CARDIOLOGY TEST REQUISITION FORM (STANDARD)

GeneDx an OPKO Health Company

PATIENT INFORMATION

First Name	Last Name	
Genetic Sex ○ Male ○ Female Gender Identification (optional): _____	Date of Birth (mm/dd/yy)	
Ancestry ○ White/Caucasian ○ Hispanic ○ Black/African American ○ Native American ○ East Asian ○ South Asian ○ Middle Eastern ○ Ashkenazi Jewish ○ Other: _____		
Email		
Address		
City	State	Zip Code
Primary Phone	Is this patient deceased? ○ Yes ○ No Deceased Date: _____	

SAMPLE INFORMATION

Date Sample Collected (mm/dd/yy) (required):	Medical Record #
○ Blood ○ Buccal Swab ○ Other (specify source): _____	
Patient has had a blood transfusion ○ Yes ○ No Date of Last Transfusion: _____ (2-4 weeks of wait time is required for some testing)	
Patient has had an allogenic bone marrow transplant ○ Yes ○ No Fibroblasts are recommended for patients who had an allogenic bone marrow transplant. See www.genedx.com/specimen-requirements for details.	
○ Treatment-Related **RUSH**	Date: _____

PATIENT CONSENT FOR GENETIC TESTING, FINANCIAL AGREEMENT AND GUARANTEE:

By signing this form, I acknowledge as the patient or relative being tested that I have read the GeneDx Informed Consent document available from my healthcare provider or at genedx.com/forms, and I authorize GeneDx to perform genetic testing as ordered. I understand that, for tests that evaluate data from multiple family members concurrently, 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 my signature below, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider. 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 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 and agree to be financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. I am aware that my insurance provider may send 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. 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 and I agree to pay any associated collection costs, including attorney fees.

More information, including the GeneDx Notice of Privacy Policies, is available on GeneDx's website: www.genedx.com

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

○ 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 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 after 60 days and it cannot be used for the studies listed above.

○ Check this box if you wish to opt out of being contacted for research studies.

Signature of Patient/Guardian (required)	**Date**
Signature of Relative A	**Date**
Signature of Relative B	**Date**

ACCOUNT INFORMATION

GeneDx Account Number		**Account Name**
Phone		Fax
Address		City
State	Zip Code	Country
Ordering Provider Name		Role/Title
NPI		Phone Number
Send Report Via ○ Fax ○ Email ○ Portal Fax #/Email: _____		
Additional Reporting Provider's Name		
Send Report Via ○ Fax ○ Email ○ Portal Fax #/Email: _____		
SEND ADDITIONAL REPORT COPIES TO:		
Provider Name		GeneDx Acct#
Fax #/Email:		

ICD-10 CODES *(Required)*

ICD-10 Codes	
Clinical Diagnosis	Age of Onset

STATEMENT OF MEDICAL NECESSISTY

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 Provider (required)	Date

PAYMENT OPTIONS *(Select One)*

○ **INSURANCE BILL** *(select all that applies)*

○ Commercial
○ Medicaid
○ Medicare
○ Tricare

FOR ALL INSURANCE CARDS PROVIDE FRONT AND BACK COPY OF CARD(S)

Patient Status
○ Hospital outpatient ○ Hospital inpatient; Date of Discharge _____
○ Not a hospital patient

Name of Insurance Carrier		Insurance ID#:	
Relationship to Insured ○ Self ○ Spouse ○ Child ○ Other: _____		Policy Holder's Name Policy Holder's Date of Birth	
Referral/Prior Authorization # (please attach)		GeneDx Benefit Investigation #	
Secondary Insurance Type:			
Insurance Carrier	Insurance ID #	Subscriber Name	Date of Birth
Relationship to Insured: ○ Self ○ Spouse ○ Child ○ Other: _____			

○ **PATIENT BILL**

Amount Due: _____

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

○ **INSTITUTIONAL BILL**

GeneDx Account #

Hospital/Lab Name

Place Sticker/Stamp Here

CLINICAL INFORMATION

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

Is this person affected: ○ Yes ○ No Clinical diagnosis:_____

Reason for testing: ○ Diagnosis ○ Presymptomatic diagnosis ○ Carrier/Familial Variant Testing

Please check all that apply. This is not a substitute for submitting clinical records.

Diagnosis

- ○ Amyloidosis
- ○ ARVC
- ○ Brugada syndrome
- ○ CPVT
- ○ DCM
- ○ Ehlers-Danlos syndrome
- ○ HCM
- ○ HHT
- ○ Hypertension
- ○ Loeys-Dietz syndrome
- ○ LQT syndrome
- ○ Noncompaction cardiomyopathy (LVNC)
- ○ Marfan syndrome
- ○ PAH
- ○ RCM
- ○ SQT syndrome
- ○ Sudden Cardiac Arrest
- ○ Sudden Death

Echocardiogram

- ○ Aortic root dimensions: _____
  - ○ Z-score: _____
- ○ EF%: _____
- ○ LVEDD: _____
  - ○ Z-score: _____
- ○ Max LV wall thickness: _____
- ○ Normal
- ○ Report Included

ECG

- ○ Prolonged QTc interval:

  Max QTc: _____
- ○ Normal
- ○ Report Included

Arrhythmia/Cardiomyopathy

- ○ Abnormal atrioventricular conduction
- ○ Atrial fibrillation
- ○ Bradycardia
- ○ Fatty replacement of ventricular myocardial tissue
- ○ Heart transplant
- ○ Syncope
- ○ Torsades de pointe
- ○ Ventricular tachycardia

HHT

- ○ Arteriovenous malformation
- ○ Epistaxis
- ○ Telangiectasia

Dislipidemias

- ○ Atherosclerosis
- ○ Corneal Arcus
- ○ LDL-C levels _____
- ○ Xanthomatosis
- ○ Other: _____

Marfan/TAAD/HDCT

- ○ Aortic/Arterial aneurysm
- ○ Aortic/Arterial dissection
- ○ Aortic root dilation
- ○ Arachnodactyly
- ○ Arterial tortuosity/ectasia
- ○ Arthralgia
- ○ Atypical scarring of skin
- ○ Beighton score_____
- ○ Bifid uvula
- ○ Blue sclerae
- ○ Bruising susceptibility
- ○ Cleft lip
- ○ Cleft palate
- ○ Craniosynostosis
- ○ Cutis laxa
- ○ Dental crowding
- ○ Dural ectasia
- ○ Ectopia lentis
- ○ Flexion contracture
- ○ High palate
- ○ Hollow organ rupture:
  - ○ Uterine rupture ○ Intestinal perforation
  - ○ Other: _____
- ○ Hypertelorism
- ○ Joint contractures
- ○ Joint dislocations
- ○ Joint hypermobility
- ○ Meets Ghent criteria
- ○ Micrognathia / Retrognathia (circle what applies)
- ○ Midface retrusion
- ○ Mitral valve prolapse
- ○ Myopia
- ○ Osteoarthritis
- ○ Pectus carinatum
- ○ Pectus excavatum
- ○ Pes Planus
- ○ Pneumothorax
- ○ Recurrent fractures
- ○ Retinal detachment
- ○ Scoliosis/Kyphosis (circle what applies)
- ○ Skin findings, Specify: _____
- ○ Stroke
- ○ Tall stature
- ○ Velvety skin

Abnormal heart morphology

- ○ Bicuspid aortic valve
- ○ Coarctation of aorta
- ○ Heart murmur
- ○ Heterotaxy
- ○ Hypoplastic left heart
- ○ Mitral valve prolapse
- ○ Patent ductus arteriosus
- ○ Patent foramen ovale
- ○ Tetralogy of Fallot
- ○ Ventricular septal defect/Atrial septal defect
- ○ Other: _____

PAH

- ○ Pulmonary hypertension

Other

- ○ Abnormality of the periventricular white matter
- ○ Angiokeratomas
- ○ Anhydrosis
- ○ Café-Au-Lait Macules
- ○ Conductive hearing impairment:
  - ○ Sensorineural ○ Conductive
- ○ Craniosynostosis
- ○ Cystic hygroma
- ○ Downslanted palpebral fissures
- ○ Dysmorphic features:

  Describe: _____
- ○ Elevated CPK
- ○ Hypotonia
- ○ Increase nuchal translucenscy
- ○ Intellectual disability
- ○ Keratoconus
- ○ Muscle weakness
- ○ Myopathy
- ○ Renal insufficiency
- ○ Short neck
- ○ Thromboembolism

  Type: _____
- ○ Other: _____

Attach pedigree and/or include additional clinical information:

_____

_____

_____

_____

_____

_____
Signature of Provider (required) Date

CARDIOLOGY TEST REQUISITION FORM (STANDARD)

GeneDx an OPKO Health Company

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

REASON FOR EXPEDITED TESTING (REQUIRED)

○ Pregnancy (gestational age____weeks) ○ Transplantation ○ Other:_____

TARGETED VARIANT TESTING AND SPECIAL SERVICES

Individual to be tested: ○ Affected/Symptomatic ○ Unaffected/Asymptomatic

○ Known Familial Variant(s) in a Nuclear Gene	○ Targeted Mosaic Variant Testing **(Insurance Billing NOT Accepted; Patient Bill or Institutional Bill MUST be selected on page 1)**
○ Known Familial Copy Number variant(s)	○ Known mtDNA Variant(s) Testing (heteroplasmy detection range 25%-100%)
○ Confirmation of Variant Identified in Research Lab	○ Known mtDNA Variant(s) Testing by NGS (heteroplasmy detection range 1.5%-100%) - Test Code 453

Proband Name: _____ Relationship to Proband: _____ Proband GeneDx Accession #: _____

Non-GeneDx Test: ○ Family member test report included (recommended if previous test was performed at another lab)
○ Positive control included/will be sent - **Positive control is recommended if previous test was performed at another lab.**
○ Positive control not available (caveat language will be included on a negative report)

Variant Information (please fill out the below information if family member report is not included)

Number of Variants:_____

Gene: _____ Coding DNA (c./m.): _____ Amino Acid (p.): _____ Transcript (NM#): _____

Gene: _____ Coding DNA (c./m.): _____ Amino Acid (p.): _____ Transcript (NM#): _____

Copy Number Variants (CNV(s) require coordinates and genome build or transcript # and exon #)

Number of Variants:_____

Gene(s): _____ Exon #: _____ Coordinates: _____ Genome Build: _____

Gene(s): _____ Exon #: _____ Coordinates: _____ Genome Build: _____

TESTING OPTIONS

CUSTOM DEL/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: _____

HISTORY

FAMILY HISTORY

○ No Known Family History ○ Pedigree Attached ○ Adopted

RELATIONSHIP TO INDIVIDUAL TO BE TESTED	MATERNAL	PATERNAL	RELEVANT HISTORY	AGE AT DX
	○	○		
	○	○		
	○	○		

TESTING HISTORY

○ Test report included (recommended)

Other relevant results (clinical, laboratory/biochemical or research):_____

CARDIOLOGY TEST REQUISITION FORM (STANDARD)

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

TEST MENU

TEST CODE	TEST NAME	# GENES	GENE LIST
ARRHYTHMIA TESTING OPTIONS			
○ 695	Arrhythmia Sequencing and Del/Dup Panel	58	*ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1^, CALM2, CALM3, CASQ2, CAV3, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, GATA4, GATA5^, GATA6, GJA5, GNB5, GPD1L, HCN4, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE1L (KCNE5), KCNH2 (HERG), KCNJ2, KCNJ5, KCNJ8, KCNQ1, LDB3, LMNA, MYL4, NKX2-5, PKP2, PLN, PPA2, RANGRF, RYR2, SCN10A, SCN1B^, SCN2B, SCN3B, SCN4B, SCN5A, SNTA1, TECRL, TGFB3, TMEM43, TRDN, TRPM4, TTN*
○ 695RE	Reflex to Rest of Combined Cardiac after Arrhythmia Panel		
○ 483	ARVC Sequencing and Del/Dup Panel	16	*CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, PKP2, PLN, RYR2, SCN5A, TGFB3, TMEM43, TTN*
○ 483RE	Reflex to Rest of Combined Cardiac after ARVC Panel		
○ TA12	SCN5A-related Brugada syndrome	1	*SCN5A*
○ 481	Brugada syndrome Sequencing and Del/Dup Panel	17	*ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNH2 (HERG), KCNJ8, PKP2, SCN10A, SCN1B^, SCN2B, SCN3B, SCN5A, TRPM4*
○ 481RE	Reflex to Rest of Arrhythmia after Brugada Syndrome Panel		
○ 482	CPVT Sequencing and Del/Dup Panel	9	*ANK2, CALM1^, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN*
○ 482RE	Reflex to Rest of Arrhythmia after CPVT Panel		
○ 727	LQTS Sequencing and Del/Dup Panel	17	*AKAP9, ANK2, CACNA1C, CALM1^, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2 (HERG), KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN*
○ 727RE	Reflex to Rest of Arrhythmia after LQTS Panel		
○ J552	SCA Arrhythmia Sequencing and Del/Dup Panel	14	*ANK2, CALM1^, CALM2, CALM3, CASQ2, CAV3, KCNE1, KCNE2, KCNH2 (HERG), KCNJ2, KCNQ1, PPA2, RYR2, SCN5A*
○ J552RE	Reflex to Rest of Arrhythmia after SCA Arrhythmia Panel		
○ J551	SQTS Sequencing and Del/Dup Panel	5	*CACNA1C, CACNB2, KCNH2 (HERG), KCNJ2, KCNQ1*
CARDIOMYOPATHY TESTING OPTIONS			
○ 694	Cardiomyopathy Sequencing and Del/Dup Panel	102	*ABCC9, ACTC1, ACTN2, AKAP9, ALMS1, ALPK3, ANKRD1, BAG3, BRAF, CAV3, CHRM2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FKRP*, FKTN, FLNC, GAA, GATA4, GATAD1, GLA, HCN4, HFE, HRAS*, ILK, JPH2, JUP, KRAS, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MIB1, MTND1*, MTND5*, MTND6*, MTTD*, MTTG*, MTTH*, MTTI*, MTTK*, MTTL1*, MTTL2*, MTTM*, MTTQ*, MTTS1*, MTTS2*, MURC, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NRAS, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SGCD, SHOC2, SOS1, TAZ^, TBX20^, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TOR1AIP1, TPM1, TTN, TTR, TXNRD2, VCL*
○ 694RE	Reflex to Rest of Combined Cardiac after Cardiomyopathy Panel		
○ TF66	Pediatric Cardiomyopathy	100	*ABCC9, ACTC1, ACTN2, AKAP9, ALMS1, ALPK3, ANKRD1, BAG3, BRAF, CAV3, CHRM2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FKRP*, FKTN, FLNC, GAA, GATA4, GATAD1, GLA, HCN4, HRAS*, ILK, JPH2, JUP, KRAS, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MIB1, MTND1*, MTND5*, MTND6*, MTTD*, MTTG*, MTTH*, MTTI*, MTTK*, MTTL1*, MTTL2*, MTTM*, MTTQ*, MTTS1*, MTTS2*, MURC, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NRAS, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SGCD, SHOC2, SOS1, TAZ^, TBX20^, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TOR1AIP1, TPM1, TTN, TXNRD2, VCL*
○ TF66RE	Reflex to Rest of Pediatric Combined Cardiac after Pediatric Cardiomyopathy		
○ 483	ARVC Sequencing and Del/Dup Panel	16	*CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, PKP2, PLN, RYR2, SCN5A, TGFB3, TMEM43, TTN*
○ 483RE	Reflex to Rest of Combined Cardiac after ARVC Panel		
○ J554	DCM/LVNC Sequencing and Del/Dup Panel	68	*ABCC9, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CHRM2, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, FKTN, FLNC, GATAD1, HCN4, ILK, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MIB1, MTND1*, MTND5*, MTND6*, MTTD*, MTTG*, MTTH*, MTTI*, MTTK*, MTTL1*, MTTL2*, MTTM*, MTTQ*, MTTS1*, MTTS2*, MYBPC3, MYH6, MYH7, MYPN, NEBL, NEXN, NKX2-5, PLN, PRDM16, RAF1, RBM20, RYR2, SCN5A, SGCD, TAZ^, TBX20^, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL*
○ J554RE	Reflex to Rest of Cardiomyopathy after DCM Panel		
○ J553	HCM Sequencing and Del/Dup Panel	42	*ACTC1, ACTN2, ALPK3, CAV3, CSRP3, FHL1, FLNC, GAA, GLA, JPH2, LAMP2, MTND1*, MTND5*, MTND6*, MTTD*, MTTG*, MTTH*, MTTI*, MTTK*, MTTL1*, MTTL2*, MTTM*, MTTQ*, MTTS1*, MTTS2*, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOZ2, PLN, PRKAG2, RAF1, RIT1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL*
○ J553RE	Reflex to Rest of Cardiomyopathy after HCM Panel		

CARDIOLOGY TEST REQUISITION FORM (STANDARD)

GeneDx an OPKO Health Company

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

TEST MENU

TEST CODE	TEST NAME	# GENES	GENE LIST
COMBINED CARDIAC PANEL			
○ 935	Combined Cardiac Panel	138	*ABCC9, ACTC1, ACTN2, AKAP9, ALMS1, ALPK3, ANK2, ANKRD1, BAG3, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1^, CALM2, CALM3, CASQ2, CAV3, CHRM2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FKRP*, FKTN, FLNC, GAA, GATA4, GATA5^, GATA6, GATAD1, GJA5, GLA, GNB5, GPD1L, HCN4, HFE, HRAS*, ILK, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE1L (KCNE5), KCNH2 (HERG), KCNJ2, KCNJ5, KCNJ8, KCNQ1, KRAS, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MIB1, MTND1*, MTND5*, MTND6*, MTTD*, MTTG*, MTTH*, MTTI*, MTTK*, MTTL1*, MTTL2*, MTTM*, MTTQ*, MTTS1*, MTTS2*, MURC, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NRAS, PDLIM3, PKP2, PLN, PPA2, PRDM16, PRKAG2, PTPN11, RAF1, RANGRF, RBM20, RIT1, RYR2, SCN10A, SCN1B^, SCN2B, SCN3B, SCN4B, SCN5A, SGCD, SHOC2, SNTA1, SOS1, TAZ^, TBX20^, TCAP, TECRL, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TOR1AIP1, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL*
○ TF67	Pediatric Combined Cardiac	136	*ABCC9, ACTC1, ACTN2, AKAP9, ALMS1, ALPK3, ANK2, ANKRD1, BAG3, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1^, CALM2, CALM3, CASQ2, CAV3, CHRM2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FKRP*, FKTN, FLNC, GAA, GATA4, GATA5^, GATA6, GATAD1, GJA5, GLA, GNB5, GPD1L, HCN4, HRAS*, ILK, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE1L (KCNE5), KCNH2 (HERG), KCNJ2, KCNJ5, KCNJ8, KCNQ1, KRAS, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MIB1, MTND1*, MTND5*, MTND6*, MTTD*, MTTG*, MTTH*, MTTI*, MTTK*, MTTL1*, MTTL2*, MTTM*, MTTQ*, MTTS1*, MTTS2*, MURC, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NRAS, PDLIM3, PKP2, PLN, PPA2, PRDM16, PRKAG2, PTPN11, RAF1, RANGRF, RBM20, RIT1, RYR2, SCN10A, SCN1B^, SCN2B, SCN3B, SCN4B, SCN5A, SGCD, SHOC2, SNTA1, SOS1, TAZ^, TBX20^, TCAP, TECRL, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TOR1AIP1, TPM1, TRDN, TRPM4, TTN, TXNRD2, VCL*
LIPIDEMIAS TESTING			
○ J556	Familial Hypercholesterolemia Sequencing and Del/Dup Panel	4	*APOB, LDLR, LDLRAP1, PCSK9*
○ TA01	Familial Dyslipidemia Sequencing and Del/Dup Panel	28	*ABCA1, ABCG5, ABCG8, ANGPTL3, APOA1^, APOA5, APOB, APOC2, APOC3, APOE, CETP, CYP27A1, CYP7A1, GCKR*, GPD1, GPIHBP1, LCAT^, LDLR, LDLRAP1, LIPA, LIPC, LMF1^, LPL, MTTP, PCSK9, SAR1B, SCARB1, STAP1*
MARFAN/TAAD AND OTHER CONNECTIVE TISSUE TESTING			
○ T999	Cutis Laxa Sequencing and Del/Dup Panel	11	*ALDH18A1, ATP6V0A2, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP4, PYCR1, RIN2, SLC2A10*
○ T998	Ehlers Danlos Sequencing and Del/Dup Panel	3	*COL3A1, COL5A1, COL5A2*
○ 918	FBN1 Sequencing and Del/Dup	1	*FBN1*
○ 919	Rest of Marfan/TAAD Sequencing and Del/Dup if Test #918 is negative	25	*ACTA2, BGN, CBS, COL3A1, COL5A1, COL5A2, FBN2, FLNA, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2*
○ 883	Marfan/TAAD Sequencing and Del/Dup Panel	26	*ACTA2, BGN, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2*
○ 883RE	Reflex to Rest of Heritable Disorders of Connective Tissue after Marfan/TAAD Panel		
○ TA02	Stickler Syndrome Sequencing and Del/Dup Panel	6	*COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2*
○ J555	Heritable Disorders of Connective Tissue (HDCT) Sequencing and Del/Dup Panel	57	*ACTA2, ADAMTS2, ALDH18A1, ATP6V0A2, ATP6V1E1, ATP7A, B3GALT6*, B3GAT3, B4GALT7, BGN, CBS, CHST14, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, COL12A1, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LOX, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, ZNF469*
OTHER CARDIAC-RELATED GENETIC TESTS			
○ 697	HHT Sequencing and Del/Dup Panel	5	*ACVRL1, ENG, GDF2, RASA1, SMAD4*
○ 696	PAH Sequencing and Del/Dup Panel	8	*ACVRL1, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNK3, SMAD9*
○ TA06	Noonan and RASopathies Sequencing and Del/Dup Panel	25	*A2ML1, ACTB^, ACTG1, BRAF, CBL, HRAS*, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*
○ 363	Cardiac Amyloidosis (TTR gene sequencing)	1	*TTR*
○ 910	Chromosomal Microarray (GenomeDx®)		
ADDITIONAL TESTS			
○	Test name:		

"Rest of" panels should be ordered at the time of an initial order and not as a stand-alone test

* 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
- ○ Get a signature for medical necessity and patient consent
- ○ Fill out sample submission form (pages 4 and 5)
- ○ Complete clinical information (page 2)
- ○ Complete payment form (page 1)

INFORMED CONSENT

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

General Information About Genetic Testing

What is genetic testing?

DNA provides instructions for our body's growth and development. Genes are distinct sequences of DNA, and are arranged on chromosomes. The DNA in a gene contains instructions for making proteins, which determine things like growth and metabolism as well as traits like eye color and blood type. Genetic disorders are caused by certain changes in DNA affecting the structure or number of chromosomes. Genetic testing is a laboratory test that tries to identify these changes in chromosomes or the DNA. Genetic testing can be a diagnostic test, which is used to identify or rule out a specific genetic condition. Genetic screening tests are used to assess the chance for a person to develop or have a child with a genetic condition. Genetic screening tests are not typically diagnostic and results may require additional testing.

The purpose of this test is to see if I, or my child, may have a genetic variant or chromosome rearrangement causing a genetic disorder or to determine the chance that I, or my child, will develop or pass on a genetic disorder in the future. 'My child' can also mean my unborn child, for the purposes of this consent.

If I/my child already know the specific gene variant(s) or chromosome rearrangement that causes the genetic disorder in my family, I will inform the laboratory of this information.

What could I learn from this genetic test?

The following describes the possible results from the test:

1) Positive: A positive result indicates that a genetic variant has been identified that explains the cause of my/my child's genetic disorder or indicates that I/my child am at increased risk to develop the disorder in the future. It is possible to test positive for more than one genetic variant.

2) Negative: A negative result indicates that no disease-causing genetic variant was identified by the test performed. It does not guarantee that I/my child will be healthy or free from genetic disorders or medical conditions. If I/my child test negative for a variant known to cause the genetic disorder in other members of my/my child's family, this result rules out a diagnosis of the same genetic disorder in me/my child due to this specific change.

3) Inconclusive/Variant of Uncertain Significance (VUS): A finding of a variant of uncertain significance indicates that a genetic change was detected, but it is currently unknown whether that change is associated with a genetic disorder either now or in the future. A variant of uncertain significance is not the same as a positive result and does not clarify whether I/my child is at increased risk to develop a genetic disorder. The change could be a normal genetic variant or it could be disease-causing. Further analysis may be recommended, including testing parents and other family members. Detailed medical records or information from other family members also may be needed to help clarify results.

4) Unexpected results: 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 tell me about the risk for another genetic condition I/my child is not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. This information may be disclosed to the ordering health care provider if it likely impacts medical care.

Result interpretation is based on currently available information in the medical literature, research and scientific databases. Because the literature, medical and scientific knowledge are constantly changing, new information that becomes available in the future may replace or add to the information GeneDx used to interpret my/my child's results. Providers can contact GeneDx at any time to discuss the classification of an identified variant. In addition, I or my/my child's health care providers may monitor publicly available resources used by the medical community, such as ClinVar (www.clinvar.com), to find current information about the clinical interpretation of my/my child's variant(s).

For tests that evaluate data from multiple family members, my spouse, or partner concurrently, results may be included in a single comprehensive report.

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 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/my child's test results. The patient report may include clinical and genetic information about a relative when it is relevant to the interpretation of the results. Relatives do not receive an independent analysis of data nor a separate report.

What are the risks and limitations of this genetic test?

- Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer. 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.
- Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in my/my child's family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results. In some cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. This includes non-paternity (the stated father of an individual is not the biological father) and consanguinity (the parents of an individual are related by blood). It may be necessary to report these findings to the health care provider who ordered the test.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to: mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, or the presence of change(s) in such a small percentage of cells that the change(s) may not be detectable by the test (mosaicism).
- This test does not have the ability to detect all of the long-term medical risks that I/my child might experience. The result of this test does not guarantee my health or the health of my child/fetus. Other diagnostic tests may still need to be done, especially when only a genetic screening test has been performed previously.
- Occasionally, an additional sample may be needed if the initial specimen 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 here: www.nsgc.org. Further testing or additional consultations with a health care provider may be necessary.

To maintain confidentiality, the test results will only be released to the referring health care provider, to the ordering laboratory, to me, to other health care providers involved in my/my child's diagnosis and treatment, or to others as entitled by law. The United States Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, I understand that I can visit www.genome.gov/10002077.

International Specimens

If I/my child 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/my child's residence.

Additional information about the specific test being ordered is available from my health care provider or I can go to the GeneDx website, www.genedx.com.This information includes the complete gene lists, the specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, the limitations of genetic testing, as well as information about how specimens and information are stored and used.

Specimen Retention

After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring heath care 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 will not be retained 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. No tests other than those authorized shall be performed on the biological sample.

Database Participation

De-identified heath history and genetic information can help health care providers and scientists understand how genes affect human health. Though {I/my child} may not personally benefit, sharing this information helps health care providers to provide better care for their patients and researchers to make discoveries. GeneDx shares this type of information with health care providers, scientists and health care databases. No personal identifying information will be shared, as it will be replaced with a unique code.

Even though only a code is used for the reporting to the database, there is a risk that {I/my child} 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/my child's} genetic or health information with public resources, such as genealogy websites.

Recontact for Research Participation

Separate from the above, GeneDx may collaborate with scientists, researchers and drug developers to advance knowledge of genetic diseases and to develop new treatments. If there are opportunities to participate in research relevant to the disorder in {my/my child's} family, and if I have consented for recontact, GeneDx may allow my healthcare provider to be recontacted for research purposes, such as the development of new testing, drug development, or other treatment modalities. In some situations, such as if my health care provider is not available, I may be contacted directly.

Any research that results in medical advances, including new products, tests or discoveries, may have potential commercial value and may be developed and owned by GeneDx or the collaborating researchers. If any individuals or corporations benefit financially from these studies, no compensation will be provided to {my/my child} or {my/my child's} heirs.