SPURRING INNOVATION **IN RARE DISEASES**

WHAT ARE RARE DISEASES?

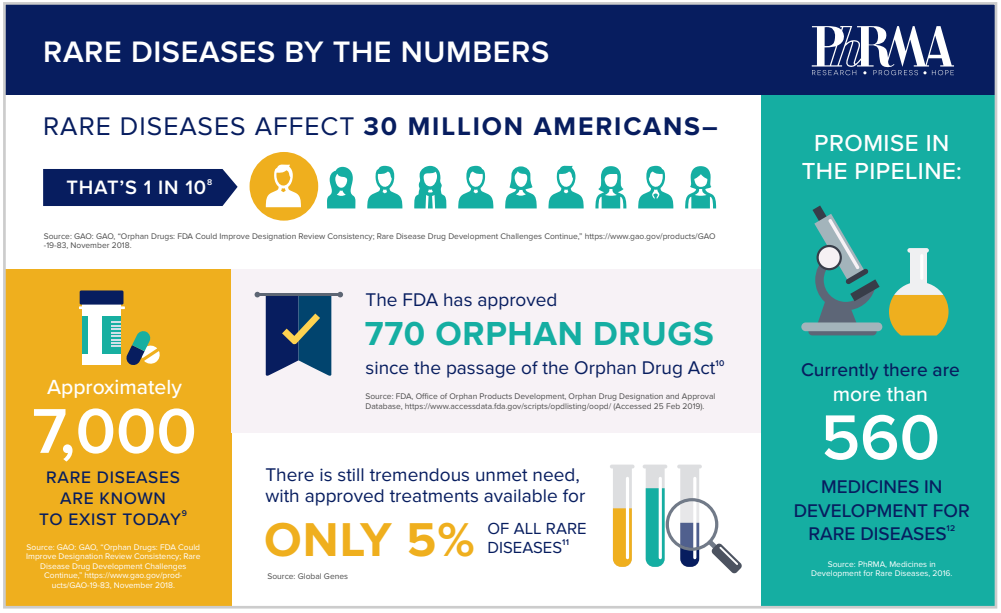
Although rare diseases each may individually only impact a relatively small number of patients, defined as less than 200,000 in the United States, their impact on public health is far-reaching.[1] While the median number of patients with each rare disease is less than 10,000, rare diseases affect 30 million Americans – about 1 in 10.[2, 3]

Approximately 7,000 different rare diseases are known today,[4] with many more still to be identified,[5] and it is estimated that 80% of rare diseases are genetic in origin.[6] Additionally, 85% to 90% of rare diseases are considered "serious or life threatening."[7]

Although researchers have made tremendous progress in advancing innovation for rare diseases, 95% of rare diseases still do not have a treatment option, representing a significant unmet need for patients. Rare diseases are often biologically complex and much remains unknown about the underlying causes and the clinical course of many individual conditions. Even within a particular rare disease, there can be many variations or subtypes resulting in different clinical manifestations and disease progressions.

RARE DISEASE DRUG DEVELOPMENT: CHALLENGES AND COMPLEXITIES

Developing medicines to treat rare diseases is particularly challenging. The complex biology of many rare diseases presents unique hurdles for scientists and, due to the inherently small population of patients with a rare disease, recruiting for and conducting clinical studies can be very difficult. Recent research found that the average orphan drug takes 2.3 years longer to develop than other medicines. [13]



Despite these challenges, America's biopharmaceutical researchers are leveraging new technologies and burgeoning scientific understanding to develop groundbreaking therapies for rare diseases.

FACILITATING RARE DISEASE DRUG DEVELOPMENT

The Orphan Drug Act (ODA) has been crucial in driving innovation in the treatment of rare diseases. Since the passage of the ODA in 1983, the FDA has approved 770 orphan drugs, in contrast to fewer than 10 medicines for rare diseases in the decade prior.[14] However, there remains a great need for new treatments.

The ODA has sought to provide incentives for R&D to develop medicines aimed at treating diseases for which there was no reasonable expectation that sales of the drug in the United States could support the development of the drug. These incentives include an R&D tax credit for 25% of clinical trial costs, as well as an exclusive right to market the drug for the orphan indication for 7 years upon FDA approval.[15] In addition, federal funding is available through grants to perform clinical trials of orphan products.

HARNESSING INNOVATION IN RARE DISEASE TREATMENT: RECENT ADVANCES

In 2018, progress continued for patients with rare diseases. Thirty-four novel new drugs approved in 2018 at FDA's Center for Drug Evaluation and Research (CDER) were for rare diseases.[16] There were also several medicines approved through the Center for Biologic Evaluation and Research (CBER) that brought significant treatment advances to patients with rare diseases.[17]

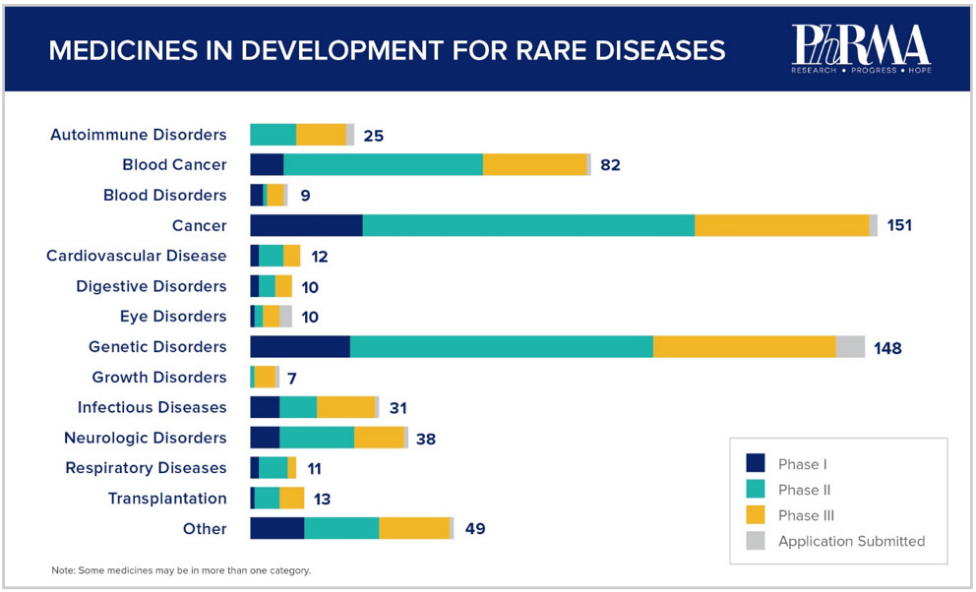
The new medicines help patients across a variety of disease areas, including genetic disorders and rare forms of cancer. Many of the new medicines offer treatment options where there were few or none previously available. Of the medicines approved at CDER to treat rare diseases, fifteen were first-in-class treatments, offering patients an entirely new way to tackle their disease.[18]

In recent years some noteworthy rare disease advances included:[19]

- The first approvals in an entirely new treatment paradigm called gene therapy, beginning with landmark advances in treating rare cancers and an inherited form of vision loss.[20]
- The first treatments directed at treating the underlying causes of cystic fibrosis, based on rare genetic mutations.[21]
- Significant advances in targeted therapies for many forms of blood cancer, including chronic lymphocytic leukemia, chronic myeloid leukemia, follicular lymphoma, acute myeloid leukemia, and multiple myeloma.[22, 23, 24]
- New medicines that can prevent or slow the impact of several extremely rare, devastating conditions, including pulmonary arterial hypertension, hereditary angioedema, and Gaucher disease.[25]
- The first therapies available to treat many rare pediatric diseases, including a specific form of a rare inherited disorder called Batten disease, a progressive, metabolic disease called hypophosphatasia (HPP), a devastating parasitic infection called Chagas disease, and neuroblastoma, a rare form of cancer that occurs in nerve cells and the brain.[26, 27, 28, 29]

SPURRING CONTINUED INNOVATION FOR PATIENTS

We've seen incredible advances in the development of medicines to treat patients with rare diseases. Despite this progress, there remains substantial unmet need for patients, as only 5% of rare diseases today have available treatment options. The biopharmaceutical industry is committed to advancing new medicines for patients with rare diseases, and the pipeline has never been more promising. There are more than 560 medicines currently in development for rare diseases.[30] Unprecedented scientific potential makes this a promising time for many patients with rare diseases. Maintaining incentives for research and development into these complex and challenging disease areas is critical to bring new medicines to patients.



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