Understanding Rare Diseases, Orphan Drugs, and the Drug Pricing Debate

AMERICANS FOR LOWER DRUG PRICES

Patients with rare diseases are often at the center of discussions about prescription drug prices. That's understandable. Families facing rare diseases frequently have fewer treatment options, fewer specialists, and fewer support resources than patients with more common conditions. Developing treatments for small patient populations can be difficult, expensive, and risky. Those realities deserve respect.

But to understand the policy debate, it helps to know what terms like "rare disease" and "orphan drug" actually mean, because they do not always describe the same thing.

WHAT IS A RARE DISEASE?

In the United States, a rare disease is generally defined as a condition affecting fewer than 200,000 people. That may sound uncommon, but there are more than 10,000 identified rare diseases. Taken together, they affect tens of millions of Americans.

Some rare diseases affect only a few dozen families worldwide. Others affect thousands of patients. Most have no approved treatment at all.

Families living with rare diseases often spend years searching for a diagnosis, navigating fragmented care, raising research funds, and advocating for scientific attention to conditions that many people have never heard of. That is the challenge Congress sought to address more than four decades ago.

WHAT IS AN ORPHAN DRUG?

An orphan drug is not a type of medicine. It is a legal designation. In 1983, Congress created the Orphan Drug Act because companies often had little financial incentive to develop treatments for very small patient populations. If only a few thousand people need a medicine, the potential market may not justify the cost and risk of development.

To encourage research, Congress created incentives for drug companies pursuing treatments for rare diseases. Those incentives can include:

- Seven years of orphan-drug exclusivity for the approved orphan indication
- Federal tax credits for certain qualified clinical testing expenses
- Waivers of certain FDA user fees, and
- Additional regulatory support during development

The policy has been remarkably successful. Before the Orphan Drug Act, very few treatments were approved specifically for rare diseases. Today, hundreds are available, and many more are in development. That success should be recognized.

SO WHERE DOES THE CONTROVERSY COME FROM?

The debate is not about whether patients with rare diseases deserve treatments. They do. The debate is about whether incentives designed for small patient populations are always being used in the way Congress originally intended.

Some medicines receive orphan designation for a rare condition and later gain approval for additional uses that affect much larger patient populations. In other cases, a drug may receive multiple orphan designations covering different rare conditions while also becoming a major commercial product. *(continued)*

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Nothing about this is necessarily illegal or improper. Companies are operating within the legal framework Congress created. But it does raise a reasonable policy question: *Should incentives designed to encourage development for very small patient populations be treated differently once a product generates substantial revenue from much larger markets?*

Reasonable people can disagree about the answer. But it is a different question than whether patients with rare diseases deserve access to treatment.

WHO ARE WE TALKING ABOUT?

Several well-known medicines have received orphan-drug designations while also becoming major commercial products. Humira, for example, received orphan designation for certain rare conditions while ultimately becoming one of the highest-grossing medicines in pharmaceutical history. Other widely used medicines have similarly received orphan designations for particular indications while also serving larger patient populations.

Researchers studying the orphan-drug program have found that some orphan-designated products are eventually approved for both rare and non-rare conditions. That does not mean the orphan designation was inappropriate. It does mean that the term “orphan drug” does not always describe a medicine serving only a small number of patients.

WHY THIS MATTERS

If you’ve followed recent debates over prescription drug prices, you may have heard warnings that greater pricing oversight could harm patients with rare diseases. Now that we've covered the basics, it's easier to evaluate those claims.

The first question is simple: *What kind of oversight are we talking about?* In many states, drug pricing review boards function primarily as transparency bodies. They gather information about how prescription drug prices are set and how those prices affect patients, employers, insurers, and public programs. They do not determine whether a medicine may be prescribed, whether a patient may receive it, or whether a manufacturer may sell it. In those states, it is difficult to see a direct pathway by which transparency and information gathering alone would restrict patient access to treatment. If anything, greater transparency may help policymakers better understand the unique challenges facing patients with rare diseases, including the economics of developing medicines for small populations.

A smaller number of states have authorized drug pricing review boards to consider upper payment limits (UPLs) on certain medicines. Those programs are newer, more limited, and remain under development. To date, very few UPLs have been established anywhere in the country. In Colorado, the first state to adopt one, implementation has been delayed by ongoing litigation involving the manufacturer of the drug under review. And it is important to note that these programs generally focus on medicines with substantial spending, significant market impact, or large numbers of patients. A treatment serving a few hundred patients presents a very different policy question than a medicine generating billions of dollars in annual revenue from broad use across large patient populations. Some states have also incorporated exemptions, protections, or review criteria that can limit how rare-disease therapies are evaluated. *(continued)*

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Reasonable people can disagree about the best approach to drug pricing policy. But discussions about rare diseases are most productive when they begin with a clear understanding of the facts: a rare disease, an orphan-drug designation, and a widely used commercial product that happens to possess one are not always the same thing.

QUESTIONS WORTH ASKING

The next time you hear that a proposal will affect "orphan drugs" or "rare disease patients," it may be worth asking a few additional questions.

- *Is the discussion about a medicine serving a few hundred patients, or a product used by millions?*
- *Is the proposal about transparency, payment, access, or something else entirely?*
- *Are policymakers discussing a rare disease, an orphan-drug designation, or a commercially successful medicine that happens to have one?*

Those distinctions matter. Understanding them is the first step toward evaluating the debate for yourself.

ONE FINAL THOUGHT

Patients living with rare diseases deserve thoughtful policy discussions grounded in evidence rather than assumptions. They deserve a system that genuinely supports innovation for small patient populations. They deserve access to the medicines they depend on. And they deserve honest explanations of how the system works. Those goals are not in conflict.