Living Proof: Teenager lives life to the fullest

By Anne Ward Ernst

Punching buttons on a video-game controller, slumped low into the sofa in his Almaden Valley living room and wearing a black-hooded sweatshirt and loose-fitting jeans, he looks like lots of kids his age. But Adam Fulton is not like lots of kids his age.

He has a rare disease that kills most children in infancy, and when Adam was just 5 months old, he almost died from it.

"He was like a rag doll," says his mother, Valerie Fulton, of the episode that sent them to the hospital. "There was no tone in his muscles."

While at the hospital it was discovered that Adam's liver was enlarged and the tissue around his heart was infiltrated with fat. It was then that he was diagnosed with a genetic syndrome called Long-chain 3-hydroxyacyl-CoA dehydrogenase, or LCHAD. **[Correction note from Valerie, Adam's mom – LCHAD is not a syndrome but a metabolic disorder whereby a specific enzyme is missing or deficient.]**

His prognosis was not good.

"Most said, 'He'll probably die of heart failure,'" she says of one of the most common results of the disease.

Unless you talk to him about his special diet or activity restrictions, you would never know of his condition.

He is now a 13-year-old seventh-grader at Castillero Middle School who is on the swim team, is an active fencer and, as a baritone in the school's concert choir and men's choir, performed in the Anaheim Heritage Festival last weekend. He's a good student and thought of highly by his teachers.

"He's one of my better male singers," says his choir instructor, David Finch. "He always gives you everything, and he's always so well-behaved. He's a great kid."

Adam is active in Children's Musical Theater San Jose and of the nine productions he's been in, he has a clear favorite: *Oliver*.

"I got to be an orphan and a thief," he says.

On the day he received his report card, he was trying to persuade his mother to pay him for his high marks—all As and Bs—but his mother would have none of it. But she is going to give him money to participate for two weeks this summer in yet another series of medical tests concerning LCHAD. So, though he leads a seemingly regular teenage life, it always circles back to LCHAD.

The one person who gave Valerie any sign of hope of survival when Adam was first diagnosed is the same person who continues to monitor Adam's progress today—his nutritionist, Elaina Jurecki.

"She told me, 'We'll treat it with diet,'" Valerie says.

Jurecki works in the Kaiser Permanente health system and says it currently follows four people in Northern California with the same disease.

LCHAD is a condition in which the body cannot oxidize fatty acids because a necessary enzyme is either missing or not functioning correctly. Jurecki and other experts in the metabolic field say there are probably fewer than 200 people afflicted with LCHAD living in the United States, and most are children.

"There are not a lot of adults because [babies] often die from this disease," says Jurecki.

Early death could be avoided, Jurecki and others say, if the state of California would adopt a requirement to test for LCHAD at birth so that parents can be armed with knowledge and treat the disease with diet, as the Fultons have. All newborns currently get five standard tests from blood taken from a prick on their heel, and LCHAD could be determined from the same sample.

"They have had that in place for years," she says. "Adding this would be just a matter of cost."

Melanie Gillingham, a researcher at the Oregon Health and Sciences University in Portland, Ore., says a lot of states, including Oregon, are starting to screen for LCHAD, but California is still in the debate stage, she says.

"Hopefully, we'll be able to pick these kids up before they get sick, and hopefully they will be able to survive into adulthood."

Some typical symptoms of this metabolic disorder include lethargy, hypoglycemia, developmental delay, cardiomyopathy—problems with the functioning of the heart—and, as in Adam's case when he was first diagnosed, poor muscle tone. The genetic mutation is unknowingly passed on by two "carrier" parents, and of babies born to such parents, there is a 25 percent chance they will be born with the disease and a 50 percent chance those children will be carriers.

Valerie has two older sons who do not have the disease, and said she had expressed concern to Adam's pediatrician when he was an infant that he was unable to hold up his head on his own and seemed to be developing more slowly than her first two babies. She said the doctor attributed those symptoms to Adam's premature birth. Valerie herself had almost died during her pregnancy with Adam because of symptoms attributed to the HELLP syndrome—which stands for *h*emolysis, *e*levated *l*iver enzyme levels and a *l*ow *p*latelet count—and she was experiencing kidney failure, so Adam was delivered by C-

section 5 ½ weeks early. At first, doctors told her that her HELLP syndrome and Adam's LCHAD were unrelated, but scientific findings since reported show otherwise. She and the baby were chemically and biologically at odds with one another.

"Basically, we were killing each other," Valerie says.

Comparing infant photos of Adam, she points out how one taken of him just before his diagnosis shows him thin-looking, with his skin pale and mottled. In a photo taken a couple of months later, he is plumper with a healthy, rosy hue. The difference was found in his diet.

Jurecki put Adam on a restrictive, very low-fat diet—specifically low in the long-chain fats commonly found in most diets, which his body cannot metabolize into energy. But he also requires a different type of fat that is not found in our foods—medium-chain fat—and without this fat in his system, his body starts searches for other energy sources by attacking and breaking down muscles.

He was given this special diet through a feeding tube in his nose until he was 2 ½ years old. Now every day he drinks 18 grams of medium-chain triglyceride oil **[Correction note from Valerie--three times a day or 54 grams]** mixed with nonfat milk. The prescription oil costs $75 per bottle and is not covered by their medical insurance plan, but Valerie says she found a less-expensive source, though it still isn't cheap at $11 per 16.7-ounce bottle. He eats regular foods, too, and loves things like rice and sushi, Valerie says, but knows that if he has a slice of pizza, he has to remove the cheese and pat off the oils first. It's a diet they can live with, but it's not always easy.

"You can't just go out the door and go out to eat," says family friend Jenny Carroll, whose 10-year-old daughter, Jane, also has LCHAD. "If you go to someone's house they might not have what she can eat. She has to have her formula, the nonfat milk with oil in it, every six hours."

The Fultons and the Carrolls, who live outside of Madison, Wis., became friends through one of the studies both youngsters participated in that measured the effects of adding fish oil to their diet to help control and contain the retinal degeneration that occurs in children with LCHAD and leads to blindness.

During his young life Adam has undergone a variety of tests, has been involved in several different studies, and has traveled nationally and internationally to be observed by scientists and doctors who will, because of his participation, better understand the disease for future generations.

"LCHAD is a newly described disease. Doctors have only been describing and looking at it since the late '80s or early '90s," says Gillingham. "We don't really know what an older person [with LCHAD] looks like. There are a lot of things we don't know about this disease or them."

The tests he will endure this summer—the ones for which his mother is going to pay him $20 a day—will keep him virtually quarantined for two weeks, and Jurecki says results of the controlled studies will be a great help to those who study this little-known disease.

"The only way we learn is by doing these collaborative studies," Jurecki says. "To have patients like Adam—and there is potentially no direct benefit for him—really helps the community as a whole."

They are not easily tolerable tests, Jurecki and Gillingham say.

"He's already had a bunch of ophthalmology tests. This one is an electroretingram, or ERG," Gillingham says. "It's not an easy test to do. If I did it, they would have to sedate me."

A contact lens placed on the retina has electrode wires coming off of it and keeps the eye open and then his face will be placed in a bowl and bright lights are flashed off and on to test visual acuity and activity. The test lasts about an hour to an hour-and-a-half, and Gillingham says they sedate younger kids, but Adam tolerates tests such as these so well that he has not received any sedatives the last couple of times he participated. They say he's an amazing patient, but to Adam it's just a way of life.