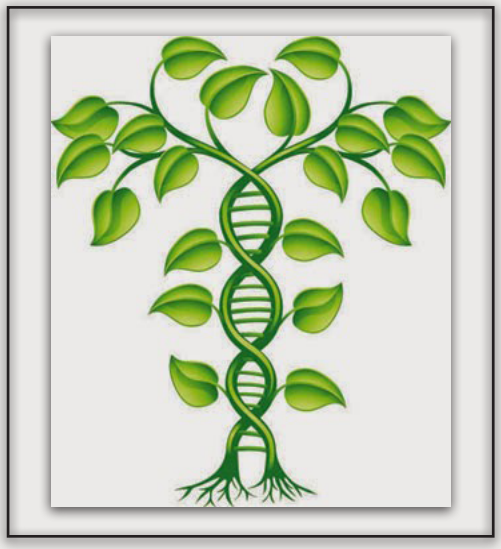
To Your Heart!

Family History and Genetic Testing: Worth the Worry?

By Narendra Singh MD



In 2000 President Clinton and Prime Minister Tony Blair jointly announced to the world that we had sequenced the human genome. Fifteen years later the promise of that accomplishment is not fully realized but we have made some important advances. Our body has 23 pairs of chromosomes. Each chromosome has multiple genes that contain DNA code that allows our body to produce enzymes and proteins that are vital to our existence.

When it comes to clogged arteries, the Interheart study showed that over 90% of your risk for this type of heart disease can be attributed to nine risk factors. Six factors increase risk - abnormal lipids, smoking, hypertension, diabetes, abdominal obesity, and psychosocial factors. Three factors decrease risk-regular physical activity, moderate alcohol intake, and regular consumption of fruits and vegetables.

A family history of a heart attack can increase the severity of presentation or age of onset for some of these risk factors but generally does not involve identifying new risk factors. Genetic tests are available that can help predict your predisposition for high cholesterol or diabetes but they usually do not alter management advice. A notable exception is a positive genetic test will help with insurance coverage for expensive meds such as the cholesterol lowering PCSK9 inhibitors or new blood thinners.

Genetic testing is very useful if there is a family history of sudden death or ventricular arrhythmias. Conditions with unusual names such as Brugada, Marfans, Long QT syndrome, hypertrophic cardiomyopathy and arrhythmogenic right ventricular dysplasia can be inherited. Early detection and the implantation of a defibrillator can be life saving!

If you were born with a congenital heart defect then genetic counseling and testing is often useful since the risk for the fetus to also have a heart defect is around 15 to 20%. Results of genetic testing are now protected against discrimination from employers or insurers.

Response to commonly used drugs such as warfarin, clopidogrel and statins can also be predicted by genetic testing however the test results presently do not routinely change our treatment decisions. Even if you have a detrimental gene, the environment you live in can affect if that gene will actually express itself. This is called epigenetics. Also, until we know what every gene does we can not provide a true estimate of individual risk. You may have 4 known genes that increase your risk for heart disease but 8 unknown genes that protect you from heart disease. If we test you for the known genes then we will have unnecessarily worried you…when in fact you were not at risk!

Earlier this year President Obama announced the Precision Medicine Initiative. The goal is to fund research that will in the future take into account an individual's genetic makeup, environment and lifestyle to determine the best course of disease prevention and treatment. This approach is already yielding much success in cancer therapy such as the Angelina Jolie BRACA gene decision to have a bilateral mastectomy. It is hoped that in the future this personalized guidance for risk will be the norm for all medical conditions.■

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