What is a Rare Disease?

Important Facts About Rare Diseases

Leadiant Biosciences

What is a Rare Disease?

A rare disease is defined as a disease that affect a small number of people compared to the general population. A disease is considered rare if it affects fewer than **200,000 people in the United States (US).**[1]

Most rare diseases are genetic, which means the disease is caused by changes in a gene or chromosome and is passed from parent to child.

Some rare diseases are more commonly known than others. Did you know that **cystinosis, cystic fibrosis, Duchenne muscular dystrophy,** and **Huntington's disease** are all considered rare diseases?

Statistics: Rare is Not Rare

More than **7,000 rare diseases** have been identified impacting more than **300 million** people worldwide; this includes **25 to 30 million Americans** or about **1 in 10 people in the US**[1]

~95% of rare diseases do not have an FDA-approved treatment[2]

72% of rare diseases are genetic[3]

3 out of 10 children who have a rare disease are unlikely to see their fifth birthday[4]

The economic burden of rare diseases in the U.S. (total direct and indirect costs associated with having a rare disease) approached **$1 trillion in 2019**[5]

Direct costs totaled **$418 billion:**

- Inpatient care
- Outpatient care
- Physician visits
- Medications
- Durable medical equipment

Indirect costs totaled **$548 billion:**

- Forced retirement
- Loss of income due to partial or full inability to work
- Home modifications
- Vehicle modifications
- Uncovered healthcare costs

Most rare diseases lack approved treatments despite major advances in research providing the tools to understand their molecular basis.[6]

Developing Therapies for Rare Diseases

There are many factors that cause rare diseases to often be underdiagnosed, misdiagnosed, or delayed in diagnosis. It's important to underscore the impact that rare disease has on not only the affected individual, but also their families, friends, and caregivers. Many individuals are not provided with information on rare diseases the same way that they are with information on more common diseases. There are many valuable resources available to those who wish to learn more about rare diseases and the rare disease community, such as NORD and Global Genes, two rare disease advocacy organizations.

Developing therapies for rare diseases is challenging for a number of reasons, including:

- *Complex biology and insufficiently studied molecular mechanisms*
- *Lack of understanding of the natural history of the disease progress*
- *Lack of engagement from general public to support research and development in the rare disease area*
- *Complexity to conduct clinical trials due to the rarity of patients*

What is Being Done to Advance Treatments for Rare Diseases?

Scientific Research

Scientific research helps to better understand rare diseases, helping lead to the development of effective treatments.

Natural History Studies

Natural history studies help researchers and medical professionals further understand how different rare diseases and their symptoms change and progress over time.

Clinical Trials

Clinical trials help develop and evaluate the safety and effectiveness of new possible treatments for different rare diseases.

Support Resources

Learn more about rare diseases and the needs of the rare disease community here:

- **Global Genes:** www.globalgenes.org
- **NORD:** www.raredisease.org
- **Rare Diseases Clinical Research Network:** www.rarediseasesnetwork.org
- **Rare Diseases at FDA:** www.fda.gov/patients/rare-diseases-fda

To learn more about clinical trials and ongoing research, visit clinicaltrials.gov.

Our Commitment

At Leadiant Biosciences, Inc., we focus on the rare. Leadiant employees work with rare dedication to provide therapies for patients with rare diseases. We partner with patient communities, physicians and researchers worldwide to overcome challenges and be a resource of hope.

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