All sections on this page are required unless otherwise specified. Important fields are highlighted. Incomplete information could result in a delay of testing.

PATIENT INFORMATION					
First Name	Last Name				
Sex Assigned at Birth: OMale OFemale	Date of Birth (mm/dd/	/уу)			
Patient Karyotype (if known):					
Gender Identification (optional):					
Email					
Address					
City	State	Zip Code			
Primary Phone	Is this patient decease	ed? O Yes O No			
	Deceased Date:				

SAMPLE INFORMATION				
Date Sample Collected (mm/dd/yy) Medical Record #				
OBlood OBuccal Swab O Other (specify source):				
Treatment-related RUSH (optional) Reason: O Transplantation O Pregnancy O Surgery O Other:				
Patient has had a blood transfusion       OYes       ONo       Date of Last Transfusion:         (2-4 weeks of wait time is required for some testing)				
Patient has had an allogeneic bone marrow transplant () Yes () No Fibroblasts are required for patients who had an allogeneic bone marrow transplant. See www.genedx.com/specimen-requirements for details.				
Patient has a personal history of a hematolo	gic malignancy or disease			
	<u>ONe</u>			

O Yes (specify diagnosis) ONo If yes, please call the lab to discuss with a genetic counselor the most appropriate sample type.

# **PATIENT CONSENT**

By signing this form, I acknowledge as the patient or relative being tested that I have read or have had read to me the GeneDx Informed Consent document at the end of this test requisition form, and understand the information regarding molecular genetics testing. I have had the opportunity to ask questions about the testing, the procedure, the risks, and the alternatives. By signing this form, I authorize GeneDx to perform genetic testing as ordered. I understand that, for tests that evaluate data from multiple family members concurrently, test results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their healthcare providers.

- By checking this box, I confirm that I am a New York State resident, and I give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing, and to be used as a de-identified sample for test development and improvement, internal validation, quality assurance, and training purposes. Otherwise, New York law requires GeneDx to destroy my sample within 60 days, and it cannot be used for test development studies.
- Check this box if you wish to opt out of being contacted for research studies

Date
Date
Date
ission to GeneDx to contact ng. Data rates may apply.
zing the genetic testing.

ACCOUNT INFORMATION				
GeneDx Account Number	Account Name			
Phone	Fax			
Address	·			
City	State	Zip Code		
Ordering Provider Name		Role/Title		
NPI	Phone Number			
Send Report Via: Fax Email Portal				
Additional Ordering Provider Name (optional)		Role/Title		
NPI				
Send Report Via: Fax Email Portal Fax #/Email:				
SEND ADDITIONAL REPORT COPIES TO (optiona	I)			
Provider Name	GeneDx Acct#			

#### STATEMENT OF MEDICAL NECESSITY

By submission of this test requisition and accompanying sample(s), I: (i) authorize and direct GeneDx to perform the testing indicated; (ii) certify that the person listed as the ordering provider is authorized by law to order the test(s) requested; (iii) certify that any custom panel and/or ordered test(s) requested on this test requisition form are reasonable and medically necessary for the diagnosis and/or treatment of a disease, illness, impairment, symptom, syndrome or disorder; (iv) the test results will determine my patient's medical management and treatment decisions of this patient's condition on this date of service; (v) have obtained this patient's and relatives', when applicable, written informed consent to undergo any genetic testing requested; and (vi) that the full and appropriate diagnosis code(s) are indicated to the highest level of specificity. **Signature of Ordering Provider** Date

### ICD-10-CM CODES

С	D-	10-	СМ	Coc	les

Fax #/Email:

Clinical Diagnosis

Age of Onset

PAYMENT OPTIONS (Select One)					
O INSURANCE BILL Select all that apply Commercial	Patient Status OHospital outpati ONot a hospital pe	- · ·	tient; Date of Dischar	ge:	
 ☐ Medicaid ☐ Medicare	Name of Insuranc	Name of Insurance Carrier Insurance ID#:			
Tricare	Relationship to Ins		• •		
	Oself Ospouse Ochild Oother Policy Holder's Name		Policy Holder's Date	of Birth	
PROVIDE FRONT AND BACK COPY OF CARD(S)	Referral/Prior Authorization # (please attach)		Hold test for cost estimate and contact patient		
	Secondary Insura	псе Туре:	if estimate is '>\$2 (commercial ins		
	Insurance Carrier	Insurance ID #	Subscriber Name	Date of Birth	
	Relationship to Insured Oself Ospouse Ochild Oother:				
O PATIENT BILL	If Patient Bill is selected, I am electing to be treated as a self-pay patient for this testing. I agree that neither GeneDx nor I will submit a claim to my insurance for this testing, if I have insurance. GeneDx will send an invoice to the patient listed above.				
	Authorized Patient/Guardian Signature				
	GeneDx Account #	-	Place Sticker/St	amp Here	
	Hospital/Lab Nam	e			

# Gene

First Name

Last Name

Date of Birth

**Gene** 

Is this person affected: O Yes O No	Clinical diagnosis:	
	ptomatic diagnosis 🛛 Carrier/Familial variant	
lease check all that apply. This is not a subst		
Diagnosis ] Amyloidosis	Marfan/TAAD/HDCT	Abnormal heart morphology Bicuspid aortic valve
	□ Aortic/Arterial dissection	Coarctation of aorta
Brugada syndrome	□ Aortic root dilation	🗆 Heart murmur
⊐ СРVТ ́	□ Arachnodactyly	🗆 Heterotaxy
DCM	🗆 Arterial tortuosity/ectasia	🗆 Hypoplastic left heart
] Ehlers-Danlos syndrome	🗆 Arthralgia	🗆 Mitral valve prolapse
	Atypical scarring of skin	Patent ductus arteriosus
	Beighton score:	Patent foramen ovale
] Hypertension	🗆 Bifid uvula	Tetralogy of Fallot
] Loeys-Dietz syndrome	Blue sclerae	Ventricular septal defect
] LQT syndrome ] Noncompaction cardiomyopathy (LVNC)	□ Bruising susceptibility □ Cleft lip	Atrial septal defect     Other:
Marfan syndrome	□ Cleft palate	
		РАН
	Cutis laxa	Pulmonary hypertension
□ SQT syndrome	Dental crowding	
Sudden Cardiac Arrest	Dural ectasia	Other
🛛 Sudden Death	🗆 Ectopia lentis	Abnormality of the periventricular white
	Flexion contracture	matter
Echocardiogram	🗆 High palate	🗆 Angiokeratomas
Aortic root dimensions:		Anhydrosis
Z-score:	Uterine rupture	Café-au-lait macules
□ EF%:	Intestinal perforation	Hearing impairment:
	Other:	□ Sensorineural
Z-score: ] Max LV wall thickness:	Hypertelorism     Joint contractures	
□ Max LV wall thickness:	□ Joint contractures □ Joint dislocations	□ Craniosynostosis □ Cystic hygroma
	□ Joint dislocations □ Joint hypermobility	Downslanted palpebral fissures
	Meets Ghent criteria	Dysmorphic features:
ECG	Micrognathia / Retrognathia (circle what	Describe:
∃ Prolonged QTc interval:	applies)	Elevated CPK
Max QTc:	□ Midface retrusion	🗆 Hypotonia
□ Normal	🗆 Mitral valve prolapse	🗆 Increase nuchal translucency
Report Included	🗆 Myopia	🗆 Intellectual disability
	Osteoarthritis	□ Keratoconus
Arrhythmia/Cardiomyopathy	Pectus carinatum	Muscle weakness
Abnormal atrioventricular conduction	Pectus excavatum	□ Myopathy
Atrial fibrillation	Pes planus     Pneumothorax	Renal insufficiency     Short neck
☐ Bradycardia ☐ Fatty replacement of ventricular	Recurrent fractures	☐ Thromboembolism
myocardial tissue	Retinal detachment	Type:
☐ Heart transplant	□ Scoliosis/kyphosis (circle what applies)	· , po
	□ Skin findings, Specify:	
Torsades de pointe	□ Stroke	
□ Ventricular tachycardia	□ Tall stature	Attach pedigree and/or include additional
·	🗆 Velvety skin	clinical information:
ННТ		
Arteriovenous malformation		
] Telangiectasia		
Dielinidensige		
Dislipidemias Atherosclerosis		
Corneal arcus		
LDL-C levels:		
Li Xanthomatosis		
🗆 Xanthomatosis 🗆 Other:		

First Name

Last Name

Date of Birth

Gene[

FAMILY HISTORY					
🗆 No Known Family History	□ Pe	edigree Att	ached 🗆 Adopted		
Relationship	Maternal	Paternal	Relevant History	Age at Dx	
1	0	0			
2	0	0			
3	0	0			

### PREVIOUS GENETIC TESTING

Personal or family history of genetic testing

O Yes (If yes, please complete all fields below)

Relation to patient (self, sibling, etc.), Genetic Test(s) and Result (e.g. positive, negative, etc.). If relative was tested at GeneDx, please also provide their accession #:

If patient or relative(s) were found to have a positive or VUS result on prior testing, please provide details below. Indicate any Variants of Interest<sup>‡</sup> via the checkbox below.

O No

Relation (self, sibling, etc.)	Gene	Transcript #	c./p. (SNV) or exon # (CNV)	Build, coordinates (CNV)	Variant of Interest‡?
1					
2					
3					
Required for sequence variants: gene, c./p., transcript # Required for CNVs: gene, transcript #, exon # <u>QR</u> build, coordinates					
Abnormal karyotype, FISH, or ot	her results:				
	_				

‡ For certain tests, GeneDx **may** be able to specifically comment upon the presence or absence of previously identified variant(s) of interest in the report. Complete variant information must be provided <u>in the table above</u> at the time the test order is placed. If you do not complete the table above and check off that a previously identified variant is a variant of interest, it will not be possible to comment upon the presence or absence of the variant in the report retrospectively. This service is not applicable to targeted variant testing.

TARGETED VARIANT TESTING					
Individual to be tested:	O Affected/Symptom	atic OUnaffecte	d/Asymptomatic		
□ Known Familial Variant(s □ Known Familial Copy Nur	·	□Confirmation of Varia □Known mtDNA Varia	ant Identified in Research Lab nt(s) Testing	□Targeted Mosaic Variant Testing (Insurance Billing NOT Accepted; Patient Bill or Institutional Bill MUST be selected on page 1)	
Proband Name		Relationship to Proband		Proband GeneDx Accession #	
	ositive control included/w ositive control not availab	ill be sent <b>- Positive contr</b> le (caveat language will k	I if previous test was performe tol is recommended if previou be included on a negative repo member report is not included)	s test was performed at another lab. ort)	
Gene	Coding DNA (o	:./m.)	Amino Acid (p.)	Transcript (NM#)	
Gene Coding DNA (c./m.)		/m.)	Amino Acid (p.)	Transcript (NM#)	
COPY NUMBER VARIA	NT		I	Number of Variants:	
Gene(s)	Exon #		Coordinates	Genome Build	
Gene(s)	Exon #		Coordinates	Genome Build	

First Name

Last Name

Date of Birth

Gene

TEST MENU						
TEST CODE	TEST NAME	TEST CODE	TEST NAME			
910	Chromosomal Microarray (MicroarrayDx)	□ TJ07	Xpanded® Congenital Heart Defects Panel			

#### FAMILY MEMBER FOR XPANDED® PANEL TESTING OPTION NO SEPARATE REPORT, ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS OF PROBAND SAMPLE. See Test Menu page for proband test selection. TJ33 Xpanded® Congenital Heart Defects, Family member testing First Name Last Name DOB O Asymptomatic **O** Symptomatic Biological O At GeneDx (Accession #: ) Mother O Not available O To be sent within 3 weeks First Name Last Name DOB O Asymptomatic **O** Symptomatic Biological O At GeneDx (Accession #: ) Father O Not available O To be sent within 3 weeks Relationship to Proband First Name Last Name DOB O Asymptomatic **O** Symptomatic Other Biological ) O At GeneDx (Accession #: Relative O Not available O To be sent within 3 weeks

	TEST MENU (continued)						
TEST CODE	TEST NAME	TEST CODE	TEST NAME				
ARRHYTHM	IA TESTING OPTIONS						
695	Arrhythmia Sequencing and Del/Dup Panel	483	ARVC Sequencing and Del/Dup Panel				
🗖 695RE	Reflex to Rest of Combined Cardiac after Arrhythmia Panel	□ 483RE	Reflex to Rest of Combined Cardiac after ARVC Panel				
481	Brugada syndrome Sequencing and Del/Dup Panel	□ J552	SCA Arrhythmia Sequencing and Del/Dup Panel				
□ 481RE	Reflex to Rest of Arrhythmia after Brugada Syndrome Panel	☐ J552RE	Reflex to Rest of Arrhythmia after SCA Arrhythmia Panel				
727	LQTS Sequencing and Del/Dup Panel						
□ 727RE	Reflex to Rest of Arrhythmia after LQTS Panel						
CARDIOMYOPATHY TESTING OPTIONS							
□ 694	Cardiomyopathy Sequencing and Del/Dup Panel	483	ARVC Sequencing and Del/Dup Panel				
🗖 694RE	Reflex to Rest of Combined Cardiac after Cardiomyopathy Panel	□ 483RE	Reflex to Rest of Combined Cardiac after ARVC Panel				
🔲 J554	DCM/LVNC Sequencing and Del/Dup Panel	🗖 J553	HCM Sequencing and Del/Dup Panel				
☐ J554RE	Reflex to Rest of Cardiomyopathy after DCM Panel	iomyopathy after DCM Panel 🛛 J553RE Reflex to Rest of Cardiomyopathy after HCM Panel					
COMBINED	ARRHYTHMIA AND CARDIOMYOPATHY TESTING OPTI	ONS					
□ 935	Combined Cardiac Panel						
LIPIDEMIAS	TESTING OPTIONS						
🗖 J556	Familial Hypercholesterolemia Sequencing and Del/Dup Panel	🗖 TA01	Familial Dyslipidemia Sequencing and Del/Dup Panel				
MARFAN/T	AAD AND OTHER CONNECTIVE TISSUE TESTING OPTIO	NS					
🗖 Т998	Ehlers Danlos Sequencing and Del/Dup Panel	□ TA02	Stickler Syndrome Sequencing and Del/Dup Panel				
883	Marfan/TAAD Sequencing and Del/DupPanel	🗖 J555	Heritable Disorders of Connective Tissue (HDCT) Sequencing				
□ 883RE	Reflex to Rest of Heritable Disorders of Connective Tissue after Marfan/TAAD Panel		and Del/Dup Pane				
918	FBNI Sequencing and Del/Dup	919	Rest of Marfan/TAAD Sequencing 25 and Del/Dup if Test #918 is negative				

GeneDx tests are frequently updated and improved based upon the most recent scientific evidence. The test codes, genes, and gene quantities listed on this test requisition are subject to change by GeneDx at any time. The most current test menu, list of genes, and technical limitations included for a specific test panel may be found on our website, genedx.com. Please note that GeneDx reserves the right to modify and upgrade any ordered panel to the version currently listed on our website.

First Name

Last Name

Date of Birth

Gene

	TEST MENU	(continued)			
TEST CODE	TEST NAME	TEST CODE	TEST NAME		
OTHER CAR	DIAC-RELATED GENETIC TESTING OPTIONS				
697	HHT Sequencing and Del/Dup Panel	□ TA06	Noonan and RASopathies Sequencing and Del/Dup Panel		
CUSTOM DE	EL/DUP TESTING				
□ 906	Deletion/Duplication Analysis of ONE Nuclear Gene	□ 703	Deletion/Duplication Analysis of 2-20 Nuclear Genes		
Write-in Desired Gene(s) to be Tested:					
WRITE-IN TEST SELECTION					
Test Code: Test Name:					
"Rest of" panels are not a stand alone tests and must be ordered in conjunction with the original parent panel.					

			c	USTOM CARI	DIOLOGY PAN	EL			
□ J779	Create your ow	n panel by choc	sing from the Cu	ustom Cardiolog	y Gene List below	I			
	CUSTOM CARDIOLOGY GENE LIST								
Primary Disease	e Genes on the Co	ardiogenetics M	enu						
<ul> <li>□ ABCC9</li> <li>□ ACTA2</li> <li>□ ACTA2</li> <li>□ ACTC1</li> <li>□ ACTN2</li> <li>□ ACVR11</li> <li>□ ADAMTS2</li> <li>□ ALPH3A1</li> <li>□ BAG31</li> <li>□ BAG3</li> </ul>	□         BGN           □         BMPR2           □         BRAF           □         CACNAIC           □         CACNAIC           □         CACNA2DI           □         CACNA2DI           □         CACNB2           □         CALM1           □         CALM2           □         CALM3           □         CASQ2           □         CAVI           □         CAV3           □         CBS           □         CHRM2           □         COLIIA1           □         COLIIA2           □         COLIA2	□ COL2A1 □ COL3A1 □ COL5A1 □ COL5A2 □ COL9A2 □ COL9A3 □ COL9A3 □ CRYAB □ CSRP3 □ CTNNA3 □ DES □ DMD □ DOLK □ DSC2 □ DSE □ DSC2 □ DSC2	□ ELN □ EMD □ ENG □ EYA4 □ FBLN5 □ FBN1 □ FBN2 □ FHL1 □ FKRP* □ FKRP* □ FLNA □ FLNA □ FLNA □ GATA4 □ GATA4 □ GATA6 □ GATAD1 □ GDF2 □ GJA5	□ GLA □ GNB5 □ GPD1L □ HCN4 □ HFE □ HRAS* □ ILK □ JPH2 □ JUP □ KCNA5 □ KCNE3 □ KCNE1 □ KCNE3 □ KCNE3 □ KCNE3 □ KCNE2 □ KCNE2 □ KCNJ2 □ KCNJ5	<pre>     KCNJ8     KCNK3     KCNQ1     KRAS     LAMA4     LAMP2     LDB3     LDLR     LDLRAP1     LMNA     LOX     LRRC10     LTBP4     MAP2K1     MAP2K1     MAP2K2     MAT2A     MAFD5     MIB1     MURC</pre>	■ MYBPC3         ■ MYH1         ■ MYH6         ■ MYH7         ■ MYL3         ■ MYL4         ■ MYL5         ■ NK25         ■ NKX2-55         ■ NKX2-55         ■ NKX2-55         ■ NRAS         ■ PCSK9         ■ PDLIM3         ■ PKP2         ■ PLN	□ PLOD1 □ PPA2 □ PRDM16 □ PRCM16 □ PRKG2 □ PRKG1 □ PTPN11 □ PYCR1 □ RAF1 □ RASA1 □ RBM20 □ RIN2 □ RIN2 □ RIN2 □ RIT1 □ RYR2 □ SCN10A □ SCN18^ □ SCN28 □ SCN38 □ SCN48	□ SCN5A □ SGCD □ SHOC2 □ SKI □ SLC2A10 □ SLC39A13 □ SMAD2 □ SMAD3 □ SMAD4 □ S	□ TMEM43 □ TMPO □ TNNC1 □ TNNI3 □ TNNT2 □ TNXB □ TOR1AIP1 □ TPM1 □ TRPM4 □ TTN □ TTR □ TTR □ TXNRD2 □ VCL □ ZNF469
Expanded Phen	/1								
□ ABCC6 □ ACADVL □ ACTA1 □ ACVR1 □ ADAMTS10	□ ADAMTS17 □ ADAMTSL4 □ AGL □ ASPH □ COL4A1	□ COX15 □ CPT1A □ CTF1* □ DNAJC19 □ ELAC2	☐ FOXC2 ☐ FOXF1 ☐ FOXRED1 ☐ GLB1 ☐ JAG1	<ul> <li>☐ KLF10</li> <li>☐ LTBP2</li> <li>☐ LZTR1</li> <li>☐ MRPS22</li> <li>☐ MTO1</li> </ul>	☐ MYOMI ☐ NPPA ☐ PLEC ☐ RRAS ☐ SCARF2	□ SCO2* □ SLC25A20 □ SLC25A3 □ SLC25A4 □ SMS	□ SOS2 □ SYNE1 □ SYNE2 □ TBX1* □ TBX5	☐ TMEM70 ☐ TRIM63 ☐ TSFM ☐ UPF3B ☐ XK	
Limited Evidence Genes									
☐ AEBP1 ☐ ATP6VOD2 ☐ BMPRIB ☐ CALR	□ CALR3 □ CBLN2 □ COA5^ □ FGF12	□ FHL2 □ FHOD3 □ FOXE3* □ KCNB2	□ KCNJ16 □ KCNT1^ □ LZTS1 □ MAP3K8	☐ MRPL3 ☐ MYO6 ☐ NKX2-6 ☐ NOS1AP	☐ PI4KA ☐ PLEKHM2 ☐ PLOD3 ☐ RASA2	□ SCN4A □ SCNNIA □ SLC2A5 □ SLMAP	□ SMAD1 □ SMAD6 □ SPRY1 □ TAB2		
* Del/Dup analysis not offered ^ Gene level resolution; may not detect exon level events									

### DID YOU REMEMBER TO ...?

□ Label specimen tube appropriately with TWO identifiers

 $\hfill\square$  Get a signature for medical necessity and patient consent

GeneDx tests are frequently updated and improved based upon the most recent scientific evidence. The test codes, genes, and gene quantities listed on this test requisition are subject to change by GeneDx at any time. The most current test menu, list of genes, and technical limitations included for a specific test panel may be found on our website, genedx.com. Please note that GeneDx reserves the right to modify and upgrade any ordered panel to the version currently listed on our website.

# **INFORMED CONSENT**

First Name	Last Name	Date of Birth

For the purposes of this consent, "I", "my", and "your" will refer to me or to my child, including my unborn child, if my child is the person for whom the healthcare provider has ordered testing.

#### PURPOSE OF THIS TEST

The purpose of this test is (a) to see if I may have a genetic variant or chromosome rearrangement causing a genetic disorder; or (b) to evaluate the chance that I will develop or pass on a genetic disorder in the future. If I already know the specific gene variant(s) or chromosome rearrangement that causes the genetic disorder in my family, I agree to inform the laboratory of this information.

### WHAT TYPE OF TEST RESULTS CAN I EXPECT FROM GENETIC TESTING?

- 1. <u>Positive</u>: A change in your DNA was found, which is very likely the cause of your features/symptoms. This is the most straightforward test result, which can be used as the basis to test other family members to determine their chances of having either the disease or a child with the disease.
- 2. <u>Negative</u>: No variants were found to explain your symptoms. This does not mean that you do not have a genetic condition. It is still possible that there is a genetic variant not found by the test that was ordered. Your healthcare provider or genetic counselor may discuss more testing either now or in the future.
- 3. Variant of Uncertain Significance (VUS): A change in a gene was found. However, we are not sure whether this variant is the cause of your symptoms/features. More information is needed. We may suggest testing other family members to help figure out the meaning of the test result.
- 4. <u>Unexpected Results</u>: In rare instances, this test may reveal an important genetic change that is not directly related to the reason for ordering this test. For example, this test may find you are at risk for another genetic condition I am not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. We may disclose this information to the ordering healthcare provider if it likely affects medical care.

Because medical and scientific knowledge is constantly changing, new information that becomes available may supplement the information GeneDx used to interpret my results. Healthcare providers can contact GeneDx at any time to discuss the classification of an identified variant.

#### WHAT IS TRIO/DUO-BASED GENETIC TESTING?

For some genetic tests, including samples from the biological parents and/or other biological relatives along with the patient's sample can help with the interpretation of the test results. These tests are often referred to as "trio tests" since they typically include samples from the patient and both parents.

Samples from relatives should be submitted with the patient's sample. Clinical information must be provided for the patient and any relative who submits a sample.

I understand that GeneDx will use the relative sample(s) when needed for the interpretation of my test results and that my test report may include clinical and genetic information about a relative when it is relevant to the interpretation of the test results. I further understand that relatives will not receive an independent analysis of data nor a separate report.

### **RISKS AND LIMITATIONS OF GENETIC TESTING**

- 1. In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology.
- 2. Accurate interpretation of test results may require knowing the true biological relationships in a family. I understand that if I fail to accurately state the biological relationships in my family, it could lead to incorrect interpretation of the test results, incorrect diagnoses, and/or inconclusive test results. If genetic testing reveals that the true biological relationships in a family are not as I reported them, including non-paternity (the reported father is not the biological father) and
- consanguinity (the parents are related by blood), I agree to have these findings reported to the healthcare provider who ordered the test. 3. Although genetic testing is highly accurate, inaccurate results may occur. These reasons include, but are not limited to mislabeled samples, inaccurate reporting of
- clinical/medical information, rare technical errors, or other reasons. 4. I understand that this test may not detect all of the long-term medical risks that I might experience. The result of this test does not guarantee my health and that
- additional diagnostic tests may still need to be done. 5. I agree to provide an additional sample if the initial sample is not adequate.

### PATIENT CONFIDENTIALITY AND GENETIC COUNSELING

It is recommended that I receive genetic counseling before and after having this genetic test. I can find a genetic counselor in my area at www.nsgc.org. Further testing or additional consultations with a healthcare provider may be necessary.

To maintain confidentiality, test results will only be released to the referring healthcare provider, the ordering laboratory, to me, to other healthcare providers involved in my care, diagnosis and treatment, or to others with my consent or as permitted or required by law. Federal laws prohibit unauthorized disclosure of this information. More information can be found at: www.genome.gov/10002077

### INTERNATIONAL SAMPLES

If I reside outside the United States, I attest that by providing a sample for testing, I am not knowingly violating any export ban or other legal restriction in the country of my residence.

### SAMPLE RETENTION

After testing is complete, my sample may be de-identified and be used for test development and improvement, internal validation, quality assurance, and training purposes. GeneDx will not return DNA samples to you or to referring healthcare providers, unless specific prior arrangements have been made.

I understand that samples from residents of New York State will not be included in the de-identified research studies described in this authorization and GeneDx will not retain them for more than 60 days after test completion, unless specifically authorized by my selection. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language. GeneDx will not perform any tests on the biological sample other than those specifically authorized.

### DATABASE PARTICIPATION

De-identified health history and genetic information can help healthcare providers and scientists understand how genes affect human health. Sharing this deidentified information helps healthcare providers to provide better care for their patients and researchers to make new discoveries. GeneDx shares this type of information with healthcare providers, scientists, and healthcare databases. GeneDx will not share any personally identifying information and will replace the identifying information with a unique code not derived from any personally identifying information. Even with a unique code, there is a risk that I could be identified based on the genetic and health information that is shared. GeneDx believes that this is unlikely, though the risk is greater if I have already shared my genetic or health information with public resources, such as genealogy websites.

Gene

# **INFORMED CONSENT**

First Name

Last Name

Date of Birth

Gene

#### EXOME/GENOME SEQUENCING SECONDARY FINDINGS

· Applicable only for full exome sequencing and genome sequencing tests

• Does not pertain to Xpanded® or Slice tests

As many different genes and conditions are analyzed in an exome or genome sequencing test, these tests may reveal some findings not directly related to the reason for ordering the test. Such findings are called "incidental" or "secondary" and can provide information that was not anticipated.

Secondary findings are variants, identified by an exome or genome sequencing test, in genes that are unrelated to the individual's reported clinical features.

The American College of Medical Genetics and Genomics (ACMG) has recommended that secondary findings identified in a specific subset of medically actionable genes associated with various inherited disorders be reported for all probands undergoing exome or genome sequencing. Please refer to the latest version of the ACMG recommendations for reporting of secondary findings in clinical exome and genome sequencing for complete details of the genes and associated genetic disorders. Reportable secondary findings will be confirmed by an alternate test method when needed.

#### WHAT WILL BE REPORTED FOR THE PATIENT?

All pathogenic and likely pathogenic variants associated with specific genotypes identified in the genes (for which a minimum of 10X coverage was achieved by exome sequencing), as recommended by the ACMG.

#### WHAT WILL BE REPORTED FOR RELATIVES?

The presence or absence of any secondary finding(s) reported for the proband will be provided for all relatives analyzed by an exome or genome sequencing test.

#### LIMITATIONS

Pathogenic and/or likely pathogenic variants may be present in a portion of the gene not covered by this test and therefore are not reported. The absence of reportable secondary findings for any particular gene does not mean there are no pathogenic and/or likely pathogenic variants in that gene. Pathogenic variants and/or likely pathogenic variants that may be present in a relative, but are not present in the proband, will not be identified nor reported. Only changes at the sequence level will be reported in the secondary findings report. Larger deletions/duplications, abnormal methylation, triplet repeat or other expansion variants, or other variants not routinely identified by clinical exome and genome sequencing will not be reported.

#### FINANCIAL AGREEMENT AND GUARANTEE

For insurance billing, I understand and authorize GeneDx to bill my health insurance plan on my behalf, to release any information required for billing, and to be my designated representative for purposes of appealing any denial of benefits. I irrevocably assign to and direct that payment be made directly to GeneDx.

I understand that my out-of-pocket costs may be different than the estimated amount indicated to me by GeneDx as part of a benefit investigation. I agree to be financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. If my insurance provider sends a payment directly to me for services performed by GeneDx on my behalf, I agree to endorse the insurance check and forward it to GeneDx within 30 days of receipt as payment towards GeneDx's claim for services rendered.

If I do not have health insurance, I agree to pay for the full cost of the genetic testing that was ordered by my healthcare provider and billed to me by GeneDx. I further understand and agree that, if I fail to make payment for genetic testing, in accordance with the payment policies of GeneDx, my account may be turned over to an external collection agency for non-payment. I agree to pay any associated collection costs, including attorney fees. By my signature on the GeneDx Test Requisition Form or at the bottom of this form, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider.

#### MEDICARE

A completed Advance Beneficiary Notice (ABN) is required for Medicare patients, when applicable. Please visit our website, www.genedx.com/billing for more information.