

RARE DISEASES quick kit

a women in government publication

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RARE DISEASES often difficult to diagnose and treat

While individual rare diseases, by definition in the United States, affect fewer than 200,000 people, around 25 to 30 million Americans are afflicted with at least one of the 6,800 known rare diseases.ⁱ

Around the world, some 350 million people are living with rare diseases, including some—like cystic fibrosis and Lou Gehrig’s disease—that are well known, and many others that are not.ⁱⁱ Each year on the last day of February, World Rare Disease Day observances are held around the world in an effort to bring awareness of rare diseases to improve access to treatments and medical representation.ⁱⁱⁱ

Genetics and environmental factors such as diet, smoking and exposure to chemicals can play a role in the cause of rare diseases. Most rare diseases are traced back to mutation in a single gene^{iv} and many are often very complex.^v Physicians often have difficulty accurately diagnosing rare diseases. A 2013 Shire report—based on surveys with physicians, patients and caregivers—found:

- » It took an average of 7.6 years for U.S. doctors to diagnose a rare disease.
- » Patients typically visited eight physicians—four primary care doctors and four specialists—before getting a correct diagnosis for a rare disease.
- » Patients were misdiagnosed two to three times before getting a correct diagnosis.
- » Compared to managing common ailments, 98 percent of physicians said it takes more office visits to diagnose a rare disease.
- » More than half—67 percent—of rare disease patients and caregivers said they needed to provide their health care professional with information about their disease.^{vi}

Even when they do get a correct diagnosis, many patients with rare diseases have either no or ineffective treatment options available to them.^{vii} Some federal programs have targeted development of treatment for rare diseases.

» Congress in 1983 passed the Orphan Drug Act to incentivize development of treatments for rare diseases.^{viii} Since then, the U.S. Food and Drug Administration has approved treatments for more than 360 rare diseases.^{ix}

» The National Institutes for Health in 2009 launched the Therapeutics for Rare and Neglected Diseases—or TRND—program with the hope of creating a drug development pipeline to treat rare and neglected diseases.^x

In addition, because health care professionals don’t deal with rare diseases regularly, they are often challenged with the diagnosis and must devote more resources—in both physician and non-physician time—to help the patient. That is just one more factor in the added costs for patients with rare diseases.

» In the Shire report, 92 percent of physicians surveyed said it is more difficult to address the needs of a patient with a rare disease in an office visit.

» 98 percent of respondent physicians said they need more office visits to adequately address a patient’s symptoms.

» 55 percent of patients responding to the Shire report had direct medical expenses not covered by insurance, even though 90 percent of those patients responding to the report were covered by insurance.^{xi}

The National Organization for Rare Disorders—or NORD—has

WORLD RARE
DISEASE DAY

FEBRUARY 29, 2016

Observances will be held around the world in an effort to bring awareness of rare diseases to improve access to treatments and medical representation.

been actively involved in expanding the federal programs to address the problems patients with rare diseases face. NORD has also recognized the role state policy plays in helping patients, and several states have addressed the needs of these patients in a number of ways, but the policies are inconsistent. A 2015 NORD report rates states on policies aimed at addressing the needs of patients with rare diseases.

- » Some patients need special nutrition items to control their rare disease, but only half the states include some mandate for private insurers to cover these foods. In those states without the mandate, patients need help paying for these medical foods.
- » Only six states set policy for coverage based on whether a given disease is part of the newborn screening panel; many states specify which conditions are covered in the law, which makes it difficult to ensure coverage for new disorders.
- » Some insurers place therapies for patients with rare diseases on a “specialty-tier” of the drug formulary, which means patients often pay a lot more for the drug. Only eight states have set caps on drug co-pays for these treatments.^{xii}

ⁱ*FAQ About Rare Diseases*. (2016, February 22). Retrieved from National Human Genome Research Institute: www.genome.gov/27531963

ⁱⁱ*RARE list*. (2016, February 22). Retrieved from <https://globalgenes.org/rarelist/>; <https://globalgenes.org/rarelist/>

ⁱⁱⁱ*World Rare Disease Day 2016*. (2016, February 23). Retrieved from Global Genes: <https://globalgenes.org/world-rare-disease-day/>

^{iv}*FAQ About Rare Diseases*

^v*Medline Plus Rare Diseases*. (2016, February 22). Retrieved from Medline Plus: <https://www.nlm.nih.gov/medlineplus/rarediseases.html>

^{vi}*Rare Disease Impact Report*. (2016, February 22). Retrieved from Global Genes: <https://globalgenes.org/wp-content/uploads/2013/04/ShireReport-1.pdf>

^{vii}*Medline Plus Rare Diseases*

^{viii}*FAQ About Rare Diseases*

^{ix}*2015 State Policy Progress Report: A Roadmap for State Improvement*. (2016, February 23). Retrieved from National Association for Rare Disorders: <http://www.rarediseases.org/assets/files/rare-action-network/2015-State-Progress-Report.pdf?email=mary.branham@gmail.com>

^x*Press Releases: NIH Announces New Program to Develop Therapeutics for Rare and Neglected Diseases*. (2016, February 23). Retrieved from National Institutes of Health: <http://www.nih.gov/news-events/news-releases/nih-announces-new-program-develop-therapeutics-rare-neglected-diseases>

^{xi}*Rare Disease Impact Report*

^{xii}*2015 State Policy Progress Report: A Roadmap for State Improvement*





FOR IMMEDIATE RELEASE

[Insert State Legislator's Name] Supports Awareness of World Rare Disease Day

[CITY], [STATE]—[DATE], 2016—Do you know that 25 to 30 million Americans are afflicted with at least one of the 6,800 known rare diseases? Around the world, some 350 million people are living with rare diseases, including some—like cystic fibrosis and Lou Gehrig's disease—that are well known, and many others that are not. Each year on the last day of February, World Rare Disease Day observances are held in an effort to bring awareness of rare diseases to improve access to treatments and medical representation.

Join rare disease patients, the medical community, caregivers and other health care advocates looking to make a difference in [insert State Name] as part of World Rare Disease Day 2016.

[Insert quote on the importance of awareness of rare diseases and why this is important to you.]

Many important decisions related to rare disease diagnosis and treatment are made at the state level, including prescription drug cost-sharing, newborn screening coverage, Medicaid and Children's Health Insurance Program (CHIP) eligibility, and Medical Foods coverage. The implementation of the Affordable Care Act has highlighted the increasingly important role of states in assuring that the health care needs of Americans are addressed.

According to the National Institutes of Health (NIH), a disease is rare if it affects fewer than 200,000 Americans. One in 10 Americans live with a rare disease and two-thirds of these patients are children. There are more than 7,000 rare diseases, 95 percent of which have no treatment. Often, research gets funded by the families and friends of patients or by patient organizations.

World Rare Disease Day is an annual awareness activity dedicated to elevating public understanding of rare diseases and calling attention to the special challenges people face. The observance takes place every year on the last day of February to underscore the nature of rare diseases and what patients face. It was established in Europe in 2008 by EURORDIS, the organization representing rare disease patients in Europe, and is now observed in more than 80 nations.

For more information about World Rare Disease Day in the U.S., go to www.rarediseaseday.us. To search for information about rare diseases, visit the National Organization for Rare Disorders' website, www.rarediseases.org or Global Genes at www.globalgenes.org. For state policymaker information, visit Women In Government at www.womeningovernment.org.

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Media contacts:

[If applicable, insert your organization's media officer with contact information]



February 29, 2016—World Rare Disease Day

WHEREAS, nearly 30 million Americans are affected by nearly 7,000 rare diseases and conditions in the United States;

WHEREAS, while rare diseases, by definition, affect fewer than 200,000 people, the sheer number of rare diseases means one in 10 Americans are affected by these diseases;

WHEREAS, many rare diseases are serious and debilitating conditions that have a significant impact on the lives of those affected;

WHEREAS, while the U.S. Food and Drug Administration (FDA) has approved treatments for more than 360 rare diseases, the vast majority of rare diseases have no or ineffective treatments;

WHEREAS, diagnosis of rare diseases often takes years and multiple visits to physicians, both general practitioners and specialists;

WHEREAS, some rare diseases are well known, many of them are not and many patients and families affected by less widely known rare diseases bear a large share of the burden of funding research and raising public awareness to support the search for treatments;

WHEREAS, thousands of residents of _____ are among those affected by rare diseases since nearly one in 10 Americans have rare diseases;

WHEREAS, Global Genes will hold a variety of awareness-raising activities starting on World Rare Disease Day, February 29th and throughout the month of March;

WHEREAS, thousands of patients and caregivers, medical professionals, researchers, companies developing orphan products to treat people with rare diseases, and others will participate in that observance;

NOW, THEREFORE, I, _____ do hereby proclaim February 29, 2016, as Rare Disease Day.



Bank of Tweets and Facebook Posts for World Rare Disease Day



Twitter

Diagnosis of rare disease can take, on average, 7.6 years. **#WRDD2016 #CareAboutRare @GlobalGenes**

Doctors need more office visits to adequately address a rare disease patient's symptoms. **#WRDD2016 #CareAboutRare @GlobalGenes**

Treatment for rare diseases can be expensive, often not covered by insurance. **#WRDD2016 #CareAboutRare @GlobalGenes**

Only 8 states cap co-pays for drugs that treat rare diseases. **#WRDD2016 #CareAboutRare @GlobalGenes**

Patients with rare diseases are often misdiagnosed 2 to 3 times before getting a correct diagnosis. **#WRDD2016 #CareAboutRare @GlobalGenes**

Orphan Drug Act of 1983 aims to incentivize development of treatments for rare diseases. **#WRDD2016 #CareAboutRare @GlobalGenes**

Only 6 states set insurance coverage for rare diseases included in newborn screening panel. **#WRDD2016 #CareAboutRare @GlobalGenes**

Rare diseases require more resources from doctors, their staffs. **#WRDD2016 #CareAboutRare @GlobalGenes**

Most rare diseases can be traced back to mutation in a single gene and many are often very complex. **#WRDD2016 #CareAboutRare @GlobalGenes**



Facebook

It's National Rare Disease Day, use **#ICareAboutRARE** and share this post if you or someone you know lives with a rare disease. Together we can spread awareness for the 350 million people worldwide living with diseases that often have yet to be cured. For more information check out—www.globalgenes.org and www.womeningovernment.org **#WRDD2016**

There are 7,000 different types of rare diseases and disorders, with more being discovered every day. On average it takes 8 years for someone suffering from a rare disease to finally get a diagnosis. To learn more about how you can help and get involved please visit www.globalgenes.org and www.womeningovernment.org **#WRDD2016 #ICareAboutRARE**