

## Fact Sheet

# 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 from the 1950s through the 1980s 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 few research laboratories capable of conducting genetic testing and few commercial genetic testing laboratories.

## Today

- Individuals with the following family history backgrounds may inquire about genetic testing:
  - Family history of an uncommon inherited disease such as Duchenne Muscular Dystrophy or sickle cell anemia;
  - A history of more common diseases such as cancer or Parkinson’s Disease that affect multiple relatives in several generations of a family;
  - Couples considering having a baby and who have a family history of inherited diseases, or who belong to an ethnic group with a higher risk of a specific inherited disease.
- 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 variation 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.

- **Pharmacogenomic testing** - gives information about how certain medicines are processed in a person's body. This type of testing can help a healthcare provider to 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.
- Direct-to-consumer genetic testing is a new approach that allows people to order certain genetic tests by sending a sample of their saliva or tissue to a laboratory. The laboratory returns the results only to the individual who sent the sample. Often, no healthcare provider is involved in this process.
- Many people are concerned about whether or not their health insurance will cover the cost of the genetic test. Not all insurance companies cover all types of genetic testing.
- Other concerns about genetic tests include the privacy of genetic test results, especially from insurers or employers.
- Resources about genetic testing available today include:
  - GeneTests - <http://www.genetests.org/>
  - National Human Genome Research Institute – [www.genome.gov/health](http://www.genome.gov/health)
  - National Cancer Institute - [www.cancer.gov/cancertopics/UnderstandingCancer/genetesting](http://www.cancer.gov/cancertopics/UnderstandingCancer/genetesting)
  - Genetics Home Reference - <http://ghr.nlm.nih.gov/handbook/testing>
  - Medline Plus/Genetic Testing - [www.nlm.nih.gov/medlineplus/genetictesting.html](http://www.nlm.nih.gov/medlineplus/genetictesting.html)
  - Genetic and Rare Diseases Information Center (GARD) - <http://rarediseases.info.nih.gov/GARD/Default.aspx?PageID=4>

### Tomorrow

- In the future, genetic testing will be used to scan all of a person's genetic material, so that disease risk variants can be identified and early intervention and treatment can be planned.
- The cost of testing an individual's entire genome will be less than \$1,000.
- We will live in a time of "personalized medicine," where many treatments for medical conditions will be chosen based upon their effectiveness in patients with a specific genetic makeup learned from genetic testing.

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