

Genetics and Cardiovascular Disease

Genetic testing has entered the mainstream in cardiovascular disease assessment and preventive management. For at least four important settings, genetic testing has begun to play a key role. These include inherited cardiac conditions such as cardiomyopathies, channelopathies, homozygous and heterozygous familial hypercholesterolemia, atrial fibrillation and other arrhythmias, and cardiopathies as aortic disorders like Marfan's Syndrome, familial thoracic aortic aneurysm, and Loeys-Dietz Syndrome.

With respect to cardiomyopathies and channelopathies, genetic testing for hypertrophic cardiomyopathy, dilated cardiomyopathy, long QT Syndrome, and Brugada, official recommendation has been received from the Heart Rhythm Society and European Society of Cardiology. New evidence has also better classified the genetic underpinnings of dilated cardiomyopathy, the most common inherited cardiac condition.

Genetic testing clearly contributes to risk assessment, and finding a genetic cause for a condition lays the foundation for more effective early intervention. In familial hypercholesterolemia (FH), both heterozygous and homozygous FH are substantially more common than had been believed until recently. Genetic testing improves the care of individuals because it can identify them pre-symptomatically and early treatment can change their long term prognosis.

Genetic testing is available, affordable, and efficient. Several commercial companies offer the testing and PRIMA HEART offers on-site blood draws and facilitation with the companies for insurance authorization and cost effective pricing. Selection of what possible testing is appropriate is incorporated into cardiovascular consultations, specifically focused on genetics, inherited disorders, and family heart disease.

Dr. Robert Superko is a nationally recognized cardiologist who has spent his career in this area of research and clinical practice and numerous publications, addressing multiple aspects of genetics and genomics in cardiovascular disease.