Rare But Not Scarce: The Struggle Faced by Millions

A single rare disease may afflict just a small number of people, but collectively up to 400 million people globally live with a rare disease.

Exactly what is a rare disease? Collectively, rare isn't scarce or infrequent. That's the message that the organizers of Rare Disease Day 2020 are striving to promote. Worldwide, there are between 300 million and 400 million patients suffering from the 6,000 to 7,000 different rare diseases. Yet millions of people with rare diseases struggle to access diagnosis, treatment, and care — indeed, 95% of rare diseases do not have a single FDA approved drug treatment. In the United States alone that means more than 30 million people living with rare diseases have few to no options.

However, there's much cause for optimism. A 2019 study published by Tufts University found that rare disease drug development is one of the fastest-growing areas in R&D, accounting for as much as one-third of products in the R&D pipeline. And in 2018, BIO reported that there were 595 companies developing orphan therapies, and almost every pharmaceutical company has, or is opening, a rare disease division. Additionally, venture capital companies are demonstrating significant interest in start-ups and orphan drugs. Indeed, the growth in the market is such that, according to the EvaluatePharma Orphan Drug Report 2019, sales of orphan drugs are expected to reach $242 billion by 2024 — one-fifth of the global pharmaceutical market.

Since the Orphan Drug Act (ODA) of 1983, more than 600 new therapies have been

Rare Disease Day Around the World

More than 100 countries are participating in Rare Disease Day 2020, Feb. 29. Over the years, a variety of countries have drawn attention to rare diseases through a variety of activities. A small sample of the worldwide activities include:

Australia: A participant since 2009, Australia has held events across the country, including an Indian dance event, a silent auction, fun runs, bake sales, a high tea event, and in 2019 a "tennathon" was held, attempting to break the Guinness World Record for the longest doubles tennis match (more than 60 hours) and raise money for charity.

Brazil: Since first participating in 2010, Brazil has held week-long celebrations in the lead up to Rare Disease Day. Activities have included a television documentary on rare diseases, Zumba classes, glucose tests, and in 2019 a photographic exhibition highlighted the characteristics of rare diseases to promote awareness and prevent misdiagnosis.

China: A participant since 2009, China's national publicity campaign impacted more than 1 million people, summits have gathered greater than 200 specialists on rare diseases, and advocacy activities have taken place across the country.

Europe: Rare Disease Day was created by EURORDIS-Rare Diseases Europe and its Council of National Alliances in 2008. Each year EURORDIS coordinates Rare Disease Day at the international level, as well as with the national alliances, patient organizations, and other groups at the national and local levels.

India: Patient organizations first became involved in Rare Disease Day in 2010. Events since then have included poster campaigns, school visits, competitions, a film festival, counseling sessions, and more. In 2019 India's annual Racefor7 event took place again, where more than 7,000 participants ran a 7km race to represent the more than 7,000 rare diseases documented so far.

Japan: Japanese associations joined Rare Disease Day in 2010 with events, including seminars, conferences, and fun events. In 2019 Rare Disease Day was celebrated in nearly 30 cities, including Tokyo, Osaka, and Kyoto with lectures, workshops, and photo exhibitions.

South Africa: Since first participating in 2010, patient organizations have launched many successful media campaigns, and have reached out to government ministers to stress the need for official policy on rare diseases. An annual "Denim Walk" takes place through the Walter Sisulu Botanical Gardens to raise awareness of those living with rare diseases.

United States: A participant since 2009, the country holds a huge variety of events, from an inclusive online music campaign to make a compilation of songs to unite a global community, to races both in real time and virtual.

Source: Rare Disease Day, https://www.rarediseaseday.org/events/world

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approved by the FDA to treat rare diseases. That compares with just 34 approvals before the act was passed. And in 2018, 34 of the 59 novel drugs approved by the agency carried orphan designations.

Tackling the Challenges

While more and more companies are pursuing therapies for rare diseases, the Tufts study found that the development process — clinical through approval — for rare disease drug development takes on average four years longer than it does for more common conditions. Another major challenge is identifying and enrolling patients for clinical trials.

EXECUTIVE VIEWPOINTS

Susan Abedi
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Best-in-Class AI-Based Analytics

The industry needs to integrate best-in-class AI-based analytics with medical and marketing expertise to accelerate the process of getting life-saving therapies to patients with rare diseases. Data scientists together with medical experts can build models which, through features, predict if a patient does or does not have the particular disease. How well an AI algorithm "interacts" with data depends on the sophistication of the feature-building process, which requires a comprehensive understanding of disease nuances.

Patient Identification

Identifying patients is only part of the battle. One needs solutions that enable yet-to-be-diagnosed patients to be mapped to actual clinician practices where they are currently being managed for other conditions. Doing so will considerably increase the actionability of the results. The ability to find undiagnosed patients along their disease journey, and to understand the HCPs who are currently treating them, has the potential to dramatically change how we identify HCPs who encounter yet-to-be-diagnosed patients and how we engage effectively with these HCPs.

Christopher Tobias, Ph.D.
President
Dudnyk

The Positive Impact of Gene Sequencing

First, making gene sequencing more affordable and accessible should increase the number of patients who can be identified with specific genetic-related diseases, thus improving R&D efforts through the availability of more individuals who could participate in clinical trials. More importantly, it should decrease the time for patients to achieve a correct diagnosis and lead to greater access to new medications on the horizon that treat their disease.

Laurie Bartolomeo
Executive VP, Creative Director
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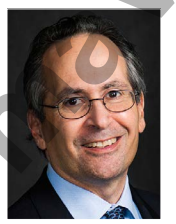
Make a Charitable Connection

Any orphan product manufacturer should establish a charitable foundation that helps supply patients in need with the life-saving therapies they otherwise could not afford. It is also best practice for our clients to advocate and assist in attaining adequate coverage of prescription costs for their patients, as well as other meaningful wrap-around support. Living with a rare disease is challenging enough without having to constantly battle for access to essential treatments and services.



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Access to Therapies

Addressing the following three critical areas is essential to assuring patients facing rare diseases have access to life-saving therapies: treatment value needs to be articulated to payers to assure coverage; patient out-of-pocket expenses need to be reduced through payer coverage, patient assistance programs, and regulatory limits; and clinical trials and labeled indications need to match appropriate treatment criteria or additional support needs to be provided to support expanded coverage.

Wrap-Around Services

Companies need to evaluate services being provided and by whom, and from this gap analysis determine what services are needed. In addition to filling identified gaps, companies should develop a process for coordinating services with any existing services. All too often, wrap-around services either overlap or conflict, which can not only confuse patients but cause suboptimum outcomes. By filling the gaps and working with existing resources, patients and other stakeholders can realize improved outcomes for patients with rare diseases.

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The good news is that there is global commitment to addressing rare diseases. In 2016, the United Nations recognized rare disease research as a global health priority, and included action on rare diseases in its sustainable development goals for 2030.

In the United States, the National Institutes of Health (NIH) set up the Rare Diseases Clinical Research Network in 2003 to tackle challenges faced when carrying out research on rare diseases. The network brings together scientists across many disciplines at clinical sites around the world to work with patients and advocacy groups. Since it began, more than 40,000 patients have been enrolled in network clinical studies, studying more than 280 rare diseases.

The EU launched a program — the European Joint Programme on Rare Diseases — with the objective of ensuring new treatments and diagnostic tools are available to those who most need them.

Perhaps the greatest opportunity for addressing the challenges presented by rare diseases is the advancement — and cost reduction — in genome sequencing. This is key because the majority of patients with a rare disease — 80% — have a faulty gene. While whole genome sequencing has its limits — partly due to a large number of variants an individual's genome has, and also limited understanding of the pathogenic mutations behind many rare diseases — it has been shown to be a valuable tool. A study published in 2019 in Genomic Medicine involving 60 patients with a suspected genetic disease, demonstrated significant value in genome sequencing, with 41 of the 60 cases producing clinically significant genomic findings. By 2030 more than 36 million rare disease patients worldwide will have their genome sequenced, up from 30,000 in 2017, thanks to the huge reduction in cost of the technology required.

There are initiatives worldwide to gather, store, and apply genomic data from at least 100,000 genomes, including Japan, the UK, China, Australia, Saudi Arabia, the United States, Estonia, France, Dubai, and Turkey.

The UK's 100,000 Genomes Project has been recruiting people with a rare genetic disease who don't have a molecular diagnosis. Scientists will look at every letter of DNA in that person's genome with the objective of finding the change that is responsible for causing the disease. Once the data of those taking part has been processed, results are sent to that patient's NHS clinical team. As knowledge about genomics grows, analysis of patient data will continue and information about an individual's health will be shared with their clinicians.

The largest initiative, the All of Us Research Program in the United States, seeks to gather health and wellness data from 1 million or more Americans, including communities previously underrepresented in research.

These initiatives create new scientific avenues of research and drug development, as well as pathways to patient access. In addition, artificial intelligence is helping to speed up improvements in research and diagnosis.

Technology and Rare Diseases

Technology advances are integral to improving understanding about rare diseases. Technology has made it easier for patient groups to build registries that provide insight to clinicians and researchers. Imaging technologies and the discovery of biomarkers related to rare diseases provide researchers with more robust tools to match genotypes and phenotypes. Advances in RNA sequencing are helping to improve diagnosis. In one study, researchers looked at the value of RNA sequencing from blood from patients with undiagnosed rare diseases across 16 disease categories. The study resulted in a 7.5% diagnostic rate and a further 16.7% improved candidate gene resolution.

One company that is using advanced technology to better understand disease-causing genetic variations is FDNA, which uses AI to analyze a comprehensive database of real-world data. The company's next-generation phenotyping (NGP) technologies capture, structure, and analyze complex human physiological data to produce genomic insights.

Another company, Centogene, has developed a multiomic-technology platform combining genomics, proteomics, and metabolomics to assist with genetic analysis. The company's CentoMD curated database for rare diseases has analyzed more than 400,000 cases, and more than 12.2 million variants across 120 countries.

A breakthrough innovation was made by a team of researchers from Kaunas University of Technology in Lithuania, who created a mobile app to help recognize early signs of Huntington's disease, a progressive brain disorder that affects around 30,000 people in the United States. Early signs can include irritability, depression, small involuntary movements, poor coordination, and trouble learning new information or making decisions.

As advances continue to be made in understanding rare diseases, the number of products to treat – and even cure — rare diseases will continue to grow, ultimately giving many more patients the care and support they need. PV

Rare Facts

1 in 10 people are affected by rare disease

1 in 2 rare diseases don't have a foundation or research support group

1 of 2 patients diagnosed with a rare disease is a child

3 of 10 children with a rare disease won't live to see their 5th birthday

400 million people suffer from a rare disease globally (greater than the population of the U.S.)

8 in 10 rare diseases are caused by a faulty gene

4.8 years is the average time it takes for rare patients to receive an accurate diagnosis

95% of rare diseases lack an FDA approved treatment

Rare diseases impact more people than cancer and AIDS combined

Source: Global Genes

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