

## Sharing The Pain

*Rare Disease Puts an AU Economist in Touch With Fellow Patients Around the World*

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It is possible, Arturo Porzecanski will tell you, to feel very lonely even when there are people all around you.

Retired from Wall Street, the renowned economist teaches scores of American University students about international economics and finance every semester. Just a few blocks from the university, he lives with a supportive wife; their two grown children visit regularly.

But in 2005, at age 56, Porzecanski was told he had an extraordinarily rare disease called systemic capillary leak syndrome (SCLS), or Clarkson's disease. Characterized by sudden attacks that cause a rapid drop in blood pressure, the disease can lead to multiple organ failure -- and death -- within hours.

Porzecanski's first attack left him disabled, and doctors told him it was probably only a matter of time before another attack killed him.

No one -- not his concerned doctors, not his loving family -- quite understood what Porzecanski was going through. So he waived his rights to privacy and asked doctors to help him locate fellow sufferers.

They did. He made contact with four other Americans with SCLS in Maryland, Ohio, New York and North Carolina.

Three have since died.

"I was very depressed and felt increasingly alone," Porzecanski recalls.

Then the economist discovered RareShare, a Web site that changed his life.

Founded last summer by an MBA student who once worked for a biotech company, RareShare (<http://www.rareshare.org>) is aimed at people like Porzecanski: men and women who needed to find somebody somewhere in the world who knew what they were going through.

It was while working for Nutra Pharma, which is developing a treatment for another rare disease, that David Isserman noticed how people with rare diseases were constantly looking for ways to connect with one another through the Internet.

In response, he helped build a site that lets patients form communities around specific diseases, create discussion boards and share resources such as links to papers and articles. Since launching a beta version last July, RareShare has grown to have more than 1,000 registered users and hundreds of communities for different rare diseases. About a third have users engaged in active discussions.

RareShare may never grow huge, Isserman said. Yet the needs it meets are becoming clear.

As Stephen Groft, director of rare diseases research at the National Institutes of Health, put it: "Few people have individual rare diseases, but collectively as a group, about 25 million people in the United States have one of the 6,000 or so rare disorders. When you add in the family members, you have a large percentage of the population that is affected."

Every week, Isserman gets five to seven requests to create new communities. In January, the site received more than 34,000 page views and had more than 13,000 unique visitors.

For Porzecanski, the site was a lifeline. "RareShare.org put me in touch with other patients," he said.

Porzecanski quickly told the only other SCLS patient he knew about the site, as well as Phillip Greipp, a hematologist at Mayo Clinic who is one of the few doctors in the world to have researched the disease. They both signed up.

Then others started joining.

They came from all across the country and all around the world: from Idaho, Maryland, Missouri, Canada, India, Ireland, Switzerland and Guatemala. And Porzecanski is now in touch with a dozen people with SCLS.



## One in a Million

With a disease as rare as SCLS, many doctors have never seen a case before and often do not immediately recognize it. There is no known cause for SCLS, and no genetic predisposition for it has been identified. A disease is generally considered rare if there are fewer than 200,000 known cases worldwide. There are only about 100 known cases of SCLS.

"If you're a trained hematologist from a good institution, you'd be aware of the syndrome," said Greipp, who sees patients from around the world. "I'd say one in a million have it, but all I can do is wildly estimate. . . . We've only seen 28 patients here with it in the last 25 years."

SCLS causes massive leakage of plasma from blood vessels into muscle compartments and body cavities. As a result, vital organs are deprived of oxygen-carrying blood circulation. The combination of the leakage and the replacement fluids often administered by doctors results in a dangerous swelling of the limbs.

That's basically what happened to Porzecanski in 2005. His blood pressure began to drop while he was in the shower, and he passed out. His wife rushed him to the emergency room, where doctors gave him too much intravenous fluid. The fluid saved his life but also caused his arms and legs to swell, damaging his muscles.

For some time, he was unable to walk. Today, he has no movement or sensation below his knees, and he wears braces on both legs. His forearms also are devoid of muscle, and he has little feeling in his hands and fingers.

You wouldn't know when you meet him. The professor is witty and disguises his disabilities well. But there are signs.

Porzecanski often leans slightly against a wall or a desk, giving the illusion that he is standing in place -- something he cannot do without support.

In his desk drawer there is a small hook that he uses to fasten any shirt buttons that might come undone.

In his briefcase he carries a handout that explains the emergency care that he needs, in case he should suddenly suffer another attack.

And he still wakes up every day knowing that he could have an attack. And that the attack could kill him.

That was a feeling his family and friends could never fully understand. But through RareShare, Porzecanski has found confidants, men and women for whom he has become the "community expert," answering questions, sometimes in his native Spanish, and sharing all the information he had.

"It gave me great pleasure to put down everything I knew: all the critical articles, the world's experts, information about custom shoes," he said.

"For me there was just a lot of support in knowing I was not alone."

## **Fellow Sufferers**

Among those who signed up were Wendy Peterson and her husband, Nolan, an SCLS patient in rural Idaho.

The couple was frustrated after Nolan was found to have the disease. In their community of 10,000, Wendy said, she felt isolated.

She was involving her son, a computer engineer, in building a Web site for people with SCLS when he found RareShare.

"It helped to know what other people are going through," Wendy Peterson said.

After signing up with RareShare, Wendy posted a thread titled "Help" on the discussion board.

"Has any one found any thing that will stop me from dropping my blood pressure and passing out? . . . How are you coping with this disease?" she wrote on her husband's behalf. (He was having attacks every five to six days and was unable to write himself.)

Porzecanski posted a response soon after.

"Have you/your doctors considered using long-term, monthly IVIg treatment, which has been tried with partial success in Europe? Have them check out the just-published articles posted under Disorder Resources . . . and discuss it with Dr. Greipp. It's risky, expensive and experimental, though. It would be good to know more details about what exactly you've tried (dosage, frequency, etc.) in such a short time."

Another patient, Judith Davis, wrote to the Petersons from Baltimore.

"It is very good to have connected with you and Nolan. I learn so much by speaking to others with SCLS. . . . There are so few of us left alive that every bit of info gathered is significant."

## **New Mission in Life**

Rejuvenated by the support he has found through RareShare, Porzecanski is making it his life's mission to serve as a patient advocate for SCLS.

"I was already doing it," he said. "But without RareShare, I was alone."

On one wall in his office at American University is a framed Wall Street Journal article hailing Porzecanski as one of the pioneers in the study of emerging markets. On another wall is a lifetime achievement award from the Legacy Laureate Society at the University of Pittsburgh, where he did his PhD.

At American University, he is a "distinguished economist-in-residence."

He is a decorated man.

And yet his proudest moments are when he makes progress, inch by inch, as an advocate for SCLS.

Recently he testified before experts at the National Institutes of Health, explaining his condition and the need for more research on SCLS.

He calls it his "big day."

"I wanted so badly to live long enough until the day NIH scientists had been exposed to SCLS and had begun to study it, and now I've achieved it," he said.

The National Institute of Allergy and Infectious Diseases has set aside a small budget to study SCLS.

Studying the disease may unlock clues to other, more common, diseases that are also characterized by leaky blood vessels, such as diabetes, bloodstream infections and the Ebola virus infections, said Kirk Druey, the NIH doctor who will lead the SCLS study.

What's more, the capillary leak is self-reversing. If a patient survives an attack, he returns to normal until the next attack. "Understanding this will help us understand the pathological leakiness in these other conditions," Druey said.

For Porzecanski, it feels like a burden has been lifted.

"I can be happy knowing I've passed the baton to the scientific community," he said.

Porzecanski's own story is a remarkable one. But he will tell you that the real story is about how a few people coming together can make a difference. And that while a disease may affect only a few, it is hardly insignificant.

"It's not about me," he said. "It's about Patient Power."

Interactive

## An International Connection

Soon after American University economist Arturo Porzecanski was diagnosed with systemic capillary leak syndrome, he yearned to find other SCLS sufferers who understood what he was going through. Thanks to [RareShare.org](http://RareShare.org), a social networking site for people with rare diseases, Porzecanski has been able to form relationships with a dozen other SCLS patients around the world.

Roll over icons on the map to find out more information on Porzecanski's fellow patients.



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