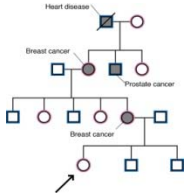
Cardiovascular Disease – Genomics to Health, February 2013

The most common form of heart disease is coronary artery disease (CAD) with polygenic (many genetic) changes in combination with environmental influences, e.g. diet, exercise, smoking. More rare cardiac conditions, Long QT syndrome or Familial Hypercholesterolemia, are caused by single gene mutations. Family history assessment is important in assessing for polygenic and single gene disorders.

Cardiovascular Disease (Selected Examples) 	Family History Risk Factors 	Assessment Red Flags 
Coronary Artery Disease (CAD) **(CAD caused by narrowed and hardened arteries to the heart. May result in angina, heart attack, heart failure, or arrhythmias. Leading cause of mortality.)**	•Premature (<50 years) family history of angina, myocardial infarction, angioplasty, or bypass surgery increases lifetime risk for heart disease and cardiovascular disease mortality by ~50%. •Sibling (brother, sister) history of heart disease (regardless of age of onset) increases the odds of heart disease by ~50%.	•↑ Lipid profile •History of smoking, obesity, diabetes, hypertension •Family history of CAD (i.e. angina, myocardial infarction, angioplasty, or bypass surgery) •Genetic testing generally not indicated
Familial Hypercholesterolemia (FH) **(Inherited tendency to ↑ total cholesterol, ↑ LDL, normal triglycerides; most commonly due to mutation on the LDLR gene.)**	•With the most common type of FH, each child of an affected parent has a 50% chance of inheriting the FH gene mutation.	•Strong family history of early heart attacks (<50-55 years of age) •LDL levels > 220 mg/dl adults: > 170-200 mg/dl children •Genetics referral/testing may be indicated
Inherited Long QT Syndrome **(Disturbance of the heart's electrical system with a prolonged QT interval; Inherited LQTS due to a mutation in a LQTS gene.)**	•With the most common type of inherited LQTS, each child of an affected parent has a 50% chance of inheriting the LQTS gene mutation.	•History of syncope •Implanted pacemaker/defibrillator •Seizure disorder •Family history of sudden unexplained death or sudden infant death syndrome •Genetic referral/testing may be indicated •Consensus statement on genomics webpage

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