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Cystinosis Foundation Fights Ultra-Rare Disease

By Sophie Braccini

True Claycombe, Legislative Assistant to Congressman David McKinley, 1st District of West Virginia; Valerie Hotz, Executive Director of the Cystinosis Foundation and Penny Hughes, whose 6-year-old granddaughter has cystinosis. Photo Timothy Lundin

In recognition of the sixth international Rare Disease Day Feb. 28 - which includes events, presentations and symposiums in 68 countries - Moraga resident and executive director of the Cystinosis Foundation, Valerie Hotz, will return to Washington, D.C. with a group of local people similarly affected by the rare disease to try to make a difference.

In 1982 Hotz's nephew Joshua, who was then 18 months old, was diagnosed with Cystinosis - a rare genetic disorder that causes the abnormal accumulation of the amino acid cystine (a building block of proteins) in all the cells of the body, forming crystals and damaging them. Approximately 1 in 150,000 newborns are diagnosed with Cystinosis each year, according to the Genetics Home Reference (ghr.nlm.nih.gov).

"When someone is diagnosed with a rare or ultra-rare disease, the whole family is impacted," says Hotz. "It took months to find what was affecting [Josh]. At first, the parents were told that nothing was wrong with him, even though they were seeing the problems, such as excessive urination and failure to thrive." When Josh was finally diagnosed, they were told that there was no treatment, that children with the disease did not survive for more than 10 years and that they should just go home.

"My mother, Josh's grandma, Jean Hobbs-Hotz, would not believe there was no treatment, and we all started looking everywhere for a solution. I found Dr. Sheldon Orloff who put us in touch with the U.S. Cystinosis specialist at UC San Diego, Jerry Schneider, who dedicated his life to researching this disease and found a treatment with professor Jeff Thoene from the University of Michigan."

A year after Josh's diagnosis in 1983 Congress passed the Orphan Drug Act, which allowed the federal government to assist in the development of medications for rare diseases. Josh was able to become part of a clinical study with the first drug available to counteract the disease, Cysteamine.

Now 30 years old, Joshua continues to take the drug. Unfortunately he lost his vision six years ago, but has continued his profession as a pianist. The eye drops that protect the eyes were developed too late for him.

Patients of rare diseases often struggle to get a diagnosis, information and treatment, notes Hotz. Her goal while in Washington will be to push for the passage of bill H.R. 460, The Patients' Access to Treatment Act, which would help alleviate costs for patients such as co-payments and coinsurance for prescription medication.

"What we are very concerned about is reimbursement, so patients can have access to new treatment." She says that California senators Diane Feinstein and Barbara Boxer, and congressman George Miller are supportive.

"The current medication has to be taken every six hours, including at night," says Hotz. "There is now a new drug coming that will be long acting and that people will take only twice a day, that way children won't have to be awoken in the middle of the night; it will change the quality of life for the patients and their families tremendously." The FDA should soon approve the drug. The Cystinosis Foundation wants it to be affordable.

Hotz's mother started the Cystinosis Foundation as a way for patients' families to talk and support each other. After Hotz raised her three kids in Moraga, she took over the direction of the foundation; her mother is still involved.

"Since 1985 we've held a national convention, and after a while we started seeing people coming from overseas," says Hotz. They were approached by French professor Michel Broyer to expand the conference in Europe. The first one was held in 2000.

"There are pockets of populations affected all over the world," says Hotz. "We've established 12 support groups in 12 different countries."

Sessions for families are held during the conventions where they get information about the disease, its cure and ask questions. "We also have professional sessions for the medical profession," says Hotz. "These sessions have reinforced international bonds between scientists. They get together, brainstorm, and it's been very fruitful."

The discovery of the gene responsible for the disease was the result of international cooperation.

In the United States, Hotz organized "Capitol Hill Days" Jan. 28 where the foundation took 10 families affected by Cystinosis to meet their congressional leaders and prepare for Rare Disease Day.

The next international convention in 2013 will be in Brazil and in 2014 in England.

For more information about Cystinosis, visit cystinosisfroundation.org; other resources recommended by Hotz include: rareconnect.org, and the Undiagnosed Disease Program at www.genome.gov/27544402.

International Rare Disease Day is Feb. 28

The Sixth Annual International Rare Disease Day - an advocacy day that includes events, presentations and symposiums in 68 countries to recognize rare diseases as a global health challenge - is Feb. 28. The European Organization for Rare Diseases (EURORDIS) estimates that as many as 5,000 to 7,000 distinct rare diseases exist (also sometimes called orphan diseases). On average (definitions vary per country), a disease is rare if it affects one out of 2,000 people; there are also ultra-rare diseases that affect one out of two million.

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