

Rare and Ultra-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.



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Approximately **300 million** people worldwide live with a rare disease.¹

In the United States, a rare disease affects fewer than **200,000 people** (or about 620 patients per million of population).^{2,3}

In Europe, a rare disease affects **1 in 2,000 people** (or about 500 patients per million of population).⁴

In Europe, a disease is generally considered to be ultra-rare if it affects **one patient per 50,000 people** (or fewer than 20 patients per million of population).⁵

There are approximately **7,000 rare diseases**, but only about 5% of them have treatment options available.^{6,7}

“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.”

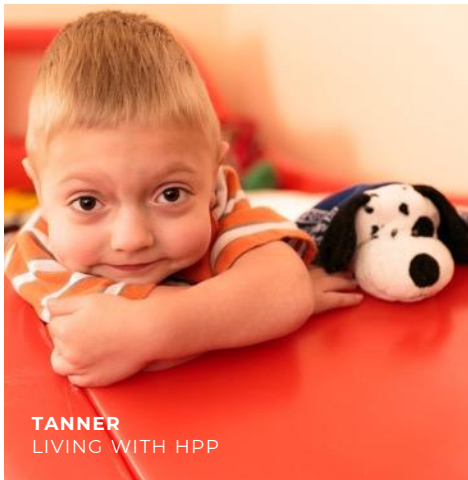
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Rare and Ultra-Rare Diseases



TANNER
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“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, 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 increases, 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 non-profit patient advocacy organization working to eliminate challenges of rare disease.
- **National Organization for Rare Disorders (NORD)** is a U.S.-based non-profit advocacy organization dedicated to helping people with rare diseases and assisting the organizations that serve them.
- **European Organisation for Rare Diseases (EURORDIS)** is a non-governmental patient-driven alliance of patient organizations representing 700+ rare disease patient organizations in 63 countries.
- **RareConnect** is an initiative of EURORDIS in which rare disease patients, families and patient organizations can develop online communities and conversations across continents and languages.
- **Orphanet** is a database of information on rare diseases and orphan drugs for the public.

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

