

## **Genetic Counseling**

### **What is genetic counseling?**

Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. The process integrates the following elements:

- Interpreting family history and medical information to assess risk of disease occurrence or recurrence
- Education about inheritance, testing, management, prevention, resources and research
- Counseling to promote informed decision-making and adaptation to the risk or condition

Trained genetics professionals include doctors who specialize as medical geneticists, genetic counselors (health professionals with specialized graduate degrees) and genetic nurses.

If you are concerned about hereditary disease, you may benefit from a genetic consultation, an important part of the decision-making process for genetic testing.

### **What to expect in your consultation**

A genetic consultation provides information, addresses a patient's specific questions and concerns, and offers support.

During a consultation, a genetics professional will:

- Assess the risk of a genetic disorder by researching your family's history and evaluating medical records.
- Help you weigh the medical, social and ethical decisions surrounding genetic testing to help you make informed, independent decisions about your health care.
- Interpret the results of genetic tests and medical data.
- Offer supportive counseling and assist you in coping with difficult situations and feelings.
- Respect your individual beliefs, traditions and feelings.
- Serve as patient advocates.
- Discuss reproductive options.
- Explore strategies for communicating information to others, especially family members who may be at risk.
- Provide written materials and referrals to support groups, other families with the same or similar condition, and local and national service agencies.

A genetics professional will not:

- Tell you which decision to make.
- Advise you not to have children.
- Recommend that a woman continue or end a pregnancy.
- Tell you whether to undergo testing for a genetic disorder.

## **What is genetic testing?**

Genetic testing identifies changes in DNA, RNA, genes, chromosomes or proteins – the blueprint

which instructs how we grow and develop. The result of a genetic test can often confirm or exclude a suspected genetic condition or help determine a person's chance of developing or transmitting a genetic disorder. Unlike routine blood tests, the results from a genetic test will not change over time and only need to be performed once in a person's life.

Approximately 20-50% of all individuals with frontotemporal dementia (FTD) have a known family history. This means that 50-80% of those affected have no known family history of the disorder. Sometimes, this is due to a misdiagnosis in the family with another condition, such as Parkinson disease, Alzheimer's or psychiatric illness. For this reason, individuals in the family may be at-risk for a "familial" disorder without prior knowledge. Genetic counseling and testing can help clarify this risk.

Genetic testing can be performed to confirm a suspected diagnosis of FTD, assess risk for extended family members or identify an individual with increased risk for developing the disease (predictive testing). Testing should first be performed on an affected individual to determine whether or not there is an identifiable genetic or sporadic form of the disease in the family. Genetic testing identifies a mutation in a gene known to cause FTD approximately 10% of the time.

It is important to remember that genetic testing is voluntary. Because testing has both benefits and limitations, this decision is personal and complex. The physical risks associated with most genetic tests are small and involve a simple blood draw. Most of the risks associated with genetic testing involve possible emotional, social or financial consequences of the test results. You may feel sad, angry, frightened, depressed, anxious or guilty after learning your results. Learning that one carries a mutation in a gene that results in a progressive, lethal disorder can be devastating and should be considered very carefully.

On the other hand, test results can give you a sense of relief from uncertainty and help you make informed decisions about managing your health care. The test results might impact life decisions, such as career choice, family planning, or insurance coverage. Your genetic counselor can explain in detail the benefits, risks and limitations of a particular test. It is important that you understand and weigh these factors before making a decision.

If you decide to proceed with genetic testing, often a sample can often be collected following your consultation. The sample is then sent to a laboratory where technicians look for the specific changes in one of the four genes associated with FTD. The laboratory will report the results to your doctor and/or genetic counselor who will discuss the results with you in person.

## **What do the results mean?**

The results of genetic tests are not always straightforward, which often makes them challenging to interpret and explain. When interpreting test results, your doctor or genetic counselor considers your medical history, family history and the type of genetic test that was done.

A positive test result means that the laboratory detected a mutation (change) in a gene known to cause FTD. Depending on your health status, this result may confirm an existing diagnosis or it may indicate that you are at increased risk for developing FTD in the future. A positive test result may have implications for other blood relatives.

A negative test result means that the laboratory did not find a mutation in the tested gene. This negative test result must be interpreted with caution. Sometimes, a negative test result can suggest a sporadic form of the disease. In this case, other family members are not thought to be at increased risk for the condition. However, in other cases, a negative test result does not entirely exclude the possibility of a familial condition. It is important to discuss the test results with a genetic counselor or doctor to better understand the implications for yourself and family members.

In rare cases, the genetic test result is inconclusive. This occurs when the test identifies a natural variation in a gene, called a polymorphism. Polymorphisms are common in the population and, generally, do not affect your health. Sometimes it is difficult to know if a variation in a person's DNA is a very rare (uncommon) polymorphism or a mutation (a change in the DNA that will cause a genetic disease). If the genetic test reveals a change that has not previously been associated with a disorder in other people, this can sometimes be difficult to determine, and, therefore, results remain uncertain.

## **The Genetic Information Nondiscrimination Act**

The Genetic Information Nondiscrimination Act of 2008 (GINA) was enacted on May 21, 2008 to protect Americans against discrimination based on their genetic information when it comes to health insurance and employment. GINA prohibits group health plans and health insurers from denying coverage or charging higher premiums to a healthy individual based solely on a genetic predisposition to developing a disease in the future. The legislation also bars employers from using individuals' genetic information when making hiring, firing or promotion decisions. This means that if you decide to undergo genetic testing, you may have to share your results with your employer or insurance company (most likely if they pay for the testing), but if the results are shared, they cannot be used to make decisions about your employment or health insurance coverage.

The insurance provisions of the bill will take effect May 21, 2009, with the employment provisions going into effect on November 21, 2009. To read the actual bill, please [click here](#) .