

PTA's Quest for Information, Support Leads To Formation of Fragile X Association

By JULIE ELLIS

When Mary Seward's second child was diagnosed with Fragile X Syndrome, she was "devastated, confused and in need of help."

She immediately tried to learn more about the genetic disorder and locate a support group of other Fragile X parents to help her through the ordeal. She became increasingly frustrated, however, at the lack of knowledge, information and support available at the local level for parents of Fragile X children.

"You can speak to a lot of professionals and go to psychologists, but talking to another parent of a Fragile X child who has been through it is the best remedy to moving on from the diagnosis and figuring out what is best for your child," said Ms. Seward, a physical therapist assistant in California. "I wasn't able to find any other parents of Fragile X children to talk to and I realized that something needed to be done."

Once she got over the initial shock of the diagnosis, Ms. Seward began forming The Fragile X Association of Southern California.

The goals of the association, according to its brochure, are to "provide vital support and information to Fragile X families and to increase public and professional awareness of the Fragile X Syndrome."

WHAT IS FRAGILE X?

"Fragile X Syndrome is the leading known hereditary cause of developmental disabilities," according to the association.

Fragile X males and females may be affected with a variety of developmental learning disabilities, mild to severe mental retardation, behavior management challenges, attention deficit



Timmy Seward at age 2.

hyperactivity disorder, sensory and perceptual problems, autism and autistic behaviors. Physical features of the disease may or may not include large ears, flat feet and hyperextensible joints.

It is estimated that Fragile X affects one in 1,000 males and one in 2,000 females. Estimates on incidence of female carriers are as high as one in 300.

The Fragile X chromosome was first discovered in 1969, but it was not associated with developmental disabilities until 1977. In 1991, a DNA test for the Fragile X gene was developed.

DIAGNOSIS

The lack of knowledge and aware-

ness of Fragile X often delays diagnosis of the condition, Ms. Seward said.

She said she first took her son, Timmy, to the doctor when he was nine months old because she suspected something was wrong, but did not get the diagnosis until he was two-and-a-half.

"He had one sort of little problem after another that didn't really quite add up to anything major, so I would take him to different specialists," she said.

They took him to a podiatrist, for example, because he had flat feet and crossed-over toes.

"It was a long road to the diagnosis but we were lucky — many families have to go through many more years of searching."

She said intervention for many Fragile X children often does not start until a speech delay is noticeable.

"It is really, really helpful to get a concrete answer to why your child is having problems," she said. "Then you can deal with it, get the right therapies and get the schools to educate the child properly."

Increasing awareness of Fragile X, she said, is beginning to make a difference.

"I am running into more and more parents, especially of children who are autistic, who have said that their physicians are testing for Fragile X," she said. About 20 percent of autistic children test positive for Fragile X.

There is still, she said, a long way to go, however.

"We have to educate the physicians,

because they still know very little about Fragile X," she said.

"It is not their fault — you can't expect doctors to know about something that didn't exist when they were in school and Fragile X is only starting to hit the curriculum now.

"My driving goal is that no one else will have to go through what I did after getting the diagnosis. So, I will keep trying to teach people about what Fragile X is and how to deal with it as I am learning how to deal with it myself on a day-to-day basis."

Mary Seward, PTA

"Even with continuing education, we still haven't made the dent we need to," she added.

TREATMENT

There is currently no cure for Fragile X Syndrome, but many treatments have proven to be beneficial.

Specific medications, behavioral management techniques and appropriate inclusion techniques combined with physical, occupational and speech therapy help maximize the potential of individuals with Fragile X Syndrome.

"Fragile X patients often have low muscle tone, balance/coordination difficulties and poor motor planning skills," Ms. Seward said. "[So], as far as physical therapy, Fragile X children need therapy balls and a lot of fun activities to help them with crawling and hand-eye coordination."

Exposing Fragile X children to positive role models is also important, Ms. Seward said.

"Research shows that Fragile X children do very well with imitation," Ms. Seward explained. "So if you put them in a program with children who have behavioral problems and other disabilities, that is what they will learn to mimic."

"If you put them in a program with normal role models, they will imitate that behavior," she continued. "We have definitely found that putting [Fragile X

children] in a regular classroom, with the proper support, really does work and is effective."

Ms. Seward said her training as a physical therapist assistant has definitely helped her with Timmy's care.

"When you have a child with a disability, there are a lot of people who are taking care of your child and you need to be able to coordinate that care," she said. "The people that treat Timmy never meet, so there are five or six experts and I am the one that teams them together."

"It definitely makes it easier because I was trained in a medical model and in working with teams," she added.

THE FRAGILE X ASSOCIATION OF SOUTHERN CALIFORNIA

Ms. Seward began the groundwork for the Fragile X Association of Southern California in December 1993, shortly after Timmy was diagnosed.

With help from staff at Children's Hospital/University Affiliated Program in Los Angeles, Calif., she was able to get in touch with some other Fragile X families.

"There is a real core group of people who are interested in developmental disabilities and families [at Children's Hospital] and they are the ones who helped me find the other families and provided all the technical and professional support I needed," Ms. Seward



The Seward family. (L to R) Mary, Elizabeth (8), Timmy (4) and Roger.

said. "I couldn't have done it on my own — not after just getting the diagnosis — it was just too much at one time."

There are currently about 60 families involved with the association, Ms. Seward said.

"These families are providing energy and support, but our numbers are not anywhere near what they should be considering how many are affected with the disorder," she said. "We should have thousands and thousands of people considering the size and population of the area we cover."

"It just shows that the diagnosis is not getting out there," she added.

The association conducts monthly informational meetings and has had two family picnics.

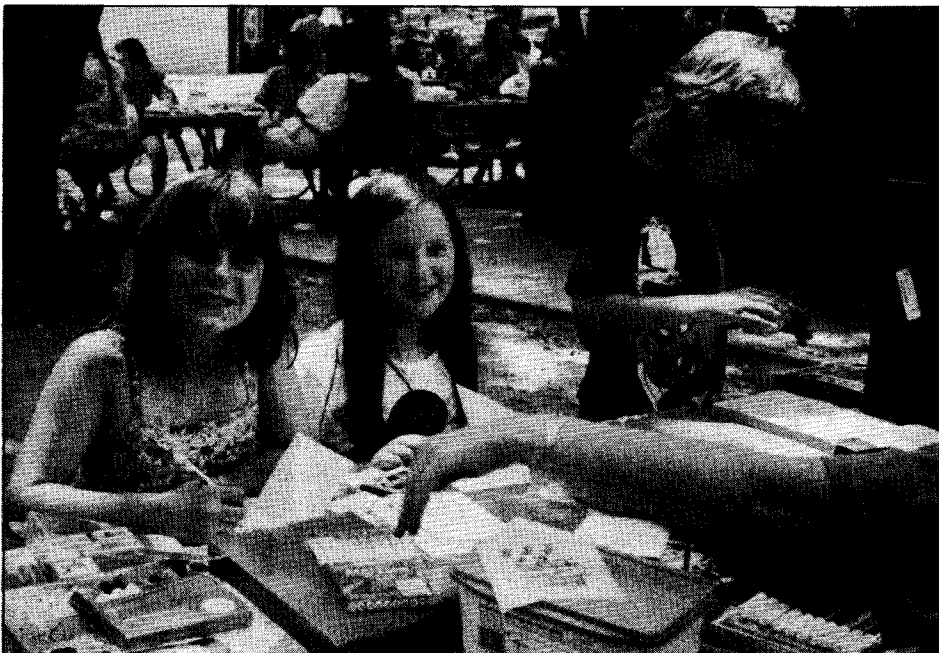
"Most Fragile X children, unless there are others in their family, have never been around other Fragile X kids so it was a lot of fun to see them together and watch how they relate," she said. "It is also very helpful for parents of young Fragile children to see adults with Fragile X and see that their families lived through it and are doing extremely well."

The association is planning a one-day symposium in February 1996 for health professionals and Fragile X families, and will continue to sponsor activities to increase awareness.

"My driving goal is that no one else will have to go through what I did after getting the diagnosis," she said. "So, I will keep trying to teach people about what Fragile X is and how to deal with it as I am learning how to deal with it myself on a day-to-day basis," she said.

For more information about the Fragile X Association of Southern California and the 1996 symposium, call the association at (818) 688-8765. ▼

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Timmy's sister, Elizabeth, plays with Stacy Reinhardt, who also has a brother with Fragile X, and Nathan Spahr at a Fragile X Association of Southern California picnic.