

People With Rare Diseases Face a Host of Unique Challenges

By Dennis Jackman, Senior Vice President, Public Affairs, [CSL Behring](#)



Dennis Jackman

People with rare and serious medical disorders encounter a host of unique challenges—from obtaining a correct diagnosis, to identifying an appropriate course of treatment and gaining affordable access to the care they need. They and their families, caretakers and health care providers face a constant struggle to identify and access available resources, away from the attention of traditional media that focuses on more common disorders such as cancer and heart disease. BIO has recognized the importance of rare disease awareness, and recently established a rare disease working group to assure focus on the critical issues facing therapies and access to them.

Barriers to diagnosis

Going undiagnosed for years or being misdiagnosed can be a serious and often life threatening problem for people with rare diseases. Why is it so difficult to detect rare disorders or diagnose them correctly? There are several factors, physician training being one. It begins in medical school where students are typically trained to look for the obvious when diagnosing a patient, in other words, if you hear hoof beats, it's most likely a horse, not a zebra.

Medical schools and physicians might reorient their training to further consider the spectrum of diagnostic possibilities, because rare disease patients do not fit the typical profile for most medical conditions; they are the “zebras.” This is the motto of the Immune Deficiency Foundation, which is the advocacy group for patients with Primary Immune Deficiencies, a body of individually rare conditions. In addition, physicians who initially treat patients with rare diseases are usually general practitioners, pediatricians and internists with less specialized training in areas such as immunology and hematology.

Availability of therapies

Even when a correct diagnosis is made, there may not be an available therapy to treat the rare condition. There are nearly 7,000 rare medical conditions, but less than 300 have therapies available to treat them. All of these factors add to the isolation of being part of a small patient group whose health care needs are often unmet.

Regulatory harmonization

Achieving greater harmonization between regulatory agencies in the US (FDA) and Europe (EMA) is essential to lowering the cost of therapies and bringing them to market more quickly. This is due to the enormous cost of conducting clinical trials, which often must be replicated because the FDA may not

accept the results of a trial conducted in Europe, or the EMA a trial in the US. The average cost of a clinical trial is \$100 million+.

Improving access to care

There are a number of ways in which patient advocacy groups, physicians and health care providers, pharmaceutical companies, government and insurers can work together to lower the cost of healthcare and improve access to care for people with rare medical conditions. The following are some examples:

- Enhance regulatory pathways for orphan drug development and licensing, including appropriate flexibility, earlier communications, expansion of accelerated review, and enhanced regulatory science. Many such provisions are in PDUFA-V and the TREAT act legislation.
- Eliminate specialty tiers that healthcare insurers or payers use to create restrictions that increase the cost to patients with rare disorders for certain prescription drugs, and prevent them from seeing specialists.
- Support the orphan drug tax credit in the US to encourage R&D for innovative therapies to treat rare diseases.
- Allow orphan designation for all orphan products, not just clinically superior ones. Each orphan product available contributes to care and should not be discouraged.
- Implement tort reform in the US with limits on liability.
- Work with regulatory agencies for a realistic pathway to clinical trials that work for rare diseases.
- Base treatment protocols on what is best for the patient, not on outdated or misinterpreted insurance coverage criteria.
- Eliminate “non tariff barriers” and other discriminatory practices that are an impediment to trading products for treating rare diseases globally.

Future outlook

Individuals with rare diseases can look forward to new and innovative products for treating rare disorders that are planned or that are in development. Much new research is being performed in partnership with academic institutions, and there will clearly be continued collaboration among patient groups, manufacturers, physicians and government agencies. The primary strategy for improving patient access to diagnosis and care remains focused on advocacy, improved knowledge and appropriate reimbursement.

At [CSL Behring](#), we are doing our part to increase the availability of biotherapies for treating rare medical conditions through our substantial and ongoing investment in R&D and manufacturing. That investment is reflected in an expanding portfolio of therapies for unmet patient needs, and cutting-edge manufacturing facilities combining state-of-the-art, industrial-scale filtration technology, highly efficient separation processes and advanced formulation to generate plasma therapies. In fact, we have received 15 regulatory approvals for new rare disease products or indications in the last eight years, and there are more in our pipeline.

We also conduct a dynamic advocacy program with stakeholder partners, that is helping to enhance access to care and protect patients' rights. We understand the many difficulties that people with rare medical disorders must live with every day. And we believe that isolation and lack of access to appropriate care should not be among them.

For more information and links to useful resources, please visit <http://www.cslbehring.com/knowledge-center>.