UVA Cardiovascular Genetics Clinic

A Life-Saving Preventive Service

Because many inherited heart conditions develop with few to no symptoms, the first recognized sign of disease is often cardiac arrest.

Although a gene mutation cannot be modified, early diagnosis is critical for improved patient outcomes. When inherited heart conditions are detected prior to symptoms, complications can often be managed successfully with medication, careful surveillance, activity restrictions or preventive surgery.

UVA Cardiovascular Genetics Clinic helps patients diagnosed with, or at risk of developing, heart conditions.

Process for Genetic Testing

When you refer a patient to UVA Cardiovascular Genetics Clinic, a genetic counselor will:

- Review the patient's medical history and relevant records
- Construct a multigenerational pedigree to identify other affected or at-risk family members
- Discuss how genetic testing may be helpful
- Handle insurance authorization for testing and obtain informed consent
- Facilitate testing at an appropriate laboratory, which requires a small blood sample
- Interpret results and communicate them to the patient and his or her providers
- Help coordinate testing for at-risk family members when necessary

Partners in Care

After the patient is informed, we report all genetic testing results to the referring physician, along with the genetic counselor's interpretation. The report includes guideline-based recommendations for how the information can be used to best manage the patient's condition and how it may impact at-risk family members.

Advanced Treatment for Heart Conditions

If a heart condition is detected, we can coordinate services with UVA Heart and Vascular Center. The center's multidisciplinary team offers the full complement of treatment options for diseases, including arrhythmia syndromes, aortic disease, congenital heart disease, Marfan syndrome, Loeys-Dietz syndrome, Ehlers-Danlos syndrome and familial thoracic aortic aneurysms. We also offer specialized care for athletes with heart problems.

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Indications for Referral

A Personal or Family History of:

Cardiomyopathy

- Hypertrophic cardiomyopathy (HCM)
- Familial or idiopathic dilated cardiomyopathy
- Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVC/D)
- Peripartum cardiomyopathy
- Left ventricular noncompaction
- Restrictive cardiomyopathy
- Familial amyloidosis

Arrhythmia

- Long QT syndrome (LQTS)
- Brugada syndrome
- Catecholaminergic polymorphic ventricular tachycardia (CPVT)
- Familial atrial fibrillation
- Progressive conduction system disease
- Unexplained sudden death
- Unexplained cardiac arrest
- Short QT syndrome

Vascular Diseases

- Aortic aneurysm and/or dissection (< 50 years old)
- Marfan syndrome
- Loeys-Dietz syndrome
- Vascular Ehlers-Danlos syndrome (EDS)

Dyslipidemia

- Familial hypercholesterolemia
- Elevated Lp(a)
- Familial combined hyperlipidemia

Features Suggestive of a Hereditary Cardiovascular Condition

- Unexplained cardiac arrest or sudden death ● ● ● ●
- Unexplained syncope, syncope with exercise or emotional distress ● ●
- Unexplained seizures or seizures with normal neurological evaluation ●
- ICD/pacemaker (age < 50 years) ● ●
- Heart failure (< 60 years) ● ●
- Heart transplant (< 60 years) ●
- Cardiomyopathy or "enlarged heart" ●
- Arrhythmia or "irregular heartbeat" ●
- Exercise intolerance ●
- Early "heart attack," coronary artery disease or stroke (males < 55 years; females < 65 years) ● ● ●
- Aortic aneurysm/dissection (< 50 years) ●
- Sudden infant death syndrome (SIDS) ●
- Unexplained accidents (i.e., drowning, single-car accident, etc.) ● ● ● ●
- Untreated LDL > 190 ●

● Cardiomyopathy
● Arrhythmia
● Vascular disease
● Familial hypercholesterolemia

To refer a patient to UVA Cardiovascular Genetics Clinic, call 434.243.1000.

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University of Virginia Health System | Heart & Vascular Center