SMA SPECIALTY MEDICAL LAB

Toll Free: (877) 697-6252 Fax: (888) 322-9524 *Phone: (954) 306-3667 Fax: (954) 306-3157*
940 Pennsylvania Blvd., Unit A, Feasterville, PA 19053
40 Exchange Place, Suite 601, New York, NY 10005
2944 SW 26th Terrace, Suite 502, Dania Beach, FL 33312
www.smalaboratory.com

GENETIC TESTING REQUISITION FORM

PATIENT INFORMATION (REQUIRED)

Last Name: _____ First Name: _____

Street Address: _____ Apt#: _____

City: _____ State: _____ Zip: _____

Phone: _____ DOB: ___/___/___ SSN: _____ Gender: F ☐ M ☐

Primary Ethnicity: ☐ African ☐ European (Finnish) ☐ Latino
☐ Ashkenazi Jewish ☐ East Asian ☐ South Asian
☐ European (Non-Finnish) ☐ Near/Middle Eastern ☐ Other

SPECIMEN INFORMATION (REQUIRED)

Date Collected: ___/___/___ Time Collected : _____

Collected and Registered By: _____ Specimen Type: ☐ Saliva ☐ Blood

ICD10 CODES *SEE BACK FOR SUGGESTED CODES*						

It is the ordering party's responsibility to order only those tests medically necessary for the diagnosis and treatment of the patient.

ADDITIONAL RESULTS RECIPIENT

Health Care Professional Name: _____

Phone: _____ Fax: _____

Email (for notification of results): _____

Mailing Address: _____

City: _____ State: _____ Zip: _____

CHART NOTES / MEDICAL NECESSITY (REQUIRED)

*attach additional supporting documentation if needed

_____

_____

_____

_____

TEST(S) REQUESTED (REQUIRED)

HEREDITARY CANCER

☐ 1250 - BREAST***Dx*** CLEAR
Multi-gene panel for inherited breast, ovarian, endometrial and pancreatic cancers

☐ 1251 - COLO***Dx*** CLEAR
Multi-gene panel for hereditary colon cancer syndromes, gastrointestinal and pancreatic cancers

☐ 1257 - PROSTATE***Dx*** CLEAR
Multi-gene panel for familial prostate, testicular, male breast and colon cancers

CARDIOLOGY

☐ 12D - CARDIOMYOPATHY PANEL
Genes associated with Hypertrophic Cardiomyopathy (HCM), Dilated Cardiomyopathy (DCM) and Left Ventricular Non-Compaction (LVNC)

☐ 14D - ARRHYTHMIA PANEL
Genes associated with inherited arrhythmias (Long and Short QT Syndromes, Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC), Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) and Brugada Syndrome)

☐ 19D - AORTIC DYSFUNCTION PANEL
For various forms of familial aortic dysfunction (Arterial tortuosity, MASS, Loeys-Dietz and Ehlers-Danlos syndromes, aortic aneurysms)

CARRIER SCREENING

☐ 1688 - CARRIER PLUS
Carrier Screening for over 100 genetic disorders and conditions, including cystic fibrosis, Bloom syndrome, Canavan disease, maple syrup urine disease 1B, glycogen storage disease Ia, galactosemia, Gaucher disease, Tay Sachs disease, familial dysautonomia, mucolipidosis IV, Niemann-Pick disease, phenylketonuria and many others

MALIGNANT HYPERTHERMIA

☐ 23D - MALIGNANT HYPERTHERMIA
A genetic test for predisposition to Malignant Hyperthermia, a severe adverse reaction to commonly used anesthetics or some muscle relaxants. Malignant Hyperthermia episode can cause coma, death, cardiac dysfunction and other complications

PATIENT PAYMENT OPTIONS (SIGNATURE REQUIRED)

☐ **OPTION 1: CREDIT CARD** (SMA Specialty Medical Lab will contact you for additional information)
☐ **OPTION 2: INVOICE PRACTICE / INSTITUTIONAL BILL / FACILITY BILL**
☐ **OPTION 3: BILL INSURANCE** (attach front and back copy of insurance card)

Insurance Company Name: _____

Policy Number / Member ID: _____

I understand that if I have enrolled in an FSA/HSA or other medical spending account with my employer or my insurance carrier, that the provision on coordination of benefits in my coverage policy may result in an automatic deduction of out of pocket costs directly from that fund by the carrier or my employer. I understand that SMA Specialty Medical Lab is in no way responsible or liable for that deduction, and will not reverse it, refund it or otherwise reimburse me for those amounts. I understand that it is my responsibility to contact my insurance carrier or employer in advance of services regarding coordination of benefits issues that may impact such an account.

➤ **Patients Initials:** _____

Patient Acknowledgement and Authorization: I acknowledge that I have provided accurate and true information to the best of my knowledge. If I have provided my insurance information for direct insurance / 3rd party billing: I hereby authorize my insurance benefits to be paid directly to SMA Specialty Medical Lab (SMA) and authorize SMA to release medical information concerning my testing, including upon request my genetic testing results, to my insurer and any business associate of insurer (TB, TPA, etc.) I authorize SMA to be my Designated Representative for purposes of appealing any denial of health benefits. I understand that I am responsible for any amounts that my insurer determines are my responsibility after calculating deductibles, co-payments and co-insurance due under my policy. **I understand that I am legally responsible for sending SMA Specialty Medical Lab any money received from my health insurance company for performance of this genetic test.**

➤ **Patients Signature:** _____ Date: _____

ORDERING HEALTH CARE PROFESSIONAL (SIGNATURE REQUIRED)

Informed Consent and Statement of Medical Necessity:

I affirm that I am legally authorized to order laboratory tests OR that I am an authorized representative of a health care professional legally authorized to order laboratory tests; and hereby order the tests requested above, which includes any collection device necessary to obtain the samples for testing. I hereby confirm that the test(s) are medically necessary for the treatment and/or plan of care for the patient, and that the information supplied on this form is accurate to the best of my knowledge. I further hereby confirm that the information has been supplied about genetic testing and that an appropriate SMA Specialty Medical Lab informed consent has been signed by the patient and is on file with a copy returned to SMA.

Did the patient opt-out for the use of their sample for research purposes in the consent? ☐ Yes ☐ No

➤ **Physicians Signature:** _____ Date: _____

SUGGESTED ICD10 CODES

*Note: The provided ICD-10 codes are listed as a convenience. Ordering practitioners should report the diagnosis code that best describes the reason for performing the test, regardless of whether the code is listed above or not.

HEREDITARY CANCER

HEREDITARY CANCER (ICD-10 Code)*	HEREDITARY CANCER (ICD-10 Code)*	HEREDITARY CANCER (ICD-10 Code)*	HEREDITARY CANCER (ICD-10 Code)*
C56.1 Malignant Neoplasm of right ovary	C50.422 Malignant neoplasm of upper-outer quadrant of left male breast	D05.91 Other unspecified type of carcinoma in situ of right breast	Z80.52 Family history of malignant neoplasm of bladder
C56.2 Malignant Neoplasm of left ovary	C50.511 Malignant neoplasm of lower-outer quadrant of right female breast	D05.92 Other unspecified type of carcinoma in situ of left breast	C25.0 Malignant neoplasm of head of pancreas
C57.01 Malignant Neoplasm of right fallopian tube	C50.512 Malignant neoplasm of lower-outer quadrant of left female breast	Z17.0 Estrogen Receptor positive status (ER+)	C25.1 Malignant neoplasm of body of pancreas
C57.02 Malignant Neoplasm of left fallopian tube	C50.521 Malignant neoplasm of lower-outer quadrant of right male breast	Z17.1 Estrogen Receptor negative status (ER-)	C25.2 Malignant neoplasm of tail of pancreas
Z80.41 Family history of malignant neoplasm of ovary	C50.522 Malignant neoplasm of lower-outer quadrant of left male breast	Z80.3 Family history of malignant neoplasm of breast	C25.3 Malignant neoplasm of pancreatic duct
C50.011 Malignant neoplasm of nipple and areola, right female breast	C50.611 Malignant neoplasm of axillary tail of right female breast	C17.0 Malignant neoplasm of duodenum	C25.4 Malignant neoplasm of endocrine duct
C50.012 Malignant neoplasm of nipple and areola, left female breast	C50.612 Malignant neoplasm of axillary tail of left female breast	C17.1 Malignant neoplasm of jejunum	C25.7 Malignant neoplasm of other parts of pancreas
C50.021 Malignant neoplasm of nipple and areola, right male breast	C50.621 Malignant neoplasm of axillary tail of right male breast	C17.2 Malignant neoplasm of ilieum	C25.8 Malignant neoplasm of overlapping sites of pancreas
C50.022 Malignant neoplasm of nipple and areola, left male breast	C50.622 Malignant neoplasm of axillary tail of left male breast	C17.3 Meckel's diverticulum, malignant	C25.9 Malignant neoplasm of pancreas, unspecified
C50.111 Malignant neoplasm of central portion of right female breast	C50.811 Malignant neoplasm of overlapping sites of right female breast	C17.8 Malignant neoplasm of overlapping sites of small intestine	Z80.0 Family history of pancreas, liver, stomach
C50.112 Malignant neoplasm of central portion of left female breast	C50.812 Malignant neoplasm of overlapping sites of left female breast	C17.9 Malignant neoplasm of small intestine, unspecified	Z85.42 Personal history of endometrial cancer, uterine cancer
C50.121 Malignant neoplasm of central portion of right male breast	C50.821 Malignant neoplasm of overlapping sites of right male breast	C18.0 Malignant neoplasm of cecum	Z85.51 Personal history of malignant neoplasm of bladder
C50.122 Malignant neoplasm of central portion of left male breast	C50.822 Malignant neoplasm of overlapping sites of left male breast	C18.1 Malignant neoplasm of appendix	Z85.850 Personal history of malignant neoplasm of thyroid
C50.211 Malignant neoplasm of upper-inner quadrant of right female breast	C50.911 Malignant neoplasm of unspecified site of right female breast	C18.2 Malignant neoplasm of ascending colon	Z80.49 Family history of cancer of the endometrium
C50.212 Malignant neoplasm of upper-inner quadrant of left female breast	C50.912 Malignant neoplasm of unspecified site of left female breast	C18.3 Malignant neoplasm of hepatic flexure	Z80.49 Family history of cancer of the genital system
C50.221 Malignant neoplasm of upper-inner quadrant of right male breast	C50.921 Malignant neoplasm of unspecified site of right male breast	C18.4 Malignant neoplasm of transverse colon	Z80.49 Family history of cancer of the uterine cervix
C50.222 Malignant neoplasm of upper-inner quadrant of left male breast	C50.922 Malignant neoplasm of unspecified site of left male breast	C18.5 Malignant neoplasm of splenic flexure	Z80.49 Family history of cancer of the uterus
C50.311 Malignant neoplasm of lower-inner quadrant of right female breast	D05.01 Lobular Carcinoma in situ of right breast (LCIS)	C18.6 Malignant neoplasm of descending colon	Z80.49 Family history of cancer of the vagina
C50.312 Malignant neoplasm of lower-inner quadrant of left female breast	D05.02 Lobular Carcinoma in situ of left breast (LCIS)	C18.7 Malignant neoplasm of sigmoid colon	Z85.028 Personal history of other malignant neoplasm of stomach
C50.321 Malignant neoplasm of lower-inner quadrant of right male breast	D05.11 Intraductal Carcinoma in situ of right breast (ICIS)	C18.8 Malignant neoplasm of overlapping sites of colon	Z85.05 Malignant neoplasm of liver
C50.322 Malignant neoplasm of lower-inner quadrant of left male breast	D05.12 Intraductal Carcinoma in situ of left breast (ICIS)	C18.9 Malignant neoplasm of colon, unspecified	Z80.0 Family history of malignant neoplasm of stomach
C50.411 Malignant neoplasm of upper-outer quadrant of right female breast	D05.81 Other specified type of carcinoma in situ of right breast	C19 Malignant neoplasm of rectogimoid junction	Z85.820 Malignant melanoma of skin
C50.412 Malignant neoplasm of upper-outer quadrant of left female breast	D05.82 Other specified type of carcinoma in situ of left breast	C20 Malignant neoplasm of rectum	
C50.421 Malignant neoplasm of upper-outer quadrant of right male breast		C21.1 Malignant neoplasm of anal canal	
		Z85.46 Malignant neoplasm of prostate	
		Z85.528 Malignant neoplasm of kidney	
		Z85.53 Malignant neoplasm of renal pelvis (part of the kidney)	
		Z85.520 Malignant carcinoid tumor of kidney	
		Z80.0 Family history of colon cancer, rectum	
		Z80.42 Family history of malignant neoplasm of prostate	
		Z80.51 Family history of malignant neoplasm of kidney	

CARDIOLOGY

CARDIOLOGY (ICD-10 Code)*	CARDIOLOGY (ICD-10 Code)*	CARDIOLOGY (ICD-10 Code)*	CARDIOLOGY (ICD-10 Code)*
I42.0 Dilated cardiomyopathy	I49.01 Unspecified atrial fibrillation	I71.01 Dissection of abdominal aorta	Q66.0 Cogenital Talipes Equinovarus ("club foot")
I42.1 Hypertrophic obstructive cardiomyopathy	I49.1 Ventricular fibrilliation	I71.1 Thoracic aortic aneurysm, ruptured	Q67.5 Cogential deformity of the spine (scoliosis)
I42.2 Hypertrophic non-obstructive cardiomyopathy	I49.3 Atrial premature depolarization (PACs)	I71.2 Thoracic aortic aneurysm, without rupture	Q67.6 Pectus excavatum
I42.5 Cardiomyopathy, other restrictive	I49.5 Ventricular premature depolarization (PVCs)	I71.3 Abdominal aortic aneurysm, ruptured	Q67.7 Pectus Carinatum
I42.8 Other cardiomyopathies	I49.8 Sick sinus syndrome	I71.8 Aortic aneurysm of unspecified site, without rupture	Q68.1 Arachnodactyly ("cogenital deformity of finger(s) and hand")
I42.8 Arrhythmogenic right ventricular dysplasia (ARVD)	Q23.8 Other unspecified cardiac arrhythmias	I71.9 Thoracic aortic ectasia	Q79.6 Ehlers-Danios syndrome
I42.9 Cardiomyopathy, unspecified	R00.1 Brugada syndrome	I77.810 Spontaneous tension pneumothorax	Q87.40 Marfan syndrome
I43 Cardiomyopathy in disease classified elsewhere	R00.2 Bradycardia, unspecified	J93.0 Primary spontaneous pneumothorax	Q87.410 Marfan syndrome with aortic dilation
I44.2 Atrioventnricular block, complete	R94.31 Palpitations	J93.11 Secondary spontaneous pneumothorax	Q87.418 Marfan syndrome with other cardiovascular manifestations
I45.81 Long QT syndrome	H27.10 Abnormal electrocardiogram	J93.81 Chronic pneumothorax	Q87.42 Marfan syndrome with ocular manifestations
I46.2 Other specified conduction disorders	H52.11 (ECG)(EKG)	J93.83 Other pneumothorax	Q87.43 Marfan syndrome with skeletal manifestations
I47.2 Cardiac arrest due to underlying cardiac conditions	H52.12 Unspecified dislocation of the lens	L90.6 Stria Atrophicae (stretch marks)	R23.3 Spontaneous Ecchymoses (easy bruising)
I48.0 Ventricular tachycardia	H52.12 Myopia, right eye	L98.8 Other specified disorders of skin and subcutaneous tissue	
I48.2 Paroxysmal atrial fibrillation	H52.13 Myopia, left eye	M35.7 Hypermobility Syndrome	
I48.91 Chronic atrial fibrillation	I34.1 Mitral valve prolapse	Q12.1 Congenital displaced lens	
	I171.00 Dissection of unspecified site of aorta		
	I71.01 Dissection of thoracic aorta		

MALIGNANT HYPERTHERMIA

FOR PATIENTS WITH PERSONAL OR FAMILY HISTORY			*FOR PATIENTS WITH NO PERSONAL OR FAMILY HISTORY*
MALIGNANT HYPERTHERMIA (ICD-10 Code)*	MALIGNANT HYPERTHERMIA (ICD-10 Code)*	MALIGNANT HYPERTHERMIA (ICD-10 Code)*	MALIGNANT HYPERTHERMIA (ICD-10 Code)*
T88.3XXA Malignant hyperthermia due to anesthesia, initial encounter	T88.3XXD Malignant hyperthermia due to anesthesia, subsequent encounter	T88.3XXS Malignant hyperthermia due to anesthesia, sequel	Z15.89 Genetic susceptibility to other disease

Informed Consent for Genetic Testing

PURPOSE AND BENEFITS

Your specimen will be analyzed for specific changes in DNA that are known to be associated with inherited diseases, such as familial cardiovascular diseases, hereditary cancer, Parkinson's Alzheimer's disease and dementia. The purpose of these tests is to help your doctor more accurately diagnose your current condition and/or future risk of disease. It can help your doctor to choose appropriate therapy and it promotes enrollment in clinical trials. The test panel(s) ordered include genes based on the guidelines from American College of Cardiology Foundation, American Heart Association, European Society of Cardiology, National Comprehensive Cancer Network, American College of Medical Genetics, American Association of Neurology, and National Institute of Aging.

WHAT TEST RESULTS MEAN AND TEST LIMITATIONS

(+) Positive (abnormal) results indicate that a genetic variant(s) associated with a hereditary disorder was detected in your DNA. However, the results should be interpreted in the context of the patient's clinical findings, and family history.

(-) Negative (normal) results mean that no disease-causing genetic variants were identified. Your risk to have the diseases tested is reduced. A "negative" result does not rule out all genetic causes of disease. It is still possible that you carry a genetic variant that this technology is unable to detect or that a condition is caused by gene(s) that are not included in the specific test ordered. Changes not targeted by these tests will not be detected. False positives, false negatives, and failed results are very rare, but possible.

DNA SAMPLE

This genetic test requires sequencing of your DNA. Your DNA samples will only be used for the specific test that is authorized by the ordering provider. If testing results are inconclusive you may be asked for an additional specimen(s). This Consent is effective for any such additional specimen(s). The original specimen(s) and isolated DNA samples are destroyed after 60 days. Some DNA samples may be retained indefinitely as deemed useful for medical research purposes in an effort to advance scientific knowledge to develop new genetic tests. To protect your identity a different unique identifier will be assigned to your specimen; all resulting research data will be recorded, handled and stored using this unique identifier. No compensation will be given to you nor will you be owed any funds due to any invention(s) resulting from research and development using your specimen(s). You can opt-out of indefinite specimen storage and specimen's use for medical research purposes by checking the box "no" in the signature section below.

GENETIC COUNSELING

It is strongly recommended that you obtain pre-testing and post-testing counseling from someone professionally trained in genetics to consider the purpose, meaning, risks, benefits, and limitations of, as well as any alternatives to, genetic testing in your particular situation, including your personal and family medical history. Further testing or additional physician consults may be warranted.

PATIENT CONFIDENTIALITY AND TEST RESULTS DISCLOSURE

Your personal information and test results are confidential. Reports are rendered only to the patient and his/her physician. Patient samples will be destroyed after testing unless the patient specifically authorizes their used in research or commercial applications. In addition to your ordering doctor, you may authorize a genetic counselor to have access to your test results. Genetic counselors are trained to provide detailed explanations about your tests, options available to you and inheritance patterns. (See check boxes below). By requesting payment by your insurance company, Medicare or other third-party payor, you specifically authorize the release of your Protected Health Information ("PHI"), including your lab test results, to such third-party payor or its authorized agents or representatives, as necessary for the purpose of determining coverage and facilitating payment.

Physician's Printed Name	**Signature of Obtaining Consent**	**Date**

PATIENT: I have read the Informed Consent and I give permission to perform genetic testing as described. My signature below acknowledges that: I understand written English sufficiently well enough, I have read and understood this Consent, all of my questions have been answered to my satisfaction, and genetic counseling has been recommended before and after testing. I understand this test is voluntary.

I allow my sample DNA and information to be provided to a genetic counselor.	☐ Yes	☐ No
I allow my sample DNA and information to be used for research.	☐ Yes	☐ No
I allow my sample and information to be used for commercial products.	☐ Yes	☐ No

Patient Name (please print)	**Signature of Giving Consent**	**Date**

Printed Name of Legally Authorized Representative	**Relationship to Patient**	**Signature of Legally Authorized Representative**	**Date**

SPECIALTY MEDICAL LABORATORY

Clinical History Questionnaire

Inherited Cardiovascular Diseases

A CAP and CLIA Accredited Laboratory | 940 Pennsylvania Blvd, Feasterville, PA 19053 | Toll Free: (877) 697-6252 • Fax: (888)322-9524

Patient Name_____ **Date of Birth**_____

Patient Signature_____ **Date** _____

Previous Genetic Testing (Please include a copy of test results if performed at another laboratory) ☐ No previous genetic testing

Patient or family member previously tested at another laboratory?: ☐ Yes ☐ No A copy of the test results provided?: ☐ Yes ☐ No

Patient previously tested at SMA Specialty Medical Lab? ☐ Yes ☐ No Family member previously tested at SMA Specialty Medical Lab? ☐ Yes ☐ No

Name: _____ DOB: _____ Relation: _____

Family History ☐ No known family history ☐ Limited Family Structure

DISEASE / CONDITION	SELF (circle response)	RELATIVES (indicate maternal or paternal side)	AGE AT DIAGNOSIS
Cardiomyopathy (cardiac muscle thickening or thinning)	Yes / No		
Arrhythmia (irregular heart beat)	Yes / No		
Aortic aneurysm (thoracic or abdominal)	Yes / No		
Heart attack (cardiac ischemia)	Yes / No		
Cardiac arrest (heart stopped)	Yes (successfully resuscitated) / No		
Heart failure (insufficient blood pumping)	Yes / No		
Heart transplant	Yes / No		
Atherosclerosis (hardening and narrowing of the arteries)	Yes / No		
Implantable Cardioverter Defibrillator (ICD) / pacemaker	Yes / No		
High cholesterol	Yes / No		
High blood pressure	Yes / No		
Fainting during exercise	Yes / No		
Fainting during normal activity	Yes / No		
Problems with exercise	Yes / No		
Muscle weakness or muscle disorders	Yes / No		
Sudden unexpected death without a known cause	Not Applicable		
Any other chronic or genetic diseases that run in your family_____	Yes / No		
Any other cardiovascular conditions not mentioned above _____	Yes / No		

Additional relevant information:

SMA Specialty Medical Lab
940 Pennsylvania Boulevard, Feasterville, PA 19053 Ph: 215-322-6590; FAX: 215-322-9524
Molecular Genetics Department

Patient Informed Consent for Genetic Testing – Cardiology

The accuracy of the genetic testing and reporting methods have been determined and verified to meet required regulatory performance standards by SMA Specialty Medical Lab ("SMA"), a licensed and CLIA (U.S. government) accredited laboratory.

PLEASE INITIAL NEXT TO EACH STATEMENT AS ACKNOWLEDGMENT THAT YOU HAVE READ AND UNDERSTAND THE INFORMATION.

____ **I understand the general risks and limitations of genetic testing** including the following:

- Saliva or blood specimens are used for testing. Side effects of having blood drawn are uncommon, but may include dizziness, fainting, soreness, pain, bleeding, bruising, and, rarely, infection.
- Genetic testing should not be used as a substitute for treating and diagnosing conditions, or the provision of health care services by a physician or other qualified healthcare professional.
- Even if a mutation or variant is present in a family, it does not mean that everyone in the family inherited this mutation or variant. The pattern of inheritance can be explained by a genetic counselor or qualified healthcare professional. Understanding this can help me and my family members prepare for varying and complicated outcomes. I understand that a genetic counselor or qualified healthcare professional can help me consider the pros and cons of speaking first with family members before being tested to find out if they want to know my results. I understand that sometimes family secrets, such as paternity, adoptions, or other difficult issues may come up.
- This testing may not provide informative results for other reasons, such as: (1) non-genetic factors; (2) individual genetic variation; (3) insufficient scientific information about the relationship between genetic information and health outcomes; (4) various laboratory and non-laboratory technical reasons; and (5) incomplete gene sequence information.
- Other risks that may be experienced as a result of this testing include: unjustified alarm and/or false reassurance that can discourage preventive measures, related emotional issues, impact on life-changing decisions, potential genetic discrimination (e.g., in employment and insurance areas) and loss of confidentiality. The testing results and information may become part of my permanent medical record and may be available to individuals and organizations with legal access to such records.

____ **I understand that it is strongly recommended that I obtain pre-testing and post-testing counseling from someone professionally trained in genetics to consider the purpose, meaning, risks, benefits, and limitations of, as well as any alternatives to, genetic testing in my particular situation, including my personal and family medical history.**

Counseling may be provided by a genetic counselor (such as those found on the National Society of Genetic Counseling website), advanced practice nurse, doctor and other qualified healthcare professional. Pre-test counseling may help me better prepare to receive the test results and allow for advance consideration of medical options and the impact test results may have on myself and my family members. Post-test counseling provides a valuable opportunity to understand the medical interpretations of detected mutations and variants, the psychological risks and benefits of learning my genetic test results, how families inherited conditions and the risk of passing an inherited variant on to my children, options for additional independent testing, and the importance of continuing regular disease surveillance and prevention activities, among other things. Further testing or additional physician consults may be warranted.

____ **I understand that if testing results are inconclusive that I may be asked for an additional specimen(s).** This Consent is effective for any such additional specimen(s).

If a minor will be tested, I understand the following: While genetic report information may be similar for adults and minors, the consequences of genetic testing of minors are relatively new and less understood. The National Society of Genetic Counselors recommends that the social and psychological risks and benefits of early identification of genetic issues from the perspective of the minor and parent/guardian be carefully considered and include genetic counseling when discussing genetic testing of children for inherited cardiovascular disease risk.

____ **I understand the following information about confidentiality and disclosure of my personal information:**

- My personal information and test results are confidential. While there can be no guarantee of privacy, SMA Specialty Medical Lab has established reasonable safeguards to protect it. This information and the test results will be released to the ordering healthcare professional. I may request a copy of my lab results from SMA Specialty Medical Lab's Client Services (see "Questions" below for contact information).
- This information and the results may also be disclosed if required by law, such as in response to a subpoena.
- I understand that if I share this information or these test results with anyone, I am responsible for any compromise of confidentiality that may result from such sharing. The original specimen(s) may be securely stored for sixty (60) days from the date of collection and any remaining isolated DNA may be securely stored in accordance with applicable laws, regulations and standards. After such storage, the specimen(s) and the isolated DNA will be properly destroyed in accordance with applicable laws and regulations and the testing laboratory's standard operating procedures.

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Molecular Genetics Department

____ **I understand the following regarding specimens for Medical Research Purposes:** I authorize that my DNA extracted from my original specimen may be retained up to 10 years by SMA Specialty Medical Lab as deemed useful for medical research purposes to develop new genetic tests. I understand that to protect my identity: a unique identifier will be assigned to my specimen; all resulting research data will be recorded, handled and stored using this unique identifier; my name will be unavailable to any member of the research team; and my identity will not be released or disclosed to others outside of SMA Specialty Medical Lab. No compensation will be given me nor will I be owed any funds due to any inventions(s) resulting from research and development using my specimen(s). I may refuse to submit my specimen for use in this way and this will not affect my results. Unless I indicate below that I do not consent to anonymous medical research, I understand that my specimen(s) may be used in this manner.

(MANDATORY for residents of New York State) Indicate your preference by checking one of the statements below:

☐ I do not wish for my specimen to be retained for Medical Research Purposes; discard within 60 days of collection.

☐ I agree to use of my de-identified biospecimen for medical research to improve genetic testing for all patients. I consent to my sample being retained beyond 60 days of collection; the retained sample will be de-identified by having all identifiers removed prior to re-testing. The de-identified sample and results obtained will remain anonymous.

____ **I understand I may withdraw my consent:** Under CLIA regulations, SMA Specialty Medical Lab cannot destroy medical records. However at my written request and according to my instructions, SMA Specialty Medical Lab can: a) destroy my DNA specimen(s) at the next regularly scheduled destruction cycle; b) delete my account; and c) move all medical information, including results report(s), into a secure, offline storage area with limited access. This means my account and results report(s) will not be searchable in SMA Specialty Medical Lab systems by regular means and I and my healthcare professional will not be able to obtain a copy of my account information and results report(s) from SMA Specialty Medical Lab. A request to withdraw my consent may be made to SMA Specialty Medical Lab' Client Services (see phone number under "Questions" below).

____ **I understand the following information regarding the general purpose of testing for inherited cardiovascular disease risk.**

- Depending upon the specific genetic testing ordered by the healthcare professional on the SMA Specialty Medical Lab's requisition form, I understand my specimen is being tested for my genetic makeup related only to my inherited cardiovascular disease risk. My DNA will be sequenced and analyzed for specific changes to find out my genetic predisposition for heritable cardiovascular diseases. The cardiovascular diseases covered by this test may include conditions such as cardiomyopathy, arrhythmia, and dysfunctions of the aorta. Many cardiovascular conditions are not inherited but occur during a person's lifetime, which is why the continued practice of heart-healthy behaviors are important. I understand what SMA includes in its reports is determined at SMA's discretion.

____ **I understand the risks and limitations of inherited cardiovascular disease testing** including the following:

- The existence of a mutation or variant does not mean I will develop a cardiovascular disease. The lack of mutations or variants does not mean I will not develop a cardiovascular disease. For some cardiovascular conditions, genetic causes have not been determined. The severity of the symptoms may vary from person to person.
- Genes are one of many things that may contribute to development of conditions in the heart and vasculature. Other factors, such as diet, exercise, obesity, personal and family medical history and lifestyle or behavioral choices, also contribute to risk for the development of cardiovascular diseases.
- Cardiomyopathy and other vascular diseases can appear to "run in families", even though they may not be caused by a mutation or variant detectable by this test. This could, for example, be caused by other shared environment/lifestyle or conditions in the family, such as diet or alcoholism.

____ **I understand that the results from genetic testing for inherited cardiovascular disease may** help a qualified healthcare professional and me learn more about my predisposition to certain heart and vasculature conditions (e.g. cardiomyopathy, aortic dysfunction, and arrhythmia) and how I may reduce my risk or slow down the development of the condition through screening and medical management. I understand that there are several types of results that can be generated, including:

- Pathogenic variant detected - A pathogenic variant could be identified in my genetic makeup that is associated with an increased risk of developing a cardiovascular disease. Knowing this information may help me and my healthcare professional make more informed choices about my health care, including screening and medical management based on what is known about the gene(s) in which a variant was found and the condition associated with it.
- Likely pathogenic variant detected - A likely pathogenic variant could be identified in my genetic makeup that could be associated with an increased risk of developing a cardiovascular disease. Knowing this information may help me and my healthcare professional make more informed choices about my health care, including screening and medical management based on what is known about the gene(s) in which a variant was found and the condition associated with it.

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Molecular Genetics Department

- Variant of uncertain significance detected – A variant of uncertain significance could be detected. This type of change may or may not be associated with an increased risk of developing a cardiovascular disease. I understand I may have at least the same likelihood of developing cardiovascular disease as the general population, and may still be at an above average risk due to a genetic predisposition that cannot be detected by this test. As clinical or scientific information evolves, I understand that I may receive updated information about the interpretation of my results.
- Negative - No variant of clinical or uncertain significance was detected. If no one in my family, including me, has ever had a heritable cardiovascular disease, I still have at least the same risk of developing a condition of the heart or vasculature as does a person in the general population. I may still be at above average risk for heritable cardiovascular disease due to a genetic predisposition that cannot be detected by this test, either in the gene(s) I am tested for or in another gene linked to hereditary cardiovascular disease.

Informed Consent Acknowledgement

____ **I understand that this testing is voluntary and freely consent to this testing.** My signature below acknowledges that:

- I understand written English sufficiently well enough, I have read and understood the front and back of this Consent, all of my questions have been answered to my satisfaction, and I agree to have the testing completed. I understand that I can receive a copy of this Consent.
- I have reached 18 years of age or older OR have the legal authority to provide this Consent and authorization for genetic testing, under all applicable laws.
- I understand SMA Specialty Medical Lab may use my DNA and clinical information in medical research studies and for publication, if appropriate, unless I opt-out by checking the appropriate box on page 2. I understand that my name or other personally identifiable information will not be used in or linked by SMA Specialty Medical Lab to the results of any studies and publications.

I **consent** to the use of my DNA extracted from my original specimen, clinical information and information provided herein for anonymized medical and research purposes. I understand this is deemed useful by SMA and explained in this Consent.

Release of Information for Insurance Claims Processing: I understand that by requesting payment by my insurance company, Medicare or other third-party payor that I specifically authorize the release of my Protected Health Information ("PHI"), including my lab test results, to such third-party payor or its authorized agents or representatives, as necessary for the purpose of determining coverage and facilitating payment. This authorization is valid for one year. I may revoke this authorization at any time by sending a written notice to SMA Specialty Medical Lab's Client Services.

_____ _____
Signature of Patient or Legally Authorized Representative Signature Date

_____ _____
Patient Name (Print) Name and Relationship (Parent/Guardian if patient is a minor)

Check one:	Self ☐	Parent ☐	Legal Guardian ☐	Durable Power of Attorney for Health Care ☐

_____ _____
Signature of Physician/Counselor/Health Personnel Signature Date

California residents only: I understand I have a right to receive a copy of the Experimental Subject's Bill of Rights from my ordering healthcare professional.

Questions: If I have further questions about this testing, I understand that I can either contact a genetic counselor, other qualified healthcare professional or SMA Specialty Medical Lab's Client Services at 1-877-697-6252, 9:00 AM to 5:00 PM Eastern Time, Monday through Friday to speak to the SMA Molecular Genetics Department.