADHB	○ LDB3	○ NKX6-2	○ PRKRA	○ SIL1	○ SUN2	○ VAC14
○ CYP27A1	○ FBLN5	○ HARS	○ LDHA	○ NOTCH3	○ PRNP	○ SLC12A6	○ SURF1	○ VAMP1
○ CYP2U1	○ FBXO38	○ HEXA	○ LIMS2	○ NT5C2	○ PRPS1	○ SLC16A1	○ SYNE1	○ VAPB
○ CYP7B1	○ FBXO7	○ HINT1	○ LITAF	○ NTRK1	○ PRRT2	○ SLC16A2	○ SYNE2	○ VCP
○ DAG1	○ FDX2	○ HMBS	○ LMNA	○ OPA1	○ PRX	○ SLC18A3	○ SYNJ1	○ VMA21
○ DCTN1	○ FGD4	○ HNRNPA2B1	○ LMOD3	○ OPA3	○ PSEN1	○ SLC1A3	○ SYT2	○ VPS13A
○ DDHD1	○ FHL1	○ HNRNPDL	○ LPIN1	○ OPTN	○ PSEN2	○ SLC22A5	○ TANGO2	○ VPS13C
○ DDHD2	○ FIG4	○ HPCA	○ LRP4	○ ORAI1	○ PYGM	○ SLC25A20	○ TARDBP	○ VPS13D
○ DES	○ FKBP14	○ HSPB1	○ LRRK2	○ PANK2	○ PYROXD1	○ SLC25A21	○ TAZ	○ VPS35
○ DGUOK	○ FKRP	○ HSPB3	○ LRSAM1	○ PARK7	○ RAB39B	○ SLC25A32	○ TBK1	○ VPS37A
○ DHTKD1	○ FKTN	○ HSPB8	○ MAG	○ PDE8B	○ RAB3GAP2	○ SLC25A46	○ TCAP	○ VRK1
○ DMD	○ FLAD1	○ HSPD1	○ MAP3K20	○ PDK3	○ RAB7A	○ SLC2A1	○ TECPR2	○ WASHC5
○ DNAJB2	○ FLNC	○ HTRA1	○ MAPT	○ PDSS2	○ RAPSN	○ SLC30A10	○ TFG	○ WNK1
○ DNAJB6	○ FOXC1	○ IBA57	○ MARS	○ PFKM	○ RBCK1	○ SLC33A1	○ TH	○ XPR1
○ DNAJC6	○ FUS	○ IGHMBP2	○ MATR3	○ PFN1	○ REEP1	○ SLC39A14	○ THAP1	○ YARS
○ DNM2	○ GAA	○ INF2	○ MCM3AP	○ PGAM2	○ REEP2	○ SLC52A1	○ TIA1	○ ZFR
○ DNMT1	○ GAN	○ ISCU	○ MECR	○ PGAP1	○ RETREG1	○ SLC52A2	○ TK2	○ ZFYVE26
○ DOK7	○ GARS	○ ISPD	○ MED20	○ PGK1	○ RNF213	○ SLC52A3	○ TMEM230	○ ZFYVE27
○ DPAGT1	○ GBA	○ ITGA7	○ MED25	○ PGM1	○ RRM2B	○ SLC5A7	○ TMEM43	
○ DPM1	○ GBA2	○ ITM2B	○ MEGF10	○ PHKA1	○ RTN2	○ SLC6A3	○ TNNT1	
○ DPM2	○ GBE1	○ KBTBD13	○ MFN2	○ PHKB	○ RXYLT1	○ SMCHD1	○ TNNT3	
○ DPM3	○ GCH1	○ KCNA2	○ MICAL1	○ PINK1	○ RYR1	○ SMN1	○ TNPO3	
○ DRD2	○ GDAP1	○ KCNJ2	○ MICU1	○ PLA2G6	○ SACS	○ SMN2	○ TOR1A	
○ DRP2	○ GFPT1	○ KCNK18	○ MME	○ PLEC	○ SBF1	○ SNAP25	○ TOR1AIP1	
○ DST	○ GJB1	○ KCNMA1	○ MORC2	○ PLEKHG5	○ SBF2	○ SNCA	○ TPM2	
○ DSTYK	○ GJC2	○ KCTD17	○ MPZ	○ PLP1	○ SCN10A	○ SOD1	○ TPM3	

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NEUROLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Developmental Brain Abnormalities			
○ 55002	Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	446	AARS, AARS2, ABAT, ABCA1, ABCD1, ACADS, ACER3, ACO2, ACOX1, ACP5, ACY1, ADAR, ADGRG1, ADK, ADSL, AGA, AHDC1, AIFM1, AIMP1, AIMP2, ALDH3A2, ALDH5A1, ALDH6A1, ALG2, AMACR, AMPD2, ANK3, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, APP, ARCN1, ARHGAP31, ARNT2, ARSA, ARX, ASNS, ASPA, ASXL1, ASXL2, ATP13A2, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, AUH, B3GALNT2, BCAP31, BCL11B, BCS1L, BMP4, BOLA3, BRAT1, C12orf65, C19orf12, CACNA1A, CARS2, CCDC88A, CHMP2B, CLCN2, CLCN7, CLN6, CLP1, CLPP, CNTNAP1, COASY, COL4A1, COL4A2, COQ2, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CP, CPLX1, CPS1, CRAT, CSF1R, CTBP1, CTC1, CTDP1, CTNS, CTSA, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DDC, DDHD2, DDOST, DEAF1, DEGS1, DGUOK, DHFR, DLL4, DNM1L, DNM2, DOCK6, DPYS, DYRK1A, EARS2, EDNRB, EGR2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ENTPD1, EPG5, EPRS, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAM126A, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGD4, FGFRL1, FH, FIG4, FKRP, FKTN, FOLR1, FOXC1, FOXG1, FOXRED1, FTL, FUCA1, GAA, GALC, GALT, GAN, GBE1, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GNAO1, GRM7, GRN, GTF2H5, GTPBP2, HEPACAM, HEXA, HIBCH, HIKESHI, HK1, HMGCL, HSD17B4, HSPD1, HTRA1, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, ISCA1, ISCA2, ITPA, JAM3, KARS, KCNJ10, KCNT1, KIAA1161, KIF1A, KIF5A, L2HGDH, LAMA1, LAMA2, LAMB1, LARGE1, LETM1, LIAS, LIPT1, LIPT2, LMNB1, LONP1, LRPPRC, LYRM7, MAG, MAN2B1, MAPT, MARS2, MAT1A, MCOLN1, MEF2C, MGP, MLC1, MLYCD, MOCS1, MOCS2, MPLKIP, MPV17, MPZ, MRPL44, MRPS16, MRPS22, MTFMT, MTHFR, MTR, MTRR, MUT, NADK2, NAXD, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NGLY1, NKX6-2, NOTCH1, NOTCH3, NPC1, NPC2, NRXN1, NUBPL, NUP62, OCRL, OSGEP, OTC, PAFAH1B1, PAH, PANK2, PARS2, PC, PCDH12, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHGDH, PHYH, PIGA, PINK1, PLA2G6, PLEKHG2, PLP1, PMP22, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPP2R1A, PPT1, PRF1, PRKDC, PRNP, PRPS1, PSAP, PSAT1, PSEN1, PTEN, PURA, PUS3, PYCR2, QARS, RARS, RBPJ, REPS1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RPIA, RPS6KC1, RRM2B, SAMHD1, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SH3TC2, SHPK, SLC13A3, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A4, SLC20A2, SLC25A1, SLC25A12, SLC25A22, SLC25A4, SLC25A42, SLC33A1, SLC35A2, SLC46A1, SLC6A8, SLC6A9, SNIP1, SNORD118, SNRPB, SON, SOX10, SPART, SPAST, SPATA5, SPG11, SPG7, SPTAN1, SQSTM1, SSR4, STAMBP, STAT1, STN1, STX11, STXBP1, STXBP2, SUCLA2, SUMF1, SURF1, SYNE1, TACO1, TAF2, TARS2, TBC1D24, TBCD, TBCK, TIMM50, TM4SF20, TMEM106B, TMEM126B, TMEM165, TMEM70, TMTC3, TPI1, TRAPPC11, TRAPPC9, TREM2, TREX1, TRMT10A, TRMT5, TSC1, TSEN54, TTC19, TUBB2A, TUBB4A, TUFM, TWNK, TYMP, TYROBP, UBE2A, UFM1, UGT1A1, UNC13D, UPB1, VARS2, VCP, VPS11, VPS33A, WARS2, WDR45, WHSC1, WWOX, ZEB2, ZFYVE26, ZNF335
○ 55006	Invitae Brain Malformations Panel	134	ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARID1A, ARID1B, ARX, ASNS, ASPM, ATP6V0A2, B3GALNT2, B4GAT1, BMP4, C19orf12, CASK, CCM2, CCND2, CDK5, CDON, CHMP1A, COASY, COL18A1, COL3A1, COL4A1, COL4A2, CP, CRADD, DAG1, DCHS1, DCX, DIAPH1, DISP1, DLL1, DMXL2, DPF2, DYNC1H1, EMC1, ERMARD, EXOSC3, FA2H, FAT4, FGFR1, FIG4, FKRP, FKTN, FLNA, FOXA2, FTL, GAS1, GLI2, GMPPB, GPSM2, GRIN2B, IER3IP1, ISPD, KATNB1, KCNMA1, KIF11, KIF1BP, KIF2A, KRIT1, LAMA1, LAMB1, LAMC3, LARGE1, LRP2, MED17, MFSD2A, MRE11, NDE1, NEDD4L, NPRL3, OCLN, OPHN1, PAFAH1B1, PANK2, PDCD10, PHGDH, PIK3R2, PLA2G6, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PTCH1, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RARS2, RELN, RERE, RTTN, RXYLT1, SEPSECS, SHH, SIX3, SLC25A19, SMARCA4, SMARCB1, SMARCE1, SMC1A, SNAP29, SON, SOX2, SRD5A3, STAG2, STAMBP, STIL, TBC1D20, TGIF1, TMTC3, TOE1, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, VLDLR, VRK1, WDR45, WDR62, YWHAE, ZIC2

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NEUROLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Developmental Brain Abnormalities (continued)			
○ 55004	Invitae Cerebral Palsy Spectrum Disorders Panel	265	ABAT, ACADM, ACADVL, ACAT1, ACBD5, ACOX1, ADAR, ADCY5, ADD3, ADSL, AHI1, AKT3, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, ALS2, AMACR, AMPD2, AMT, AP4B1, AP4E1, AP4M1, AP4S1, APTX, ARG1, ARHGEF9, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ATAD1, ATM, ATP13A2, ATP1A3, ATP7A, ATP7B, ATP8A2, AUH, BCKDHA, BCKDHB, BTD, C19orf12, CACNA1A, CBS, CDKL5, CEP290, CHRNA1, CLN2 (TPP1), CLN3, COASY, COL4A1, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ9, CP, CPS1, CTNNB1, CTSB, CYP27A1, CYP2U1, DBH, DBT, DCAF17, DDC, DGKZ, DHFR, DLAT, DLD, DMD, DNAJC12, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPHA4, ETFA, ETFB, ETFDH, ETHE1, FAM126A, FH, FOLR1, FOXG1, FRRS1L, FTL, FUCA1, GABRA2, GAD1, GALC, GAMT, GATM, GCDH, GCH1, GFAP, GJC2, GLB1, GLDC, GLRA1, GLRB, GM2A, GNAO1, GNB1, GPHN, GPR88, GRIN1, HESX1, HEXA, HLCS, HMGCL, HPRT1, HSD17B10, HSD17B4, IFIH1, ITPA, ITPR1, KANK1, KCNC3, KCNJ6, KDM5C, KIDINS220, KIF1A, KMT2C, L1CAM, L2HGDH, LAMA2, LIAS, LMBRD1, MAOA, MCCC1, MCCC2, MCEE, MECP2, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOCS3, MPC1, MTHFR, MTOR, MTR, MTRR, MTTP, MUT, NAA10, NAGS, NBAS, NGLY1, NIPA1, NKX2-1, NPC1, NPC2, NPHP1, OTC, PAFAH1B1, PAH, PAK3, PALM, PANK2, PCBD1, PCCA, PCCB, PCDH12, PDE10A, PDE2A, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PIK3CA, PLA2G6, PLP1, PLXNA2, PNP, PNPO, POLG, POLR3A, PPT1, PROSC, PSAT1, PSPH, PTS, QDPR, RANBP2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCN1A, SCN2A, SCN3A, SCN8A, SETD5, SHH, SIX3, SLC16A2, SLC17A5, SLC18A2, SLC19A3, SLC1A4, SLC25A15, SLC25A22, SLC2A1, SLC30A10, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SPAST, SPG11, SPR, SPTBN2, SQSTM1, ST3GAL5, STXBP1, SUCLA2, SUCLG1, SUOX, SURF1, TBC1D24, TBCK, TBL1XR1, TCF4, TGIF1, TH, TMEM67, TREX1, TSEN54, TTPA, TUBA1A, TUBB4A, UBE3A, WDR45, WDR62, ZFYVE26, ZIC1, ZIC2
○ 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
○ 03402	Invitae Early Infantile Epileptic Encephalopathy Panel	59	ALDH7A1, ARHGEF9, ARX, BRAT1, CACNA2D2, CASK, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, FARS2, FOLR1, FRRS1L, GABBR2, GABRA1, GABRB3, GNAO1, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, IER3IP1, KCNA2, KCNB1, KCNMA1, KCNQ2, KCNQ3, KCNT1, PCDH19, PIGA, PIGN, PIGO, PLCB1, PNKP, PNPO, PURA, SCN1A, SCN2A, SCN8A, SCN9A, SIK1, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SMC1A, SPTAN1, STXBP1, SYNGAP1, SZT2, TBC1D24, WDR45, WWOX
	○ 03402.1 Add-on preliminary-evidence genes	10	ARHGEF15, ATP1A2, COQ4, GPHN, KCNH5, MTOR, NECAP1, NEDD4L, SCN1B, ST3GAL3
○ 04424	Invitae Holoprosencephaly Panel	6	FGFR1, GLI2, SHH, SIX3, TGIF1, ZIC2
	○ 04424.1 Add-on preliminary-evidence genes	4	CDON, FOXH1, NODAL, PTCH1
○ 03404	Invitae Rett and Angelman Syndromes and Related Disorders Panel	24	ADSL, ALDH5A1, ATRX, CDKL5, CNTNAP2, DYRK1A, EHMT1, FOXG1, GABBR2, IQSEC2, KANSL1, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN8A, SLC9A6, STXBP1, TCF4, UBE3A, WDR45, ZEB2
	○ 03404.1 Add-on preliminary-evidence genes	4	GABRD, HDAC8, JMJD1C, TBL1XR1
○ 01721	Invitae Tuberous Sclerosis Complex Panel	2	TSC1, TSC2

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NEUROLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

Test code	Test name	# gene(s)	Gene list
Developmental Brain Abnormalities (continued)			
○ 03401	Invitae Epilepsy Panel	304	AARS, ABAT, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG1, ALG12, ALG13, ALG6, AMACR, AMT, AP2M1, AP3B2, ARG1, ARHGEF15, ARHGEF9, ARSA, ARX, ASAH1, ASNS, ATAD1, ATP1A2, ATP1A3, ATP6AP2, ATRX, BRAT1, C12orf57, CACNA1A, CACNA1E, CACNA1H, CACNA2D2, CAD, CAMK2B, CARS2, CASK, CCDC88A, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CLCN6, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLTC, CNTN2, CNTNAP2, COG5, COL18A1, CSTB, CTNNB1, CTSD, CYFIP2, CYP27A1, DDC, DDX3X, DEAF1, DEPDC5, DHDDS, DHFR, DIAPH1, DNAJC5, DNM1, DNM1L, DOCK7, DYNC1H1, DYRK1A, ECHS1, EEF1A2, EHMT1, EMC1, EPM2A, FAR1, FARS2, FASN, FBXO11, FGF12, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GAMT, GATAD2B, GATM, GCH1, GLDC, GLRA1, GLRB, GNAO1, GNB1, GOSR2, GPAA1, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, GRIN2D, GTPBP3, GUF1, HCN1, HEXA, HNRNPU, HTT, IDH3A, IER3IP1, IFIH1, IQSEC2, ITPA, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCND2, KCNH1, KCNH2, KCNH5, KCNJ10, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD7, KIF1A, KIF2A, KIF5A, KPNA7, LAMC3, LGI1, LIAS, LMNB2, MBD5, MDH2, MECP2, MEF2C, MFSD8, MOCS1, MOCS2, MOCS3, MTOR, NACC1, NAGLU, NECAP1, NEDD4L, NEXMIF, NGLY1, NHLRC1, NPC1, NPC2, NPRL3, NRXN1, NTRK2, NUS1, PACS1, PACS2, PAFAH1B1, PCDH19, PCLO, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PIGA, PIGG, PIGN, PIGO, PIGP, PIGQ, PIGV, PIGW, PIK3AP1, PLAA, PLCB1, PNKD, PNKP, PNPO, PNPT1, POLG, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRIMA1, PRRT2, PSAP, PSAT1, PSPH, PTPN23, PURA, QARS, QDPR, RAB11A, RAB11B, RAI1, RALA, RANBP2, RBFOX1, RBFOX3, RELN, RFT1, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGDI, RORB, RUSC2, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SCP2, SERPINI1, SETBP1, SGCE, SGSH, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMC1A, SNAP25, SNIP1, SNX27, SPATA5, SPTAN1, ST3GAL3, ST3GAL5, STAG2, STRADA, STX1B, STXBP1, STXBP2, SUMF1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TANGO2, TBC1D24, TBCK, TBL1XR1, TCF4, TH, TK2, TPK1, TREX1, TSC1, TSC2, TSFM, TUBA8, TUBB2A, UBA5, UBE3A, UNC80, WDR45, WWOX, YWHAG, ZDHHC9, ZEB2, ZSWIM6
○ 03401.1	Add-on FLNA gene	1	FLNA
○ 03401.2	Add-on PTEN genes	1	PTEN

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES

○ AARS	○ ADAR	○ ALG1	○ APOPT1	○ ASPA	○ ATRX	○ CACNA1E	○ CHMP1A	○ CLTC
○ AARS2	○ ADCY5	○ ALG12	○ APP	○ ASPM	○ AUH	○ CACNA1H	○ CHMP2B	○ CNTN2
○ ABAT	○ ADD3	○ ALG13	○ APTX	○ ASS1	○ B3GALNT2	○ CACNA2D2	○ CHRNA1	○ CNTNAP1
○ ABCA1	○ ADGRG1	○ ALG2	○ ARCN1	○ ASXL1	○ B4GAT1	○ CAD	○ CHRNA2	○ CNTNAP2
○ ABCD1	○ ADK	○ ALG6	○ ARFGEF2	○ ASXL2	○ BCAP31	○ CAMK2B	○ CHRNA4	○ COASY
○ ACADM	○ ADSL	○ ALS2	○ ARG1	○ ATAD1	○ BCKDHA	○ CARS2	○ CHRNB2	○ COG5
○ ACADS	○ AGA	○ AMACR	○ ARHGAP31	○ ATM	○ BCKDHB	○ CASK	○ CLCN2	○ COL18A1
○ ACADVL	○ AHDC1	○ AMPD2	○ ARHGEF15	○ ATP13A2	○ BCL11B	○ CBS	○ CLCN4	○ COL3A1
○ ACAT1	○ AHI1	○ AMT	○ ARHGEF9	○ ATP1A2	○ BCS1L	○ CCDC88A	○ CLCN6	○ COL4A1
○ ACBD5	○ AIFM1	○ ANK3	○ ARID1A	○ ATP1A3	○ BMP4	○ CCM2	○ CLCN7	○ COL4A2
○ ACER3	○ AIMP1	○ AP1S2	○ ARID1B	○ ATP6AP2	○ BOLA3	○ CCND2	○ CLN2 (TPP1)	○ COQ2
○ ACO2	○ AIMP2	○ AP2M1	○ ARNT2	○ ATP6V0A2	○ BRAT1	○ CDK5	○ CLN3	○ COQ4
○ ACOX1	○ AKT3	○ AP3B2	○ ARSA	○ ATP7A	○ BTD	○ CDKL5	○ CLN5	○ COQ6
○ ACP5	○ ALDH3A2	○ AP4B1	○ ARX	○ ATP7B	○ C12orf57	○ CDON	○ CLN6	○ COQ7
○ ACTB	○ ALDH5A1	○ AP4E1	○ ASAH1	○ ATP8A2	○ C12orf65	○ CEP290	○ CLN8	○ COQ8A
○ ACTG1	○ ALDH6A1	○ AP4M1	○ ASL	○ ATPAF2	○ C19orf12	○ CERS1	○ CLP1	○ COQ9
○ ACY1	○ ALDH7A1	○ AP4S1	○ ASNS	○ ATRN	○ CACNA1A	○ CHD2	○ CLPP	○ COX10

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NEUROLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES (continued)

○ COX14	○ DEPDC5	○ EPM2A	○ FUCA1	○ GNB1	○ IFIH1	○ KPNA7	○ MLYCD	○ NDUFAF2
○ COX15	○ DGKZ	○ EPRS	○ GAA	○ GOSR2	○ IQSEC2	○ KRIT1	○ MMAA	○ NDUFAF3
○ COX20	○ DGUOK	○ ERCC2	○ GABBR2	○ GPAA1	○ ISCA1	○ L1CAM	○ MMAB	○ NDUFAF4
○ COX6B1	○ DHDDS	○ ERCC3	○ GABRA1	○ GPHN	○ ISCA2	○ L2HGDH	○ MMACHC	○ NDUFAF5
○ COX7B	○ DHFR	○ ERCC6	○ GABRA2	○ GPR88	○ ISPD	○ LAMA1	○ MMADHC	○ NDUFAF6
○ COX8A	○ DIAPH1	○ ERCC8	○ GABRB1	○ GPSM2	○ ITPA	○ LAMA2	○ MOCS1	○ NDUFB3
○ CP	○ DISP1	○ ERMARD	○ GABRB2	○ GRIA3	○ ITPR1	○ LAMB1	○ MOCS2	○ NDUFB8
○ CPLX1	○ DLAT	○ ETFA	○ GABRB3	○ GRIN1	○ JAM3	○ LAMC3	○ MOCS3	○ NDUFB9
○ CPS1	○ DLD	○ ETFB	○ GABRD	○ GRIN2A	○ JMJD1C	○ LARGE1	○ MPC1	○ NDUFS1
○ CRADD	○ DLL1	○ ETFDH	○ GABRG2	○ GRIN2B	○ KANK1	○ LETM1	○ MPLKIP	○ NDUFS2
○ CRAT	○ DLL4	○ ETHE1	○ GAD1	○ GRIN2D	○ KANSL1	○ LGI1	○ MPV17	○ NDUFS3
○ CSF1R	○ DMD	○ EXOSC3	○ GALC	○ GRM7	○ KARS	○ LIAS	○ MPZ	○ NDUFS4
○ CSTB	○ DMXL2	○ FA2H	○ GALT	○ GRN	○ KATNB1	○ LIPT1	○ MRE11	○ NDUFS6
○ CTBP1	○ DNAJC12	○ FAM126A	○ GAMT	○ GTF2H5	○ KCNA1	○ LIPT2	○ MRPL44	○ NDUFS7
○ CTC1	○ DNAJC5	○ FAR1	○ GAN	○ GTPBP2	○ KCNA2	○ LMBRD1	○ MRPS16	○ NDUFS8
○ CTDP1	○ DNM1	○ FARS2	○ GAS1	○ GTPBP3	○ KCNB1	○ LMNB1	○ MRPS22	○ NDUFV1
○ CTNNB1	○ DNM1L	○ FARSB	○ GATAD2B	○ GUF1	○ KCNC1	○ LMNB2	○ MTFMT	○ NDUFV2
○ CTNS	○ DNM2	○ FASN	○ GATM	○ HCN1	○ KCNC3	○ LONP1	○ MTHFR	○ NECAP1
○ CTSA	○ DOCK6	○ FASTKD2	○ GBE1	○ HDAC8	○ KCND2	○ LRP2	○ MTOR	○ NEDD4L
○ CTSB	○ DOCK7	○ FAT4	○ GCDH	○ HEPACAM	○ KCNH1	○ LRPPRC	○ MTR	○ NEXMIF
○ CTSD	○ DPF2	○ FBXL4	○ GCH1	○ HESX1	○ KCNH2	○ LYRM7	○ MTRR	○ NFU1
○ CYFIP2	○ DPYS	○ FBXO11	○ GDAP1	○ HEXA	○ KCNH5	○ MAG	○ MTTP	○ NGLY1
○ CYP27A1	○ DYNC1H1	○ FDX2	○ GFAP	○ HIBCH	○ KCNJ10	○ MAN2B1	○ MUT	○ NHLRC1
○ CYP2U1	○ DYRK1A	○ FGD4	○ GFM1	○ HIKESHI	○ KCNJ6	○ MAOA	○ NAA10	○ NIPA1
○ CYP7B1	○ EARS2	○ FGF12	○ GFM2	○ HK1	○ KCNK4	○ MAPT	○ NACC1	○ NKX2-1
○ D2HGDH	○ ECHS1	○ FGFR1	○ GJA1	○ HLCS	○ KCNMA1	○ MARS2	○ NADK2	○ NKX6-2
○ DAG1	○ EDNRB	○ FGFRL1	○ GJB1	○ HMGCL	○ KCNQ2	○ MAT1A	○ NAGLU	○ NODAL
○ DARS	○ EEF1A2	○ FH	○ GJC2	○ HNRNPU	○ KCNQ3	○ MBD5	○ NAGS	○ NOTCH1
○ DARS2	○ EGR2	○ FIG4	○ GLA	○ HPRT1	○ KCNQ5	○ MCCC1	○ NAXD	○ NOTCH3
○ DBH	○ EHMT1	○ FKRP	○ GLB1	○ HSD17B10	○ KCNT1	○ MCCC2	○ NAXE	○ NPC1
○ DBT	○ EIF2B1	○ FKTN	○ GLDC	○ HSD17B4	○ KCTD7	○ MCEE	○ NBAS	○ NPC2
○ DCAF17	○ EIF2B2	○ FLNA	○ GLI2	○ HSPD1	○ KDM5C	○ MCOLN1	○ NDE1	○ NPHP1
○ DCHS1	○ EIF2B3	○ FOLR1	○ GLRA1	○ HTRA1	○ KIAA1161	○ MDH2	○ NDRG1	○ NPRL3
○ DCX	○ EIF2B4	○ FOXA2	○ GLRB	○ HTT	○ KIDINS220	○ MECP2	○ NDUFA1	○ NRXN1
○ DDC	○ EIF2B5	○ FOXC1	○ GLRX5	○ IBA57	○ KIF11	○ MED17	○ NDUFA10	○ NTRK2
○ DDHD2	○ ELOVL4	○ FOXG1	○ GLUL	○ IDH2	○ KIF1A	○ MEF2C	○ NDUFA11	○ NUBPL
○ DDOST	○ EMC1	○ FOXH1	○ GLYCTK	○ IDH3A	○ KIF1BP	○ MFSD2A	○ NDUFA12	○ NUP62
○ DDX3X	○ ENTPD1	○ FOXRED1	○ GM2A	○ IDS	○ KIF2A	○ MFSD8	○ NDUFA2	○ NUS1
○ DEAF1	○ EPG5	○ FRRS1L	○ GMPPB	○ IDUA	○ KIF5A	○ MGP	○ NDUFA9	○ OCLN
○ DEGS1	○ EPHA4	○ FTL	○ GNAO1	○ IER3IP1	○ KMT2C	○ MLC1	○ NDUFAF1	○ OCRL

*If an order is placed using an outdated test requisition form, Invitae reserves the right to upgrade ordered tests to their current versions. View current requisition forms online at **www.invitae.com/forms** or consider placing your order online in the Invitae portal. Please note: Test IDs containing add-on codes will include the original panel as well as the add-on.*

NEUROLOGY TEST CATALOG

CLINICAL AREA: PEDIATRIC AND RARE DISEASE

PEDIATRIC AND RARE DISEASE INDIVIDUAL GENES (continued)

○ OPHN1	○ PEX2	○ POLR1C	○ RAB11B	○ SCN1B	○ SLC25A15	○ SPTBN2	○ TH	○ TYMP
○ OSGEP	○ PEX26	○ POLR3A	○ RAB18	○ SCN2A	○ SLC25A19	○ SQSTM1	○ TIMM50	○ TYROBP
○ OTC	○ PEX3	○ POLR3B	○ RAB3GAP1	○ SCN3A	○ SLC25A22	○ SRD5A3	○ TK2	○ UBA5
○ PACS1	○ PEX5	○ POMGNT1	○ RAB3GAP2	○ SCN5A	○ SLC25A4	○ SSR4	○ TM4SF20	○ UBE2A
○ PACS2	○ PEX6	○ POMGNT2	○ RAD21	○ SCN8A	○ SLC25A42	○ ST3GAL3	○ TMEM106B	○ UBE3A
○ PAFAH1B1	○ PEX7	○ POMK	○ RAI1	○ SCN9A	○ SLC2A1	○ ST3GAL5	○ TMEM126B	○ UFM1
○ PAH	○ PGAP1	○ POMT1	○ RALA	○ SCO1	○ SLC30A10	○ STAG2	○ TMEM165	○ UGT1A1
○ PAK3	○ PGK1	○ POMT2	○ RANBP2	○ SCO2	○ SLC33A1	○ STAMBP	○ TMEM67	○ UNC13D
○ PALM	○ PHGDH	○ PPP2CA	○ RARS	○ SCP2	○ SLC35A2	○ STAT1	○ TMEM70	○ UNC80
○ PANK2	○ PHYH	○ PPP2R1A	○ RARS2	○ SDHA	○ SLC46A1	○ STIL	○ TMTC3	○ UPB1
○ PARS2	○ PIGA	○ PPP2R5D	○ RBFOX1	○ SDHAF1	○ SLC6A1	○ STN1	○ TOE1	○ VARS2
○ PC	○ PIGG	○ PPP3CA	○ RBFOX3	○ SDHB	○ SLC6A19	○ STRADA	○ TPI1	○ VCP
○ PCBD1	○ PIGN	○ PPT1	○ RBPJ	○ SDHD	○ SLC6A3	○ STX11	○ TPK1	○ VLDLR
○ PCCA	○ PIGO	○ PRDM8	○ RELN	○ SEPSECS	○ SLC6A5	○ STX1B	○ TRAPPC11	○ VPS11
○ PCCB	○ PIGP	○ PRF1	○ REPS1	○ SERAC1	○ SLC6A8	○ STXBP1	○ TRAPPC9	○ VPS33A
○ PCDH12	○ PIGQ	○ PRICKLE1	○ RERE	○ SERPINI1	○ SLC6A9	○ STXBP2	○ TREM2	○ VRK1
○ PCDH19	○ PIGV	○ PRICKLE2	○ RFT1	○ SETBP1	○ SLC9A6	○ SUCLA2	○ TREX1	○ WARS2
○ PCLO	○ PIGW	○ PRIMA1	○ RHOBTB2	○ SETD5	○ SMARCA4	○ SUCLG1	○ TRMT10A	○ WDR45
○ PDCD10	○ PIK3AP1	○ PRKDC	○ RMND1	○ SGCE	○ SMARCB1	○ SUMF1	○ TRMT5	○ WDR62
○ PDE10A	○ PIK3CA	○ PRNP	○ RNASEH2A	○ SGSH	○ SMARCE1	○ SUOX	○ TSC1	○ WHSC1
○ PDE2A	○ PIK3R2	○ PROSC	○ RNASEH2B	○ SH3TC2	○ SMC1A	○ SURF1	○ TSC2	○ WWOX
○ PDHA1	○ PINK1	○ PRPS1	○ RNASEH2C	○ SHH	○ SNAP25	○ SYN1	○ TSEN2	○ YWHAE
○ PDHB	○ PLA2G6	○ PRRT2	○ RNASET2	○ SHPK	○ SNAP29	○ SYNE1	○ TSEN34	○ YWHAG
○ PDHX	○ PLAA	○ PSAP	○ RNF13	○ SIK1	○ SNIP1	○ SYNGAP1	○ TSEN54	○ ZDHHC9
○ PDP1	○ PLCB1	○ PSAT1	○ RNF216	○ SIX3	○ SNORD118	○ SYNJ1	○ TSFM	○ ZEB2
○ PDSS1	○ PLEKHG2	○ PSEN1	○ ROGDI	○ SLC12A5	○ SNRPB	○ SZT2	○ TTC19	○ ZFYVE26
○ PDSS2	○ PLP1	○ PSPH	○ RORB	○ SLC13A3	○ SNX27	○ TACO1	○ TTPA	○ ZIC1
○ PDYN	○ PLXNA2	○ PTCH1	○ RPIA	○ SLC13A5	○ SON	○ TAF2	○ TUBA1A	○ ZIC2
○ PET100	○ PMP22	○ PTEN	○ RPS6KC1	○ SLC16A2	○ SOX10	○ TANGO2	○ TUBA8	○ ZNF335
○ PEX1	○ PNKD	○ PTPN23	○ RRM2B	○ SLC17A5	○ SOX2	○ TARS2	○ TUBB2A	○ ZSWIM6
○ PEX10	○ PNKP	○ PTS	○ RTTN	○ SLC18A2	○ SPART	○ TBC1D20	○ TUBB2B	
○ PEX11B	○ PNP	○ PURA	○ RUSC2	○ SLC19A3	○ SPAST	○ TBC1D24	○ TUBB3	
○ PEX12	○ PNPO	○ PUS3	○ RXYLT1	○ SLC1A2	○ SPATA5	○ TBCD	○ TUBB4A	
○ PEX13	○ PNPT1	○ PYCR2	○ SAMHD1	○ SLC1A4	○ SPG11	○ TBCK	○ TUBG1	
○ PEX14	○ POLG	○ QARS	○ SATB2	○ SLC20A2	○ SPG7	○ TBL1XR1	○ TUBGCP6	
○ PEX16	○ POLG2	○ QDPR	○ SCARB2	○ SLC25A1	○ SPR	○ TCF4	○ TUFM	
○ PEX19	○ POLR1A	○ RAB11A	○ SCN1A	○ SLC25A12	○ SPTAN1	○ TGIF1	○ TWNK	

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Frequently asked questions

What is a genetic counselor?

Genetic counselors are healthcare providers specifically trained in medical genetics; they are experts at explaining complex genetic concepts and analyzing family history to understand disease risk. They also help people understand what their genetic test results mean for themselves and for their family members.

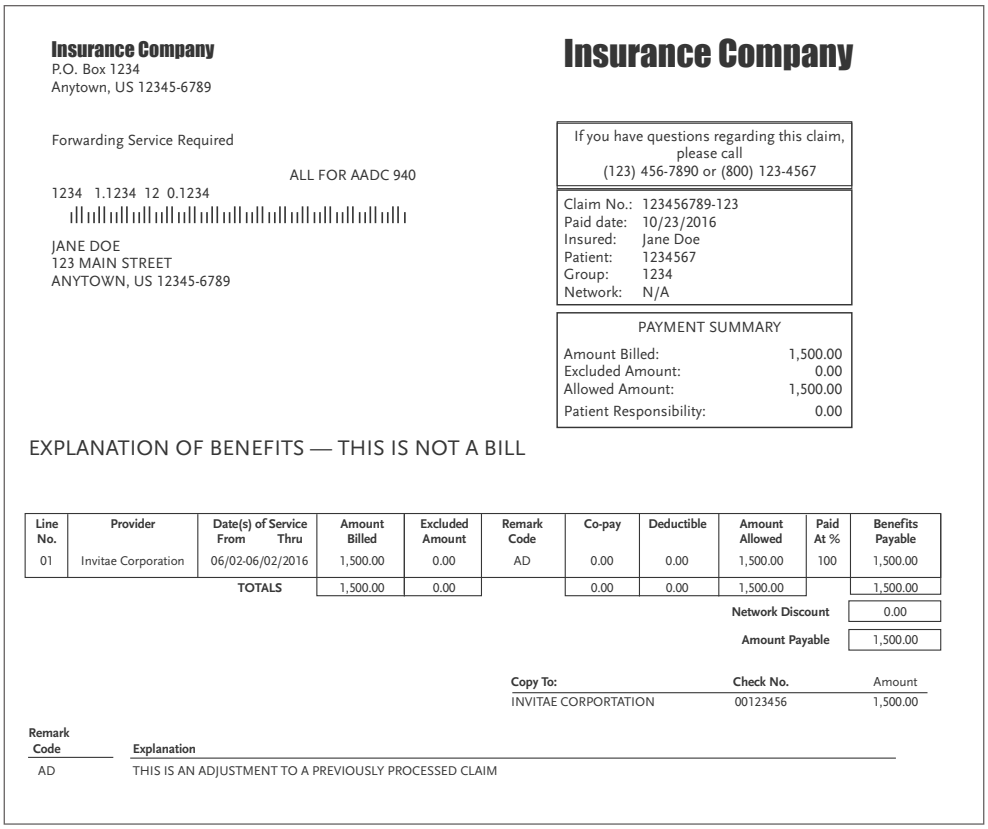
What should I expect when I call Invitae?

The genetic counselor will review your genetic test results (after you have received them from your healthcare provider) and answer any initial questions. If desired, you can also ask the genetic counselor to schedule a comprehensive post-test genetic counseling session to discuss in-depth what your genetic test results mean for you and for your family members.

What happens during a comprehensive post-test genetic counseling session?

An Invitae genetic counselor will review your personal and family medical history and then explain what your genetic test results may mean for you and your family members. If your test was positive, genetic testing may be recommended for your family members. Invitae offers Family Variant Testing for blood relatives at no additional charge within 90 days of your test report date for diagnostic panel and proactive health screens.

Learn more at **www.invitae.com/patients/genetic-counseling**



Insurance Company
P.O. Box 1234
Anytown, US 12345-6789

Forwarding Service Required

ALL FOR AADC 940

1234 1.1234 12 0.1234

JANE DOE
123 MAIN STREET
ANYTOWN, US 12345-6789

Insurance Company

If you have questions regarding this claim, please call
(123) 456-7890 or (800) 123-4567

Claim No.: 123456789-123
Paid date: 10/23/2016
Insured: Jane Doe
Patient: 1234567
Group: 1234
Network: N/A

PAYMENT SUMMARY

Amount Billed:	1,500.00
Excluded Amount:	0.00
Allowed Amount:	1,500.00
Patient Responsibility:	0.00

EXPLANATION OF BENEFITS — THIS IS NOT A BILL

Line No.	Provider	Date(s) of Service From	Thru	Amount Billed	Excluded Amount	Remark Code	Co-pay	Deductible	Amount Allowed	Paid At %	Benefits Payable
01	Invitae Corporation	06/02-06/02/2016		1,500.00	0.00	AD	0.00	0.00	1,500.00	100	1,500.00
		TOTALS		1,500.00	0.00		0.00	0.00	1,500.00		1,500.00
									Network Discount		0.00
									Amount Payable		1,500.00

Copy To:	Check No.	Amount
INVITAE CORPORTATION	00123456	1,500.00

Remark Code	Explanation
AD	THIS IS AN ADJUSTMENT TO A PREVIOUSLY PROCESSED CLAIM

EXPLANATION OF BENEFITS

The amount Invitae bills your insurance company will be reflected on your "explanation of benefits" letter. If you receive such a letter from your insurance company, please know that it is not a bill. Invitae will also receive this letter and will handle any appeals processes.

MORE INFORMATION

If you have questions about Invitae's payment options, our Client Services team is available to help. You can reach them at clientservices@invitae.com or 800-436-3037. Additional information can also be found at www.invitae.com/patients.

This flyer describes billing options for Invitae's single-gene and panel tests. To see billing options for Invitae's exome tests, please visit www.invitae.com/exome.

CLINICAL HISTORY

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ◯ Yes ◯ No If yes, describe below and attach pedigree and/or clinical notes.

Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis	Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

PERSONAL HISTORY

Is/was this patient affected or symptomatic?† ◯ Yes ◯ No

Provide details in the required clinical history questions (if applicable).

† Symptomatic means this patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

REQUIRED CLINICAL HISTORY

Clinical history	YES	NO	UNKNOWN
Syncope with stress	◯	◯	◯
Syncope without stress	◯	◯	◯
History of aborted SCD	◯	◯	◯
Congenital deafness	◯	◯	◯
Skeletal muscle weakness	◯	◯	◯

List other relevant history:

_____

_____

_____

Histological and biochemical findings	YES	NO	UNKNOWN
Fibrofatty replacement of myocardium	◯	◯	◯
Amyloid-positive tissue biopsy	◯	◯	◯
Elevated creatine kinase	◯	◯	◯

Imaging findings	YES	NO	UNKNOWN
CMRI delayed enhancement	◯	◯	◯
LV noncompaction	◯	◯	◯
Dilation of the right ventricle	◯	◯	◯
Dilation of the left ventricle	◯	◯	◯
Myocardial scarring	◯	◯	◯
LV outflow tract obstruction	◯	◯	◯
LV ejection fraction	_____ %		◯
RV ejection fraction	_____ %		◯
Maximum LV wall thickness	_____ **mm**		◯
LV end systolic diameter	_____ **mm**		◯
LV end diastolic diameter	_____ **mm**		◯

List other relevant abnormalities:

_____

_____

_____

ECG findings	YES	NO	UNKNOWN
Normal ECG	◯	◯	◯
AV Block	◯	◯	◯
Ventricular fibrillation	◯	◯	◯
Ventricular tachycardia	◯	◯	◯
Bidirectional VT	◯	◯	◯
Torsade de pointes	◯	◯	◯
T wave alternans	◯	◯	◯
Notched T wave in 3 leads	◯	◯	◯
Positive exercise stress test	◯	◯	◯
Low heart rate for age	◯	◯	◯
Cardiac conduction defects	◯	◯	◯
QTc interval	_____ **ms**		◯

List other relevant abnormalities:

_____

_____

_____

OPTIONAL - REQUESTED VARIANTS FOR THIS PATIENT'S REPORT, IF KNOWN

To have the presence or absence of specific variants commented on in this patient's report, provide the details below. For gene-specific family follow-up see ***Note*** *under Test Selection.*

Was the proband (individual with variant) tested at Invitae? ◯ Yes, Invitae Order ID: RQ# _____ ◯ No: Attach copy of lab results (*required*)

Variant(s) *(e.g. GENE c.2200A>T (p.Thr734Ser) NM_00012345) If left blank, all variants identified in the proband will be commented on.*

This patient's relationship to proband:

◯ Parent ◯ Sibling ◯ Grandchild
◯ Child ◯ Self ◯ Other: _____

TEST SELECTION – Select test(s) from either option 1 or 2 below:

1. DETECT CARDIOMYOPATHY AND ARRHYTHMIA PROGRAM – Indicate test(s) to be performed below:

Test code	Test name		# of genes	Gene list
○ 02101	Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel		100	ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BRAF, CACNA1C, CACNA1D, CALM1, CALM2,CALM3, CASQ2, CBL, CDH2, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GATA4, GATA5, GJA5, GLA, HCN4, HRAS, JUP, KCNE1, KCNH2, KCNJ2, KCNQ1, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3,MYL4, MYLK3, NF1, NKX2-5, NRAS, PCCA, PCCB, PKP2, PLN, PPA2, PPCS, PPP1CB, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RYR2, SCN5A, SDHA, SGCD, SHOC2, SLC22A5,SOS1, SOS2, SPRED1, TAZ, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL
	○ 02101.1	Add-on preliminary-evidence genes	57	A2ML1, AKAP9, ANK2, ANKRD1, CACNA2D1, CACNB2, CALR3, CAV3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA6, GATAD1, GPD1L, HAND1, ILK, JPH2, KCNA5, KCND3, KCNE2, KCNE3, KCNE5, KCNJ5, KCNJ8,KCNK3, KIF20A, KLF10, LAMA4, LDB3, LRRC10, MAP3K8, MED12, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NPPA, PDLIM3, PLEKHM2, PRDM16, RANGRF, RASA2, RRAS, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SLMAP, SNTA1, TMPO, TXNRD2
	○ 02101.4	Add-on sudden unexpected death in epilepsy (SUDEP) genes for arrhythmia and cardiomyopathy	11	DEPDC5, KCNA1, KCNQ2, KCNQ3, KCNT1, PCDH19, PRRT2, SCN1A, SCN8A, SCN9A, SLC2A1

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING *For relatives of a program participant ('proband') who received a Pathogenic/Likely Pathogenic result or approved VUS.*

○ Family follow-up testing for Proband's Invitae Order ID: RQ#_____	**This patient's relationship to proband:** ○ Parent ○ Sibling ○ Grandchild ○ Child ○ Other: _____	**Gene(s) to be tested in this patient:**

***NOTE**: The presence or absence of all variants identified in the proband for the gene(s) ordered for gene-specific family follow-up will be commented on in this patient's report unless a limited selection is specified in the **Requested Variants** section above. Invitae will report any Pathogenic/Likely Pathogenic variants found in this patient for the gene(s) ordered.*

Invitae continually updates its panels based on the most recent evidence. If an order is placed using an outdated test requisition form, Invitae reserves the right to upgrade ordered tests to their current versions. Test IDs containing add-on codes will include the original panel as well as the add-on.

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/forms). The medical professional will retain evidence that the patient consented to genetic testing. The Patient 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) and has been informed that deidentified (also referred to as pseudonymized) patient data may be used and shared with third parties in connection with the Program, for research and commercial purposes. For orders originating outside the United States, the Patient has been informed that their personal information and specimen will be transferred to and processed in the United States. The medical professional warrants that (i) he/she will not seek reimbursement for this no-charge test from any third party, including but not limited to government healthcare programs; (ii) participation in the Program will not influence the his/her medical decisions; (iii) he/she is not obligated to purchase or prescribe any product or service offered by a sponsor of the Program; (iv) he/she is not obligated to participate in or to encourage patients to participate in any clinical trial or other research program conducted by a sponsor; and (v) he/she will participate in the Program in accordance with applicable laws. The medical professional consents to the sharing of organization and clinician contact information with third parties, including commercial organizations, who may contact the medical professional directly in connection with the Program. A list of third party partners will be provided upon request. I attest that I am authorized under applicable law to order this test.

Medical professional signature (required)	**Date (MM/DD/YYYY)**

CLINICAL HISTORY

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ◯ Yes ◯ No If yes, describe below and attach pedigree and/or clinical notes.

Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis	Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

PERSONAL HISTORY

Is/was this patient affected or symptomatic?† ◯ Yes ◯ No

Provide details in the required clinical history questions (if applicable).

† Symptomatic means this patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

OPTIONAL - REQUESTED VARIANTS FOR THIS PATIENT'S REPORT, IF KNOWN

*To have the presence or absence of specific variants commented on in this patient's report, provide the details below. For gene-specific family follow-up see **Note** under Test Selection.*

Was the proband (individual with variant) tested at Invitae? ◯ Yes, Invitae Order ID: RQ# _____ ◯ No: Attach copy of lab results (*required*)

Variant(s) *(e.g. GENE c.2200A>T (p.Thr734Ser) NM_00012345) If left blank, all variants identified in the proband will be commented on.*		**This patient's relationship to proband:**
		◯ Parent ◯ Sibling ◯ Grandchild
		◯ Child ◯ Self ◯ Other: _____

TEST SELECTION – Select test(s) from either option 1 or 2 below:

1. ALNYLAM ACT® (hATTR AMYLOIDOSIS) PROGRAM – Indicate test(s) to be performed below:

Test code	Test name	# of genes	Gene list
◯ 02251	Invitae Cardiomyopathy Comprehensive Panel	82	ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BRAF, CACNA1C, CBL, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GLA, HCN4, HRAS, JUP, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYLK3, NF1, NRAS, PCCA, PCCB, PKP2, PLN, PPCS, PPP1CB, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RYR2, SCN5A, SDHA, SGCD, SHOC2, SLC22A5, SOS1, SOS2, SPRED1, TAZ, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL
◯ 03200	Invitae Comprehensive Neuropathies Panel	102	AARS, AIFM1, APOA1, ASAH1, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CHCHD10, COX6A1, CYP27A1, CYP7B1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EXOSC9, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GLA, GNB4, GSN, HARS, HEXA, HINT1, HMBS, HSPB1, HSPB8, IGHMBP2, INF2, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, POLG, POLG2, PRDM12 , PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN11A, SCN9A, SEPT9, SH3TC2, SIGMAR1 , SLC12A6, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SMN2, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, VRK1, WNK1, YARS
◯ 02265	Invitae Hereditary Transthyretin-mediated Amyloidosis (hATTR Amyloidosis) Test	1	TTR

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING *For relatives of a program participant ('proband') who received a Pathogenic/Likely Pathogenic result or approved VUS.*

◯ Family follow-up testing for Proband's Invitae Order ID: RQ#_____	**This patient's relationship to proband:** ◯ Parent ◯ Sibling ◯ Grandchild ◯ Child ◯ Other: _____	**Gene(s) to be tested in this patient:**

***NOTE**: The presence or absence of all variants identified in the proband for the gene(s) ordered for gene-specific family follow-up will be commented on in this patient's report unless a limited selection is specified in the **Requested Variants** section above. Invitae will report any Pathogenic/Likely Pathogenic variants found in this patient for the gene(s) ordered.*

Invitae continually updates its panels based on the most recent evidence. If an order is placed using an outdated test requisition form, Invitae reserves the right to upgrade ordered tests to their current versions. Test IDs containing add-on codes will include the original panel as well as the add-on.

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/forms) and in connection with the Alnylam Act® program, the Patient 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) and has been informed that de-identified Patient data may be used and shared with third parties, including Alnylam Pharmaceuticals, Inc. ("Alnylam"), for research and commercial purposes to contact their medical professional. For orders originating in Canada, the Patient has been informed that their personal information and specimen will be transferred to and processed in the U.S. and that de-identified Patient data may be used and shared for research and commercial purposes in the U.S. and elsewhere. The medical professional warrants that he/she will not seek reimbursement for this sponsored test from any third party, including but not limited to U.S. federal healthcare programs. The medical professional also hereby acknowledges that organization and clinician contact information provided in the order may be shared with third parties, including Alnylam, that may contact the medical professional directly in connection with the Alnylam Act® program, or Alnylam products. The medical professional understands that the use of this sponsored test is not intended to be, nor should it be construed as, either express or implied, an obligation or inducement for the medical professional to recommend, purchase, order, prescribe, promote, administer or otherwise support any Alnylam product or any other Invitae product or service. I attest that I am authorized under applicable state law to order this test.

Medical professional signature (required)	**Date (MM/DD/YYYY)**

Could it be **FABRY DISEASE?**

QUESTIONS TO ASK YOUR PATIENT	YES	NO
1. Do you have chronic tingling or burning in your hands or feet?	☐	☐
2. Have you ever experienced episodes of extreme pain in your hands and/or feet of unknown cause, possibly accompanied by fever?	☐	☐
3. Do you have trouble sweating or exercising?	☐	☐
4. Do you find heat or cold hard to tolerate?	☐	☐
5. Do you frequently have gastrointestinal problems such as pain and bloating after eating, or nausea, cramps, or diarrhea?	☐	☐
6. Do you have small raised reddish-purple spots on your skin, especially in the "bathing trunk" area?	☐	☐
7. Do you have a family history of early cardiac or valvular disease, renal failure, or stroke?	☐	☐

Clusters of signs and symptoms could help distinguish Fabry disease–a progressive, potentially life-threatening disorder–from more common conditions.

PROGRESSIVE SIGNS AND SYMPTOMS

Fabry disease is progressive and affects multiple organ systems. This chart indicates signs and symptoms that may appear at various stages of life.[1]

Most males with the defective gene are subject to significant morbidity and mortality.[2] While females with the defective gene demonstrate a wide range of disease severity, most develop symptoms.[3,4]

SYMPTOMS	Childhood	Adolescence	Adulthood
Hearing loss and tinnitus	●	●	●
Episodic pain crises	●	●	●
Neuropathic pain	●	●	●
Hypohidrosis/anhidrosis	●	●	●
Corneal and lenticular opacities	●	●	●
Recurrent fever	●	●	●
Heat and cold intolerance	●	●	●
Psychosocial manifestations	●	●	●
Gastrointestinal distress	●	●	●
Proteinuria		●	●
Angiokeratomas		●	●
Fatigue		●	●
Renal insufficiency			●
Neurological complications			●
Cerebrovascular disease			●
Cardiac dysfunction			●

TAKE ACTION

DIAGNOSING MALES:

- Alpha galactosidase enzyme assay is diagnostic.
- Males typically have <1% normal alpha-galactosidase in plasma and leukocytes.[2]

DIAGNOSING FEMALES:

- Enzyme assay alone is frequently insufficient for diagnosis.
- DNA-based diagnosis is required in females with normal to low-normal enzyme activity levels, and is advisable in all suspected patients.

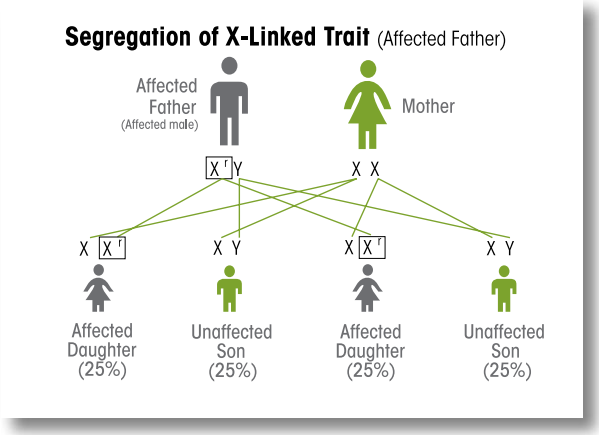
OCULAR ASSESSMENT:

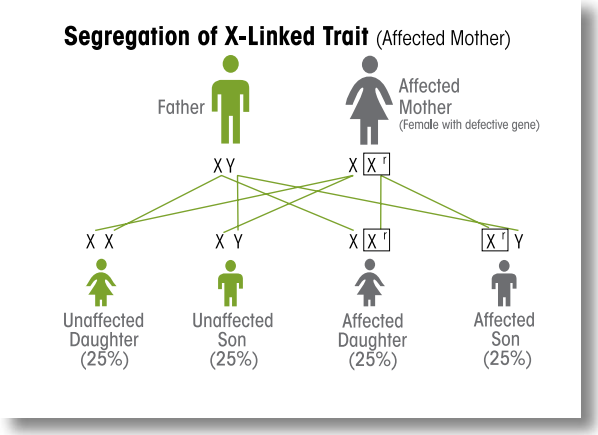
- Corneal whorling, visible through slit lamp ophthalmoscopy, is present in >90% of Fabry disease patients.[5]
- A slit lamp exam by an eye care professional may help establish the need for further testing.

HOW FABRY DISEASE IS INHERITED

Fabry disease is an X-linked genetic disorder. Males with the defective gene pass it on to all of their daughters and none of their sons. Females with the defective gene have a 50% chance with each pregnancy of passing the gene to each of their offspring.

Because females have two X chromosomes in every somatic cell, Fabry disease symptoms are more variable in females than they are in males. However, potentially life-threatening complications can develop, even in females whose presentation may suggest a more moderate disease course.





WHAT TO DO IF YOU SUSPECT FABRY DISEASE

If you suspect that a patient has Fabry disease, refer to a geneticist. A geneticist can help establish a definitive diagnosis and provide information on disease management.

SANOFI GENZYME RESOURCES

For Providers:
Sanofi Genzyme
Medical Information
1-800-745-4447, option 2

For Patients:
Connect with a Sanofi Genzyme Case Manager online at www.careconnectpss.com or call 1-800-745-4447, Option 3, Monday through Friday, 8:00 AM to 6:00 PM ET

www.discoverfabry.com

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3. Wang RY, Lelis A, Mirocha J, Wilcox WR. Heterozygous Fabry women are not just carriers, but have a significant burden of disease and impaired quality of life. Genet Med 2007;9:34-45.
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5. Franceschetti A. Fabry disease: ocular manifestations. In: Bergsma D, Bron AJ, Cotlier E (eds). The Eye and Inborn Errors in Metabolism. Vol. 12, No. 3. New York: AR Liss Co., 1976;195-208.

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Fabry Disease Recommended Schedule of Assessments

The Recommended Schedule of Assessments represents the core Fabry disease-related assessments that allow evaluation of a patient's disease progression over time. Physicians will determine the actual frequency of necessary assessments according to a patient's individualized need for medical care and routine follow-up.

These recommendations were developed by the Fabry Registry Board of Advisors, a group of physicians who have experience in managing patients with Fabry disease. The Fabry Registry is sponsored and administered by Sanofi Genzyme.

Pediatric Patients (Under 18 Years of Age*)

	Upon Enrollment	Every 6 – 12 months[a]	Every 24-36 months	At time of an event or therapy change
GENERAL				
Medical History, with particular focus on:				
Gastrointestinal Symptoms				
Pain	■	■		■
Sweating				
Heat & Cold Intolerance				
Family History	■		■	
Physical Exam	■	■		■
Vital Signs, Height and Weight	■	■		■
Blood Pressure[b]	■	■		■
Enzyme Activity and Genotype	■			
Concomitant Medication Assessment	■	■		■
Pediatric Quality of Life Assessment – PedsQL™ Pediatric Quality of Life Inventory	■	■		■
Pediatric Quality of Life Assessment – PedsQL™ Multidimensional Fatigue Scale	■	■		■
Pediatric Pain Assessment – PedsQL™ Pediatric Pain Questionnaire™	■	■		■
LABORATORY TESTS				
Glomerular Filtration Rate (GFR)[c]	■		■	■
Albuminuria and Proteinuria[d]	■	■		■
OTHER STUDIES				
Audiologic Evaluation[e]	■		■	■
Cranial MRI – T1, T2 and FLAIR	■		■ [f]	■ [f1]
Electrocardiogram[g]	■		■	■
Echocardiogram[h]	■		■	■
Cardiac MRI[i]	■		■	■
Ophthalmology – Slit Lamp Exam[j]	■		■	

* Initiation of Laboratory Tests, Imaging, and Other Studies: There is variability in the clinical complications and progression of Fabry disease. Children are at risk for life threatening complications. There are no biomarkers available to discern mildly affected from severely affected patients. In children with a family history of early presenting or severe disease, complete evaluations should be done at the time of diagnosis. Other patients should be completely evaluated at no later than 5 years of age.

[a] Patients are recommended to undergo these evaluations every 6 months; for those with milder disease, once per year may be sufficient

[b] Blood pressure should be measured 3 times at each assessment; only the last 2 measurements should be recorded.

[c] GFR should be measured directly every 24-36 months until age 15, and annually thereafter. If direct measurement is not possible, serum creatinine levels should be obtained at the recommended intervals for an estimation of GFR, which is a less sensitive method.

[d] First morning voided urine for protein, albumin and creatinine in order to calculate a protein/creatinine ratio and albumin/creatinine ratio. Protein, albumin, and creatinine measurements can also be performed on timed samples (e.g. 24 hours).

[e] Audiologic evaluation should be performed at the earliest age that is practical.

[f] Cranial MRIs should be performed at ages 10, 15, and 18 years.

[f1] At the time of a cerebrovascular event, a cranial MRI should also include diffused weighted images and apparent diffusion coefficient (DWI/ADC).

[g] Electrocardiogram should be performed starting at age 10–12 years. If abnormal and/or clinical symptoms arise, Holter monitoring is recommended.

[h] Echocardiogram should be performed starting at age 10–12 years.

[i] Cardiac MRI is recommended to be performed in patients under age 25 if cardiac hypertrophy or significant arrhythmia is present.

[j] Monitor yearly if retinal vessel tortuosity noted.

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Fabry Disease Recommended Schedule of Assessments

Adult Patients (≥18 Years of Age)

	Upon Enrollment	Every 6 months	Every 12 months	Every 24-36 months	At time of an event or therapy change
GENERAL					
Medical History	■	■			■
Family History	■			■	
Physical Exam	■	■			■
Vital Signs, Height and Weight	■	■			■
Enzyme Activity and Genotype	■				
Concomitant Medication Assessment	■	■			■
Quality of Life (SF-36®, BPI)	■	■			■
LABORATORY TESTS					
Serum Creatinine[a] and BUN	■	■			■
Urine Protein Excretion[b]	■	■			■
Lipid panel	■		■		
OTHER STUDIES					
Audiologic Evaluation	■			■	■
Cranial MRI – T1, T2 and FLAIR	■			■	■[c]
Electrocardiogram[d]	■		■		■
Echocardiogram	■		■		■
24-Hour Holter Monitoring[e]	■		■		■
Cardiac MRI[f]	■		■[f1]	■[f1]	■[f2]
Respiratory – Spirometry Exam[g]	■			■	
Ophthalmology – Slit Lamp Exam[h]	■				

[a] Directly measuring glomerular filtration rate (GFR) is recommended if a more precise evaluation is desired.

[b] 24 hour or first morning void urine for protein, creatinine and albumin.

[c] At the time of an event, a cranial MRI should also include diffused weighted images and apparent diffusion coefficient (DWI/ADC).

[d] If electrocardiogram is abnormal and/or clinical symptoms arise, Holter monitoring is recommended.

[e] Annual 24-hour Holter monitoring is recommended for males 30 years of age or older and females 40 years of age or older.

[f] Cardiac MRI is recommended at Fabry diagnosis for patients ages 25 and older. It is recommended to be performed under age 25 if cardiac hypertrophy or significant arrhythmia is present.

[f1] If first MRI is abnormal: 1) patients with moderate or severe LVH receiving ERT should have MRI annually; 2) patients with significant arrhythmia should have MRI at least every 2 years or at frequency factoring cardiac disease severity and the physician's clinical judgment; 3) males with no or mild LVH receiving ERT should have MRI every 2 years.

[f2] If first MRI is normal, repeat every 5 years or earlier if ECG/ECHO results are abnormal on annual exam.

[g] If spirometry is abnormal, perform yearly.

[h] Monitor yearly if retinal vessel tortuosity noted.

For more information on Fabry disease, contact Sanofi Genzyme Medical Information at 800-745-4447, option 2.

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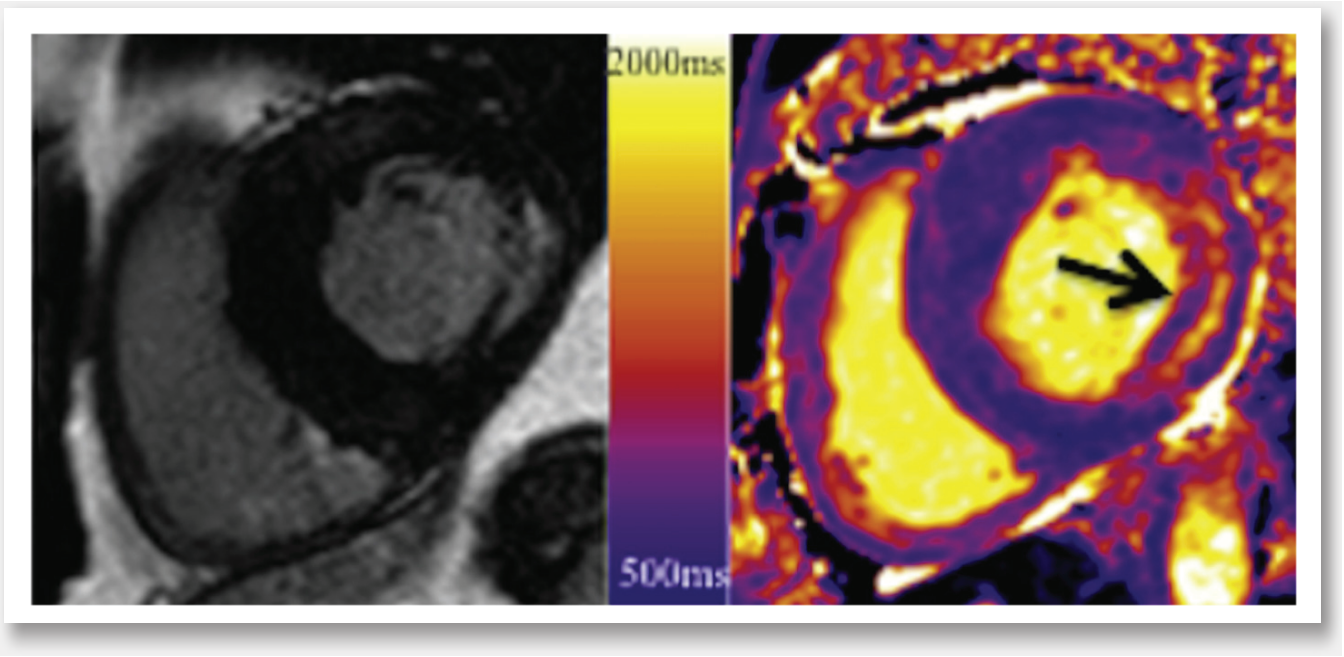
Have you seen unexplained hypertrophic cardiomyopathy (HCM)?

It could be Fabry disease



Phase sensitive inversion recovery (PSIR) image showing severe left ventricular hypertrophy (LVH) and late gadolinium enhancement (LGE) in basal inferolateral (BIFL) wall in midmyocardium (pictured left).[1] T1 mapping shows low T1 in most of the myocardium due to GL-3 deposition except in area of fibrosis in BIFL where it is elevated (pictured right).[1]

Fabry prevalence in idiopathic HCM is up to ~300x higher in females and ~1200x higher in males than in the general population.[2-5]

Cardiologists can play a critical role in the early detection of Fabry disease

Discover more at
medscape.com/infosites/281991.1

References: 1. Nordin et al., *J Am Coll Cardiol.* 2016;68:1707-8. **2.** Laney D. *J Genet Couns*. 2008;17:79-83. **3.** van der Tol L. *J Med Genet.* 2014;51(1):1-9. **4.** Nakao S. *N Engl J Med*. 1995;333(5):288-93. **5.** Desnick RJ et al., Chapter 150. In: Valle D et al., eds. The Online Metabolic and Molecular Bases of Inherited Disease. New York, NY: *McGraw* Hill; 2014.

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