



## Basic Information about Genetics: What's It All About? Care4Dystonia, Inc.

Over the past ten years new scientific methods have been developed to locate and identify the genetic aspects of inherited diseases including common disorders such as colon cancer. The genes responsible for causing hemophilia, breast cancer, several neuromuscular diseases, and a form of blindness have also been identified by medical researchers in recent years. Diagnostic tests are now available to identify a person's potential risk for the development of a number of diseases that may "run" in their family. This is true of several movement disorders.

This is very promising news for all of us involved in the field of medical genetics. This brochure has been specifically developed to answer questions you may have concerning genes, genetic disorders, diagnostic tests and genetic counseling services that are available to you and your family.

To fully comprehend genetic disorders and the services that your Movement Disorder Center can provide to you and your family, let's review some important aspects of cell biology first.

We would be happy to answer any questions that you may have as you read this information.

### Reviewing Cell Biology....

Your body is made up of billions of cells.

Each of these cells has a specific function- nerve cells (neurons) found in your brain help to coordinate and transmit messages to other areas of your body. Specialized nerve cells, found in the basal ganglia region of your brain, help to coordinate movement. Bone cells provide structural support for your body. Thus a cell or a group of cells are highly specialized in relation to function.

Although highly specialized, many of the cells in your body share common features. One of these features is a structure called the nucleus. The nucleus is the command center for each cell. This is similar to the brain being the "command center" of your body.

The nucleus in each cell contains structures called chromosomes.

Chromosomes contain genetic information that is packaged into structures we call DNA (deoxyribonucleic acid).

DNA is made up of molecules of sugar, phosphate and four different types of proteins. Portions of DNA carry "coded instructions that allow a cell to perform its highly specialized function in the body.

The portions carrying the "coded instructions" are called GENES.

There are over 100,000 genes that determine our traits. Many of the cells in your body divide and replicate. During cell replication genetic information is copied and "passed" on to new cells.

Sometimes, due to an "error" in replication, portions of DNA (GENES) may not be copied and "passed" on to new cells correctly. These errors are called mutations. Thus, as these new "mutated" cells divide and replicate, mutations are inherited by new cells. A mutation does not always cause an adverse effect on a person. However, some mutations can be harmful to a person. For example, a mutation in a gene for a protein required for effective vision can result in color blindness. It is important to understand that a mutation occurring in a specific cell type does not necessarily effect all of the other cells in your body.

You have inherited specific traits from your parents. These traits include hair and eye color, body build, and many others. The genes for these traits came from your parents.

Thus heredity is the passing on of traits from parents to offspring. The concepts of heredity are not new to us. The roots of genetics originally began in a small pea garden in Austria in 1865. Experiments conducted by an Austrian monk, Gregor Mendel, formed the major principles of inheritance that continue to be applied in the study of genetics today.

Some traits appear more often in males rather than in females. If a trait occurs more frequently in one sex, the trait is said to be a sex-linked trait.

Other traits can be sex-linked but not “expressed” in the individual. They may be dominant or recessive in nature.

A woman may “carry” the gene for a medical disorder but she may not necessarily develop the disorder during her lifetime. This is then a recessive sex-linked trait. The ability to pass on the gene that carries the specific disorder to her offspring certainly exists. If she were to marry a man who also carried the gene for the same medical disorder, it is highly likely that their children could develop the medical condition sometime during their lifetimes.

You should keep in mind that many things (ie. race, gender, environmental factors etc.) can modify the expression of a gene because not all people inheriting a gene that encodes for a disease actually develop the disease. This certainly applies to dystonia and other movement disorders such as tremor.

### **Genetic Testing...**

The gene is the fundamental unit of heredity. When an alteration in a gene occurs, a clinical disease may occur in a person. As of July 1995 genes for over 250 disorders have already been identified. Knowledge of the location and presence of a specific gene can be very important to you especially if there is a family history of a specific medical disorder in your family. In 1995 researchers identified the DYT1 gene located on chromosome 9 for early-onset childhood dystonia.

One purpose for genetic testing is for pre-pregnancy planning. A gene test can identify if you are a carrier of a gene that can be passed on to your children. This type of testing is available through Athena Diagnostics, Inc. or other genetic-focused pharmaceutical company. A second purpose for testing is to identify and make a diagnosis of a condition in a person. Some medical conditions can now be diagnosed before the presence of disease symptoms. If this is the case, effective treatment can be initiated at an earlier time for you by your physician. Thus you can continue to lead a productive and healthy life.

Genetic testing is a complex laboratory process. Genetic testing is usually coordinated through a genetic counseling clinic. Costs include a fee (s) for the laboratory test and counseling services. These fees are not always reimbursed by health insurance organizations. You should explore what your health insurance policy “covers” prior to genetic testing process.

It takes approximately two weeks for actual genetic testing to be completed. However, the entire genetic testing process may occur over a matter of months. A series of steps are involved in genetic testing.

## **The Genetic Testing Process...**

- *Initial phone contact*

Genetic testing usually begins with someone wanting to know whether or not a disease is likely to recur in other family members. Most counseling is initiated by a phone call to a movement disorders division. Initial contact may include a pre-screen interview over the phone with one of our staff members or our genetic counselor.

- *Decision-making to complete genetic testing*

Your decision to undergo genetic testing should be informed carefully considered, and freely chosen. It is ultimately your choice to complete this process. The support of family and friends is important when making this decision. Do not be afraid to ask any questions. You should never feel coerced into genetic testing. We have included space in this brochure for your questions. Write them down there.

- *Pretest session(s)*

These hourly session(s) may include psychological and neurological evaluations as well as genetic counseling. Taking a family history will be an important part of at least one of these sessions. Special data forms will be utilized to ensure clarification of the information you provide. A pedigree or family tree will also be developed to help trace the presence of genetic disease in previous generations.

## **The Actual Process**

- *Actual Genetic Testing*

The blood test is obtained and sent to the laboratory for evaluation. You should always sign a written consent form that provides an explanation about the blood test that is to be performed. To minimize fears and anxiety it is helpful to bring a family member or friend with you on the day of the test. For many people this can be a very emotional day; thus it is important that you express your feelings as best as you can to others.

- *Disclosure of results*

The results of the test are provided to you in person. Your physician will also receive a copy of your test results. As a patient you have the right to receive a copy of the test results. Test results are never given to anyone without your consent. Test results include a detailed interpretation of the results including in-depth comments and follow-up recommendations. If you do not understand your test results, clarify any uncertainties.

- *Post-test Counseling Session(s)*

These sessions will focus on providing you with the help to adjust to the personal and emotional impact of the disorder. Information on the many types of therapeutic and support services pertinent to your needs will also be provided to you and your family.

WRITE ANY QUESTIONS HERE...

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In 1997, the DYT1 (dystonia) gene for childhood-onset dystonia was identified. There is a screening test available for this gene. Talk with your Team about this Test. For more information about genetics and disease visit the website <http://www.geneclinics.com>.