Familial Cardiomyopathy

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- Cardiomyopathies are diseases in which the heart muscle becomes enlarged, weakened, or stiff, preventing it from working as well as it should. There are different types of cardiomyopathies that run-in families.
  - Dilated cardiomyopathy: pumping chambers of the heart are abnormally large and weak
    - Between 30-50% of people with idiopathic dilated cardiomyopathy have inherited a genetic mutation for the disease.
  - Hypertrophic cardiomyopathy: the pumping chambers of the heart become abnormally thick
    - Men and women are equally likely to inherit hypertrophic cardiomyopathy.
    - If a child does inherit the mutation, the most likely period in which the disease will develop (i.e. the heart wall muscle thickening) is during puberty, although the severity of disease cannot be predicted.
  - Amyloidosis and Amyloid Transthyretin (ATTR) Cardiomyopathy
    - Amyloid protein deposits in the heart muscle, causing stiffening of the heart muscle. When the heart stiffens (also known as restrictive cardiomyopathy), the heart is unable to pump as efficiently as usual.
    - called familial or hereditary amyloidosis.
    - Patients are born with a genetic mutation causing accelerated amyloid protein deposits.
    - Most commonly deposits are on the heart and the nerves.
    - More common in African American patients.
- People who have one or more family members who experienced sudden cardiac death or developed unexplained heart failure before age 60 should be evaluated by a genetics center that is experienced with cardiovascular diseases. A genetics counselor can help to determine if genetic testing would be helpful.
- For more information about Genetic testing see [www.nsgc.org](http://www.nsgc.org); click on "Find a counselor"