

Local documentary shines light on gene disorder

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SANTA CRUZ -- Fragile X syndrome is a rare but devastating gene disorder that can leave its victims without the ability to walk, talk or think normally. Simple things like getting dressed and going to the bathroom alone are often impossible.

Kathy Elder, a Santa Cruz film producer and writer, has watched her 30-year-old twin cousins Ryan and Conor Kahl struggle through life with Fragile X and the heartache their parents have suffered as they've worked to come to grips with the disorder, which usually causes mental retardation and autism.

Elder has poured her compassion and experience with the disorder into a documentary called "Living with Fragile X," an hour-long film that reveals what it's like to live with the syndrome and how doctors hope for a breakthrough in gene therapy and gene repair.

"It's important to know about because it's genetic," Elder said. "This is an emotional documentary, not something you'd find on 'Nova.' It's about how we come to accept the people we love."

Elder and her business partner Greg Mishey, a freelance editor who lives in Seabright, spent the past seven years working on the documentary. They shot 70 hours of interviews with Fragile X families and experts as well as snippets of kids and adults living with the disorder.

They interviewed expert Fragile X doctors and scientists at the UC Davis MIND Institute and Emory University in Atlanta studying the disorder.

The goal of "Living with Fragile X" is to educate audiences about the syndrome and show a new side of those who are traditionally considered "flawed" or "abnormal," Elder said.

The Kahl twins, who are featured in the film, may never drive, handle money or date, but they can live a productive life, their father Bucky Kahl said.

"They're normal in a different way," Kahl said.

Like most parents of children with Fragile X, Kahl and his wife, Sara, worry about who will take care of the boys when they're not here.

An estimated 100,000 in the U.S. have been diagnosed with the disorder, which affects the X chromosome, according to Robert Miller, executive director of the National Fragile X Foundation in Walnut Creek.

Chromosomes are packages of genes passed from generation to generation. Most people have 46 chromosomes, two of which are sex chromosomes. Women have two Xs, and men have an X and a Y.

Typical Fragile X characteristics include a long narrow face with a large forehead and ears. People with Fragile X are often flat-footed and double-jointed, and men tend to have enlarged testicles, Miller said.

Fragile X is the most common mental impairment passed through the genes. Down syndrome is more common, but is not inherited, he said.

"Either the mother or father can be a carrier," Miller said. "If the mom has it, she has a 50 percent chance of passing it to each pregnancy. If the father has it, all of the daughters will get it but not the sons.

"One of the problems with preventing the disorder is most people don't know they carry the mutated gene, Miller said. A basic blood test determines if someone carries the mutated gene.



Dan O'Connell
Greg Mishey
about the Rio
the Rio
at

'LIVING WITH FRAGILE X'

what: Documentary about gene disorder that causes mental retardation and autism.
when: 7 p.m. Jan. 12.
where: Rio Theatre, 1205 Soquel Ave., Santa Cruz.
details: 60-minute film followed by Q & A with Fragile X experts Dr. Randi Hagerman, medical director of the UC Davis MIND Institute, and Robert Miller, executive director of the National Fragile X Foundation.
cost: \$5.
contact: 423-8209 or visit
www.thinkmediaonline.com or www.fragilex.org.

Elder's partner Mishey hopes people come away from the film with a fresh perspective on how they look at people who don't fit the status quo.

"Before you cast judgment, think about what that other person is going through," Mishey said. "We're not all on the same wavelength."

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