Genetics Home Reference

Your Guide to Understanding Genetic Conditions

Handbook

Help Me Understand Genetics

Genetic Testing

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Chapter 6

Genetic Testing

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What is genetic testing?

Genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. More than 1,000 genetic tests are currently in use, and more are being developed.

Several methods can be used for genetic testing:

- Molecular genetic tests (or gene tests) study single genes or short lengths of DNA to identify variations or mutations that lead to a genetic disorder.
- Chromosomal genetic tests analyze whole chromosomes or long lengths of DNA to see if there are large genetic changes, such as an extra copy of a chromosome, that cause a genetic condition.
- Biochemical genetic tests study the amount or activity level of proteins; abnormalities in either can indicate changes to the DNA that result in a genetic disorder.

Genetic testing is voluntary. Because testing has benefits as well as limitations and risks, the decision about whether to be tested is a personal and complex one. A geneticist or genetic counselor can help by providing information about the pros and cons of the test and discussing the social and emotional aspects of testing.

For general information about genetic testing:

MedlinePlus offers a list of links to information about genetic testing (http://www.nlm.nih.gov/medlineplus/genetictesting.html).

The National Human Genome Research Institute provides an overview of this topic in its Frequently Asked Questions About Genetic Testing (http://www.genome.gov/19516567). Additional information about genetic testing legislation, policy, and oversight (http://www.genome.gov/10002335) is available from the Institute.

The National Institutes of Health fact sheets Genetic Testing: What It Means for Your Health and for Your Family's Health (http://www.genome.gov/Pages/Health/PatientsPublicInfo/GeneticTestingWhatItMeansForYourHealth.pdf) and Genetic Testing: How it is Used for Healthcare (http://www.nih.gov/about/researchresultsforthepublic/genetictesting.pdf) each provide a brief overview for people considering genetic testing.

Educational resources related to genetic testing (http://geneed.nlm.nih.gov/topic_subtopic.php?tid=41&sid=42) are available from GeneEd.

The Genetics and Public Policy Center also offers information about genetic testing (http://www.dnapolicy.org/science.gt.php).

You can also search for clinical trials involving genetic testing. ClinicalTrials.gov (http://clinicaltrials.gov/), a service of the National Institutes of Health, provides easy access to information on clinical trials. You can search for specific trials or browse by condition or trial sponsor. You may wish to refer to a list of studies related to genetic testing (http://clinicaltrials.gov/search?term=%22genetic+testing%22) that are accepting (or will accept) participants.

What are the types of genetic tests?

Genetic testing can provide information about a person's genes and chromosomes. Available types of testing include:

Newborn screening

Newborn screening is used just after birth to identify genetic disorders that can be treated early in life. Millions of babies are tested each year in the United States. All states currently test infants for phenylketonuria (a genetic disorder that causes mental retardation if left untreated) and congenital hypothyroidism (a disorder of the thyroid gland). Most states also test for other genetic disorders.

Diagnostic testing

Diagnostic testing is used to identify or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms. Diagnostic testing can be performed before birth or at any time during a person's life, but is not available for all genes or all genetic conditions. The results of a diagnostic test can influence a person's choices about health care and the management of the disorder.

Carrier testing

Carrier testing is used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. This type of testing is offered to individuals who have a family history of a genetic disorder and to people in certain ethnic groups with an increased risk of specific genetic conditions. If both parents are tested, the test can provide information about a couple's risk of having a child with a genetic condition.

Prenatal testing

Prenatal testing is used to detect changes in a fetus's genes or chromosomes before birth. This type of testing is offered during pregnancy if there is an increased risk that the baby will have a genetic or chromosomal disorder. In some cases, prenatal testing can lessen a couple's uncertainty or help them make decisions about a pregnancy. It cannot identify all possible inherited disorders and birth defects, however.

Preimplantation testing

Preimplantation testing, also called preimplantation genetic diagnosis (PGD), is a specialized technique that can reduce the risk of having a child with a particular genetic or chromosomal disorder. It is used to detect genetic changes in embryos that were created using assisted reproductive techniques such as in-vitro fertilization. In-vitro fertilization involves removing egg cells from a woman's ovaries and fertilizing them with sperm cells outside the body. To perform preimplantation testing, a small number of cells are taken from these embryos and tested for certain genetic changes. Only embryos without these changes are implanted in the uterus to initiate a pregnancy.

Predictive and presymptomatic testing

Predictive and presymptomatic types of testing are used to detect gene mutations associated with disorders that appear after birth, often later in life. These tests can be helpful to people who have a family member with a genetic disorder, but who have no features of the disorder themselves at the time of testing. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer. Presymptomatic testing can determine whether a person will develop a genetic disorder, such as hemochromatosis (an iron overload disorder), before any signs or symptoms appear. The results of predictive and presymptomatic testing can provide information about a person's risk of developing a specific disorder and help with making decisions about medical care.

Forensic testing

Forensic testing uses DNA sequences to identify an individual for legal purposes. Unlike the tests described above, forensic testing is not used to detect gene mutations associated with disease. This type of testing can identify crime or catastrophe victims, rule out or implicate a crime suspect, or establish biological relationships between people (for example, paternity).

For more information about the uses of genetic testing:

The National Center for Biotechnology Information (NCBI) provides information about the types of genetic testing (http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerusesof.shtml).

A Brief Primer on Genetic Testing (http://www.genome.gov/10506784), which outlines the different kinds of genetic tests, is available from the National Human Genome Research Institute.

Educational resources related to patient genetic testing/carrier screening (http://geneed.nlm.nih.gov/topic_subtopic.php?tid=52&sid=55) are available from GeneEd.

The Centre for Genetics Education offers fact sheets about types of testing used for prenatal diagnosis (http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/Prenatal-Testing-Overview-FS17), preimplantation genetic diagnosis (http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/Preimplantation-Genetic-Diagnosis-FS18), and the medical applications of genetic testing and screening (http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/DNAGeneticTestingTestingforGeneticConditionsandGeneticSusceptibilityFS21).

The National Newborn Screening and Genetics Resource Center (http://genes-r-us.uthscsa.edu/) offers detailed information about newborn screening. Additional information about newborn screening (http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/NewbornScreeningforGeneticConditionsFS20), particularly in Australia, is available from the Centre for Genetics Education.

For information about forensic DNA testing, refer to the fact sheet DNA Forensics (http://www.ornl.gov/sci/techresources/Human_Genome/elsi/forensics.shtml) from the U.S. Department of Energy Office of Science and the fact sheet about forensic genetic testing (http://www.genetics.edu.au/Information/Genetics-Fact-Sheets/DNAGeneticTestingPaternityandForensicUseFS22) from the Centre for Genetics Education.

How is genetic testing done?

Once a person decides to proceed with genetic testing, a medical geneticist, primary care doctor, specialist, or nurse practitioner can order the test. Genetic testing is often done as part of a genetic consultation.

Genetic tests are performed on a sample of blood, hair, skin, amniotic fluid (the fluid that surrounds a fetus during pregnancy), or other tissue. For example, a procedure called a buccal smear uses a small brush or cotton swab to collect a sample of cells from the inside surface of the cheek. The sample is sent to a laboratory where technicians look for specific changes in chromosomes, DNA, or proteins, depending on the suspected disorder. The laboratory reports the test results in writing to a person's doctor or genetic counselor.

Newborn screening tests are done on a small blood sample, which is taken by pricking the baby's heel. Unlike other types of genetic testing, a parent will usually only receive the result if it is positive. If the test result is positive, additional testing is needed to determine whether the baby has a genetic disorder.

Before a person has a genetic test, it is important that he or she understands the testing procedure, the benefits and limitations of the test, and the possible consequences of the test results. The process of educating a person about the test and obtaining permission is called informed consent.

For more information about genetic testing procedures:

The National Center for Biotechnology Information (NCBI) explains the testing process and informed consent (http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerordertest.shtml).

Scientific Testimony, an online journal, provides an introduction to DNA testing techniques (http://www.scientific.org/tutorials/articles/riley/riley.html) written for the general public.

What is direct-to-consumer genetic testing?

Traditionally, genetic tests have been available only through healthcare providers such as physicians, nurse practitioners, and genetic counselors. Healthcare providers order the appropriate test from a laboratory, collect and send the samples, and interpret the test results. Direct-to-consumer genetic testing refers to genetic tests that are marketed directly to consumers via television, print advertisements, or the Internet. This form of testing, which is also known as at-home genetic testing, provides access to a person's genetic information without necessarily involving a doctor or insurance company in the process.

If a consumer chooses to purchase a genetic test directly, the test kit is mailed to the consumer instead of being ordered through a doctor's office. The test typically involves collecting a DNA sample at home, often by swabbing the inside of the cheek, and mailing the sample back to the laboratory. In some cases, the person must visit a health clinic to have blood drawn. Consumers are notified of their results by mail or over the telephone, or the results are posted online. In some cases, a genetic counselor or other healthcare provider is available to explain the results and answer questions. The price for this type of at-home genetic testing ranges from several hundred dollars to more than a thousand dollars.

The growing market for direct-to-consumer genetic testing may promote awareness of genetic diseases, allow consumers to take a more proactive role in their health care, and offer a means for people to learn about their ancestral origins. At-home genetic tests, however, have significant risks and limitations. Consumers are vulnerable to being misled by the results of unproven or invalid tests. Without guidance from a healthcare provider, they may make important decisions about treatment or prevention based on inaccurate, incomplete, or misunderstood information about their health. Consumers may also experience an invasion of genetic privacy if testing companies use their genetic information in an unauthorized way.

Genetic testing provides only one piece of information about a person's health—other genetic and environmental factors, lifestyle choices, and family medical history also affect a person's risk of developing many disorders. These factors are discussed during a consultation with a doctor or genetic counselor, but in many cases are not addressed by at-home genetic tests. More research is needed to fully understand the benefits and limitations of direct-to-consumer genetic testing.

For more information about direct-to-consumer genetic testing:

The American College of Medical Genetics, which is a national association of doctors specializing in genetics, has issued a statement on direct-to-consumer genetic

testing (http://www.acmg.net/AM/Template.cfm?Section=Policy_Statements&Template=/CM/ContentDisplay.cfm&ContentID=2975).

The American Society of Human Genetics, a professional membership organization for specialists in genetics, has also issued a statement on direct-to-consumer genetic testing in the United States (http://ashg.org/pdf/dtc_statement.pdf).

The Federal Trade Commission (FTC) works to protect consumers and promote truth in advertising. The FTC offers a fact sheet for consumers (http://www.consumer.ftc.gov/articles/0166-home-genetic-tests) about the benefits and risks of at-home genetic tests.

An issue brief on direct-to-consumer genetic testing (http://www.dnapolicy.org/policy.issue.php?action=detail&issuebrief_id=32) is available from the Genetics & Public Policy Center.

The Genetic Alliance also provides information about the promotion of genetic testing services directly to consumers (http://www.geneticalliance.org/issues.testing.consumers).

Additional information about direct-to-consumer marketing of genetic tests (http://www.genome.gov/12010659) is available from the National Human Genome Research Institute.

How can consumers be sure a genetic test is valid and useful?

Before undergoing genetic testing, it is important to be sure that the test is valid and useful. A genetic test is valid if it provides an accurate result. Two main measures of accuracy apply to genetic tests: analytical validity and clinical validity. Another measure of the quality of a genetic test is its usefulness, or clinical utility.

- Analytical validity refers to how well the test predicts the presence or absence of a particular gene or genetic change. In other words, can the test accurately detect whether a specific genetic variant is present or absent?
- Clinical validity refers to how well the genetic variant being analyzed is related to the presence, absence, or risk of a specific disease.
- Clinical utility refers to whether the test can provide information about diagnosis, treatment, management, or prevention of a disease that will be helpful to a consumer.

All laboratories that perform health-related testing, including genetic testing, are subject to federal regulatory standards called the Clinical Laboratory Improvement Amendments (CLIA) or even stricter state requirements. CLIA standards cover how tests are performed, the qualifications of laboratory personnel, and quality control and testing procedures for each laboratory. By controlling the quality of laboratory practices, CLIA standards are designed to ensure the analytical validity of genetic tests.

CLIA standards do not address the clinical validity or clinical utility of genetic tests. The Food and Drug Administration (FDA) requires information about clinical validity for some genetic tests. Additionally, the state of New York requires information on clinical validity for all laboratory tests performed for people living in that state. Consumers, health providers, and health insurance companies are often the ones who determine the clinical utility of a genetic test.

It can be difficult to determine the quality of a genetic test sold directly to the public. Some providers of direct-to-consumer genetic tests are not CLIA-certified, so it can be difficult to tell whether their tests are valid. If providers of direct-to-consumer genetic tests offer easy-to-understand information about the scientific basis of their tests, it can help consumers make more informed decisions. It may also be helpful to discuss any concerns with a health professional before ordering a direct-to-consumer genetic test.

For more information about determining the quality of genetic tests:

The Centers for Disease Control and Prevention (CDC) provides an explanation of the factors used to evaluate genetic tests (http://www.cdc.gov/genomics/gtesting/ACCE/index.htm), including analytical validity, clinical validity, and clinical utility, as part of their ACCE project. Additional information about the ACCE framework (http://www.phgfoundation.org/tutorials/acce/) is available in an interactive tutorial from the PHG Foundation.

A brief overview of the regulation of genetic testing (http://www.dnapolicy.org/policy.issue.php?action=detail&issuebrief_id=10) is available from the Genetics & Public Policy Center.

The Genetic Alliance offers information about the quality of genetic tests and current public policy issues (http://www.geneticalliance.org/issues.testing.quality) surrounding their regulation.

An interactive tutorial about clinical utility (http://www.phgfoundation.org/tutorials/clinicalUtility/) is available from the PHG Foundation.

The U.S. Centers for Medicare and Medicaid Services (CMS) provide an overview of the Clinical Laboratory Improvement Amendments (CLIA) (http://www.cms.gov/Regulations-and-Guidance/Legislation/CLIA/).

Additional information about the oversight of genetic testing in the United States is available from a Report of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) (http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf).

What do the results of genetic tests mean?

The results of genetic tests are not always straightforward, which often makes them challenging to interpret and explain. Therefore, it is important for patients and their families to ask questions about the potential meaning of genetic test results both before and after the test is performed. When interpreting test results, healthcare professionals consider a person's medical history, family history, and the type of genetic test that was done.

A positive test result means that the laboratory found a change in a particular gene, chromosome, or protein of interest. Depending on the purpose of the test, this result may confirm a diagnosis, indicate that a person is a carrier of a particular genetic mutation, identify an increased risk of developing a disease (such as cancer) in the future, or suggest a need for further testing. Because family members have some genetic material in common, a positive test result may also have implications for certain blood relatives of the person undergoing testing. It is important to note that a positive result of a predictive or presymptomatic genetic test usually cannot establish the exact risk of developing a disorder. Also, health professionals typically cannot use a positive test result to predict the course or severity of a condition.

A negative test result means that the laboratory did not find a change in the gene, chromosome, or protein under consideration. This result can indicate that a person is not affected by a particular disorder, is not a carrier of a specific genetic mutation, or does not have an increased risk of developing a certain disease. It is possible, however, that the test missed a disease-causing genetic alteration because many tests cannot detect all genetic changes that can cause a particular disorder. Further testing may be required to confirm a negative result.

In some cases, a negative result might not give any useful information. This type of result is called uninformative, indeterminate, inconclusive, or ambiguous. Uninformative test results sometimes occur because everyone has common, natural variations in their DNA, called polymorphisms, that do not affect health. If a genetic test finds a change in DNA that has not been associated with a disorder in other people, it can be difficult to tell whether it is a natural polymorphism or a disease-causing mutation. An uninformative result cannot confirm or rule out a specific diagnosis, and it cannot indicate whether a person has an increased risk of developing a disorder. In some cases, testing other affected and unaffected family members can help clarify this type of result.

For more information about interpreting genetic test results:

The Department of Energy, Office of Science offers information about evaluating gene tests (http://www.ornl.gov/sci/techresources/Human_Genome/resource/testeval.shtml).

The National Center for Biotechnology Information (NCBI) provides a brief discussion of interpreting test results (http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerordertest.shtml#testresult) and a sample laboratory report (http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/labreport.shtml) for a genetic test.

The National Women's Health Resource Center offers a list of questions about genetic testing (http://www.healthywomen.org/condition/genetic-testing#hc-tab-1), including the meaning of test results, that patients and families can ask their healthcare professional.

What is the cost of genetic testing, and how long does it take to get the results?

The cost of genetic testing can range from under $100 to more than $2,000, depending on the nature and complexity of the test. The cost increases if more than one test is necessary or if multiple family members must be tested to obtain a meaningful result. For newborn screening, costs vary by state. Some states cover part of the total cost, but most charge a fee of $15 to $60 per infant.

From the date that a sample is taken, it may take a few weeks to several months to receive the test results. Results for prenatal testing are usually available more quickly because time is an important consideration in making decisions about a pregnancy. The doctor or genetic counselor who orders a particular test can provide specific information about the cost and time frame associated with that test.

For more information about the costs and turnaround time for genetic tests:

The National Center for Biotechnology Information (NCBI) provides a list of factors that influence the turnaround time and costs of genetic testing (http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerordertest.shtml#choosing). Scroll down to the sections called “Turn-Around Time” and “Cost.”

Will health insurance cover the costs of genetic testing?

In many cases, health insurance plans will cover the costs of genetic testing when it is recommended by a person's doctor. Health insurance providers have different policies about which tests are covered, however. A person interested in submitting the costs of testing may wish to contact his or her insurance company beforehand to ask about coverage.

Some people may choose not to use their insurance to pay for testing because the results of a genetic test can affect a person's health insurance coverage. Instead, they may opt to pay out-of-pocket for the test. People considering genetic testing may want to find out more about their state's privacy protection laws before they ask their insurance company to cover the costs. (Refer to What is genetic discrimination? (http://ghr.nlm.nih.gov/handbook/testing/discrimination) for more information.)

For more information about insurance coverage of genetic testing:

The U.S. Department of Energy Office of Science provides a brief discussion of insurance coverage for genetic testing (http://www.ornl.gov/sci/techresources/Human_Genome/medicine/genetest.shtml#insurance).

What are the benefits of genetic testing?

Genetic testing has potential benefits whether the results are positive or negative for a gene mutation. Test results can provide a sense of relief from uncertainty and help people make informed decisions about managing their health care. For example, a negative result can eliminate the need for unnecessary checkups and screening tests in some cases. A positive result can direct a person toward available prevention, monitoring, and treatment options. Some test results can also help people make decisions about having children. Newborn screening can identify genetic disorders early in life so treatment can be started as early as possible.

For more information about the benefits of genetic testing:

The National Cancer Institute provides a brief discussion of the benefits of genetic testing (http://www.cancer.gov/cancertopics/understandingcancer/genetesting/page29).

Additional information on this topic is available in the fact sheet Gene Testing (http://www.ornl.gov/sci/techresources/Human_Genome/medicine/genetest.shtml#procon) from the U.S. Department of Energy Office of Science.

What are the risks and limitations of genetic testing?

The physical risks associated with most genetic tests are very small, particularly for those tests that require only a blood sample or buccal smear (a procedure that samples cells from the inside surface of the cheek). The procedures used for prenatal testing carry a small but real risk of losing the pregnancy (miscarriage) because they require a sample of amniotic fluid or tissue from around the fetus.

Many of the risks associated with genetic testing involve the emotional, social, or financial consequences of the test results. People may feel angry, depressed, anxious, or guilty about their results. In some cases, genetic testing creates tension within a family because the results can reveal information about other family members in addition to the person who is tested. The possibility of genetic discrimination in employment or insurance is also a concern. (Refer to What is genetic discrimination? (http://ghr.nlm.nih.gov/handbook/testing/discrimination) for additional information.)

Genetic testing can provide only limited information about an inherited condition. The test often can't determine if a person will show symptoms of a disorder, how severe the symptoms will be, or whether the disorder will progress over time. Another major limitation is the lack of treatment strategies for many genetic disorders once they are diagnosed.

A genetics professional can explain in detail the benefits, risks, and limitations of a particular test. It is important that any person who is considering genetic testing understand and weigh these factors before making a decision.

For more information about the risks and limitations of genetic testing:

The National Cancer Institute provides a brief discussion of the limitations of genetic testing:

- Limitations of Gene Testing (http://www.cancer.gov/cancertopics/understandingcancer/genetesting/page30)
- Major Limitations of Gene Testing (http://www.cancer.gov/cancertopics/understandingcancer/genetesting/page31)

The National Center for Biotechnology Information (NCBI) outlines points to consider for each type of genetic testing (http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerusesof.shtml).

What is genetic discrimination?

Genetic discrimination occurs when people are treated differently by their employer or insurance company because they have a gene mutation that causes or increases the risk of an inherited disorder. People who undergo genetic testing may be at risk for genetic discrimination.

The results of a genetic test are normally included in a person's medical records. When a person applies for life, disability, or health insurance, the insurance company may ask to look at these records before making a decision about coverage. An employer may also have the right to look at an employee's medical records. As a result, genetic test results could affect a person's insurance coverage or employment. People making decisions about genetic testing should be aware that when test results are placed in their medical records, the results might not be kept private.

Fear of discrimination is a common concern among people considering genetic testing. Several laws at the federal and state levels help protect people against genetic discrimination; however, genetic testing is a fast-growing field and these laws don't cover every situation.

For more information about privacy and genetic discrimination:

The National Human Genome Research Institute provides a detailed discussion of genetic discrimination and current laws that address this issue:

- Genetic Discrimination in Health Insurance or Employment (http://www.genome.gov/11510227)
- Privacy and Discrimination in Genetics (http://www.genome.gov/10002077)
- NHGRI Policy and Legislation Database (http://www.genome.gov/PolicyEthics/LegDatabase/pubsearch.cfm)

The Genetic Alliance offers links to resources and policy statements on genetic discrimination (http://www.geneticalliance.org/issues.discrimination).

Additional information about policy and legislation related to genetic privacy (http://www.ornl.gov/sci/techresources/Human_Genome/elsi/legislat.shtml) is available from the U.S. Department of Energy Office of Science.

The Australian Research Council's Genetic Discrimination Project (http://www.gdproject.org/) is studying the impact of genetic discrimination on consumers, third parties (such as insurers), and the legal system in Australia.

How does genetic testing in a research setting differ from clinical genetic testing?

The main differences between clinical genetic testing and research testing are the purpose of the test and who receives the results. The goals of research testing include finding unknown genes, learning how genes work, and advancing our understanding of genetic conditions. The results of testing done as part of a research study are usually not available to patients or their healthcare providers. Clinical testing, on the other hand, is done to find out about an inherited disorder in an individual patient or family. People receive the results of a clinical test and can use them to help them make decisions about medical care or reproductive issues.

It is important for people considering genetic testing to know whether the test is available on a clinical or research basis. Clinical and research testing both involve a process of informed consent in which patients learn about the testing procedure, the risks and benefits of the test, and the potential consequences of testing.

For more information about the differences between clinical and research testing:

The National Center for Biotechnology Information (NCBI) outlines the major differences between clinical tests and research tests (http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/concepts/primer/primerwhatistest.shtml). Scroll down to the sections “What is a Clinical Test?” and “What is a Research Test?”

What is genetic ancestry testing?

Genetic ancestry testing, or genetic genealogy, is a way for people interested in family history (genealogy) to go beyond what they can learn from relatives or from historical documentation. Examination of DNA variations can provide clues about where a person's ancestors might have come from and about relationships between families. Certain patterns of genetic variation are often shared among people of particular backgrounds. The more closely related two individuals, families, or populations are, the more patterns of variation they typically share.

Three types of genetic ancestry testing are commonly used for genealogy:

- Y chromosome testing: Variations in the Y chromosome, passed exclusively from father to son, can be used to explore ancestry in the direct male line. Y chromosome testing can only be done on males, because females do not have a Y chromosome. However, women interested in this type of genetic testing sometimes recruit a male relative to have the test done. Because the Y chromosome is passed on in the same pattern as are family names in many cultures, Y chromosome testing is often used to investigate questions such as whether two families with the same surname are related.
- Mitochondrial DNA testing: This type of testing identifies genetic variations in mitochondrial DNA. Although most DNA is packaged in chromosomes within the cell nucleus, cell structures called mitochondria also have a small amount of their own DNA (known as mitochondrial DNA). Both males and females have mitochondrial DNA, which is passed on from their mothers, so this type of testing can be used by either sex. It provides information about the direct female ancestral line. Mitochondrial DNA testing can be useful for genealogy because it preserves information about female ancestors that may be lost from the historical record because of the way surnames are often passed down.

- Single nucleotide polymorphism (http://ghr.nlm.nih.gov/handbook/genomicresearch/snp) testing: These tests evaluate large numbers of variations (single nucleotide polymorphisms or SNPs) across a person's entire genome. The results are compared with those of others who have taken the tests to provide an estimate of a person's ethnic background. For example, the pattern of SNPs might indicate that a person's ancestry is approximately 50 percent African, 25 percent European, 20 percent Asian, and 5 percent unknown. Genealogists use this type of test because Y chromosome and mitochondrial DNA test results, which represent only single ancestral lines, do not capture the overall ethnic background of an individual.

Genetic ancestry testing has a number of limitations. Test providers compare individuals' test results to different databases of previous tests, so ethnicity estimates may not be consistent from one provider to another. Also, because most human populations have migrated many times throughout their history and mixed with nearby groups, ethnicity estimates based on genetic testing may differ from an individual's expectations. In ethnic groups with a smaller range of genetic variation due to the group's size and history, most members share many SNPs, and it may be difficult to distinguish people who have a relatively recent common ancestor, such as fourth cousins, from the group as a whole.

Genetic ancestry testing is offered by several companies and organizations. Most companies provide online forums and other services to allow people who have been tested to share and discuss their results with others, which may allow them to discover previously unknown relationships. On a larger scale, combined genetic ancestry test results from many people can be used by scientists to explore the history of populations as they arose, migrated, and mixed with other groups.

For more information about genetic ancestry testing:

The British Broadcasting Company offers an introductory article on genetic genealogy (http://www.bbc.co.uk/history/familyhistory/next_steps/genetic_genealogy_01.shtml).

The University of Utah provides video tutorials (http://learn.genetics.utah.edu/content/extras/molgen/) on molecular genealogy.

The International Society of Genetic Genealogy (http://www.isogg.org/) promotes the use of DNA testing in genealogy.

The American Society of Human Genetics (ASHG) developed a position paper on ancestry testing (http://www.ashg.org/pdf/ASHGAncestryTestingStatement_FINAL.pdf).

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http://ghr.nlm.nih.gov/

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Department of Health & Human Services

Handbook
Help Me Understand Genetics

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