Disease affecting Cajuns studied
Families suffer CMT disorder in S. Louisiana

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When William Adams burned his feet a few years ago, he was amazed to discover it hurt. Then, he lost all feeling in his feet.

"I went to the foot doctor, then a neurologist and learned what I had suspected all along. He said I had Charcot-Marie-Tooth (CMT) disease. My mother had it though she didn't know it at the time. Both of my sisters got it and several of their children have been diagnosed with it," said the 75-year-old Prairieville resident.

Adams was only diagnosed with CMT seven years ago. Retired as a parish clerk from the highway department, he considers himself fortunate to have developed the disease so late in his life. Doctors say the genetic disorder usually manifests itself in patients in their teens.

Charcot-Marie-Tooth disease, named for the 19th century French and English scientists who described it, is a nerve disease affecting muscles. Also called hereditary muscular atrophy, the disease causes outlying muscles in the hands and feet to fall into disuse because they do not do proper signals from the nerves.

The genetic disorder is passed from one generation to the next. Each child born to an affected parent has a 50 percent chance of getting the disease.

CMT causes foot and hand deformities resulting from the wasting of muscles. It can cause loss of sensation in the peripheral nerves of the hands and feet. Normal physical activity may be hindered and, in some cases, victims may be confined to wheelchairs.

CMT patients tend to have hand and foot deformities, including short feet with high arches and balance problems. Symptoms vary in severity from person to person.

However, the disease does not affect the intellect or lifespan, according to Dr. Carlos Garcia of the LSU Medical School in New Orleans. Disability is generally not too great and most victims lead a fairly normal life.

Scientists at Baylor College of Medicine in Houston have determined that CMT is caused by a duplication of DNA on chromosome 17 and are developing a blood test to diagnose the disorder.

There is an unusually high concentration of CMT among Cajun families. CMT patients like Adams are being treated, as well as monitored for research, at Muscular Dystrophy Association clinics in New Orleans, Baton Rouge and Lafayette.

Garcia is one of the physicians seeing area CMT patients and helping conduct research studies. "Cajuns are a gold mine for genetic studies of all types, not just CMT," Garcia said. "They have been isolated for many years, and that has been inbreeding in many of the families. They are also good subjects because they are friendly, cooperative and rooted to their homes." At one point, researchers went to a family reunion in Houma, took blood and did nerve conduction studies on a number of family members related to identified CMT patients.

Five large ancestral lines in South Louisiana have been identified so far in the CMT studies, he said. The disease can be traced five generations in several families and nearly 100 members of a family in the Thibodaux area have been identified as having CMT. In all, some 350 CMT patients have been diagnosed in Louisiana.

In the past, the disorder has been diagnosed through clinical diagnosis, family history and nerve conduction tests, Garcia said. The blood test may be available in the near future as a more definitive diagnostic test.
physical therapy may be prescribed to avoid deformity by keeping the muscles active and stretching the heel cord. Plastic braces are used to hold the feet up. In extreme cases of disability, surgeons can transplant tendons and fix drooping ankles with bone fusions. Pins may be used to straighten toes.

When hands are affected, occupational therapy is helpful in retraining people to use special tools for small tasks, such as closing buttons, opening jars and zipping zippers.

However, the future holds promise for much more exciting and effective treatment of CMT, Garcia said. Because scientists have pinpointed the chromosome that causes the disorder, he said, there is hope scientists can, through genetic therapy, give the patient relief.

“Seven years ago, I would have said there was no hope” of curing Charcot-Marie-Tooth, said Garcia. “Today, I am very optimistic. Technology is advancing so fast, and I believe there is real hope for the future.”