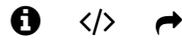


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**Parents share challenges they face with children who have rare genetic mutation** 2:25

In August 2017, families with children who have a rare genetic mutation gathered with scientists at a hotel in suburban Washington, DC, at the invitation of Sacramento lobbyist Joe Lang, whose daughter Jordan has the disorder. Will and Lisa McCarty of Fresno came with their son, Liam. The mutation is on a gene known as PPP2R5D. **Dan Morain** - The Sacramento Bee

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DAN MORAIN

Their son has a rare genetic disorder. But here's why they don't give up hope.

BY DAN MORAIN

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At about the time their baby Liam was undergoing heart surgery, Lisa and Will McCarty saw a movie about the first child to undergo the operation 50 years earlier. The child's parents had agreed to the experiment so that other children might live.

"What a gift that family gave to the rest of us," Lisa said, thinking about what her family might offer future children.

OPINION

When they were in high school in Fresno, Lisa asked Will to the prom, and they got married a year later. Will served in the Army National Guard, came home in one piece from

Iraq and got hired by the Fresno Police Department. Lisa got a special education teaching credential. They had a bright and lovely daughter, Ayiana, 11 years ago, followed three years later by Liam.

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Heart surgeons at Valley Children's Hospital in Madera worked their miracle, but Liam's problems went deeper. He couldn't nurse, wouldn't look people in their eyes and didn't hit milestones. At their doctor's suggestion, they had his genome mapped in 2015. The result showed a mutation on one gene, known as PPP2R5D.

In the evenings, Lisa would type PPP2R5D into Google, searching for some information and hope. She got a hit when a mom in Arizona whose child has the same mutation posted on Facebook. Now there are about three dozen who share their successes and frustrations on a Facebook group.

"That's been my job for 12-plus years, fixing problems," Will said. "People have a problem. I come, I try to figure out how to fix it, or teach them to fix it. With Liam, I have no way to fix it."

Last week, I wrote about Joe and Cynthia Lang and their daughter, Jordan, who has the same mutation. One of Sacramento's most successful lobbyists, Lang helps to fix issues for the liquor industry, race tracks, tobacco, oil, and many other interests. Now, he's on quest to fix his child. Toward that end, he paid for PPP2R5D families from as far away as Israel, Norway and New Zealand to gather at a Marriott in Virginia, where they met one another and conferred with scientists who are researching the gene.

Good cop that he is, Will McCarty was skeptical. Why would a guy he never met put his family and him up in suburban Washington D.C., for three nights? A scam to bilk desperate parents, perhaps. McCarty's sleuthing showed Lang checked out. So there he was last weekend with his wife, daughter and Liam in a hotel room telling Liam's story to one of the nation's top genetic researchers, Dr. Wendy K. Chung of Columbia Medical School.

Unlike most of the kids with the defect, Liam can repeat the sound of each letter, as Lisa and Liam demonstrated for Chung. Lately, she has him reading Pippi Longstocking and he laughs in appropriate places. A cousin is diagnosed with autism, as is Liam. Chung took notes.

"If you had to put your finger on the one or two things that you're looking for a solution for, what would it be?" Chung asked. Communication, they said. "I think he knows so much more than he can communicate," Lisa said.

Many of the families who gathered in Virginia came from countries that paid for genetic testing, but not therapy. Not so in California. As a junior lawmaker in the late 1950s, Frank Lanterman, a Republican assemblyman from the Pasadena area, was appalled at the bedlam he witnessed in state hospitals, and made it his mission to improve patients' lives. In 1977, long before Lisa and Will McCarty were born, he carried the final bill that bears his name.

The Lanterman Act, signed by Gov. Jerry Brown, entitles every child in California who has an intellectual disability to cradle-to-grave therapy. The Department of Development Services' budget of \$6 billion grows as the incidence of autism rises. The caseload is more than 300,000 people.

From age 1, Liam received 20 and sometimes 30 hours a week of therapy, though they've scaled back lately. He makes eye contact now, and listens to his parents when they tell him to stop before he injures himself. The bills would total as high as \$70,000 a year. A school teacher and a cop never could afford it. It's all funded by the state.

"I don't know where Liam would have been without it," Lisa said.

In 2015, President Barack Obama announced the precision medicine initiative, the notion that science could tailor treatment and prevention to an individual's unique characteristics, including their genome sequence. It's relevant to treatment for the 7,000 known rare diseases, including PPP2R5D, otherwise called Jordan's Syndrome. Unfortunately, President Donald Trump urged a 20 percent cut in the funding for the National Institutes of Health. If approved by Congress, that would impede the research and cures.

A government grant for PPP2R5D would be nice. But Lang is not waiting. He has set a goal of raising \$20 million to fund at least the initial research into the syndrome. In Virginia, the researchers gathered blood from each child who has the mutation. They intend to use the blood to grow brain cells, in a search for some way to interrupt and maybe alter the mutation. Chung believes that whatever drug they develop could have applications beyond the rare disease.

Lisa and Will McCarty have no idea whether any of this will help their son. But she thinks of the movie about the pioneering heart surgery, "Something the Lord Made." If the research helps other families, Will, Lisa, Ayiana and Liam will – like Joe and Cynthia and Jordan Lang, and Frank Lanterman before them – have given a greater gift than most. "We think that is pretty incredible." It is.

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