UF doctors cast net to help muscular dystrophy patients

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GAINESVILLE, Fla. — Patients and families affected by muscular dystrophy urgently need to take practical steps to help slow the destructive course of the disease, say University of Florida researchers who today (Sept. 2, 2005) have launched a Web-based effort to collect information from people coping with the illness.

“The reason that it is so important to identify things that may offer a margin of benefit is that the realistic time horizon for a real treatment or cure is within decades, maybe sooner,” said Dietrich Gravenstein, M.D., an anesthesiologist associated with UF’s College of Medicine. “I have experience with individuals that suffer from rare diseases. I see how stoic the parents and patients are that face these challenges and I have been frustrated by how little scientifically validated advice there is to simply help them confidently make the many choices they face.”

Muscular dystrophies, genetic diseases characterized by progressive weakness of the muscles that control movement, affect between 50,000 to 250,000 people at any one time, according to the Muscular Dystrophy Family Foundation. In Duchenne muscular dystrophy, which affects 1 out of every 3,500 to 5,000 boys, the disease starts to tear down muscles at birth and is typically diagnosed before age 6. Most patients are wheelchair-dependent by 12. By their late teens, most require a respirator to breathe. Victims of the disease do not live past their 20s.

The Web site — www.UFanswers.org — will collect voluntary submissions about personal experiences, lifestyles and the progression of the disease. Even families who have lost loved ones can contribute their experiences and help others living with muscular dystrophy. The Web site officially launches just before the annual Jerry Lewis Labor Day Telethon for Muscular Dystrophy.

Users will give their names for verification purposes, but individual information will be kept strictly confidential.

Questions about diet and medication — even the moment a wheelchair is brought into a household — will be asked through the site. The Web site differs from blogs or traditional sites that might feature chat rooms, personal experiences or lists of resources in that it is a data warehouse that researchers with approved protocols can mine for strategies to impact the disease. It uses advanced information science techniques to collect and classify data.

“Anecdotal information is unscientific, but by gathering it in great numbers, we hope to pinpoint factors that are causally related to an improved disease course,” said Gravenstein, who has a son with muscular dystrophy.
“Right now, we are forced to speculate. It’s terribly disheartening. For example, conventional wisdom is to let children regulate their own play; that when they tire they will rest. Yet strenuous exercise and contact activities are discouraged because this accelerates muscle loss. So, we are left to ask: are other activities such as stair climbing harmful or harmless? And with the respiratory or cardiac failure all patients eventually develop, are these accelerated if someone in your home smokes? What are the effects of surgery and how the anesthetics are managed? Parents anguish over such questions — for which there are currently no answers — and we hope to shed light on them.”

Additional sets of clinical and anecdotal data would be useful to the muscular dystrophy community, according to Pat Furlong, president of Parent Project Muscular Dystrophy, an Ohio-based, not-for-profit organization that works to help families, support research and build international collaborations.

“Over the last 20 years, I have spoken with more than 1,000 families of boys with Duchenne or Becker muscular dystrophy,” Furlong said. “Without exception, the families, researchers, physicians and the health-care community would benefit from the ability to access information relevant to a certain condition. It is certainly critical for the muscular dystrophy community to work together to better understand what positively or negatively impacts individuals affected by neuromuscular disorders, as well as the value of specific intervention.”

Funding to build the database is through support from the Florida-based I. Heermann Anesthesiology Foundation. Anesthesiologists are particularly interested because they participate in tendon release and spine stabilization procedures, common in muscular dystrophy patients, Gravenstein said. The disease causes muscles to become infiltrated with fibrous tissue, making the muscles appear large even as the kids become weaker. Patients begin to walk on their toes and their backs bow. Surgeons cut the tendons in the legs and correct the curvature of the spine to enable the children to walk or sit for a while longer, or barring that, to rest more comfortably. The muscle contractions also occur in the diaphragm and heart.

“Children with muscular dystrophy and their parents don’t have a rosy future since there is still no cure for this disease,” said Hans van Oostrom, Ph.D., an associate professor of anesthesiology and biomedical engineering who designed the www.UFanswers.org Web site. “No single institution that provides care collects enough experiences or data, but we’ve built a mechanism to create a large database at a single source to catalog a geographically broader experience. Our objective is to do statistical analysis on these data and report our validated results back to participants. Ultimately, we would like to make these data available to anyone who has a legitimate interest, but only in a way that guarantees the absolute confidentiality of the people who contributed their experiences.”