FACT SHEET

NATIONAL INSTITUTES OF HEALTH

Genetic Testing: How it is Used for Healthcare

Yesterday

- Chromosomes – which are units of heredity inside cells - were first discovered in the late 1800s.
- In the early 1900s, inherited diseases were first linked to chromosomes.
- Discoveries starting in the 1950s have helped scientists to develop genetic tests for genetic conditions such as Down syndrome, cystic fibrosis, and Duchenne muscular dystrophy.
- Genetic testing was initially used to make or confirm a diagnosis of a genetic condition, and to screen newborns for conditions such as phenylketonuria (PKU), so that early interventions and treatments could be administered.
- There were some research laboratories capable of conducting genetic testing and few commercial genetic testing laboratories.

Today

- Genetic testing is available for over 2000 rare and common conditions.
- Genetic testing is available from over 500 laboratories.
- There are a number of different types of genetic tests available today, including:
    - **Diagnostic testing** - identifies a genetic condition or disease that is making or in the future will make a person ill. The results of diagnostic testing can help in treating and managing the disorder.
    - **Predictive and pre-symptomatic genetic testing** - finds genetic variations that increase a person's chance of developing specific diseases. This type of genetic testing may help provide information about a person's risk of developing a disease, and can help in decisions about lifestyle and health care.
    - **Carrier testing**–tells people if they "carry" a genetic change that can cause a disease. Carriers usually show no signs of the disorder; however, they can pass on the genetic variation to their children, who may develop the disorder or become carriers themselves.
    - **Prenatal testing** - is offered during pregnancy to help identify fetuses that have certain diseases.
    - **Pre-implantation genetic testing** –is done in conjunction with *in vitro* fertilization to determine if embryos for implantation carry genes that could cause disease.
    - **Newborn screening** - is used to test babies one or two days after birth to find out if they have certain diseases known to cause problems with health and development.
    - **Pharmacogenetic testing** - gives information about how certain medicines are processed in a person's body. This type of testing can help a healthcare provider choose the medicines that work best with a person's genetic makeup. For example, genetic testing is now available to guide treatments for certain cancers.
    - **Research genetic testing** – helps scientists learn more about how genes contribute to health and disease, as well as develop gene-based treatments. Sometimes the results do not directly help the research participant, but they may benefit others in the future by helping researchers expand their understanding of the human body.
- Some types of testing can be purchased directly by the health care consumer. Often, no healthcare provider is involved in this process.
- Though an increasing number of genetic tests are covered by insurance, not all insurance companies cover all types of genetic testing.
- The Genetic Information Non-discrimination Act (GINA) (http://www.genome.gov/24519851) provides national protections against discrimination for health insurance and employment purposes based on genetic test results. Some states offer additional protections for health care consumers that undergo genetic testing.

- Resources about genetic testing available today include:
    - GeneTests - www.genetests.org
    - National Human Genome Research Institute – www.genome.gov/health
    - National Cancer Institute – www.cancer.gov/cancertopics/UnderstandingCancer/genetesting
    - Genetics Home Reference – www.ghr.nlm.nih.gov/handbook/testing
    - Medline Plus/Genetic Testing – www.nlm.nih.gov/medlineplus/genetictesting.html
    - Genetic and Rare Diseases Information Center (GARD) www.rarediseases.info.nih.gov/GARD/Default.aspx?PageID=4

Tomorrow

- In the future, genetic testing will be an important part of health care for many individuals.
- NIH is in the process of building the Genetic Test Registry (www.ncbi.nlm.nih.gov/gtr/), an online resource that will provide a centralized location for test developers and manufacturers to submit information on their tests. Patients, healthcare providers, researchers, and others will be able to search the registry for information on indications of use, clinical validity and utility, and so on. It is expected that the Genetic Test Registry will be available in 2011.
- The cost of genetic testing will continue to decline. Eventually the cost of the ultimate genetic test - sequencing an individual's entire genome - will be less than $1,000.
- We will live in a time of more effective "personalized medicine". Information from genetic testing will help to detect risk for disease, guide strategies for maintaining health, offer more accurate diagnosis, and guide treatment choices for a wide variety of conditions.

For additional information contact:

The Communications and Public Liaison Branch, NHGRI, at (301) 402-0911

National Human Genome Research Institute (NHGRI)
http://www.genome.gov/