THE GENETIC DISEASE PREVALENT AMONG CAJUNS ROBS PEOPLE OF THE ABILITY TO MOVE. ATAXIA: A MATER OF HEREDITY

When the early settlers arrived here in the mid-1700s from Canada they were a small population living in isolation from the rest of the world. When children came of age, they found only a small number of eligible candidates for marriage. So it was unusual to have any days for counsel or to marry counsel.

It was just part of life in the wilderness. But then, the effects of the practice have reached into present-day generations. "We have a high incidence of the genetic disorders here," said New Orleans neonatologist Dr. Michael Wiensky. "Part of the reason is because of the intermarriage among Cajuns during the early years. It enhances a recessive gene. It is not until later that it becomes illegal to marry a first cousin.

Among the genetically transmitted diseases prevalent in the Acadian population is a neuromuscular degenerative disease called ataxia. The Louisiana Chapter of the National Ataxia Foundation.

The word, ataxia, means "without sense," Wiensky explained. "If you're drunk, you're ataxic. But being drunk is a temporary disease. These are progressive degenerative diseases." There are several forms of ataxia. Among the more common forms of the condition found in south Louisiana are Friedreich ataxia, Charcot-Marie-Tooth, French Settlement disease and Ataxia Triplex type.

Denise Drake of New Orleans, state chapter president of the National Ataxia Foundation, first noticed symptoms of Friedreich ataxia when she was about 10 years old. "You slowly lose your balance," she said. "Very often our parents think faster than our bodies will move and that is a big aggravation.

"So, Drake is wheelchair-bound and needs help to wash, feed and bathe, and to get around. She lives in a nursing home," Wiensky said. "50 percent of these patients have all their mental faculties. They are one of the worst aspects of this disease.

Although the symptoms of ataxia can be related to some degree with medication, there is no cure. The only defense is prevention and education.

"The only good news is that the state chapter of the Ataxia Foundation periodically holds screenings all over the state to try to spread awareness," Wiensky said. Among the professionals who work with ataxia patients are geneticists, physiotherapists, speech therapists and nurses. In addition to looking for symptoms of the onset of the disease, geneticists look for blood samples for genetic testing.

Geneticists have isolated a defective gene in ataxia patients that prevents them from producing adequate amounts of a certain defectively synthesized protein called frataxin. Dr. Robert K. Wiensky holds a doctorate in genetics and serves as the director of the Center for Human Genetics at LUSM Medical School in New Orleans. The center is also the director of the Center for Acadian Diseases and Healthy Care Research.

The disease is a threat for both parents and children. Normally, a child born to both parents who have the disease will get the disease. But if one parent has the disease, there's a 50-50 chance of having the disease.

How is ataxia inherited?

In recessive types of ataxia, like Friedreich ataxia, parents rarely exhibit symptoms, but they each carry a recessive gene which may cause ataxia in their offspring. The recessive gene is not harmful to the parents, but it is inherited into the offspring, where it can cause serious problems. This is a problem because if both parents carry the recessive gene, their children will have a 50-50 chance of developing the disease.

Dominant hereditary ataxia

Domiant ataxia is passed on in a hereditary disease through defective genes directly from parent to child. Each of the parents is a carrier of the defective gene, and one child in four will inherit the disease.

What to do if you think your child or family member may have ataxia?

If you think your child or family member may have ataxia, it is important to consult with a healthcare professional. They can help you determine if the symptoms you are experiencing are consistent with ataxia and refer you to a specialist for further evaluation.

For more information, you can contact the National Ataxia Foundation at 504-822-2265 or visit their website at www.caadiensfoundation.org. The foundation offers resources and support for those affected by ataxia, and can provide information on genetic counseling and prevention.

The disease is a threat for both parents and children. Normally, a child born to both parents who have the disease will get the disease. But if one parent has the disease, there's a 50-50 chance of having the disease.

What can be done to treat ataxia?

Currently, there are no cures for ataxia, but there are treatments available to manage symptoms and improve quality of life. These include physical therapy, occupational therapy, and speech therapy. In addition, medications can help alleviate some of the symptoms, such as tremors, difficulty walking, and dizziness.

Friedreich ataxia is a rare, degenerative neurological disorder that affects the brain and spinal cord. It is caused by a mutation in the FXN gene, which codes for a protein called frataxin. The disease typically presents in childhood or adolescence and is characterized by progressive weakness, ataxia, and sensory disturbances.

Diagnosis of Friedreich ataxia is usually made through genetic testing, which can identify the specific mutation in the FXN gene. However, some cases may be diagnosed based on symptoms alone if the genetic testing is not available.

While there is no cure for Friedreich ataxia, there are several treatments available to help manage symptoms and improve quality of life. Physical therapy, occupational therapy, and speech therapy can all play important roles in managing the disease.

Physical therapy can help improve mobility and balance, while occupational therapy can help with tasks such as dressing and feeding. Speech therapy can help improve swallowing and communication skills. Medications, such as vitamin B12, can also be used to treat some of the symptoms of Friedreich ataxia.

In addition to these treatments, supportive care is also an important aspect of managing Friedreich ataxia. This can include regular medical check-ups, botox injections for dystonia, and pain medication for seizures.

Ataxia is a complex and varied group of disorders, and the specific treatments used will depend on the individual case and the symptoms present. Early diagnosis and prompt treatment can help improve quality of life and prevent complications.

In conclusion, Friedreich ataxia is a rare and progressive genetic disorder that can significantly impact an individual's quality of life. With early diagnosis and appropriate treatments, however, many people with Friedreich ataxia are able to lead fulfilling lives. It is important for individuals with ataxia to work closely with their healthcare providers to develop an individualized treatment plan that meets their specific needs.