

Genetic Counseling

- What is a genetic consultation?
- What happens during a genetic consultation?
- What is genetic testing?
- What do the results mean?

What is genetic counseling?

A genetic consultation is a health service that involves evaluation of an individual or family for one or more of the following:

- Confirming, diagnosing or ruling out a genetic condition
- Identifying medical management issues
- Calculating and communicating genetic risks
- Providing or arranging for support

Trained genetics professionals include doctors who specialize as medical geneticists, genetic counselors (certified healthcare workers) and genetic nurses.

If you are concerned about hereditary human prion disease, you may benefit from a genetic consultation. A genetic consultation is 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 support and help with feelings of guilt, grief and fear.
- 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 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.

In genetic prion disease, diagnostic testing can identify the presence of the mutations in the prion protein gene that confirm the diagnosis of the genetic form of prion disease. A negative genetic test can rule out the possibility that a patient has a genetic form of prion disease. Predictive genetic testing can identify an increased risk of developing prion disease, even if you are not yet showing symptoms. Genetic testing can be performed with a simple blood sample. A family history of prion disease is usually present in only 40% of cases of genetic prion disease. This means that in 60% of genetic cases of prion disease there is not a known family history! In these cases, however, a careful review of all biologically related family members often reveals a history of "Alzheimer's," "Parkinson's", or psychiatric illness that was likely misdiagnosed. Therefore, it is generally necessary to do genetic testing if you want to exclude the possibility of genetic disease. Because some families may have a genetic mutation in their family, but not know it, they may not know that they have a "familial" disease.

Genetic testing is voluntary. Because testing has both benefits and limitations, the decision about whether to be tested is a personal and complex one. The physical risks associated with most genetic tests are very small. Many of the risks associated with genetic testing involve the emotional, social or financial consequences of the test results. You may feel angry, depressed, anxious, or guilty about your results. Learning that one carries a gene for a fatal disease 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 help you make 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 be collected during your consultation. The sample is then sent to a laboratory where technicians look for the specific changes in the prion gene. The laboratory will report the results to your doctor or genetic counselor.

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 considers your medical history, family history and the type of genetic test that was done.

A positive test result means that the laboratory found the expected changes in the prion gene. Depending on your health status, this result may confirm a diagnosis, indicate that you are a carrier of the genetic mutation or identify an increased risk of developing CJD in the future. A positive test result may have implications for blood relatives too.

A negative test result means that the laboratory did not find an abnormal change (mutation) in your gene. This negative test result indicates that you do not have genetic prion disease, because you do not have a genetic mutation in the prion protein gene.

In some rare cases, an inconclusive test result sometimes occurs because everyone has common, natural variations in their DNA, called polymorphisms, which 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 genetic prion disease). If the 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 rare, natural polymorphism or a disease-causing mutation.