Utah-led group gives hope to sufferers of rare skin disease

Genetics » DNA "typo" makes walking a challenge.

By JULIA LYON
The Salt Lake Tribune

When Mary Schwartz's son fell in love with a young woman whose painful foot condition often forced her to walk on her knees, the mother knew she had to do something to make a difference.

The genetic skin disorder meant her new daughter-in-law constantly battled calluses and blisters on her feet Her future grandchildren had a 50-50 chance of doing the same.

The ultra-rare disease strikes so few people that her daughter-in-law

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Janice Schwartz, who suffers from a rare skin disease, uses a wheelchair because of the pain caused by the blisters and calluses on her feet.

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>
> **JANICE SCHWARTZ** | *who suffers from pachyonychia congenita, a rare skin disease*

Disorder

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didn't meet a fellow pachyonychia congenita sufferer until she was 36 years old.

"I felt like I wasn't alone in the world," said Janice Schwartz, now 43, of the encounter. "Instead of thinking I'm just this bizarre person, this freak, there are other people who have it."

And now two of her four children, both teenage sons, have it, too. But their lives could change thanks to the Global Rare Diseases Patient Registry and Data Repository, a new effort from the National Institutes of Health.

Pachyonychia Congenita Project, the nonprofit Mary Schwartz founded, is one of the groups chosen to participate in the pilot program. The plan is to grow the existing patient registry developed by the nonprofit, place it online in additional languages and use standardized questions that will allow researchers to analyze data across diseases.

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"They do get sidelined" » Doctors diagnosed Janice Schwartz with pachyonychia congenita as a baby when her fingernails and toenails were thicker than normal. The real problems began when she started to walk.

Blisters and calluses sprouted on the bottoms of her soft toddler feet and continue to plague her. The pain changes: Sometimes it's a burning sensation like her feet are on fire, other times they just ache or pulse. She can't stand or walk without pain. Even the blankets on her bed can make her feet hurt.

As far as Schwartz knows, there's no treatment other than staying off her feet and managing the pain, which she does with the help of crutches, an electric wheelchair and a rolling office chair in the kitchen. Or walking on her knees, which are so battered she guesses she might qualify for a knee replacement.

No one in Schwartz's family had the disorder before she was born with what she calls a "typo" in her DNA. She knows some people with PC have chosen not to have kids. But about half of the cases of the disorder are spontaneous, meaning there is no family history.

"It was a big decision to have children," the Cottonwood Heights mom said. "But even with the pain you can have a really good life."

Her teenage boys, now 15 and 17, have had to give up sports like skiing, soccer and baseball because the pain to their feet was too intense. One is a camp counselor and relies on a walking stick while working with kids. The other has a job as a computer programmer. At the end of the day, the honors students come home and sit down as soon as possible.

"I think about my boys, and I would love for them to walk pain-free," the mother said. "They still try to have great attitudes, but they do get sidelined."

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Discovering community » Individuals with the roughly 7,000 rare diseases in the world are often so scattered that little progress can be made on treatment. Only six people in Utah have pachyonychia congenita, Mary Schwartz said.

Less than 20 percent of those diseases are known to have patient registries, according to Yaffa Rubinstein, the director for patient resources at the NIH Office of Rare Diseases Research.

"So if you want to do any study or clinical trial in a meaningful way you have to locate these patients," she said.

The global registry pilot program is working with 19 organizations that already have their own registries and 15 that do not — assisting those to develop ones. The ultimate goal is treatment and cures.

The PC Project started collecting patient information in 2004 for researchers around the world. Free genetic screening is available. The International PC Research Registry has already produced a new classification system for the disorder and higher participation in clinical studies. About 1,100 people in more than 50 countries have been identified so far.

Schwartz has had a massive education in scientific research since starting the project a decade ago. She knew fundraising was key.

"I just thought, 'OK, in about 10 years I'll be ready and I'll go find a researcher and that researcher will go find a cure,'" she recalled. A scientist later told her: "That's not how science works."

The first money spent was on a ski retreat in Park City for 25 scientists who came together to set research goals for the disorder. Until then, no research was being done at all. Since that retreat, a full scale clinical trial has taken place and another is in the works.

"Right now we're still in the hope stage," she said.

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Online

To learn more or to contribute, visit **pachyonychia.org**. Donations are matched two to one.

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Janice Schwartz approaches her daughter Rebecca, 9, on the living room floor in their home. Schwartz sometimes gets around on her knees due to blisters and calluses on her feet.