

Rare Diseases

Rare diseases can be difficult to diagnose and devastating to live with. Although rare diseases affect a small number of people, the impact on patients, their families, and society is profound, as many are severe, chronic, and progressive. Patients often live without hope, as they may face premature death without effective treatment options.



Approximately **400 million** people worldwide live with a rare disease.¹

In the United States, a disease is considered rare if it affects less than **200,000 people**.²

In Europe, a disease is considered rare when it affects **1 in 2,000 people**.³

In Japan, a rare disease is defined as affecting fewer than **50,000 people**.⁴

There are more than **7,000 rare diseases**; over 90% of them don't have an approved treatment option.^{5,6}

“When I shared my story, I realized it's important to know you are not alone and that there is hope. It gave me a sense of comfort and encouragement. Now, I know that others can benefit from my experience, and I want to be an inspiration to them.”

VICTOR
LIVING WITH PNH

References:

1. Global Genes. RARE Disease Facts. Accessed January 18, 2023. <https://globalgenes.org/rare-disease-facts/>
2. U.S. Food and Drug Administration. Orphan Drug Act - Relevant Excerpts. Accessed January 18, 2023. <https://www.fda.gov/industry/designating-orphan-product-drugs-and-biological-products/orphan-drug-act-relevant-excerpts>
3. Orphanet. About Rare Diseases. Accessed January 18, 2023. https://www.orpha.net/consor/cgi-bin/Education_AboutRareDiseases.php?lng=EN.
4. Japan Ministry of Health, Labour and Welfare. Accessed January 18, 2023. https://www.mhlw.go.jp/english/policy/health-medical/pharmaceuticals/orphan_drug.html.
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6. The Power of Being Counted | RARE-X. Accessed February 14, 2023. <https://rare-x.org/wp-content/uploads/2022/05/be-counted-052722-WEB.pdf> [Alexion co-authored this report]

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TANNER
LIVING WITH HPP

“My hope for Tanner in the future is that he enters into adulthood. I think that he's very smart, and I think that he is definitely going to go to college. I kind of hope that he will be a doctor and maybe help kids like doctors have helped him.”

RENÉ
TANNER'S MOM



Jill
LIVING WITH aHUS

Challenges of Diagnosis and Drug Development and Treatment

Diagnosis

- Often, very few physicians are familiar with diagnosing and treating these illnesses, leading to missed, delayed, or inaccurate diagnoses.
- Few researchers or companies explore the disease, due to the very small number of patients affected.
- Enhanced diagnostic tools and dissemination of knowledge are needed to improve treatments.

Drug Development & Treatment

- It can be difficult for investigators to identify appropriate patients who qualify for enrollment in ongoing rare disease clinical trials.
- The cost and risk associated with manufacturing orphan drugs increase, since most are complex biologics requiring living cells (production is simpler and less expensive with chemical drugs).

Helpful Resources

When a patient is diagnosed with a rare or ultra-rare disease, having a support system can be just as important as having the right physician and treatment plan. Several groups provide resources and support:

- **Global Genes** is a leading non-profit organization dedicated to overcoming challenges facing the more than 400 million people worldwide living with rare diseases.
- **National Organization for Rare Disorders (NORD)** is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them.
- **EURODIS—Rare Diseases Europe** is a unique, non-profit alliance of over 1000 rare disease patient organisations from 74 countries that work together to improve the lives of millions of people living with a rare disease globally.
- **RareConnect** is a safe, easy to use platform where rare disease patients, families, and patient organizations can develop online communities and conversations across continents and languages.
- **Orphanet** is a unique resource, gathering and improving knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases.

Visit [Alexion.com](https://www.alexion.com) for more information about the rare diseases we focus on.