

## Rare Disease Facts for Medical Students

### What is a rare disease?

Any disease affecting fewer than 200,000 Americans is considered rare.

### How many rare diseases are there?

According to the National Institutes of Health (NIH), there are approximately 7,000 rare diseases.

### Who is affected by rare diseases?

Approximately 30 million Americans have rare diseases. Most rare diseases are genetic and 2/3 affect children, but others have adult onset.

### What special problems do people with rare diseases have?

People with rare diseases are a medically underserved population. Challenges include:

- *Difficulty obtaining an accurate diagnosis — many patients see multiple specialists prior to diagnosis*
- *Limited treatment options — only about 350 of the 7,000 rare diseases have FDA approved treatments*
- *Little or no research being done on many of these diseases*
- *Difficulty finding physicians or treatment centers with experience in treating a particular rare disease*
- *Treatments are generally more expensive than those for common diseases*
- *Reimbursement issues related to private insurance, Medicare, and Medicaid*
- *Difficulty accessing medical, social, or financial services or assistance because those making the decisions are not familiar with the disease*
- *Feelings of isolation and of having been abandoned or “orphaned” by our healthcare system*

### Why is it important for medical students to be aware of these problems?

Medical students can't possibly learn all aspects of all 7,000 rare diseases. However, it's important for



**VanHoutan Family, two children with Batten disease**

medical students to be aware of rare disease challenges so that they can adopt a way of thinking that leaves them open to “zebras” when they hear hoof beats.

### What is NORD?

The National Organization for Rare Disorders (NORD) is a nonprofit organization established in 1983 to provide advocacy, education, patient/family services, and research to improve the lives of people with rare diseases.

NORD is a unique federation of patient advocacy organizations dedicated to helping people with rare diseases and assisting the organizations that serve them. NORD is committed to the diagnosis, treatment, and cure of rare disorders.

### What can medical students do to help?

- Promote awareness among your peers about challenges associated with living with a rare disease
- Support Rare Disease Day (last day of February each year) by visiting the national website ([www.RareDiseaseDay.US](http://www.RareDiseaseDay.US)) to learn about ways to get involved and take a leadership role at your school
- Become a student member of NORD by contacting NORD's Educational Initiatives Department ([education@rarediseases.org](mailto:education@rarediseases.org)) to request a free Student Membership and receive a monthly eNews and other news of special interest to students

## 1 in 10 Americans Have Rare Diseases



**Anneliese, undiagnosed rare disease**

Anneliese is a sweet, smiley 2-year-old who has been given a difficult journey in life. MRIs of her brain show progressive cerebellar atrophy, which means her condition is getting worse. Despite extensive medical testing, Anneliese has no diagnosis. Her mother wrote: “We live with no prognosis, no treatment options, and little hope...”



**Estle Lawson, idiopathic pulmonary fibrosis**

Estle Lawson was diagnosed with idiopathic pulmonary fibrosis, a devastating disease in which lung tissue becomes thick and scarred over time and the brain and other organs can't get the oxygen they need. Researchers are studying possible treatments and two were approved in 2014. Continued research is important for patients with this deadly disease and thousands of other rare diseases.



**Drew Thortensen, Noonan syndrome, with his family**

Drew Thortensen was diagnosed with Noonan syndrome before he was born, as a result of genetic testing after doctors identified a problem just 12 weeks into his mother's pregnancy. Drew's family joined the Noonan Syndrome Foundation and have actively promoted awareness of this rare syndrome. Rare disease patients often have a very close relationship with their medical experts, and the Thortensens have met Dr. Noonan through their Foundation activities.



**Daniel Wong, NORSE syndrome**

Daniel Wong was a healthy 22-year-old who suddenly fell ill and was taken to a major medical center. He was diagnosed with NORSE (New-Onset Refractory Status Epilepticus), a little known rare disease that was first identified less than 10 years ago. While his medical team tried to save his life, Daniel's family struggled to find information on NORSE. Daniel lost his battle but medical experts in Belgium and at Yale University School of Medicine helped NORD develop a report on this condition for its Rare Disease Database to help other families seeking information.