

Genetic Testing

Genetic testing is a medical/laboratory test used to determine if a person has genetic abnormalities. According to the Human Genome Project there are approximately between 20,000 and 25,000 genes within a human. Of the genes that can be tested and identified for defects, here some tests that can be done.

Preimplantion testing is done when a woman conceives through in-vitro fertilization. After her eggs are fertilized, cells are extracted and tested for abnormalities. The fertilized eggs without any genetic defects are then implanted back within the woman to carry out the pregnancy.

Prenatal testing is usually done when a woman is in her 16th to 18th week of pregnancy. Amniotic fluid is taken from the placenta (amniocentesis) and the cells within the fluid are tested. This test is not often performed unless there is a history of child having a genetic defect and/or the mother's age or a family history of a genetic disorder.

Newborn screenings: there are two tests required on all newborns shortly after birth. The PKU or phenylketonuria and congenital hypothyroidism test. All states require these two tests but many states do additional screenings or tests.

Diagnostic and Predictive & Presymptomatic testing. Diagnostic testing is done when a genetic disorder is suspected. The test cannot identify all genes or abnormalities but they are a good start to identifying or excluding a genetic disorder. Predictive and presymptomatic testing is done to determine if a person has a genetic disorder before signs of the disorder begin to show.

Forensic testing does not test for an abnormality within a gene. These simple tests are used in cases where a person needs to be identified or excluded using their DNA. Two examples are criminal cases and paternity cases.

The costs of the test can range from \$200 to \$3000. It all depends on the type of laboratory procedure being used, the cost of labor, the size of the gene being tested, patent costs, whether a commercial or research lab is conducting the test. The cost can be high also because of the many people that review the test before the results are sent, shipping costs, doctors and counselors fees and the fact that these tests are still rare.

It is always best to confer with your insurance company before any medical procedure and genetic testing is no exception. Insurance companies may cover the costs if the test is ordered/requested by a doctor. Some detractors to this will cite that if a test comes back and there is a genetic disorder the person will be discriminated against. On April 24, 2008 the Senate passed a bill called GINA. The Genetic Information Nondiscrimination Act prevents employers and health insurance companies from using a person's genetic results against them. On May 1, 2008 the House passed GINA and it is on its way for the President to sign.

Another issue with privacy is what they call DTC or Direct to Consumer testing. If a person wishes to have themselves tested, there are commercial labs providing this service, for a fee. One example is for genealogy purposes, they can test the mother's line (mitochondrial – this test costs more) and the father's line. Vital information can be lost or stolen in the mail and there are virtually no government regulations over DTC as opposed to government sanctioned research testing.

Last, although this is actually one of the first steps, one should visit a Genetic Counselor. They can be a counselor, nurse, doctor, or a geneticist. In addition to the wealth of knowledge now available on the Internet a

genetic counselor can help someone every step of the genetic testing process. Counselors are not always seen as medically necessary by insurance standards, so it is best to check with the insurance company first. It would not be unheard of to pay for many of these things yourself. Genetic testing has advanced since the Human Genome Project began their research in 1990, but sadly the health insurance industry has yet to advance themselves.

The websites given on the fact sheet and in the bibliography are a good place to start. The Genetics Home Reference website gives information on cells and DNA with pictures (Biology 101 revisited) and information on the Human Genome Project in addition to genetics and genetic testing. The National Human Genome Research Institute is a good website concerning government policy and ethics regarding genetic testing and also is a good source for ongoing and upcoming research and grants the government is sponsoring.

References

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Fact Sheet

Genetic testing is a medical/laboratory procedure used to determine abnormalities within a person's genetic makeup.

A single person carries 46 chromosomes, 23 from their mother and 23 from their father. Within the 46 chromosomes there are between 20,000 and 25,000 genes.

- ***Types of Genetic Testing:***

Preimplantation or PGD (Preimplantation Genetic Diagnosis). Used with in-vitro fertilization. After a woman's eggs are fertilized, they are tested for genetic problems. Those without any problems are implanted back into to the woman.

Prenatal testing. Using the amniotic fluid from the placenta, this test is done during pregnancy to identify genetic abnormalities with the fetus. Usually done between the 16th and 18th week of pregnancy.

Newborn Screening. After a baby is born (in a hospital), all babies are tested for PKU (phenylketonuria) and Congenital Hypothyroidism in all states.

Diagnostic testing. Done to identify or exclude a certain genetic abnormality.

Predictive and presymptomatic testing. Done to determine if someone has a genetic abnormality before symptoms begin to start.

Carrier testing. Done when there is a family history of genetic disorders.

Forensic testing. Not used to identify genetic abnormalities but just to identify a person in instances of crimes or paternity.

- ***How a sample is collected:***

Samples are taken from the blood, skin cells, hair follicles and amniotic fluid.

- ***Costs:***

They can vary from \$200-\$3000 depending on the type and amount of tests performed, insurance coverage and out of pocket expenses.

- ***Excellent websites to get started:***

<http://www.genome.gov> (Human Genome Research Institute)

<http://www.dnapolicy.org> (Genetics & Public Policy Center)

<http://ghr.nlm.nih.gov> (Genetics Home Reference – US National Library of Medicine)

<http://www.genetichealth.com> (Genetic Health Institute)

