

## Patient Information

Patient First Name: \_\_\_\_\_ MI \_\_\_\_\_ Last Name \_\_\_\_\_  
 Gender:  Male  Female Date of Birth: (mm/dd/yy) | Date of Death: (mm/dd/yy)  
 Patient Address: Street \_\_\_\_\_ Apt # \_\_\_\_\_  
 City/State/Zip Code \_\_\_\_\_  
 Patient Phone: Home \_\_\_\_\_ Work Phone \_\_\_\_\_  
 Medical Record Number \_\_\_\_\_

## Ordering Physician Information

Physician/Genetic Counselor \_\_\_\_\_  
 NPI # \_\_\_\_\_ Specialty (Example: Genetics or Cardiology) \_\_\_\_\_  
 Institution \_\_\_\_\_  
 Street Address \_\_\_\_\_  
 City/State/Zip Code \_\_\_\_\_  
 Phone \_\_\_\_\_ Fax \_\_\_\_\_  
 Signature \_\_\_\_\_ Email \_\_\_\_\_

## Additional Reporting Address

Physician/Genetic Counselor \_\_\_\_\_  
 Mailing address \_\_\_\_\_  
 City \_\_\_\_\_ State \_\_\_\_\_ Zip code \_\_\_\_\_  
 Phone \_\_\_\_\_ Fax (important) \_\_\_\_\_  
 Email \_\_\_\_\_ Beeper \_\_\_\_\_

## ICD 9 Codes

- 425.11: HCM, Obstructive
- 425.18: HCM, Non-obstructive
- 425.40: Cardiomyopathy, Familial
- 427.70: Metabolic Cardiomyopathy
- 425.80: Syndromic Cardiomyopathy
- 426.82: Long QT syndrome
- 427.9: Cardiac dysrhythmia, unspecified
- 746.89: Brugada Syndrome
- 759.82: Marfan Syndrome
- 794.31: Abnormal EKG
- V18.9: Family member is a carrier of a genetic disease
- Other: \_\_\_\_\_

## Specimen Information

- Date obtained: \_\_\_\_\_
- 2-5 ml whole blood in EDTA in one lavender/purple top tube
  - DNA from \_\_\_\_\_ (tissue type?) (Concentration: \_\_\_\_\_ µg/mL)
  - Saliva (not accepted for Marfan syndrome)
  - Other: \_\_\_\_\_
  - Sample tubes labeled with patient information

## Patient Consent

I have read the Informed Consent document and I give permission to GeneDx to perform genetic testing as described.  
 I also give permission for my specimen and clinical information to be used in cardiology and/or genetic research studies. My name will not be used in any research studies, and it will not be possible to link results of research studies back to my specimen.  
 Check this box, if you wish to opt out of research studies.  
 I will cooperate fully with GeneDx by providing all necessary documents needed for insurance billing and appeals. I understand that I am responsible for sending GeneDx any and all of the money that I receive directly from my insurance company in payment for this test. Reasonable collection and/or attorney's fees, including filing and service fees, shall be assessed if the account is sent to collection but said fees shall not exceed those permitted by state law.  
 Patient Sign Here: \_\_\_\_\_

## Clinical Information

Age at diagnosis: \_\_\_\_\_ Years Ethnicity: \_\_\_\_\_  
 Family member(s) affected:  Yes  No  
 Relationship(s): \_\_\_\_\_

Syncope  Yes  No Number of Episodes \_\_\_\_\_  
 Cardiac Arrest/SCD  Yes  No

## Patient's Diagnostic Tests (Please provide copies)

Electrocardiogram	Maximum QTc interval _____ msec
Echocardiogram	Maximum LV wall thickness: _____ mm EF% _____ Left ventricular internal diastolic dimension: _____ mm
Cardiac MRI	Maximum LV wall thickness: _____ mm EF% _____ Left ventricular internal diastolic dimension: _____ mm
RV fatty infiltrate	<input type="checkbox"/> Yes <input type="checkbox"/> No

## Cardiology Genetic Testing Panels

### Sequencing Tests

- 333: **HCM (18 Genes)** ACTC1, CAV3, GLA, LAMP2, MTTG, MTTI, MTTK, MTTQ, MYBPC3, MYH7, MYL2, MYL3, PRKAG2, TNNC1, TNNI3, TNNT2, TPM1, TTR
- 350: **DCM/LVNC (27 Genes)** ACTC1, DES, LAMP2, LMNA, MTND1, MTND5, MTND6, MTTD, MTTT, MTTI, MTTK, MTTL1, MTTL2, MTTM, MTTQ, MTTSI, MTTSS2, MYBPC3, MYH7, PLN, SGCD, TAZ, TNNI3, TNNT2, TPM1, TTR, ZASP
- 360: **LQTS (12 Genes)** AKAP9, ANK2, CACNA1C, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN4B, SCN5A, SNTA1
- 423: **SQTS (3 Genes)** KCNH2, KCNJ2, KCNQ1
- 384: **BrS (7 Genes)** CACNA1C, CACNB2, GPD1L, SCN1B, SCN5A, KCNE3, SCN3B
- 385: **ARVC (7 Genes)** DSC2, DSG2, DSP, JUP, PKP2, RYR2, TMEM43
- 386: **CPVT (3 Genes)** CASQ2, KCNJ2, RYR2
- 510: **Marfan Syndrome/LDS (3 Genes)** FBN1, TGFBRI, TGFBR2
- 356: **Noonan Syndrome (11 Genes)** BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SOS1, SHOCK2 (S2G mutation only)
- 511: **Loeys-Dietz Syndrome (2 Genes)** TGFBRI, TGFBR2
- 401: **Supravalvular Aortic Stenosis (1 Gene)** ELN
- 1004: **Alagille Syndrome (1 Gene with Del/Dup Testing)** JAG1
- 459: **SCA Arrhythmia Panel (10 Genes)** ANK2, CASQ2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, SCN5A (Please call before ordering this test)
- 363: **Cardiac Amyloidosis (1 Gene)** TTR

### 901: Testing for a previously identified familial mutation

Gene: \_\_\_\_\_ Mutation: \_\_\_\_\_  
 Proband Name: \_\_\_\_\_  
 Proband GeneDxAccession #: \_\_\_\_\_  
 Relationship to proband: \_\_\_\_\_

### Deletion/duplication tests

- 484: **LQTS Del/Dup (12 Genes)** AKAP9, ANK2, CACNA1C, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN4B, SCN5A, SNTA1
- 485: **Cardiomyopathies Del/Dup (20 Genes) Research use only\*** ACTC1, CAV3, GLA, LAMP2, MYBPC3, MYH7, MYL2, MYL3, PRKAG2, TNNC1, TNNI3, TNNT2, TPM1, TTR, DES, LMNA, PLN, SGCD, TAZ, ZASP
- 486: **ARVC Del/Dup (7 Genes) Research use only\*** DSC2, DSG2, DSP, JUP, PKP2, RYR2, TMEM43
- 487: **Other Arrhythmic disorders Del/Dup (7 Genes) Research use only\*** CACNA1C, CACNB2, GPD1L, SCN1B, SCN5A, CASQ2, RYR2
- 458: **Marfan Syndrome/LDS Del/Dup (3 Genes)** FBN1, TGFBRI, TGFBR2
- 910: **Congenital Heart Disorder due to Micro Del/Dup Syndromes** Whole genome array CGH

\*Except for Long QT syndrome and Marfan syndrome, the clinical significance of del/dup testing is currently unknown.

## For office use only:

## Institutional Bill

GeneDx Account # \_\_\_\_\_

Hospital/Lab Name \_\_\_\_\_

Contact Name \_\_\_\_\_

Address \_\_\_\_\_

City \_\_\_\_\_ State \_\_\_\_\_ Zip Code \_\_\_\_\_

Phone \_\_\_\_\_ Fax \_\_\_\_\_

Please send a duplicate report to this address

BILLING STAMP

## Commercial Insurance Bill

I have prior authorization # \_\_\_\_\_

Name of Insured \_\_\_\_\_ Date of Birth \_\_\_\_\_

Relationship to Insured  Self  Spouse  Child  Other

Insurance Carrier \_\_\_\_\_

ID # \_\_\_\_\_

Group # \_\_\_\_\_ Policy Name \_\_\_\_\_

SSN # \_\_\_\_\_

**Please include a copy of the front and back of the patient's insurance card.**  
 GeneDx will bill insurance and appeal for payment on the patient's behalf. A patient will be responsible for any co-pay, co-insurance and unmet deductible amounts that their policy dictates. Patient's out-of-pocket cost will be limited to \$100 per test, except in FL and CO. For insurance billing questions, please contact our Patient Advocacy department at (301) 519-2100 x6727.

### Credit Card Information

**Credit card information is required for all Blue Cross, Blue Shield and related plans, GHI, Healthnet, State Health Plan of North Carolina and Teamsters Benefit Trust.**

Name as it appears on card \_\_\_\_\_

MasterCard  Visa  Discover  American Express

Account Number \_\_\_\_\_ Expiration date \_\_\_\_\_ CVC \_\_\_\_\_

Signature \_\_\_\_\_ Date \_\_\_\_\_

## Patient Bill

I understand that my credit card will be charged the full amount for the testing.

**Please bill my credit card (all major cards accepted)**

Name as it appears on card \_\_\_\_\_

MasterCard  Visa  Discover  American Express

Account Number \_\_\_\_\_

Expiration date \_\_\_\_\_ CVC \_\_\_\_\_

Amount \_\_\_\_\_

Signature \_\_\_\_\_ Date \_\_\_\_\_

**GeneDx provides a financial assistance program or hardship discount for patients who do not have commercial insurance and cannot afford to pay out of pocket. Please call us at (301) 519-2100 x 6106**

### Internal use only

BI CCI MHC II MP EX

**My signature of informed consent certifies that I have been provided with the following facts about the cardiology genetic test for which I am giving permission. I have had the opportunity to discuss the benefits, risks and limitations of this testing and I have had any questions answered.**

## Why is this test performed?

1. Many genetic diseases are caused by mutations in one or more genes in an individual. These mutations may involve small changes, such as single nucleotide alterations, or deletions and duplications that affect part of or an entire gene. Genetic diseases can be caused by either small or large mutations.
2. In the cardiac genetic sequencing test [my/my child's] DNA will be studied to see if a small change or mutation has occurred in a gene or genes associated with heart structure and function. Sequencing tests do not identify large deletions or duplications in the genes tested, or any changes (small or large) in genes not included in this test. Large deletions and duplications at exon-level can be detected by a deletion/duplication test.
3. An array CGH test examines the human genome for loss or gain of DNA material on chromosomes. Many genetic disorders are caused by a deletion (loss) or duplication (gain) of one or more genes.
4. I understand that this test is performed to determine if I and/or members of my family have a mutation in a gene or a loss or gain of genetic material associated with a specific genetic disease, and whether I am/we are affected with, or at an increased risk to someday be affected with this genetic disease.
5. This test is not the only way to look for changes in my DNA, and my physician may recommend this test before or after ordering other genetic and/or cardiology tests.

## What might I find out from this test?

6. I might learn that no change (mutation) was found in the gene(s) studied or that no loss or gain of genetic material was detected. This outcome does not mean that [I do not/my child does not] have a genetic disease.
7. I might learn that a specific gene or genes do have a change (mutation), is deleted (lost) or is duplicated (gained), thus explaining the cause of the disorder with which [I have/my child has] been diagnosed.
8. I might learn that a gene change or a loss or gain of genetic material was found in my gene(s) that is associated with possible long-term medical problems that I do not already know about. My physician will be informed of any long-term risks that may be associated with the findings of this test, according to current medical understanding.
9. I might learn that I have a gene change (gene variant) or genomic imbalance (copy number variant) whose clinical consequences, or relationship to medical problems is currently unknown. The variant could be a normal change in the gene, or could be disease-causing mutation. Some areas of the human genome can exist in less or more than the normal quantity (copy number) and not cause medical problems. Without further information, the effects of the variant may not be known and an inconclusive result may be reported.
10. When a gene variant or copy number variant is found, it is important to find out if the gene or copy number variant is also found in other family members. Testing the biological parents of the affected individual, and/or testing other affected family members may be necessary to determine if the gene or copy number variant is disease-causing or a normal genetic change.

## What are the limitations of the test?

11. In some cases, genetic tests are unable to identify a change in a gene even though the change is present. This may be due to the current lack of knowledge about a gene's complete structure or due to the inability of the current technology (test method), to identify certain types of changes in the gene(s). In addition there may be other genes associated with a disease that have not yet been identified which are not included in the genes tested.
12. Array CGH cannot detect certain chromosomal imbalances in which the amount of DNA is unaltered. Very small changes that are beyond the resolution of the array can also go undetected.
13. An error in the diagnosis of a disease may occur if the true biological relationships of the family members being tested are not as stated. This is particularly important when parents of an affected child are submitted to the laboratory for testing. For example, non-paternity means that the stated father of an individual is not the true biological father. This test may detect non-paternity, and it may be necessary to report this finding to the individual(s) who requested testing. Non-disclosure regarding any non-biological relationships may result in incorrect result interpretation, incorrect diagnoses in the family members and/or inconclusive test results.
14. I understand that the genetic test(s) performed by GeneDx is specific for this disease and in no way guarantees my health or the health of my living or unborn children. This test does not have the ability to detect all of the long-term medical risks that [I/my child] might experience. The accuracy of genetic testing is dependent on the clinical diagnosis made elsewhere, and GeneDx cannot be responsible for incorrect clinical diagnoses.

## Permission for research studies

I also give permission for my specimen and clinical information to be used in cardiology and/or genetic research studies. GeneDx participates in the International Standard Cytogenetic Array (ISCA) consortium. Anonymized array CGH data and clinical information may be submitted to a HIPAA-compliant, de-identified public database as part of the National Institute of Health's effort to improve diagnostic testing and enhance our understanding of the relationships between genetic changes and clinical symptoms. Visit the consortium website at <https://isca.genetics.emory.edu> for more information. My name will not be used in any research studies and it will not be possible to link results of research studies back to my specimen.

Check this box if you wish to opt out of research studies.

Because of the complexity of genetic testing and the important implications of the information received from testing, results will only be reported to me through a physician, genetic counselor or another certified genetics/cardiology professional. The result reports are confidential and will only be released to other medical professionals or other parties with my express written consent. Participation in genetic testing is completely voluntary.

## Informed Consent

**Sign here or on Page 1 of the Sample Submission (Order) Form.**

**Patient or Guardian's Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_