





PARADE

COULD YOU HAVE A RARE DISEASE? by Dr. Ranit Mishori

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The names themselves sound otherworldly: Cat eye syndrome. Stiff person syndrome. Jumping Frenchmen of Maine condition. Blue diaper syndrome. Prune belly syndrome. There's a condition known as SCID. And another one called SCAD. And a roll call of illnesses that sound like names of distant planets: Porphyria. Alkaptonuria. Monilethrix. And Pinta.

Otherworldly but not funny—not if you or somebody you know is suffering from one of these or any of the more than 6,000 other diseases in the database of rare illnesses kept by the National Institutes of Health (NIH).

There are so many ways human beings can get sick, but the majority of diseases affecting Americans can be classified under relatively few and quite familiar categories: heart disease, cancer, diabetes and so on—diseases that are targeted for billions of dollars in research and that already have produced an arsenal of medications by a scientific and pharmaceutical community that is forever trying to find the perfect “cure.”

Then there are the rare diseases: If fewer than 200,000 Americans suffer from a disease, it is considered rare. More than 25 million Americans have a disease that fits this definition, says Dr. Stephen Groft, director of the NIH's Office of Rare Diseases. So, in a way, “rare” is not that rare after all.

Some of these conditions actually are quite familiar, including Lou Gehrig's disease (or amyotrophic lateral sclerosis), cystic fibrosis and hemophilia. But others are so rare that they may afflict only a few hundred people and are unheard of by most Americans. For the afflicted, unfortunately, there are often few experts to turn to and drug trials to sign up for. More tellingly, there may not be many people who understand what they are going through, though the Internet is beginning to make it easier to find fellow sufferers and hard-to-discover resources.

Consider Desiree Lyon Howe of Houston. She was only 17 when her severe belly pain started. “It felt like a thousand flaming swords embedded in my abdomen,” she recalls. Over the next 10 years, the pain recurred with regularity.

Her doctors were unable to find any explanation for her condition. “Nobody believed me, not even my family,” she recalls. She was labeled an “overanxious, melodramatic, young hypochondriac.”

It took several more years of pain, suffering and depression, as well as repeat hospitalizations, before one doctor and one test changed everything. The diagnosis: acute intermittent porphyria. Getting a name for her ailment was a breakthrough in itself. Says Howe: “It took away my fear, even though I knew that I could have another attack.”

What Doctors Look For

Most doctors are trained to put rare diseases last on the list of possibilities when figuring out what is ailing a patient. There's a saying in medical school: “When you hear hoofbeats, think horses, not zebras.” In other words, consider the obvious first, because most illnesses are going to be the commonplace, ordinary ones.

“It is extraordinarily difficult to navigate the process of standard care for the 10 most common conditions and still try to keep in mind rare conditions,” says Dr. Daniel Wattendorf, a family physician and clinical geneticist.

That may explain why nearly one in seven patients with a rare disorder goes undiagnosed for more than six years. On average, an accurate diagnosis takes nearly three years. Most crucially, it is lost time—time in which treatments could have been started. But that assumes that treatments exist. “Many times there aren't a lot of investigators who are interested” in the study of rare disorders, says the NIH's Dr. Groft.

Abbey Meyers, president of the National Organization for Rare Disorders, is the mother of three children with a neurological disorder called Tourette's syndrome, which is characterized by involuntary sounds and movements. Years ago, Meyers and others helped push legislation through Congress that would specifically promote the development of treatments for rare diseases by giving incentives to pharmaceutical companies. The result: 290 new drugs for rare diseases have been developed—helping more than 14 million Americans, says Dr. Marlene Haffner of the Food and Drug Administration. An additional 1,400 drugs continue to be studied.

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