



Not So Rare

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According to the National Institutes of Health Office of Rare Diseases, rare diseases affect nearly 25 million people in the United States. Having a rare disease is not so rare! And many of these 6,000-7,000 different diseases have serious or life-threatening consequences.

In this issue of *Neurology Now*, we review some of the progress that has been made in the diagnosis and treatment of several rare neurologic diseases. “Too Rare for Research” (page 29) features people who have advocated for more research and better treatment for these diseases.

By definition, a rare (or “orphan”) disease in the United States affects fewer than 200,000 Americans. Given the high cost of drug development, researchers and drug companies often focus their efforts on treatments that will improve health for a large number of people, which means rare diseases are often ignored. Fortunately, the Orphan Drug Act creates incentives for the development of treatments for rare diseases (and for non-orphan diseases in cases where companies don’t expect to recover the costs of developing and marketing the treatment). Some diseases that are considered rare in the United States are common in other parts of the world, so new treatments developed here can have far-reaching effects.

There are hundreds of rare diseases that can attack the nervous system—in people of all ages. Some rare diseases—such as amyotrophic lateral sclerosis (ALS, also known as Lou Gehrig’s disease), Duchenne muscular dystrophy, and Huntington’s disease—are reported on frequently by the media. Many have celebrity spokespeople who help raise awareness.

Other rare diseases are familiar only to the people who have them as well as their family and friends. Some of these include narcolepsy, which is characterized by sleep attacks during the day, vivid dreams, episodes of collapse due to loss of muscle tone, and temporary paralysis upon waking; progressive supranuclear palsy, a rare movement disorder that causes symptoms similar to Parkinson’s disease; glioma, a very aggressive and malignant form of brain

cancer; and Creutzfeldt-Jakob disease (CJD), a rapidly progressive dementia with jerking movements caused by the buildup of very toxic proteins in the brain called prions.

Thankfully, more treatments for rare diseases are now being studied. One reason for this is the huge investment in research and development stimulated by the Orphan Drug Act. Since 1983, when the act was passed, more than 350 medications have been approved for the treatment of rare diseases. A recent publication from the Pharmaceutical

Research and Manufacturers of America reports that the number of drugs in the pipeline for rare diseases increased from 303 in 2007 to 460 in 2011.

In addition, scientists have made significant advances in understanding what causes many human diseases. Thanks to initiatives such as the Human Genome Project, we have identified the genetic origins of many health problems. We have new tools to determine what has gone wrong, which is the first step in developing strategies to fix a problem.

This is all good news. Still, people with rare diseases often feel extremely isolated. Even getting an accurate diagnosis can be difficult. No neurologist could possibly have experience diagnosing and treating all the rare neurologic diseases. As a result, patients and families may need to search hard for an expert—in some

cases by scouring the Internet, as was the case for the Soeby family in our story.

Do you have a rare neurologic disease or know someone who does? Please share your experience of getting a diagnosis, any involvement in clinical research, and advice for coping with a rare neurologic disease by emailing us at neurologynow@lwwny.com.

Take good care,

Robin L. Brey, M.D.
Editor-in-Chief



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