

Genetics 101: The Egg, the Yolk and You

Researchers around the world are focusing on gene mapping and DNA sequencing with the hopes that personal genomics (the study of an individual's genetic makeup) will help in the determination of personalized medical treatments. Healthnetwork's Medical Director, Susan Locke, MD, shares some basics about genetic testing.

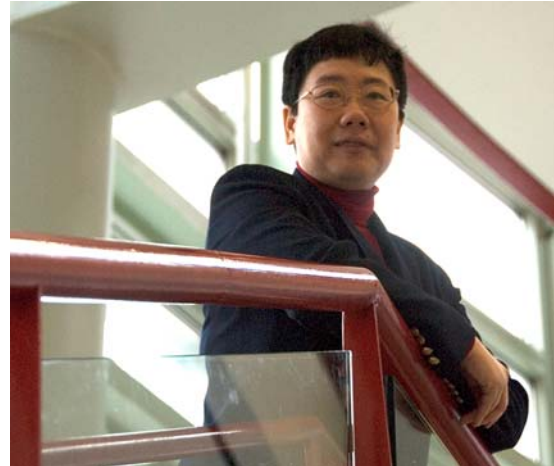
Genetic testing examines your DNA which is the chemical database that carries "instructions" for your body's functioning. Genetic testing looks for changes in your genes that may account for illness or disease.

Typical genetic tests:

- Prenatal testing detects abnormalities in the genes of a fetus.
- Newborn screenings is one of the most common forms of testing.
- Carrier testing identifies people who carry a gene for a disease and their likelihood of passing it on to their children.
- Pre-symptomatic testing for genetic diseases in adults with a family history determines one's risk for developing that condition.
- Diagnostic testing in a person who has disease symptoms will confirm if the individual has a suspected disease.

Most genetic tests are done by either a blood sample or a swab of the inside of the cheek. An exception to this is prenatal testing which is done by obtaining a sample of amniotic fluid or the placenta. Genetic testing is available in most hospitals and in general, it takes about 3-4 weeks to receive test results.

The results of genetic testing are not always straightforward and a person's medical history, family history and type of genetic test must be taken into account. No matter the result, the goal remains the same; to empower patients through knowledge and allow them to be proactive about their health.



Charis Eng, MD, PhD, FACP

For more specific information we turned to Charis Eng, MD, PhD, FACP, recipient of the 2009 Healthnetwork Service Excellence Award and Chair and Founding Director of the Genomic Medicine Institute (GMI) at the Cleveland Clinic. The GMI, which Dr. Eng formed a little over four years ago, comprises 6 lab-based patient-oriented genetics research teams focusing on cancer genetics. A true leader in her field, recently, Dr. Eng was appointed by Kathleen Sebelius to the U.S. Department of Health and Human Services' Secretary's Advisory Committee on Genetics, Health and Society (2009-2013).

Dr. Eng shares some details of her genetic research and the value of genetic testing as it relates to the patient.

We are made up of cells; I tell my patients a cell is like an egg with a yolk and white. The yolk is the nucleus of the cell or the command center. It holds the DNA and the genes. I also tell my patients that there are 30,000 different genes in humans and they are like 30,000 encyclopedias.

These genes or encyclopedias can be small with only one volume or very large with 80 volumes. For the

cell and body to be well, all 30,000 encyclopedias have to be arranged in order, and read well without typographical or grammatical errors. When a bad typographical error occurs in one sentence of a volume of an encyclopedia, a mutation has occurred. Mutations in different genes mean different risks of different diseases. So finding the affected gene allows for very accurate diagnoses and begins the process of personalizing healthcare. Once a family-specific mutation is uncovered, then we can look for that one specific typo in every single member of the patient's family in the setting of genetic counseling. This is called predictive testing because it can predict which family member who has yet to develop a disease has a high likelihood of doing so.

It is important to keep in mind that genetic testing is only one small part of the entire genetics evaluation which includes genetic counseling and risk assessment as well. Based on a patient's history, physical exam and the family health history, a tentative list of genetic diagnoses are formed in the genetic professionals mind. This tentative list guides which gene to offer testing (after all, there are 30,000 genes and we cannot and must not offer testing indiscriminately, at this time).

Healthnetwork Foundation partners with over 30 top-tier medical institutions across America. If you have questions about genetic testing or if you are interested in scheduling an appointment with a genetics counselor, contact Healthnetwork.

One Call Starts It All: 866-968-2467, 440-893-0830
or email help@healthnetworkfoundation.org



Susan Locke, MD
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Have a health question you want to ask Dr. Locke? Send an email to help@healthnetworkfoundation.org. All inquiries are kept private and confidential.

If you have questions about Healthnetwork or you would like a referral to one of our centers of excellence, please call our office: **866-968-2467** or **440-893-0830**.