

Genetic Testing & Screening

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Definition: What is Genetic Testing and Screening?

Genetic testing determines the presence or absence of genetic markers linked to specific diseases or disorders. When symptoms, family history, or exposure to toxins put a patient at risk of having or developing a genetic disease or condition, a physician may order a genetic test to determine whether the suspected genetic marker is actually present. A genetic marker can confirm a suspected diagnosis of an illness or indicate a potential to develop a disease or disorder in the future. However, the presence of a particular marker does not necessarily predict the future health of the individual.

Genetic screening is the application of genetic testing to an entire population. The most common form of genetic screening is newborn screening, which began in 1962 and is now used in every state to test nearly 4 million babies per year in the U.S.

Genetic disorders (diseases or conditions) result when there is a mutation or change in the gene, the particular section of DNA, that codes for a protein. The mutation can be as simple as one nucleotide, one letter in the code, being deleted or out of order, but can also result from an entire section of the sequence being deleted, moved, or repeated. As a result of the mutation, the DNA will direct the production of a protein with a different number or order of amino acids. Or in some cases too much or too little of the protein will be made. But when there is a genetic mutation, the protein may not be able to function properly, and one of the body's essential functions may be disrupted, causing symptoms in the patient. In addition to being the correct shape, proteins must also be produced in the correct location, at the correct time, and in the correct amount.

More than one gene can direct production of a single protein. Thus, many genetic disorders are not caused by a single, easily-detectable, variation in just one gene, but rather by a combination of variations across several different areas of the chromosomes.

Types of Genetic Tests and Screens Include:

Carrier Screening: Individuals are tested not because they have symptoms themselves, but to determine if they may carry a gene that could be inherited by a child. The results of carrier screening may be important to reproductive choices. Because every individual has two copies of each chromosome (one inherited from each parent), an alteration on one chromosome may be compensated for by a normal gene on the other chromosome, preventing the individual from developing the disorder. (This is what happens when the gene is recessive: both chromosomes must be altered for symptoms to occur) However, a parent who carries the alteration on one chromosome may pass that chromosome on to a child. If two parents have a disease-causing alteration in the same gene and both are passed to the child, the child will develop the disorder even if neither parent showed symptoms. Common recessive genetic disorders include sickle cell anemia, Tay-Sachs, and cystic fibrosis.

Newborn Screening: Newborns are tested shortly after birth for various genetic disorders. In most hospitals newborn screening is automatically performed unless the mother specifically requests otherwise (a blood sample is usually obtained by a heel prick shortly after birth). Newborn screening can save lives through early diagnosis of diseases such as phenylketonuria (PKU) in which a careful diet limiting phenylalanine consumption can easily prevent serious symptoms, including brain damage.

Genetic Traits: Dominant vs. Recessive --

Each cell in each person contains two copies of each of the 23 chromosomes, one from each parent. The two copies carry codes for all of the same proteins, and all of these codes are in the same places. However, the code on one chromosome from one parent may not be exactly identical to the code on the same chromosome from the other parent. So which code does the cell use? The answer is determined by whether that particular gene, that section of the code, is dominant or recessive. Dominant traits, such as brown hair and blue eyes, are those that appear when only one copy of the chromosome carries the code. Recessive traits, such as blonde hair and blue eyes, are traits that appear only if both copies of the chromosome carry the code.

Prenatal Testing: Testing for genetic disorders is done while the child is still in the womb. Physicians will order this test when there is an increased risk of the child having a genetic disorder due to maternal age, family history, maternal exposure to toxic substances, or abnormal fetal ultrasounds. Testing is done by removing either amniotic fluid (using amniocentesis) or a small piece of the placenta (through chorionic villus sampling), to obtain fetal cells.

Predictive Testing: Individuals are tested for a specific genetic condition even though they are not currently displaying symptoms. Such testing may be undertaken due to a family history of a particular genetic disorder. Often, these disorders do not appear until later in life. Predictive testing can be done for disorders in which eventual development is certain if one is found to have the mutation (called presymptomatic testing), such as Huntington's disease, or for genetic alterations that make a particular disorder more likely but not certain (called predispositional testing), such as cancer. Predispositional testing can determine the likelihood of disease or development, usually expressed as a certain percentage increase in risk, from the general population.

Cancer Genetics --

Cancer is caused by rapid growth and proliferation of cells. Although the process is very complex, there is a significant relationship between the occurrence of cancer and an individual's DNA. Proteins that regulate how quickly cells grow and divide are called oncogenes. An alteration in the DNA sequence of one of these oncogenes can cause cells to grow and divide at an increased rate or without one or more of the important controls.

For more information on the genetics of cancer, please see [links](#) and [further readings](#).

Confirmational Testing/Diagnostic Testing: Individuals are tested to confirm the diagnosis of a genetic disorder for which they are already displaying symptoms. Genetic testing in this case can assist doctors in providing the most effective treatment by ensuring an accurate diagnosis.

Preimplantation Genetic Diagnosis (PGD): Genetic tests are performed on fertilized eggs prior to implantation using in vitro fertilization (IVF). This is often done when a couple that wants to have a child but knows that their offspring are at high risk for a genetic condition. First, the eggs are first fertilized outside of the body. A cell is then removed from each egg and a genetic test is conducted. Then, only those fertilized eggs that do not carry the gene of concern are then implanted into the mother's womb. A few families have also used PGD to select an embryo that will produce a child that is a perfect match to an older sibling with a life-threatening condition. The newborn is then able to provide life-saving cord blood, to the older sibling, and possibly supply bone marrow, tissue, or organs later in life.

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Ethical Issues

The rapidly increasing number of genetic tests--and the growing number of applications for such tests--presents a number of ethical issues, many of them unique to the field of genetic testing and screening. Below are some important facts about genetic testing and screening, and some resulting ethical questions.

- Genetic testing and screening involve complicated science and may have complicated repercussions (such as psychological effects on the patient, or implications for employment or insurance):
 - Should a doctor ever be allowed to test a patient for a genetic condition without their knowledge and consent?
 - How do we ensure that patients have a sufficient understanding of genetic testing in order to give valid consent?
- Researchers have developed, and continue to develop, genetic tests for diseases for which there is not yet an adequate treatment or cure:
 - Should we test for diseases that we have no treatment or cure for?
 - At what age should we test for diseases that will not occur until later in life?
- Genetic testing is often very expensive:
 - Should insurance cover the costs of genetic tests?

- Does it matter if the disease being tested for is treatable?
- Should individuals that can afford it be able to use commercial kits that offer genetic testing results without genetic counseling?
- The results of a genetic test may include information regarding the patient's health status in the distant future. In addition, genetics necessarily involve familial relationships and the result of one family member's test may have important implications for other family members:
 - Who should have access to such information? Only the patient? Family members that may also be affected? Insurance companies? Employers?
 - What if a genetic test reveals unrelated or unintended information such as false paternity? Who is entitled to that information?
- Although environmental factors also play an important role, our genes are often instrumental in the development of our personalities. A genetic test may one day tell us if an individual has a high risk of things like insanity, gambling, or obesity:
 - What impact does such information have on our understanding of such conditions?
 - Will conditions such as these be thought of as mere variations, like hair color, or genetic disorders that should be treated?

Preimplantation Genetic Diagnosis (PGD) also raises a number of additional ethical questions:

- It is possible to test fertilized eggs for genetic disorders, and thus possible to avoid implantation of fetuses with unwanted genetic conditions, including life-threatening or debilitating diseases:
 - Is this a form of eugenics?
 - What about selecting for non-medical conditions like athletic ability or hair color?
- The use of PGD to create savior siblings, children born for the purpose of saving the life of an older sibling, also presents a number of ethical dilemmas:
 - Is the child a means rather than an end? Is this acceptable?
 - Will the child be expected to provide whole organs to the older child later in life if that is necessary?
 - What psychological affect will this have on the child? On the older sibling? On the rest of the family?

Employer Access to Employee Genetic Information

“Most of us get our health insurance through our employers. As a condition of that, your employer actually has access to your medical records...It's not like they go through every person's medical record, but they legitimately and legally have access. So, if there is genetic test information in your record, your employer has access to it. Whether they do anything with it is anybody's guess. But it's not protected ...and that's the point. There has been legislation proposed at the federal level—something called the Genetic Information Non-Discrimination Act (GINA). One of the houses of Congress has passed it. The other hasn't taken it up yet. But that would provide some blanket protection for employees.”

—Dr. Jeffrey Kahn, Director, Center for Bioethics at the University of Minnesota , in an interview with Wendy Wilde on 950 AM Air America Radio MN, on October 26, 2005.

Links

General Resources:

Genetics and Public Policy Center provides a great deal of information on many topics related to genetic testing and screening:

<http://www.dnapolicy.org/>

The United States Department of Energy (DOE) Office of Science maintains a website on genome projects supported by the DOE, especially the Human Genome Project:

<http://www.doegenomes.org/>

The National Library of Medicine maintains current links to several genetics resources:

<http://www.ncbi.nlm.nih.gov/genome/guide/human/resources.shtml>

Biology of Genetics:

The Biology Project website, by the University of Arizona , offers tutorials and problem sets on genetics, forensics, and related topics:

http://www.biology.arizona.edu/human_bio/human_bio.html

BioTeach offers a basic yet thorough explanation of oncogenes, the genes related to cancer development:

<http://www.bioteach.ubc.ca/CellBiology/Oncogenes/index.htm>

"Student Guide to the Human Genome Project" is provided by the United States Department Of Energy:

http://www.ornl.gov/sci/techresources/Human_Genome/education/students.shtml

GeneTest offers a great deal of information include GeneReviews, online expert-authored information on a vast number of genetic disorders, searchable by disease name:

<http://www.geneclinics.org/>

Testing/Screening Programs and Information:

The National Newborn Screening & Genetics Resource Center offers a great deal of information on newborn screening and state-by-state screening programs:

<http://genes-r-us.uthscsa.edu>

The public resource, Universe of Genetic Testing; by Lab Tests Online, offers information and links for various types of genetic screening and testing:

<http://www.labtestsonline.org/understanding/features/genetics.html>

BioTeach also offers a basic yet thorough explanation of preimplantation genetic diagnosis:

<http://www.bioteach.ubc.ca/Bioethics/PreimplantationGeneticDiagnosisAndOurFuture/index.htm>

MedlinePlus offers many resources on both prenatal and genetic testing:

<http://www.nlm.nih.gov/medlineplus/genetictesting.html>

<http://www.nlm.nih.gov/medlineplus/prenataltesting.html>

Genetic Health offers a case study on the genetic testing process:

http://www.genetichealth.com/GT_Genetic_Testing_Case_Study_The_Testing_Process.html

Ethics:

"Choosing the Future: Genetics and Reproductive Decision Making," by the Human Genetics Commission offers a comprehensive look at views of the U.K. on these issues:

<http://www.hgc.gov.uk/UploadDocs/DocPub/Document/ChooseFuturefull.pdf>
(pdf, 195KB, 43sec@56k)

"Your Genes, Your Choices: Exploring the Issues Raised by Genetic Research" by Catherine Baker is available for free download or by order from AAAS:

http://www.ornl.gov/sci/techresources/Human_Genome/publicat/genechoice/index.html

"Minorities, Race, and Genomics" is provided by the United States DOE:

http://www.ornl.gov/sci/techresources/Human_Genome/elsi/minorities.shtml

"A Fact Sheet on Genetic Discrimination in Health Insurance" is provided by the National Human Genome Research Institute:

<http://www.genome.gov/10002328>

New Developments:

Nature Magazine's Genome Gateway provides information on breaking news and new research related to the Human Genome Project:

<http://www.nature.com/genomics/index.html>

To find out how closely related we are to chimpanzees, read "Chimp Genetic Code Opens Human Frontiers," from MSNBC, online at <http://www.msnbc.msn.com/id/9136200/> .

Suggested Reading

"Chinese Geneticists' Views of Ethical Issues in Genetic Testing and Screening: Evidence for Eugenics in China "

Xin, M. *American Journal of Human Genetics* 63 (1998): 688-695.

"Creating a Stem Cell Donor: A Case Study in Reproductive Genetics"

Kahn, J. & Mastroianni, A., *Kennedy Institute of Ethics Journal* 14.1 (2004): 81-96.

"Disclosure of Genetic Tests for Health Insurance: Is It Ethical Not to?"

Raithatha, N. & Smith, R. *The Lancet* January 363 (2004): 395-396.

"Do Races Differ? Not Really, DNA Shows"

Anger, N. *New York Times* , August 22, 2000.

"Down's Syndrome Screening is Unethical: Views of Today's Research Ethics Committees"

Reynolds, TM, *Journal of Clinical Pathology* 56 (2003): 268-70.

"The 'Duty to Warn' a Patient's Family Members About Hereditary Disease Risk"

Offit, K. et al, *JAMA* 292.12 (2004): 1469-1473.

"Ethical Issues Concerning Genetic Testing and Screening in Public Health"

Hodge, J. *American Journal of Medical Genetics* 125C (2004): 66-70.

"Ethical Issues in Genetic Screening for Cancer"

Vineis, P. *Annals of Oncology* 8.10 (1997): 945-949.

"Genetic Discrimination: The Clinician Perspective"

Nedelcu, R. et al., *Clinical Genetics* 66 (2004): 311-317.

"Pharmacogenetic Testing, Informed Consent and the Problem of Secondary Information"

Netzer, C. & Biller-Andorno, N. *Bioethics* 18.4 (2004): 344-360.

"When a Doctor Stumbles on a Family Secret"

Lerner, B. *New York Times* , September, 16, 2003.

Facts & Statistics

The Human Genome is over 3 billion base pairs long. (To be exact, it is 3,076,781,887 nucleotide base pairs long.)

The Human Genome Project was complete approximately two years ahead of schedule.

According to the March of Dimes, 1 out of every 100 American babies is born with a serious genetic defect.

Over 4 million babies are tested in the United States each year through newborn screening programs, and serious diseases are detected in over 3000.

Genetic testing has been used to confirm the presence of the Cohen modal haplotype--associated with the paternally inherited Jewish Priesthood, in the African Lemba tribe--confirming their Jewish heritage.

Tay-Sachs disease is highly prevalent in the Ashkenazi Jewish and French Canadian populations; sickle cell anemia is highly prevalent in African American, African, Mediterranean , and Hispanic populations; and cystic fibrosis is most commonly found in Caucasian populations.

Most breast cancer is thought to be non-hereditary and occurs in individuals with no known risk factors. Women have a 12-13% chance of developing breast cancer, and for every one-hundred cases in women, one man also develops breast cancer. However, a mutation in the BRCA gene increases the risk of developing breast cancer up to 36% to 85%, depending on the mutation.

Over 80 prisoners have been exonerated by DNA evidence since 1992; ten of them were on death row.

Executive Order 13145 prohibits discrimination based upon genetic information in federal employment in the United States . There is no federal law that specifically prohibits discrimination based upon genetic information in private employment.