Drugs

Orphan Products: Hope for People With Rare Diseases

Jumping Frenchmen of Maine sounds like an uproarious, modern-day stage show or even a new wave rock group. But it's neither. It's the name of an unusual disorder that causes an extreme startle reaction to unexpected noises or sights. Though little is known about Jumping Frenchmen of Maine, the disorder and more than 6,000 other rare or "orphan" diseases are receiving increasing attention from the government, patient groups, and the pharmaceutical industry.

FDA Rare Disease Patient Advocacy Day, 2012[1]

An orphan disease is defined as a condition that affects fewer than 200,000 people nationwide. This includes diseases as familiar as cystic fibrosis, Lou Gehrig's disease, and Tourette's syndrome, and as unfamiliar as Hamburger disease, Job syndrome, and acromegaly, or "gigantism." Some diseases have patient populations of fewer than a hundred. Collectively, however, they affect as many as 25 million Americans, according to the National Institutes of Health (NIH), and that makes the diseases--and finding treatments for them--a serious public health concern.

Most Inherit Orphan Diseases

New rare diseases are discovered every year. Most are inherited and caused by alterations or defects in genes (mutations). Others can be acquired as a result of environmental and toxic conditions. Genes are pieces of DNA, part of the code that determines the traits and individual characteristics of all living things. Each human cell contains around 30,000 genes. Besides influencing features such as eye and hair color, genes also can play a role in the development of diseases and in their transmission from parent to child.

As disparate as rare diseases are, patients share many common frustrations. For example, for one-third of people with a rare disease, getting an accurate diagnosis can take one to five years. And people often are so isolated that they may never know anyone else with the same disease. Patients often must travel long distances to visit the few doctors knowledgeable about their illnesses, and the costs involved with diagnosis, treatment, and other related expenses can be exorbitant.

Managing Rare Diseases

Many rare diseases or conditions can be difficult to diagnose and manage because in their early stages, symptoms may be absent or masked, misunderstood, or confused with other diseases.

For example, adrenomyeloneuropathy (AMN), one of a group of genetically determined progressive disorders known as leukodystrophies that affect the brain, spinal cord, and peripheral nerves, is often misdiagnosed as multiple sclerosis (MS), according to the United Leukodystrophy Foundation. Since diagnosis of neurological conditions relies on subtle and circumstantial evidence, even the most experienced clinicians may have difficulty distinguishing between the two. For rare disease patients, there may be no cures, but treatments of the symptoms can help. Participating in a clinical trial may be a way to receive the most advanced care for some diseases. People who experience unexplained symptoms, recurrent infections, and pain that have gone undiagnosed for a long period of time might want to visit a referral center that is experienced in diagnosing patients with rare diseases. Some rare diseases do not have clearly defined treatment guidelines and require the specific skills of an expert physician. Be sure to go to a hospital that is familiar with treating people with multiple problems.

Adopting the 'Orphans'

Before the passage of rare disease laws in the United States, patients diagnosed with a rare disease were denied access to effective medicines because prescription drug manufacturers rarely could make a profit from marketing drugs to such small groups. Consequently, the prescription drug industry did not adequately fund research for orphan product development. Other potential sources, such as research hospitals and universities, also lacked the capital and business expertise to develop treatments for small patient groups. Despite the urgent health need for these medicines, they came to be known as orphans because companies were not interested in adopting them.

This changed in 1983 when Congress passed the Orphan Drug Act (ODA). The ODA created financial incentives for drug and biologics manufacturers, including tax credits for costs of clinical research, government grant funding, assistance for clinical research, and a seven-year period of exclusive marketing given to the first sponsor of an orphan-designated product who obtains market approval from the Food and Drug Administration for the same indication. At the same time, federal programs at the FDA and the NIH began encouraging product development, as well as clinical research for products targeting rare diseases.

Since 1983, the ODA has resulted in the development of more than 250 orphan drugs, which now are available to treat a potential patient population of more than 13 million Americans. In contrast, the decade before 1983 saw fewer than 10 such products developed without government assistance. As a result of the ODA, treatments are available to people with rare diseases who once had no hope for survival.

"A lot of people are affected," says Marlene E. Haffner, M.D., M.P.H., director of the FDA's Office of Orphan Products Development (OOPD). "That makes it a major public health impact, and in time, we're going to see even more rare diseases requiring treatment."

Despite the success of the ODA, however, rare disease advocacy groups argue that the plight of people with orphan diseases deserves even more attention.

Patient Support Groups

Rare diseases affect so few people that information about them may be difficult to find, making the situation more traumatic and stressful. Before Congress enacted the ODA, families coping with a rare disease usually struggled alone. Support could only be found through telephone calls to other families suffering with similar diseases, and only if the names were provided by doctors.

Support groups such as the National Organization for Rare Disorders (NORD) have worked aggressively in the last 20 years to draw attention to people with rare diseases, and especially to the lack of treatment options. Paramount in NORD's ongoing cause are efforts to promote legislation, such as the ODA, that encourages further research and continuing development of products that are necessary--and often life-saving--and to provide easier access to such treatments.

The role of the support group is evolving. Recent trends at the FDA and the NIH in encouraging scientists to become involved with patient support groups has brought research even further.

New Web-based support groups continue to proliferate. Not only are people receiving comfort from others with the same conditions, but they are learning from each other's experiences as well. By the late 1990s, most nonprofit organizations had Web sites where people could ask questions and get immediate responses.

Nevertheless, people diagnosed with a rare disease often are vulnerable to misguided assistance. While Stephen C. Groft, Pharm.D., director of the NIH's Office of Rare Diseases, encourages people to use the Internet to find information, he also warns that it is dangerous to rely solely on the computer for medical advice.

"Be cautious about unproven remedies and miracle cures being touted over the Internet," he says. Too often misleading or inaccurate information is given out that can do more harm than good. In addition, one person's experience may vary greatly from another's. The Internet, he adds, should

complement the communication between patients and their doctors, not replace it.

For More Information

Food and Drug Administration
Office of Orphan Products Development
(301) 796-8660
http://www.fda.gov/ForIndustry/DevelopingProductsforRareDiseasesConditions/default.htm[2]

National Institutes of Health
Office of Rare Diseases, 6100 Executive Blvd., Room 3B01, Bethesda, MD 20892-7518
(301) 402-4336
http://rarediseases.info.nih.gov/[3] National Cancer Institute

Cancer Information Service
(800) 4-CANCER (422-6237)
www.cancer.gov[4]

National Organization for Rare Disorders
55 Kenosia Ave., PO Box 1968, Danbury, CT 06813-1968
(800) 999-6673
www.rarediseases.org[5]

The Genetic and Rare Diseases Information Center
PO Box 8126, Gaithersburg, MD 20898-8126
(888) 205-2311TTY: (888) 205-3223
www.genome.gov/health/[6]

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