Taylor & Francis
Taylor & Francis Group

Expert Opinion on Orphan Drugs

ISSN: (Print) 2167-8707 (Online) Journal homepage: informahealthcare.com/journals/ieod20

A hidden priority: the paradox of rarity (NORD perspective)

Peter L Saltonstall

To cite this article: Peter L Saltonstall (2014) A hidden priority: the paradox of rarity (NORD perspective), Expert Opinion on Orphan Drugs, 2:11, 1127-1128, DOI: 10.1517/21678707.2014.971012

To link to this article: https://doi.org/10.1517/21678707.2014.971012

EXPERT OPINION

A hidden priority: the paradox of rarity (NORD perspective)

Peter L Saltonstall
NORD, Boston, MA, USA

Expert Opinion on Orphan Drugs (2014) 2(11):1127-1128

I appreciate the opportunity to introduce this special issue of Expert Opinion, which is dedicated to rare diseases and orphan products. I am especially interested in the theme which notes the paradox of the concept of 'rarity.'

We have been using the word 'rare' for many decades to refer to diseases that, under a law passed by the US Congress in 1983, affect 200,000 or fewer Americans. The definition is different in Europe, where a 'rare' disease is a disease that affects fewer than 1 in 2000 people.

I know, through my professional work and personal life, so many people with a 'rare' disease, that I no longer think of 'rare' diseases as 'rare'. Indeed, there are about 7000 'rare' diseases, affecting about 30 million Americans. And as we learn more about the sub-categories of diseases, there will be many more diseases that technically will be defined as 'rare'.

So there is irony in using the word 'rare' to refer to so many diseases that affect such a large percentage of people.

My goal as head of the National Organization for Rare Disorders (NORD) is to help make sure that patients with 'rare' diseases have a strong voice in the public policy arena, especially with regard to how new therapies are developed and made available. We also are concerned with making sure that patients in need have access to medical products and services.

I am encouraged by the present environment for the following reasons:

- So many new therapies for 'rare' diseases are being developed. For the past few years, about a third of the new chemical entities approved by the US FDA have been for 'rare' diseases.
- There is strong commitment at the FDA and the National Institutes of Health (NIH) and other US government agencies, as well as corresponding agencies in Europe, to patients with 'rare' diseases.
- New discoveries being made virtually every day will lead to better treatments for patients with 'rare' diseases. Advances in genetics are truly remarkable and holds out great promise for the future. The genomic revolution promises to identify innumerable opportunities for new therapies.
- Investors have recognized that there is a business model for a company that makes drugs intended for patients with rare diseases. Many major drug companies have created divisions devoted to orphan drugs.

At the same time, there is a long way to go. Despite the progress to date, only a few hundred 'rare' diseases currently have FDA-approved treatments.

The patient advocacy community still faces many challenges:

It is ironic that we are on the cusp of so many scientific advances at a time when research dollars are becoming scarcer in both the US and Europe. More research investment would result in an acceleration in new therapies.

The timely diagnosis of 'rare' diseases remains an issue. Unless a physician specializes in diagnosing and treating rare diseases – and comparatively few do – most practicing physicians in the US and Europe have little experience in diagnosing one. I hear story after story from parents about how challenging it was

informa
healthcare

10.1517/21678707.2014.971012 e-ISSN 2167-8707

for them to secure an accurate and reliable diagnosis for their child. For many diseases, it can take half a dozen years or longer before an accurate diagnosis is made.

Recognition of the need for clinical testing and regulatory flexibility also is a challenge. We are fortunate that officials at the FDA who deal with orphan drugs understand that, sometimes, it is not feasible to test a new treatment for a 'rare' disease in the same way that a treatment for a common disease would be tested. The FDA has demonstrated great flexibility in assessing new treatments for 'rare' diseases. Two recent articles in major US medical journals have criticized the FDA for its policy. We need to explain better why flexibility is needed, and works.

We need to maintain incentives for the development of new orphan medical products. A proposal made public recently by a prominent US congressman would repeal the Orphan Drug Tax Credit, a crucial incentive that has stimulated the development of treatments for Americans with 'rare' diseases. My organization fought back aggressively against the idea that such a successful incentive would be repealed.

Finally, the healthcare delivery and reimbursement systems in the US are undergoing dramatic changes. More patients are entering the system, and better and more expensive treatments are being developed. We are challenged to assure that new treatments reach patients quickly after approval and are reimbursed appropriately. At NORD, we are working with all insurers to assure that treatments become available in a timely way.

I am encouraged that many of our efforts are being coordinated on an international scale. 'Rare' diseases do not recognize geographic boundaries. We have collaborated with EURORDIS, our European counterpart, as well as with organizations in other parts of the world, on many efforts to bring the patient community together. We work together, for example, to sponsor Rare Disease Day, the last day in February, to focus international attention on rare diseases.

Affiliation
Peter L Saltonstall
NORD, Boston, MA, USA
E-mail: psaltonstall@rarediseases.org

As I look to the future, I see still more progress – more personalized drugs, for example, which are created for an individual patient. As personalized medicine becomes a reality, patient involvement in the development process becomes ever more important. The word that I use is 'patient-centricity'. We have always been advocates for patients and are committed to making the patient the center of the healthcare system.

There are many reasons why everyone should be attuned to what is happening in the 'rare' disease community, but one particular reason is especially compelling: 'rare' disease research and orphan-drug development very often have provided the knowledge that leads to the development of drugs for more common diseases.

This edition of Expert Opinion will help us understand better the paradoxical nature of using the word 'rare' to define 'rare' diseases and also help us all focus on the advances made and the challenges remaining. I am optimistic that scientific advances will enable us to learn more about 'rare' diseases and how to treat them, and our public policies will create a friendly environment. Perhaps someday, as we have done with some very common diseases such as smallpox and polio, we will be able to witness the ability to treat successfully many the 'rare' diseases. This is the vision that we at NORD are working towards.

Declaration of interest

The author is President and CEO of the National Organization for Rare Disorders (NORD), which represents organizations of patients with rare diseases. The author has no relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in the manuscript. This includes employment, consultancies, honoraria, stock ownership or options, expert testimony, grants or patents received or pending, or royalties.