



# Indications for referral to cardiovascular genetics



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 non-compaction
- Restrictive cardiomyopathy
- Familial amyloidosis

## **Vascular diseases:**

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

## **Arrhythmia:**

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

## **Other:**

- Familial hypercholesterolemia
- Heritable heart defects (e.g. conotruncal defects, left ventricular outflow tract defects)
- Multiple relatives with congenital heart defects
- Familial or idiopathic pulmonary arterial hypertension

**> 1/200 people have an inherited cardiovascular disease**

**A cardiovascular genetics evaluation is recommended for families with hereditary heart conditions by HRS, HFSA, AHA, and ACC.**

# Family history suggestive of a hereditary cardiovascular condition:

- Unexplained cardiac arrest(s) or sudden death ★★★
- Unexplained syncope, syncope with exercise or emotional distress ★★
- Unexplained seizures, seizures with normal neurological evaluation ★
- ICD/Pacemaker (<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  $\geq$ 190 ★

★ Cardiomyopathy ★ Arrhythmia ★ Vascular disease ★ Familial hypercholesterolemia

Inherited cardiovascular diseases are a leading cause  
of sudden death in young people

Identifying at-risk patients and family members can be life-saving



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