

THE ORPHAN DRUG ACT: MAKING THE IMPOSSIBLE HAPPEN FOR RARE DISEASE PATIENTS

The **Orphan Drug Act** was enacted in 1983 to incentivize innovative drug development for the nearly **7,000 identified rare diseases**. Prior to 1983, only a small number of therapies had been approved to specifically treat rare diseases, leaving millions of patients within the United States and the rest of the world without treatment.

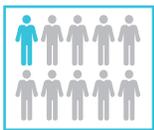


Enacting the Orphan Drug Act in 1983 with its financial incentives and other inducements was an important start to enabling more investment and development of treatments targeted to rare diseases.

— *Dr. Scott Gottlieb, Commissioner, FDA. February, 2018*



Although the number of individuals suffering from a single **rare disease** is small, the total number of individuals suffering from a rare disease is **significant**.



1 in 10 people

1 in 10 people are affected by rare diseases. Half are children.



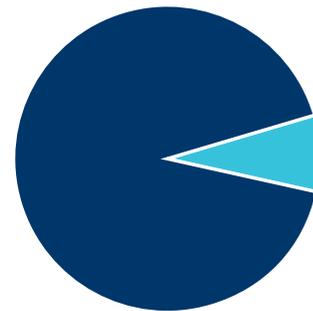
25–30 million

The National Institute of Health (NIH) estimates that between 25–30 million people suffer from rare diseases in the United States.

reports.nih.gov/nihfactsheet

TREATMENT FOR RARE DISEASES

95%
Orphan diseases without treatment



5%
Orphan diseases with treatment

491 different drugs for **676** disease indications* have been brought to patients with rare diseases.

www.accessdata.fda.gov | FDA Orphan Drug Designations & Approval

* To treat 302 diseases

The **Orphan Drug Act** is critical to ensuring that rare disease drug development continues as new rare diseases are identified.

TODAY THERE ARE APPROXIMATELY

7,000 rare diseases, affecting **25 to 30 million** Americans and **95%** do not have treatments available.

As a result of scientific advances and precision medicine, approximately **30 new rare diseases** are identified each year.

As more rare diseases are identified, it is critical that the incentives provided by the Orphan Drug Act are sustained to encourage development of therapies for rare diseases.

reports.nih.gov/nihfactsheet | October 2017, NORD/QuintilesIMS Report, "Orphan Drugs in the United States: Providing Context for Use and Cost"

While hundreds of medicines for rare diseases have been developed, only a small percentage of the nearly 7,000 identified rare diseases have treatments.

Rare Disease Patient Stories with Orphan Drugs



In the late 90's after delivering her third child, Alex Flipse began experiencing shortness of breath, severe exhaustion, and the inability to walk very far. After seeing a doctor in the small town where she lived, Alex was diagnosed with depression and prescribed an antidepressant. Sadly, it didn't help.

After passing out about 30 times in a three-month period, she went to a new doctor who conducted an ecocardiogram which showed that her heart was enlarged. Further testing confirmed that Alex had Idiopathic Pulmonary Arterial Hypertension (PH).

At the time, there was only one intravenous medication to treat PH. But after several years, Alex became involved in a clinical study which would reveal that she was misdiagnosed and that new treatments for Pulmonary Veno Hypertension — caused by heart disease — would help her tremendously. "I'm now on the right heart medication, which has made all the difference," Alex said. "I feel like I've gotten my life back."

If it wasn't for drug discovery or the new treatments given to her, Alex might not be alive. "I always wanted to see my kids grow up and at some points, didn't think I would," she added. "Now, I have three grandkids."

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In 2012, our four-year-old son, Beckett, who had a history of developmental delay was diagnosed with a rare genetic condition. Testing at Texas Children's Hospital revealed that Beckett had a change in SYNGAP1 — only five other cases had been diagnosed at the time. Mutations in the SYNGAP1 gene had been linked to intellectual disability and, in some cases, autism, but little else was known about its impact. Doctors presented a grim and hazy picture of Beckett's future, but one thing was certain: our journey to find a miracle medicine had just begun.

Doctors at first prescribed physical and occupational therapy for our son, but his symptoms continued. Over the next few years we saw many specialists and underwent a slew of tests, including CAT scans, MRIs, and EEGs. At five years old, a 24-hour EEG test finally confirmed what we had suspected for several years. Beckett was having absence seizures, as well as problems with sensory perception, and trouble sleeping.

We tried administering several medications to help Beckett before finding an off-label treatment that ultimately changed his life for the better. Over the years, the collaboration between his physicians and the scientists studying SYNGAP1 created a path to finding more answers about how this gene functions. And thanks to a patient group, our family was introduced to the drug discovery process that has helped us stay optimistic about our son's future.

For additional NORD patient stories visit:

<http://bio.org/patientexperience>