

Media Contact: Kelli Stauning  
E-mail: [kelli.stauning@cshs.org](mailto:kelli.stauning@cshs.org)  
Telephone: (310) 423-3674  
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## **ENZYME REPLACEMENT THERAPY FOUND TO EFFECTIVELY TREAT PATIENTS WITH FABRY DISEASE – AN INHERITED GENETIC DISORDER**

LOS ANGELES, CA (October 24, 2002) - Raul Hernandez was no stranger to sports activity, and was active in Little League by the time he was 10 years old. But one day while running a short race at school, Raul experienced an intense burning sensation in his feet that turned his world upside down. From that day forward, he would experience severe pain in his feet any time he engaged in physical activity or, strangely, when the weather was hot or it rained. The situation worsened when the pain spread to his hands. His doctors, however, were unable to find anything wrong with him, telling his parents that the pain was all in his mind.

Seventeen years later, Raul was officially diagnosed with Fabry disease – a rare genetic disorder that causes severe pain in the hands and feet, eventually destroying vital organs in the body. Yet even after Raul was diagnosed, no therapy was available to treat the disease. It was not until two-and-a-half years ago that Raul learned that a clinical trial at Cedars-Sinai Medical Center was testing a new drug designed to replace the enzyme that he was missing. The next thing he knew, he was on a plane to Los Angeles from his hometown in Salinas, California to take part in the clinical trial. Since then, Raul commutes every two weeks to receive treatment. He says that he is once again exercising without pain and leads as normal life as anyone else.

An update of the clinical trial at Cedars-Sinai Medical Center and at 19 other centers throughout the country and Europe, was presented this month at the annual meeting of the American Society of Human Genetics in Baltimore, Maryland by William Wilcox, M.D., Ph.D., a medical geneticist at Cedars-Sinai. The findings show that patients receiving enzyme replacement therapy for a near-two-year period via infusion with a drug called r-h $\alpha$ GAL (Fabrazyme™) continue to benefit from reduced pain and prevention of further organ damage.

“This study essentially confirms the long-term safety and effectiveness of enzyme replacement therapy for patients with Fabry,” said Dr. Wilcox.

Named Fabry disease after the dermatologist who first noted the symptoms back in the 19<sup>th</sup> century, it was only recently discovered that the disease is an inherited disorder caused by the lack of a particular enzyme called  $\alpha$ -galactosidase A or  $\alpha$ -GAL. The enzyme is needed to break down a fatty substance in cells called globotriaosylceramide or GL-3. But when  $\alpha$ -GAL is lacking, GL-3 builds up in blood vessel walls

and does increasing damage to organs such as the heart, kidney and brain. By the time that the disease is diagnosed, the organs have often sustained damage, ultimately leading to an early death.

“Raul’s bout with the disease is similar to many other patients with Fabry, as even now, the disease is often undiagnosed until adulthood when organs have started being affected,” said Dr. Wilcox. “Now we have a drug that replaces the deficient enzyme so that patients can live longer and better.”

In the study, Raul was one of 58 patients selected at random to receive r-h $\alpha$ GAL or a placebo by infusion every two weeks for a 20-week period. After completing 20 weeks of the study, all 58 patients have been receiving an infusion of r-h $\alpha$ GAL every two-weeks for over 18 months. Patients’ response to the drug was monitored via kidney and heart function tests. Tissue biopsies were also performed to assess organ function and a specialized questionnaire was used to assess patient pain levels. The investigators found that pain was significantly improved overall, while pathology studies confirmed that GL-3, or the fatty substance in cells, was consistently reduced throughout the study period. Kidney function remained stable throughout treatment during the 18-month period indicating that the disease was not causing further damage.

Although the investigators found that the majority of patients began producing antibodies in response to the drug after a three-month period, this did not impact the continued effectiveness or safety of the drug.

“Interestingly, we found that enzyme replacement therapy continues to unplug blood vessels despite the presence of antibodies,” commented Dr. Wilcox.

As one of the first patients to participate in the clinical trial, Raul says the treatment has changed his life. He can exercise and doesn’t have to worry about the weather.

Cedars-Sinai Medical Center is one of the largest nonprofit academic medical centers in the Western United States. For the fifth straight two-year period, Cedars-Sinai has been named Southern California’s gold standard in health care in an independent survey. Cedars-Sinai is internationally renowned for its diagnostic and treatment capabilities and its broad spectrum of programs and services, as well as breakthrough in biomedical research and superlative medical education. Named one of the 100 “Most Wired” hospitals in health care in 2001, the Medical Center ranks among the top 10 non-university hospitals in the nation for its research activities.

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