

OceanScene

SANTA MONICA • PACIFIC PALISADES

Life with a Coastal Point of View

Vol. 1, No. 1, 2005

PREMIERE!



Bobi Leonard
**Business Woman
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Palisadian Residents Lead the Crusade Against Familial Dysautonomia

by Maryjo Faith Morgan

What do you *do* when you don't know *what* to do?

"You search and you learn, that's what you do." Mavis Feinberg, president of The Cure FD Foundation of Pacific Palisades, speaks from personal experience. "My grandson was born with a terrible medical condition that had doctors scratching their heads, and my family traipsing from one doctor to another in search of relief and an answer. After almost two years a diagnosis finally came through." The devastating truth hit home. Her grandson had Familial Dysautonomia.

Pronounced "disauto-NO-mia", Familial Dysautonomia, (FD) is a progressive, degenerative, neurological, fatal genetic disease carried by 1 in 27 people of Central or Eastern European (Ashkenazi) Jewish descent. It is now as prevalent as Tay Sachs once was. Children with FD have unstable heart rate and blood pressure. They cannot eat by mouth and stomach tubes provide nourishment; because they are very apt to aspirate into their lungs, they get pneumonia frequently. Their eyes do not tear, often become ulcerated, and need frequent eye drops. Children with FD tend to be undersized and have very low muscle tone, which causes them to walk and talk differently from other children. A common symptom is prolonged, unprovoked retching. When in a retching crises, the exhausted and dehydrated child may retch once every minute or two for as long as three days!

FD is a progressive disease, ultimately leading to the degeneration of all organs and bones. Many children die before reaching their eighteenth birthday, after a life punctuated by crises, fatigue, and alienation from other children. Invariably, the family suffers along with the child, and quality of life is a huge issue.

Once she was finally armed with a diagnosis, Mavis, a retired college professor, began her search in earnest. She



Paul Coretz discusses a bill with Al Feinberg.

found that it was not uncommon for FD to be misdiagnosed or missed altogether, and learned that there may be hundreds of children in this country alone who have undiagnosed FD. However, since worldwide, numbers of those affected are not huge, there is little work being done toward a cure for FD. Her relentless search ultimately led her to an amazing researcher, Berish Rubin, who is dedicated to, in his own words, "... not allow another child to die on our watch."

Although the situation appears bleak, and more research is urgently needed, Berish Y. Rubin, Ph.D., Head of the Laboratory for Familial Dysautonomia Research at Fordham University, and Sylvia L. Anderson, Ph.D., Director of the same lab, are on the job, often toiling 60 or more hours a week each. Thanks to their untiring efforts, hope is on the horizon and children are already experiencing some relief. They have already charted a road map; they need the funds to get the job done.

Effective treatment, still in its infancy, is making a very real difference for children living with FD. In the summer of 2000, Dr. Rubin identified the genetic cause of FD; the mutations responsible for FD were found to occur in a gene encoding a protein termed IKAP. Genetic testing was then established so that carrier status and the risk of having children with FD can be determined, which helps couples make

informed choices, ultimately reducing the number of children born with FD.

In 2003 Dr. Rubin's lab identified two compounds, a form of vitamin E called tocotrienol and a chemical component of green tea called epigallocatechin gallate (EGCG) that actually reduce symptoms and dramatically improve the quality of life for children with FD.

Dr. Rubin states emphatically that not only have they made great strides, but they have some truly promising things on their drawing board — they simply need the funds to proceed. "Our lab at Fordham is funded entirely through donations. The progress we can make is in a direct relationship to the research funds we have available. It is very challenging, especially when we see children so dramatically improved through our efforts already."

He speaks of one child in Chicago who had been in constant crises, hospitalized for six months. Eventually Dr. Rubin was called in, and after only two weeks on tocotrienol, the boy was released from the hospital. "I told his mother not to worry, that I'd be there for his Bar Mitzvah. That was over a year and a half ago, and his symptoms have decreased. I am happy to tell you, Dr. Anderson and I just returned from Chicago, from that boy's Bar Mitzvah."

Dr. Rubin sites improved motor function in children treated with tocotrienols, such as being able to play the piano, or being able to walk and leave the wheel chair behind. One child in Florida had missed a quarter of the school year due to repeated crises last year; this year with tocotrienol treatment, not one day of school was missed. He is excited by their progress, and is utterly determined to relieve the suffering of children with FD and their



Daniela Snooke, her mother, and the Feinbergs.

families. "Since the population affected by FD is small, it is easy that they might be overlooked. I cannot let that happen. Just one child suffering and dying is one too many."

Board member Brian White saw the need for professionally designed materials

and willingly gives of his time and expertise to design and maintain the website for this small grass-roots organization. "I was raised in a family that put a great deal of value on helping out in the community and with children in need. Although staying involved is a challenge due to a demanding business, no amount of time constraints could ever outweigh the pain that these children go through on a daily basis. I guess that's why I stay involved, because these children need help." Indeed, board members, advisors, and foundation volunteers all seem driven by this same need to help the children.

On April 29, 2005, two representatives of the Cure FD Board of Directors, Mavis and Al Feinberg, met with California State Assemblyman Paul Koretz of the 42nd Assembly District to discuss introducing a bill to the State Legislature which would include FD carrier testing along with the Tay Sachs testing, which is already recommended for certain populations when couples take out a state marriage license. Such a measure would prevent many cases of FD in California in the future and would

save the state massive amounts of money which would otherwise be spent on lifetime care and treatment for those severely disabled and ill with FD. Assemblyman Koretz has made assurances that he will work to see such a bill introduced to the Assembly at the coming legislative session in fall of 2005.

There is an urgency in everyone associated with the Cure FD Foundation, an excitement fueled by hope. You may find, as Mavis did, that once you know about FD you cannot sit still.

To learn more about FD and hear why Mavis Feinberg is so committed to the Cure FD Foundation, log onto: www.curefd.org. Be sure to watch the video featuring Daniela Snooke and her mother Marcy, and hear Dr. Rubin speak about the strides toward a cure he is anxious to take. See details of his research on: www.fordham.edu/biology/rubin/rubin.htm.

Mail donations to: Cure FD Foundation, 1029 Via de la Paz #128, Pacific Palisades, CA 90272; ph: (310) 459-1056; email: info@curefd.org.