Genes are the fundamental units of heredity. They are the instructions for the development of particular traits such as eye or hair color. Individual genes are packaged into structures called chromosomes. Humans have a total of 46 chromosomes, which are located in every cell of the body. We receive 23 chromosomes (half of our genetic information) from our mother and 23 chromosomes from our father. Twenty-two of the 23 pairs of chromosomes are identical in men and women and are called autosomes. The last pair, the sex chromosomes, differ in men and women. Men have one X and one Y chromosome, and women have two X chromosomes. Because our chromosomes come in pairs, the genes they carry come in pairs as well.

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Each gene’s chemical instructions make a specific protein, and each protein has a particular function in the body. For example, a gene may carry the instructions to make a protein called an enzyme, which is necessary for a specific biochemical process in the body.

Genetic disorders arise when one or both copies of a particular gene have undergone an alteration known as a mutation, which affects the production of a specific protein. When an alteration in just one copy of a gene causes a genetic disorder, the disorder is considered recessive. When alterations in both copies of a gene are required for the disorder to be present, the disorder is considered dominant. While these conditions vary in severity and age of onset, most of the Jewish genetic disorders are debilitating, and some are fatal. Many of the disorders lack effective treatment options at this time. This brochure provides an overview of genetics and the clinical features of Jewish genetic disorders. It also highlights the value of prevention in decreasing the frequency of birth defects among the Jewish community.

The “Jewish” genetic disorders are a group of conditions that are more common among Jews of Eastern European (Ashkenazi) descent. Although these disorders can occur in the general population, they do so at a significantly higher frequency in the Ashkenazi population. Every person of Ashkenazi Jewish descent has a 1 in 5 chance of being a carrier for one of the Jewish genetic disorders.

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The Chicago Center for Jewish Genetic Disorders is a cooperative effort of the Jewish United Fund/Jewish Federation of Metropolitan Chicago and Children’s Memorial Hospital. The Center is supported by the Michael Reese Health Trust.
About the Disorders

Bloom’s syndrome is a condition characterized by infections and a predisposition to cancer. Intelligence is typically normal, although mild mental retardation has been reported in some cases. The mean age of death is 27 years and the cause is usually related to cancer. The carrier frequency is 1 in 110.

Cystic fibrosis (CF) is a progressive disorder that causes the body to produce abnormally thick, sticky mucus in the lungs and digestive system. Patients with CF can exhibit a variety of symptoms including frequent respiratory infections, poor weight gain, and progressive lung damage. Treatment of CF depends upon the stage of the disease and the organs involved. The median age of survival is in the mid-30s. CF is one of the most common genetic disorders among Jews and non-Jews alike, with both groups having a carrier frequency ranging from 1 in 25 to 1 in 29.

Familial dysautonomia is a disorder that affects bodily functions such as swallowing, temperature regulation, sensitivity to heat and pain, and tear production. This condition occurs almost exclusively in Ashkenazi Jews. Some treatments are available which can improve the length and quality of life. The carrier frequency is 1 in 30.

Glycogen storage disorder, type Ia is a metabolic disorder order that, if untreated, results in severe low blood sugar, growth failure, enlarged liver, and bleeding disorders. Disease management involves lifelong diet modification. The carrier frequency is 1 in 71.

Maple syrup urine disease is a metabolic disorder that causes the accumulation of certain amino acids in the blood. If untreated, it can lead to mental retardation, physical disabilities, seizures, and death. Disease management involves lifelong diet modification. The carrier frequency is 1 in 81.

Mucolipidosis IV is a progressive disorder that affects the brain and nervous system. Individuals lack a chemical that normally breaks down a fatty substance, causing the fat to accumulate and be stored in the central nervous system. Most reported cases of this disease have been severe, with onset during the first year of life, but some milder cases have also been seen. The carrier frequency is 1 in 100.

Niemann-Pick disease, type A is caused by a deficiency of an enzyme which is responsible for breaking down a specific fat in the body. Lack of this enzyme causes fat to accumulate in various organs. In type A, which is seen with increased frequency in individuals of Ashkenazi Jewish ancestry, accumulation of the fat also occurs in the brain and nervous system and causes rapid deterioration and death by two to three years of age. The carrier frequency is 1 in 90.

Tay-Sachs disease is caused by a deficiency of an enzyme that affects the brain and the nervous system, causing rapid and progressive deterioration. Babies appear normal at birth, but begin to lose developmental skills at three to six months of age. This condition is life shortening, with death usually occurring by age six. A late-onset form of Tay-Sachs also occurs, though it is rare. The carrier frequency for Tay-Sachs disease is 1 in 30.

Torsion dystonia is a progressive movement disorder characterized by sustained, twisting muscle spasms. With time, the severity of these spasms increases, leading to progressive disability. Individuals with torsion dystonia have normal early development and normal intelligence. There is no cure, but there has been progress in treating dystonia with a variety of interventions. Dystonia is the only one of these disorders inherited in an autosomal dominant fashion, but only one in 20 people with the gene manifest dystonic symptoms.

About Screening

There is no definitive set of Jewish genetic disorders. The list of Jewish genetic disorders for which testing can be done is constantly changing and varies from lab to lab. Screening is available for all of the disorders listed above. Carrier status for any of these disorders can be determined from a blood sample. A genetic counselor (a health professional with a specialized graduate degree and experience in the areas of both medical genetics and counseling) or other qualified healthcare provider can assist a couple in deciding whether screening is appropriate for them and which disorders to consider screening for.

The American College of Obstetricians and Gynecologists (ACOG) considers it standard of care for obstetricians and gynecologists to offer carrier screening for Canavan disease, cystic fibrosis, familial dysautonomia and Tay-Sachs disease to couples where at least one member is of Ashkenazi Jewish descent.

If an individual is found to be a carrier, a genetic counselor can help explain the implications of carrier status. When both members of a couple are found to be carriers of a gene mutation for the same disorder(s), a genetic counselor can provide important information and support which can be helpful in making family planning decisions. Given the array of new technologies, detection of carrier status prior to pregnancy allows carrier couples to exercise a wide range of reproductive options.

Please consult your healthcare provider and visit the “Jewish Genetic Disorders” section of the Center’s website at www.jewishgenetics.org to learn more about these disorders and additional disorders for which screening may be available.

About Inheritance

When a person is a carrier for a recessive disorder, that person does not have the disorder. Carriers are not affected because they still have one fully functioning copy of the gene. Only when both copies of the gene are not functioning will an individual be affected with a recessive disease.

When two carrier parents have a child there is a 25%, or 1 in 4, chance in each pregnancy that this may occur.

The information below outlines how both dominant and recessive inheritance work. All of the disorders listed are inherited through autosomal recessive inheritance except for torsion dystonia.

Autosomal Recessive Inheritance

Autosomal Dominant Inheritance

This brochure does not address the genetic predisposition to hereditary cancers (breast, ovarian and colon) which is found with greater frequency in the Ashkenazi Jewish population. For information on hereditary cancers, visit the "Cancer Genetics" section of the Center’s website at www.jewishgenetics.org.