Film explores struggles with rare diseases

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Dr. Maren Grainger-Monsen (left) and Nicole Newnham collaborated on the film, initially intended for doctors and medical students. Photo: Erik Verduzco, Special To The Chronicle / SF

There may be only a few thousand people in the United States who suffer from Hermansky-Pudlak syndrome, a disease so rare that most doctors have never even heard of it, much less know how to treat it.

But this very unusual condition is the focus of a documentary airing on public television this weekend, made by two Stanford filmmakers who are trying to draw attention to the plight of patients who suffer from rare diseases.

Here's the irony: there are a lot of those patients. While the number of people who suffer any specific disease is small, more than 30 million Americans are thought to have a rare disease. And though their symptoms and prognoses may be vastly different, they all share the isolation and lack of medical support endemic to suffering a disease few people have and no one knows anything about.

"Rare diseases, when you take them all together, are really common," said Dr. Maren Grainger-Monsen, a Stanford bioethicist who produced the film, called "Rare," with Nicole Newnham. The movie was released earlier this year and will be widely shown in the Bay Area for the first time Sunday night on KQED.

Finding hope
"When we've shown this movie, we've heard from a lot of people who said they had a rare disease, or someone in their family does or they know someone with a rare disease," Grainger-Monsen said. "I think people have taken a lot of
hope from that, from knowing there are others like them."

Many patients with rare conditions lack adequate, disease-specific treatments - assuming they've even been diagnosed at all. They will often go through years of uncertainty and multiple misdiagnoses before finally learning the name for their disease.

Once diagnosed, patients will sometimes seek out others like them, but finding anyone can be next to impossible when there may be no one in the same county or even state who shares the condition. The Internet has helped patients form communities.

A major goal of those patient communities, aside from simply providing support to their members, is to draw attention to their disease. That can be a tough task for patients with any medical condition, but it's far more difficult for those with rare diseases, who have to compete against the advocacy machines for conditions like heart disease and breast cancer. As a result, many sufferers of rare diseases, along with their friends and family, have united to form groups that lobby on behalf of all patients with unusual diagnoses. At the same time, disease-specific advocacy groups also have sprung up.

The Hermansky-Pudlak Syndrome Network was created by the parent of a patient in 1992, to find other people with the condition and to work with doctors and scientists to seek treatments. Hermansky-Pudlak is a genetic condition that is marked by albinism, bleeding disorders and, in some cases, lung and bowel diseases.

**The mother's cause**

The Stanford film follows parent Donna Appell as she tries to find HPS patients to participate in a clinical trial to test a new drug for the disease. Over the course of the 50-minute film, Appell and a scientist at the National Institutes of Health form a team of sorts - Appell trying to supply the patients so the scientist can conduct his research.

That's yet another problem associated with rare diseases - research can be extraordinarily difficult because so few people have the condition. When Appell's daughter Ashley was diagnosed with HPS, there were only a couple of dozen known cases in the United States. There are now several hundred in the network.

"Getting people diagnosed is so important. You don't find a cure for a disease that you've never heard about," said Heather Kirkwood, director of outreach for the HPS Network who has the condition herself. "I feel like I'm really lucky to work with the HPS Network. I know we're making things better."

Without spoiling the film, the results of the clinical trial become apparent by the end of the movie. Kirkwood, who is in the film, said she hopes the movie will help people understand how such research can both cause stress and offer hope for patients, especially those with rare diseases who may not have many options for new therapies.

**The ties that bind**

Newnham, the film's co-producer, said it's not just the patients who experience the emotional highs and lows of a clinical trial, and she was surprised by how close the doctors were to their work and the people they were treating.

"The most compelling part of the film for me is the beautiful collaboration between researchers and patients," Newnham said. "I went into the experience feeling like it must be necessary for scientists to distance themselves from patients. But they're colleagues. There's this huge respect between them."

Newnham and Grainger-Monsen initially had in mind a smaller audience for their film, namely medical school students and doctors taking courses in medical ethics. Now, they're eager to share it with the broader public, both as a tool to educate those who aren't familiar with rare diseases, and a means of providing information and encouragement to those who do.

"I hope people see the strength of the community and how these patients and doctors inspired each other," Grainger-Monsen said. "That's an incredibly powerful force."

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The documentary "Rare" airs at 6 p.m. Sunday on KQED. For more information about the film, and to see a preview, go to [www.rarefilm.org](http://www.rarefilm.org).

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