Rare Disease Policy: Improving Patient Access to Treatment

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Executive Summary:

While fewer than 200,000 Americans are affected by any individual rare disease, the category collectively affects up to 30 million Americans, who as a group often struggle to access affordable, disease-specific treatments. While treatments can be costly and difficult to develop and specialists can be sparse; policies to improve insurance coding, telemedicine, and discrimination of orphan drugs inside the FDA can improve treatment access for rare disease patients.

Visualized: About 1 in 10 Americans suffer from a rare disease.

Background on Rare Diseases:

In America, rare diseases are those health conditions classified as affecting less than 200,000 people. More than 7,000 have been recognized, and up to thirty million Americans are affected (Genetic and Rare Disease Information Center, 2020). While most rare diseases remain without identified treatment, their research has improved since the passing of the Orphan Drug Act in 1983 (Gabay, 2019). While the act has improved the issues in funding for rare diseases, issues of access still remain for many patients, and affordability for treatments also remains an issue. Additionally, many of these diseases are often chronic, resulting in lifetime effects and treatment costs.

Myths and Misconceptions:

•Myth: Rare diseases are not significant or pervasive health problems.
•Fact: Up to 30 million Americans, or about 10% of the population, are affected by a rare disease (Genetic and Rare Disease Information Center, 2020).
•Fact: Nearly a quarter (22%) of cancer diagnoses are for rare forms of cancer (Bergertot et al, 2018).
•Myth: The Orphan Drug Act has made treatment affordable for rare disease patients.
•Fact: The ODA is meant to improve research on these treatments, not access. Many patients still have significant costs to pay for treatments, when they are even available (National Organization for Rare Disorders, 2020).

7,000+

The number of rare diseases currently recognized, for which treatment access may be limited.

40%

The percentage of treatment costs many patients have to pay, often for expensive treatments.

The Problem: Access

While rare diseases as a category are common, each individual ailment is rare, affecting fewer than 200,000 people (Genetic and Rare Disease Info Center, 2020). The rarity of each ailment creates difficulty for rare disease patients in accessing treatment.

Additionally, there is a barrier to many patients in even being identified—most general practitioners do not have experience or knowledge with specific rare diseases, and cannot effectively diagnose or treat them.(Barghi and Liu, 2015). Consequently, many patients have to wait for access to specialists to determine the underlying causes of symptoms, before even entering treatment for their rare disease.

Once a rare disease is diagnosed, there is still difficulty in treatment. Fewer than one in ten rare disease patients receives a disease-specific treatment (Feltmate et al. 2015). This means that while patients may receive treatment, it is often symptom focused or based on similar conditions, and was not developed with the patient's specific condition. Additionally, most of these rare diseases lack cures, meaning treatment for the condition will often be life-long (Global Genes 2014). And, due to health insurance pricing for some rare disease or orphan drugs, patients are responsible for up to 40% of their often-expensive treatments (National Organization for Rare Disorders, 2020).

Increased funding for the NIH, better ICD coding, and access to telemedicine are both potential solutions to improve patient access.

5%

The percentage of rare diseases with FDA approved treatments (National Institutes of Health, 2019).

Solutions

Rare disease policy is complicated, with a lot of unknowns for science and patients alike. But with the difficulty in receiving affordable, effective treatments, there are several steps that policy makers can take to improve access:

•Increase funding to the National Institutes of Health, to improve research on rare disease with and treatments. (National Organization for Rare Disorders, 2020)
•Expansion of health insurance coding and the International Classification of Diseases coding to improve the knowledge and logistic flow in treating rare diseases (National Organization for Rare Disorders, 2020; Rodwell and Ayme, 2015).
•Expand access to telemedicine, both to allow home-bound rare disease patients access to treatment, and to reduce the travel costs for those patients living significant distances from specialists (National Organization for Rare Disorders, 2020).
•Better discrimination and evaluation of rare disease definitions inside the FDA for the purposes of orphan drug funding (Herder, 2017).

These solutions will all make access to treatment easier for rare disease patients and begin to improve the treatment course as a whole through their further development.

Citations and Sources

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