



Rare Disease Fast Facts

Alport Syndrome is a rare genetic disease that leads to kidney failure, hearing loss, and vision problems. It is estimated to affect between 30,000 and 60,000 patients in the U.S. Because of the way the disease is inherited, young boys in their teens and young adult years are hit the hardest.

Alport Syndrome is one of thousands of known rare diseases in the world. Below are some basic facts on rare diseases:

1. In the U.S., any disease affecting fewer than 200,000 people is considered rare. It is estimated that nearly 30 million Americans are affected by a rare disease. That means 1 in 10 people have a rare disease.
2. There are approximately 7,000 rare diseases. Some patients have less extensive symptoms while for others, managing their care is a full-time job.
3. According to the National Institute of Health (NIH), 80% of rare diseases are genetic in nature. Other rare diseases are the result of infections, allergies, and environmental causes. Two-thirds of rare diseases impact children.
4. Rare diseases include a wide variety of disorders and symptoms that vary not only from disease to disease, but also from patient to patient suffering from the same disease.
5. Rare diseases are usually chronic, progressive, degenerative, and often life-threatening.
6. People with rare diseases struggle to get a proper diagnosis, find information about their disease, and get treatment. Relatively common symptoms can hide underlying rare diseases, leading to misdiagnosis and delayed treatment. Many patients spend 5 years or more seeking an accurate diagnosis.
7. Rare disease patients often feel alone in facing the challenges of their medical condition.
8. Treatment for rare diseases is very expensive. More often, there is no effective treatment for a rare disease. In fact, only about 5% have FDA-approved treatments.
9. Providers treat over 90% of rare disease patients "off-label," meaning they prescribe medications or treatments approved for different diseases with similar characteristics in order to provide some level of care. Often this "off-label" care is not covered by insurance, causing a financial burden for the family.
10. Medical research is difficult because of the rarity of many diseases. Most of the push for research on rare diseases comes from patients and other grassroots efforts.

To learn more about Alport Syndrome, go to www.alportsyndrome.org.

To learn more about rare diseases, go to www.rarediseases.org (US) or www.eurordis.org (Europe).