HEALTH

Event raises awareness for World Rare Disease Day

Rare diseases may affect more than you realize

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SANTA CRUZ » Communities around the world Saturday will participate in World Rare Disease Day in order to shed a light on diseases that are less common, but of no less importance.

Nearly 7,000 different rare diseases exist today, each of these with less than 200,000 people diagnosed. According to The XLH Network, an international organization led by a Santa Cruz local, "training clinicians about rare disorders can minimize the possibility of misdiagnosis and delay in therapy."

Susan Faitos, a lifelong Santa Cruz resident and now executive director of The XLH Network, said she is looking forward to the chance to tell her story. It wasn't until Faitos was 55 years old that she finally met a doctor who understood the disease that she had been living with all of her life. "It's so rare, people with it often have to educate doctors about it," she said.

At 18 months of age, Faitos' parents approached Stanford University medicine specialists, but at that time, in the early 1960s, the ailment that kept Faitos' vitamin D and phosphorus levels constantly low and her legs oddly curved remained a mystery. It wasn't until a surgery when she was 14 years old left her bones unhealed months later that doctors realized what had been ailing Faitos since birth: X-linked Hypophosphatemia, or "XLH."

XLH is a lifelong disorder caused by a mutation, or random change, in one gene. Faitos explained. Genes hold the genetic building blocks of life — adenine (A), thymine (T), cytosine (C) and guanine (G). The Human Genome Project estimates that humans have somewhere from 20,000 to 25,000 genes. It's the arrangement of these four items that determine how proteins are formed and function in the body.

In the case of XLH, a different arrangement of A, T, C and G leads to a change in the protein called "PHEX." This ultimately causes the body to dispose of more phosphorus than normal in the urine, leading to an overall deficiency that affects bone density and growth. Aside from bowed legs, as Faitos experienced at a young age, other symptoms of XLH include a shorter stature or decreased growth rate, a waddling gait, and muscle pain or weakness, among other ailments.

According to Thomas Carpenter, a professor of pediatrics at Yale University School of Medicine who specializes in XLH treatment, just one in 20,000 people have XLH. But for Faitos, a lifelong XLH patient, learning how to deal with this rare disease has been frustrating and lonely. "There was so much I didn't know about my disease the only one made it embarrassing and almost shameful," she said.

With the invention of the internet and its widespread use across the world, Faitos found an online group of people who also lived with this rare disease. "After years of dealing with it by myself, it felt weird to suddenly start talking about it," she explained. This online forum eventually ma-

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terialized into a physical meeting in San Francisco in 2015.

"People with XLH look different ... but walking into a room with others who also have XLH was a life-changing experience," Faitos said.

Now 60 years old, Faitos recently became more invested in spreading the word about this rare disease by leading The XLH Network. In preparation for Rare Disease Day, she has been working to plan inclusive events online in an effort to raise awareness about XLH and other rare diseases.

To those who are interested in participating, a Facebook Live chat will take place Saturday, and Chris Younger, an XLH Network board member will be releasing a song he wrote titled "XLH Fight Song" on YouTube.