

COPING WITH A RARE DISEASE

By Claire Sykes

At first, doctors told her nothing was wrong and that she was overreacting. Fortunately, Dayna Fladhammer found a pediatrician who finally gave her an answer. Her six-month-old daughter had something called primary immune deficiency (PID). Soon, the then-pregnant mother would learn that all of three of her children have this rare disease.

Fladhammer never felt more alone. “Having a rare disease is very isolating,” says the former stay-at-home mom turned patient advocate in Santa Clarita, California. “It’s hard to find accurate, up-to-date information on it, unless there’s a strong patient advocacy organization for the disease, or a doctor who knows enough about it.

“Because it’s rare, the people around you either don’t have any point of reference for the disease, or it’s one that’s a worst-case scenario. So it’s hard for them to understand what you might be going through. And then there’s grieving the loss of a life you thought you would live.”

Fladhammer joins about 25 million Americans who face nearly 7,000 rare diseases. A rare disease, as defined by the National Organization for Rare Disorders (NORD), affects less than 200,000 U.S. residents. It’s also called an “orphan” disease because it’s not “adopted” by the pharmaceutical industry. The small numbers affected

by rare diseases aren't enough for companies to make and market medicines to treat or prevent them.

The first challenge, though, is to get a diagnosis. "Getting an accurate one is a big challenge," says Mary Dunkle, NORD spokesperson. "About a third of the people with rare diseases go five years or longer without a diagnosis." Lack of research on many of the diseases means that even doctors know little about them.

"Meanwhile, you spend money traveling long distances to see different doctors, and live an emotional roller coaster until you can put a name to your disease," Dunkle adds. Once you have one, there may be no treatment. Fewer than ten percent of rare diseases have a remedy. If there *is* a drug, it's likely to be very expensive, and your insurance may not cover it.

"It's so easy to get overwhelmed," says Fladhammer. She and Dunkle offer these six steps to cop with a rare disease:

Connect with others. "Find people who understand, and can lead you to information," says Fladhammer. Search the Internet for the 1,300-plus patient organizations for various rare diseases, and join online chat rooms and support groups.

Talk about it. Express your anger, fear, sadness and other feelings. Share with others who can empathize, and you won't feel so alone. You'll also trade useful information, as you swap your research and doctor-visit stories.

Educate yourself. Ask people and search the Internet for trustworthy information about your disease. Dunkle suggests the National Institutes for Health (NIH) or a reputable patient organization with medical advisors. Learn as much as you can.

Find a good doctor. You want one who listens, takes your concerns seriously, and stays open to appropriate diagnostic tests and options. Choose someone you trust and can communicate with well.

Feed your optimism. It's easy to fall prey to frustration. "But the better you keep up your own positive attitude, the better you'll do, in general," says Fladhammer. "It's a conscious decision you make."

Give back. Contribute money to medical research for your rare disease, speak to groups about your illness, or volunteer in your community. "Offering your support is one of the most positive things that comes out of having a rare disease," she adds.

You're more likely to have positive results when you deal with your illness constructively. Meanwhile, don't forget about the rest of your life. As Fladhammer puts it, "Don't let your disease define you. Your disease is what you have, not who you are."

© Claire Sykes. All rights reserved.

FIVE WAYS TO HELP OTHERS

If a friend or loved one has a rare disease, patient advocates Dayna Fladhammer and Mary Dunkle suggest you:

Support. Serve as the person's health care advocate. He or she may be too sick to do so.

Listen. Just let the person talk. Keep an open ear without passing judgment or trying to fix anything.

Learn. Find out as much as you can about the disease and the person's experience with it. That way, you'll know best how to lend a hand.

Help. Offer something specific. Say you'll babysit next weekend, shop for groceries or clean the house.

Contribute. Rare disease research lacks in funding. Your monetary gift could help find a treatment or cure.

To learn more:

* National Organization for Rare Disorders

www.rarediseases.org

(203) 744-0100.)

* National Institutes of Health Office of Rare Diseases

<http://rarediseases.info.nih.gov>

© Claire Sykes. All rights reserved.

BIBLIOGRAPHY

Dunkle, Mary, spokesperson, National Organization for Rare Diseases, Washington, DC.
Interview.

Fladhammer, Dayna, mother of children with rare disease, Santa Clarita, CA. Interview.