

Local Rotaries' Gift Enhances Treatment for Patients with Mitochondrial Disease

November 2, 2006

Debra Kain

Steve Achard (kneeling in front) and fellow Rotary Club members toured the research lab of Dr. Richard Haas, (in white coat), Professor of Neurosciences and Pediatrics at UCSD and co-director of UCSD's Mitochondrial and Metabolic Disease Center on October 27. Several Rotarian chapters, represented here, raised almost \$29,000 for Haas' Center.

It took Steve Achard's wife, Rita, nearly four years to get a diagnosis for the puzzling health condition that had the couple consulting heart specialists in Scottsdale, Arizona, Santa Barbara and San Francisco. It was only after a neurologist at the University of California, Davis recommended they see Dr. Richard H. Haas, co-director of UCSD's Mitochondrial and Metabolic Disease Center at the UCSD School of Medicine, that the Achards finally learned that Rita had a form of mitochondrial disease -- a disease many people have never heard of, but which is nearly as common as childhood cancer.

"During the course of Rita's treatment with Dr. Haas we learned more and more about mitochondria, which mimics a number of other diseases," said Steve Achard. Haas currently has Rita on treatment that has stabilized the progression of her disease, but at this stage, there is no cure.

"I have a "vested interest in fund raising for Dr. Haas and Rita," said Achard, who spearheaded a fund raising effort with Rotarian chapters in the San Diego and Tijuana region. The Rotarians raised nearly \$29,000 for Haas' lab. "It is too late for her, but perhaps some of his research and treatment options might benefit future generations." This gift contributes to the \$1 billion fundraising goal of The Campaign for UCSD: *Imagine What's Next*.

Left to right: Dr. Richard Haas, UCSD; Jack Wood, President, Fallbrook Rotary; Steve Achard, Secretary, Fallbrook Village Rotary; Paul Leehey; President, Fallbrook Village Rotary; and Roberto Cazaras, President, Club Rotario Tijuana Playas, Tijuana, Mexico.

Steve and fellow Rotarians who contributed to a Rotary matching grant gathered for a tour of the UCSD Hillcrest Medical Center facilities on October 27, where they presented Haas with a check for \$28, 804. Fallbrook Village Rotary Club, where Steve is a member, and its International partner, Club Rotario Tijuana Playas, were the sponsoring clubs. Other contributing chapters included the Fallbrook Rotary and Bonsall, with additional matching funds from Rotary International. The funds will be used to purchase a diagnostic machine for Haas' lab called the Oroboros Oxygraph 2K.

"The machine allows for needle biopsies for many patients, avoiding the larger procedure, an open muscle biopsy, which we have commonly used for most children and adults," said Haas. "We are very grateful for the Rotarian's gift, which will enable us to provide mitochondria patients with much better and more rapid results in diagnosing their condition."

Mitochondrial diseases can be confusing to diagnose and complex to manage, according to Haas. In Rita's case, a tooth implant which became infected may have triggered onset of the disease. Mitochondria are inherited from the mother, so in many patients mito disease is a genetic disease transmitted by the female side of the family. Symptoms can skip generations or can, as in Rita's case, lay dormant until stimulated by some stress, often as simple as a mild infection. In some cases, mito disease symptoms are present from birth. Nearly 4,000 children are born each year with the disease and mortality ranges from 10 to 50% of patients, depending on the particular disease.

Responsible for creating a large percentage of the energy need by the body to sustain life and growth, mitochondria are the cellular structures that function to convert the potential energy provided by food into usable energy. They are also essential for many of the body's metabolic functions. Mitochondrial dysfunction results in cell injury and organ failure. Symptoms range from loss of motor control and muscle weakness to cardiac and liver disease, gastrointestinal disorders, breathing difficulties, seizures and hearing or visual problems.

Mitochondrial and metabolic medical conditions are now referred to as *mitochondrial cytopathies*. Mitochondrial cytopathies actually include more than 40 different identified diseases that have different genetic features. The common factor among these diseases is that the mitochondria are unable to completely burn food and oxygen in order to generate energy.

UCSD's Mitochondrial and Metabolic Disease Center was founded in 1996 to meet the growing need for treatment, diagnosis and research in the emerging field of mitochondrial medicine, bringing together the talents of physicians and researchers with special training in human genetics and metabolism. More information on the Center and its fight against this disease can be found at <http://biochemgen.ucsd.edu/mmdc/index.htm>

Media Contact: Debra Kain, 619-543-6163

