SOUTHWESTERN NEWS

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LITTLE-KNOWN DISEASE GREATLY LIMITS ABILITY TO EXERCISE

DALLAS — February 6, 1995 — One of the first things Helen Galletly did when she learned from doctors at UT Southwestern Medical Center at Dallas that she suffered from McArdle's disease — a neuromuscular condition that causes victims to experience fatigue and extraordinary pain after minimal exercise — was phone her former junior high school gym teacher.

"I called him up and said, See! I wasn't faking it. There was a real reason I couldn't run those laps."

Galletly, naturally, was distressed to learn that she was a victim of a neuromuscular condition with no known cure, but she also felt a peculiar sense of relief at finally hearing scientific confirmation of what she'd told classmates and coaches for years. Her body would not allow her to exercise the way other people do.

A research clinic dedicated to studying McArdle's disease and related diseases that block the metabolism of fuels necessary for normal muscle-energy production is a joint effort of UT Southwestern and Presbyterian Hospital of Dallas. Overseen by Dr. Ronald Haller, the clinic is unique in the United States and perhaps the world in its use of state-of-the-art techniques to monitor muscle metabolism and the delivery of oxygen during exercise in order to identify causes and potential treatment of disorders that result in muscle fatigue and pain after exertion. The center sees patients from around the globe, having received referrals from Iceland, Brazil and Sweden. The clinic operates in Presbyterian Hospital's Institute for Exercise and Environmental Medicine.

McArdle's disease is caused by a genetic defect that results in deficiency of the muscle form of the enzyme glycogen phosphorylase. The result is an inability of muscle to break down glycogen (a form of carbohydrate stored in muscle) to lactic acid during exercise. Patients report feeling extreme pain and a hardening of their muscles during activities that would be easily tolerated by unaffected individuals. It is not currently possible to replace the missing muscle enzyme in McArdle's disease, but doctors can control symptoms by recommending specific

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patterns of exercise and diet.

Haller, professor of neurology and internal medicine at UT Southwestern, is eager to increase both physicians' and the public's awareness of muscle energy defects like McArdle's disease because the mental anguish suffered by victims can match the physical pain. "The kind of psychological torture that these people have to endure is unreal," Haller said.

McArdle's disease was first described by the English physician Dr. Brian McArdle in 1951. In 1959, the enzyme defect responsible for the disease was identified. In the late 1980s, a review of the disease indicated there were only approximately 100 known cases in the world. Doctors now know of at least 20 cases in the Dallas-Fort Worth area alone and believe that many more people may be suffering from the disease but have gone undiagnosed. In the Dallas clinic, affected patients have first been diagnosed in their 40s, 50s and 60s, testimony that these disorders are easily missed.

"Many times when the parents of patients find out about McArdle's, they'll comment on how the patient always wanted to be picked up as a little child. They thought the child just didn't want to walk," Haller said.

The stories told by patients themselves reflect the same type of misunderstanding. "I always noticed that whenever we had to exercise, I always got very winded or was in pain," said Galletly, a 29-year-old former advertising professional who is now in graduate school at the University of North Texas in Denton. "For a long time, I thought I was just out of shape, but no matter how much I dieted and tried to exercise I couldn't keep up.

"The best thing I've learned since being diagnosed is to listen to my body," she said. "I know now that if I push it, I could do real damage."

Pushing themselves too far may result not only in serious muscle injury but also can cause renal failure due to the toxic effects that products released from damaged muscle can have on the kidneys. Haller tells his patients to contact him whenever they notice a darkening of their urine, which could represent the excretion of myoglobin — a pigmented protein released from damaged muscle. McArdle's patients are able to do certain types of exercise, but they must know their limits.

"Back in the P.E. days, I remember teachers saying, 'No pain, no gain,' and 'Do it until

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it hurts,' " Galletly said. "But whenever the rest of the class would do 60 sit-ups and be fine, I would do 10 and not be able to move anymore."

Her problems usually were attributed to a lack of motivation. She'd make straight A's on her report cards except for physical education, for which she sometimes received a C. Before her condition was diagnosed, doctors suggested she eat carbohydrates before class. She'd load up on raisins or bread, but nothing helped.

One particular instructor, whom she had in third grade and junior high, thought Galletly was trying to manipulate him. After being frustrated by her inability to complete the daily jogging warm-up, the teacher tried to use peer pressure. He said that if Galletly couldn't make it all the way around the track once, the whole class would have to run a second lap. Her classmates were pleading with her to try, and she ended up struggling after a short distance, almost crawling, but couldn't finish the lap.

"The other kids were pretty hacked off," she said. "They told me that they didn't understand why I couldn't do better."

After graduation, Galletly had bad experiences on skiing trips and when she tried to go swimming. Once she almost drowned when the symptoms hit while on a scuba-diving excursion. Even walking to and from the mall after parking in an outer lot can be an ordeal.

Galletly's sister suffered many of the same symptoms. She was sent to Haller's clinic after a physician who was evaluating her for an unrelated illness elicited the history of exercise-induced pain and fatigue. Galletly followed her to the clinic four months ago and got the same diagnosis — McArdle's disease.

She encourages other people who experience similar symptoms to investigate the possibility that they may be suffering from McArdle's or a related disorder. "It really was such a relief just to know," she said.

Haller notes that while cures are unusual in these diseases, appropriate treatment can make dramatic differences in the quality of life of affected individuals.

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