A neuro-muscular disease called Charcot-Marie-Tooth is attacking people of Acadian descent more than most other populations. No one knows why CMT is so prevalent among Cajuns.

"It's probably the largest population of CMT in the world," says Dr. Carlos Garcia, a professor of neurology and pathology at the LSU Medical Center in New Orleans. He estimates that there are probably 400 patients in South Louisiana. Garcia has studied the disease for the past 15 years and first discovered the connection between the Cajun population and CMT sufferers.

"For some reason, it seems to be concentrated here," he says. Over 30 percent of the people who are treated at the Muscular Dystrophy Association clinic in Lafayette have CMT. Garcia visits the clinic at Our Lady of Lourdes Regional Medical Center two days every month for diagnostic testing and treatment of the disease.

CMT most often attacks the hands and feet, often causing mobility and balance problems. The hands may take on a claw-like appearance as muscles atrophy or waste away. It is primarily, however, a nerve-related disease that affects muscles.

"Since the nerve doesn't give the signal to the muscle, it atrophies," explains Dr. James Lupski of the Baylor College of Medicine in Houston. "It is secondary to a problem with nerves."

CMT has been classified as a muscular dystrophy disease, and as such, MDA funds much of the research, testing and treatment that's associated with it. "It's a nerve disease more than a muscle disease," says Garcia. "The nerve commands muscles and if the nerves are impaired, the muscles are impaired."

CMF most often attacks the hands and feet, often causing mobility and balance problems. The hands may take on a claw-like appearance as muscles atrophy or waste away.

The condition was first described a century ago by the three physicians whose names it bears. Lupski's research began three years ago. The quest is personal as well as professional because he is a victim of CMT. The disease has not, however, deterred him from obtaining a Ph.D. in biomedical sciences.
molecular biology and clinical training in pediatrics and medical genetics. He has no Acadian connections that he is aware of, though he acknowledges that the link is a crucial one for his purposes.

“It’s fantastic from the research standpoint,” Lupski says. “CMT occurs frequently in the French Acadian population. We know that there’s a lot in the French Acadian and a lot in the French Canadians.” The connection has been instrumental in furthering his research and localizing the responsible gene.

“A very great deal has been accomplished. This is the difference between medicine and medical science,” he says. “We have gone from not knowing where we are, but knowing the specific region of where we are.”

He refuses, however, to venture a guess as to when a cure may be possible. “I’d be a fool to try to answer that,” Lupski says. “It takes a lot of time.”

The Cajuns’ propensity toward CMT is unexplainable, but not unusual, according to Lupski. “The fact is you give me a genetic disease and I’ll give you a population.” He points to certain genetic tendencies inherent in the Amish, for example.

The effects of CMT vary greatly, even within the same family. Some people have it without even knowing it, while those with severe cases may be confined to wheelchairs.

“Some members of the same family are very mildly affected and some are very bad,” says Garcia.

continued on page 31
continued from page 30

CMT is not life threatening, although it can be disabling and painful at times, he says. "They lead a normal life span and can do most everything everybody else does."

"It is a progressive disease but it is self-limited in that it doesn't go beyond the hands and the feet," he adds. "At this time, we cannot control the progression of the disease."

Surgery is usually not recommended, except in severe cases, such as Lupski's. He underwent 11 operations as an adolescent to correct mobility problems. Normally, leg braces are used to remedy gait imperfections that result when the foot drops flat due to useless muscles.

If a person has CMT, there is a 50 percent chance that offspring will inherit the condition. Even so, this does not usually deter those who are affected from having children, says Garcia.

Lupski is convinced that CMT is more widespread than originally thought and that many undiagnosed people have it. "I think the disease is a lot more common than what's being reported," he says. "There's a lot more people that have the disease."

The first symptoms include foot deformities and gait problems, often followed by cramping and numbness. "It can be painful," he says, but adds, "Longevity is not affected and it doesn't affect the central nervous system." He urges anyone who thinks they may have the disease to consult a doctor.

"Some members of the same family are very mildly affected and some are very bad."

—Dr. Carlos Garcia

continued on page 32
be tested at the MD clinic at Lourdes.

Lupski stresses how very important the Cajuns’ role is in eradicating the disease. “You can’t do these kinds of studies without the patients being cooperative,” he says. “We need to get the most people we can from the families. Investigators are good, but families are most important.”

Those who provide blood samples and family histories are doing their part to solve the jigsaw puzzle that Lupski and Garcia collaborate on. “We give our blood, sweat and tears, but they truly have to give their blood,” says Lupski.