Gifts in action

Spanish mother’s search

JEFF MILLER, UW-MADISON, UW COMMUNICATIONS
So begins this story of a mother and the little boy she wants to grow. She is Maria Antonia “Toni” Fenoy Ramon, who lives in Spain. “You will carry in your soul, the soul of all Spain,” she wrote to 4-year-old Juan Manuel, or Juanma. He was a healthy 22 months old on Valentine's Day 2006, when his leg trembled and then he couldn’t stand. Toni Ramon’s odyssey from then until now saw her spend desperate hours on the Internet looking for answers to her son’s illness, searching Europe for a conclusive diagnosis, battling her own advanced breast cancer and, ultimately, mounting a plea for help that has raised more than $1.3 million for research into Alexander disease at the University of Wisconsin-Madison.

“I am a simple woman whose main objective in life, just like all of the mothers in the world, is to be able to see their children grow happy and healthy,” Ramon said.

Her journey toward hope hinges on the work of UW-Madison Professor Albee Messing, Department of Comparative Biosciences at the School of Veterinary Medicine, Waisman Center, who in 2001 discovered that nearly all cases of Alexander disease are caused by mutations in just a single gene, called GFAP. The progressive disease, a form of leukodystrophy, causes the loss of physical and mental abilities, an abnormal increase in head size and seizures. About two-thirds of patients have

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an early-onset form, beginning before age 2, and usually survive only three or four years after diagnosis. Although children may remain stable for a while, they will begin to lose skills they have acquired, Messing said. From walking, they go to a wheelchair. From talking to no speech. From swallowing to a feeding tube.

An MRI usually indicates the possibility of Alexander disease; a genetic test confirms it. “The pain (the diagnosis) produced also took away a little of my life each day, until I became sick with cancer,” Ramon said. “Nevertheless, and in spite of everything, I never resigned myself to accept the death of my son. I revolted against the illness of my son and my own. I rejected the pain in my soul and instead installed the hope of the fight.”

Messing’s findings about GFAP were quickly confirmed, and he began looking for treatments. In 2005-2007, he received a $100,000 pilot grant from the National Institutes of Child Health and Development to look at already-approved drugs to see if one might reduce the expression of the gene. The Wisconsin Alumni Research Foundation helped Messing gain access to the University’s Carbone Comprehensive Cancer Center’s drug library and robotic equipment to look for possible answers. Messing plans to submit the first results of this study for publication later this year.

When the Ramons found Messing through the Internet in early 2007, he had little to tell them. By that summer, he presented early findings to the United Leukodystrophy Foundation, which recorded and distributed his talk. It gave the Ramons new hope, and Toni Ramon mounted what Messing called an incredible fundraising effort. Thousands of Spaniards have contributed to Ramon’s cause by attending events such as tennis matches and bullfights, participating in everything from online auctions to raffles and making direct contributions.

“We come from a country thousands of miles away,” Ramon told Messing when she met him in April 2008. “I want you to know my dear professor that, although physically the three of us are here, in our hearts with us have traveled thousands of people who have made it...
possible so that you can continue your dauntless, valiant fight to find a cure for this cursed disease.”

The Ramons arrived in the United States during the spring of 2008 to attend the first conference ever devoted to Alexander disease and sponsored by the United Leukodystrophy Foundation. They–and a Spanish television documentary crew–also visited Messing’s lab in the Waisman Center, where he is trying to identify drug treatments and is searching for biomarkers to chart the progress of the disease.

“It takes a lot of faith on their part to have done this,” Messing said of the family’s fundraising. The Ramons placed few restrictions on how their gift will be spent, and they have no guarantee of results to help their son.

“The family is driven,” said Marsha Mailick Seltzer, Waisman Center director. “The way to achieve their goal is to, essentially, come here where the genetic link for this disease was discovered and support ongoing research. When one has a rare disease, it often gets little attention unless someone is devoted to it.” The Waisman Center makes rare diseases a priority.

Five months after Juanma’s diagnosis, Toni Ramon was fighting for her life after an advanced breast cancer diagnosis. Juanma’s father left his job to care for her and the children, including Juanma’s older sister. After a year of chemotherapy and radiation treatment, Toni Ramon dedicated herself to raising money for a disease too rare for pharmaceutical companies to pursue. “I was aware from the beginning that the principal deterrent to developing a treatment for Alexander disease was and is the lack of funding,” she told Messing.

Ramon, who called herself a mother who refused to stay home waiting for her son to die, credited the Spanish mass media for spreading word of her cause. She challenged Spaniards to contribute so her son could walk again and even run. “These are thousands of gift givers,” Seltzer said. A Spanish television reporter told Seltzer that even prisoners are donating one euro a week from their prison cells to Juanma. The bullfight raised 80,000 euros from those who attended. “These are thousands of givers who believe in what we call translational research,” she said. “This is a great example of translational research—to take basic science and turn it into treatments.”

Pictures on Ramon’s Web site, www.ayudajuanma.es, show her smiling son as she details her pain and her hope. “Lorenzo’s Oil” (a movie based on the true story of a family’s struggle to find help for their son who also suffered from a form of leukodystrophy) showed Ramon how another mother confronted the pain of knowing her son would die. “I wanted to know how to be capable of watching your son every day laughing, running and playing, while knowing that in a short period of time, he’ll lose everything,” she wrote. “How to be capable of looking in your son’s eyes day after day, always with the same thought in mind: he’s going to die. To wake him up every morning wondering if today will be the day that he goes blind. To prepare his food wondering when he’ll need a feeding tube. How to live each day, imagining a code to use to communicate with my son when he becomes completely tetraplegic. All of this while trying not to go crazy.”

When Ramon met Messing in Madison, she was so overwhelmed she could, at first, only squeeze his hand hard. If Messing can make a breakthrough in time, Ramon believes her son can recover some of the function he’s lost. “This miracle is going to happen,” Ramon writes on the Web site. “I know all of you are not going to abandon us.”

To Messing she said: “I am grateful for your fight without respite to obtain a cure for Alexander disease. In the film ‘Lorenzo’s Oil,’ (actress) Susan Sarandon was able to find a doctor who, by means of a chemical composition of oil, could save the life of her little Lorenzo. I remember perfectly the scene in which she pleaded to the doctor: ‘Please, hurry up.’

“Today, Dr. Messing, I am pleading to you emphatically, urgently... Please, doctor, hurry up.”