Genetic testing

What does genetics testing analyze?

Genetic testing usually refers to the analysis of DNA to identify changes in gene sequence (deletions, additions or misspellings) or expression levels. Genetic testing can also refer to the analysis of RNA to determine gene expression, biochemical tests for the presence of gene products (proteins) and for microscopic analysis of chromosomes.

Types of genetic testing

Thousands of genetic tests are available to aid physicians in the diagnosis and therapy of many diseases. Genetic testing is performed for the following reasons:

- Conformational diagnosis of a symptomatic individual
- Presymptomatic testing for estimating risk of developing disease
- Presymptomatic testing for diagnosing a disease that will manifest later
- Prenatal screening and diagnosis
- Newborn screening
- Preimplantation genetic diagnosis
- Carrier screening
- Forensic testing
- Paternal testing

Genetics testing and clinical care

Genetic testing can be used in the following ways:

- **Diagnostic genetic testing**: Identifies whether an individual has a certain genetic disease. This test detects a specific gene alteration, but is often not able to determine disease severity or age of onset. Thousands of diseases are caused by a mutation in a single gene. Examples include cystic fibrosis and Huntington’s disease.
- **Predictive genetic testing**: Determines whether an individual has an increased risk for a particular disease. Test results indicate probability and are therefore less definitive since
disease susceptibility may also be influenced by other genetic and nongenetic, environmental factors. Diseases that can be identified include certain forms of breast cancer and colorectal cancer.

**Screening genetic tests**: Tests performed on a large segment of the asymptomatic population to determine whether they are in need of more definitive testing to diagnose a genetic disease. Examples are newborn screening and noninvasive prenatal screening.

**Pharmacogenomics**: Identifies variations in an individual’s genetic makeup to determine whether a drug is suitable for that patient, and if so, what would be the safest and most effective dose.

**Whole-genome and whole-exome sequencing**: Examines the entire genome or exome to discover genetic alterations that may be the cause of disease. This test is most often used in complex diagnostic cases, but it is being explored for use in asymptomatic individuals to predict future disease.

**Tumor analysis**: Examines genetic markers in a tumor to determine which genetic alterations are driving tumor growth and which therapies would be most effective.

Evaluating genome sequencing in clinical settings.

**Information on genetic tests for clinical use**

Two resources, the Genetic Testing Registry and GeneTests, list information about specific genetic tests offered by clinical laboratories.