RARE AND ULTRA-RARE DISEASES

RARE AND ULTRA-RARE DISEASES OVERVIEW

Rare and ultra-rare diseases, often referred to as orphan and ultra-orphan diseases, affect very small numbers of patients. In the United States, a disease is defined as rare if it affects fewer than 200,000 people (or, approximately 620 patients per million of population).[1] The European definition of a rare disease is one that affects fewer than five people per 10,000 of the population (or, approximately 500 patients per million of population).[2] In contrast, 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)[3]–and most ultra-rare diseases affect far fewer than this–as few as one per million or less.

Despite the very small number of patients they affect, the impact that ultra-rare diseases have on patients, their families, and society is profound, as many are severe, chronic and progressive, with high mortality rates. Patients with severe and life-threatening ultra-rare diseases often live without hope as they have no effective treatment options and may face premature death.

CHALLENGES OF DIAGNOSIS, DRUG DEVELOPMENT AND TREATMENT

A severe and ultra-rare disease often presents unique public health challenges:

- Typically, few researchers or companies explore the disease, given the very small number of patients affected.
- Often, very few physicians are familiar with diagnosing and treating the illness, leading to missed, delayed or inaccurate diagnoses even when an approved, effective therapy is available.

Biopharmaceutical companies face unique challenges as they seek to develop new, innovative therapies for patients with ultra-rare diseases. While nearly all drug development entails high levels of risk and investment, this is especially true in ultra-rare diseases. These challenges include:

- The need to identify sufficient numbers of patients to participate in clinical trials, which often results in a large number of trial sites in multiple countries since each may only enroll one or two patients—or none at all.
- Increased regulatory risks since there is usually no approved therapy for a given ultra-rare disease, and no well-established road map for regulatory approval.
- Increased cost and risk for manufacturing ultra-orphan drugs, since most are complex biologics that require living cells for production (versus chemical drugs whose production is typically simpler and less expensive).
- The need to engage in significant physician education, patient support and post-marketing research once an ultra-orphan therapy is approved.

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ALEXION®

Alexion has a proven track record in overcoming these hurdles on behalf of patients, and has significant experience in developing and delivering breakthrough therapies for patients with ultra-rare diseases.

ACCESS TO THERAPIES FOR ULTRA-RARE DISEASES

Alexion's objective is that every patient who can benefit from our therapies will have access to them. Alexion is committed to working with private healthcare organizations, policymakers and governments around the world so that patients with ultra-rare diseases have access to Alexion therapies. Ongoing collaboration between governments and companies like Alexion can support continuing medical innovation and facilitate access to drug therapies once they are available.

HELPFUL RESOURCES

When a patient is diagnosed with a rare or ultra-rare disease, having a support system can be as important as having the right physician and treatment plan. There are several groups and resources to which a patient can turn, including:

- **Global Genes**, a non-profit patient advocacy organization working to eliminate the challenges of rare disease by building awareness, educating the global community, and providing critical connections and resources that equip advocates to become activists for their disease. Visit **globalgenes.org**.
- **National Organization for Rare Disorders (NORD)**, a U.S.-based non-profit patient advocacy organization dedicated to helping people with rare diseases and assisting the organizations that serve them. NORD is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research and service. Visit **rarediseases.org**.
- **Orphanet**, a database of information on rare diseases and orphan drugs for the public. Its aim is to contribute to the improvement of the diagnosis, care and treatment of patients with rare diseases. Visit **orpha.net**.
- **RareConnect**, a platform where rare disease patients, families and patient organizations can develop online communities and conversations across continents and languages. RareConnect is an initiative of EURORDIS. Visit **rareconnect.org**.
- **European Organisation for Rare Diseases (EURORDIS)**, a non-governmental patient-driven alliance of patient organisations representing over 700 rare disease patient organisations in 63 countries. EURORDIS represents the voice of an estimated 30 million people living with a rare disease in Europe. Visit **eurordis.org**.

For more information about Alexion, visit alexion.com.

References

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