TOOL KIT CONTENTS

About Women in Government ................................................................. 2
Rare Diseases Present Challenges for Patients, Families ...................... 3
Rare Diseases Often Difficult to Diagnose and Treat ............................. 13
Rare Diseases Legislation 2016 ............................................................. 15
Press Release .......................................................................................... 18
Sample Resolution .................................................................................. 19
Suggested Social Media Posts ............................................................... 20
ABOUT WOMEN IN GOVERNMENT

Mission Statement
Women In Government Foundation, Inc., headquartered in Washington, D.C., is a national non-profit, non-partisan organization of women state legislators that provides leadership opportunities, expert forums, and educational resources to address and resolve complex public policy issues to all 1,808 women state legislators.

Vision
Women In Government leads the nation with a bold, courageous and passionate vision that empowers and mobilizes all women legislators to effect sound policy.

History
Women In Government is soon to enter its 29th year. Since its founding, WIG has successfully implemented leadership and educational activities to support informed policy decisions at the state level. With over 28 years of experience working with state legislators, state agency representatives and the public, Women In Government proudly hosts educational conferences, state briefings, and other policy events annually to address the nation’s public policy issues.

Women In Government ensures that members and partners are provided with up-to-date publications and resource materials on cutting-edge policy topics. Beginning in 2004, Women In Government launched Policy Resource Centers that provide educational materials and resource tools, including legislative toolkits, while identifying important policy issues and supporting state legislative activity.

Women In Government’s Board of Directors is composed of 13 female state legislators who guide the organizational activities, help recruit new members and provide support to implement WIG’s strategic action plan. The Board is made up of both Democrats and Republicans, reflecting Women In Government’s non-partisan nature.

Through the support of sponsors, including corporations, foundations, and state and federal grants, Women In Government provides its members with legislative scholarships. These scholarships provide conference participants with opportunities to hear presentations from nationally noted expert speakers, receive comprehensive issue-based information and network with fellow policymakers, sharing best practices and developing policy solutions.
When baseball great Lou Gehrig started faltering in the game, his teammates and fans knew there was something wrong. He was misdiagnosed with gall bladder problems and continued to grow weaker. It wasn’t until after he had quit the game for good in 1939 that doctors at the Mayo Clinic finally diagnosed Gehrig with a very rare form of degenerative disease—amyotrophic lateral sclerosis, ALS, which is now called Lou Gehrig’s disease.¹

While most people have heard of ALS, or its more common name referring to the famous first baseman, they may not have heard of many of the nearly 7,000 rare diseases patients around the world are dealing with. That’s likely because these rare diseases individually affect less than 200,000 people, as defined in the United States.² Collectively, however, rare diseases affect 25 to 30 million Americans—and more than 350 million people around the world.³

Research Efforts

While many rare diseases have been identified for a long time, it’s only been in the last four decades that rare diseases, in general, have gained more attention. The National Organization for Rare Disorders was established in 1983 when several rare disease patient organizations formed it as an ad hoc committee to gain support for the Orphan Drug Act of 1983, which created incentives for development of treatments for rare diseases.⁴ But even after 30 years on the radar of state and federal policymakers, as well as medical researchers around the globe, many of the rare diseases that have been identified have no cure and no effective treatment.⁵

Virginia Kimonis, MD, MCRP, knows that all too well. Dr. Kimonis is a professor in the Department of Pediatrics, Division of Genetics and Genomic Medicine at the University of California—Irvine Medical Center. She’s worked under funding from the National Institutes of Health for her research into rare diseases since 1990, but will lose funding this year. She worries about her work on genetic causes of muscle disease and has been creative with her patients in generating funding for research. “It’s all about the money,” she said. “You can’t have research relying on NIH dollars in an academic setting to have an impact on rare diseases.”⁶

One family involved in her research—Rick and Cristy Spooner of California—held a fashion show to raise money in the hopes of finding at least a treatment for Mitochondrial Complex 1 Deficiency, which two of their three daughters are fighting. That led to the creation of the Spooner Girls Foundation to raise money for research into the disease. “The fact that we have to go to our friends and family and raise money on our own, the pressure that we’re really running the show is really hard,” said Cristy Spooner. “We need the support and the research and the doctors need the funding to be able to support us.”⁷

But research funding is not the only thing affected by the lack of information about rare diseases. “Because these rare diseases are not so well known, there’s not a widespread recognition of the treatment and needs of this community. Health care policy decisions are made with a broader population in mind,” said Tim Boyd, associate director of state policy for NORD.⁸

NORD and other groups are working to change that. Individual rare diseases have long had their champions and methods to draw attention. For many years, however, they essentially stood alone. NORD represented the joining of efforts for a common cause with the Orphan Drug Act of 1983. That legislation provided three basic incentives to pharmaceutical
“The fact that we have to go to our friends and family and raise money on our own, the pressure that we’re really running the show is really hard. We need the support and the research and the doctors need the funding to be able to support us.”

—Cristy Spooner
Spooners Girls Foundation

manufacturers to develop therapeutic products for rare diseases—federal funding for grants and clinical trials, a tax credit for 50 percent of the costs for clinical testing and an exclusive right to market the drug for seven years. Before passage of the Orphan Drug Act, the U.S. Food and Drug Administration had approved only 38 orphan-designated medications; that has grown to more than 360 rare diseases with an approved treatment resulting from orphan designation. The Orphan Drug Act also allowed the FDA to award research grants, as well as review and approve requests from manufacturers for orphan drug status, and garnered attention for the multitude of diseases and disorders for which there is no cure or even an effective treatment. But research into these types of diseases and disorders takes years to bear fruit. Dr. Kimonis, for instance, has been working with the gene mutation of VCP—Vasolin Containing Protein—identified since 2003. She began working with families on diseases affected by that gene mutation years before that.

The federal government, through the National Institutes of Health, provides funding for research into rare diseases in a number of programs, including clinical research to help identify and diagnose rare diseases and pharmaceutical research to speed development of a treatment for rare diseases. The Rare Diseases Clinical Research Network encompasses 2,600 researchers in hundreds of sites around the world studying more than 200 rare diseases. These teams work with patient advocacy groups and have included 40,000 patients participating in clinical studies since the inception of the program in the National Center for Advancing Translational Sciences in 2003.

NCATS also supports the Therapeutics for Rare and Neglected Disease—or TRND—program. This program works to speed the development of new drugs for rare and neglected diseases through collaborative partnerships in which TRND provides project management and drug development operational support. It also supports the Global Rare Diseases Patient Registry Data Repository—or GRDR—aimed at providing a single repository for rare disease data from around the world to assist in therapeutic development and improve the quality of life for people with rare diseases.

Marshall L. Summar, MD, chief of the Division of Genetics and Metabolism at Children’s National Medical Center in Washington, D.C., told Medscape in 2014 that registries “are one of the most important things we can do in this field.” That’s because data on all patients with a specific rare disease will be in one central location, giving researchers easy access to historical clinical information on a lot of patients that might provide a critical link in developing treatments.

In addition to administering the Orphan Drug Designation program, the FDA also focuses on treatments such as biologics, devices or medical foods through its Office of Orphan Products Development. In addition to evaluating scientific and clinical data from participants in the program and administering two grant programs, the Office of Orphan Products Development works with stakeholder groups including the medical community, researchers, industry and rare disease patient groups, as well as other government agencies.

Dr. Kimonis said because rare diseases are often tied to a gene mutation, research on one disease could pay off with information on another. For instance, Dr. Kimonis’ research has led to the discovery of a link between the NUBP1 gene responsible for Mitochondrial Complex 1 Deficiency and Parkinson’s disease. She said the research on rare diseases influence the understanding and development of new therapies for common disorders. Dr. Kimonis believes drug companies can use the Orphan Drug Act to develop new treatments for rare disorders, but can extrapolate that information for treatments for more common disorders. Drug companies, she said, “are going for the rare disorders but knowing there’s a bigger target after that.”

Diagnosis and Newborn Screening
While the rare disease community is growing as more research is conducted and more patients are diagnosed, getting an initial diagnosis for a rare disease can be difficult. Making the information readily available to researchers is important, but oftentimes that doesn’t hit the frontline clinicians who are dealing with patients suffering from difficult-to-diagnose diseases because they don’t develop in clusters. NCATS manages the Genetic and Rare Disease—or GARD—Information Center, which provides current and reliable information about rare or genetic diseases. So once a diagnosis is made, patients and their families can learn more about the disease.

Getting to that accurate diagnosis is often difficult and frustrating. In fact, a 2013 Shire report—based on surveys with physicians, patients and caregiver—found it took an average of nearly eight years for U.S. doctors to diagnose a rare disease. Patients typically see an average of four primary
With Two Daughters Fighting Rare Disease, Couple Develops Foundation for Research

Shortly after Cristy and Rick Spooner’s third child, Ryann, was born, the couple had a sense of déjà vu, and not in a good way.

Ryann, now 6, was exhibiting symptoms similar to those her older sister, Cali, had exhibited more than a decade earlier. Ryann, too, wasn’t reaching milestones. The couple had gone through years of tests on Cali and wondering to more tests and wondering, all to no avail. They went to the Mayo Clinic. They had every test performed, even a cerebellum biopsy when Cali was 1. The baby wasn’t showing any physical malformations and the family was told whatever was causing Cali’s problems wasn’t genetic.

When Ryann began having issues, the Spooners immediately requested an MRI and an EEG. The symptoms were just too similar. Cristy Spooner said the light pattern in Ryann’s MRI had the exact pattern as one Cali had had years before. The family went to a geneticist and had every possible test performed.

“It was literally just a guessing game,” said Cristy Spooner. “The geneticist would look at this panel of diseases and it showed nothing. We would go every few months and get tested for a different panel of diseases. We had no idea what was going on.”

Finally, the Spooners reconnected with a geneticist, Virginia Kimonis, MD, MCRP, a professor in the Department of Pediatrics, Division of Genetics and Genomic Medicine at the University of California-Irvine Medical Center. Genetic testing had advanced to a point that the Spooners could finally get an answer.

Fourteen years after Calyn’s birth, she and her younger sister were diagnosed with a very rare form of Mitochondrial Complex 1 Deficiency. Cristy Spooner said the girls inherited mutations in the NUBPL gene that caused a defect in an important enzyme from both parents, who carry the recessive gene. The couple’s middle daughter, Raelyn, inherited one copy of the mutated gene and doesn’t have the disorder.

By chance, a symposium on mitochondrial disease was being held at Newport Beach, Calif., soon after the Spooners got the diagnosis. The couple lives about 20 minutes from Newport Beach and decided to go.

Although the entire conference was filled with physicians who specialize in mitochondrial disease, “not one in the whole conference had anything like ours,” Cristy Spooner said.

Cellular disease can affect different parts of the body. For the Spooner girls, it affected their cerebellum.

“We got some information and discovered that this diagnosis is still very rare,” Cristy Spooner said.

Dr. Kimonis developed a “Mito Cocktail” of vitamins and enzymes as therapy that has improved their daily lives, though it is far from a cure. Because the vitamins are available over the counter, insurance doesn’t cover it in California. The couple must pay the full cost of $300 a month for treatment for both girls. Cristy Spooner would like to see that covered. Kentucky legislators in April passed a bill with a floor amendment to cover the Mito Cocktail for a young girl recently diagnosed with the same disease as the Spooners.

Dr. Kimonis has received funding from the National Institutes of Health for decades, but it still doesn’t cover all the costs that could speed the research efforts. So the Spooners decided to lend a helping hand. They held a special needs fashion show in 2014 and donated money from the fundraiser to the University of California Irvine Hospital for research.

After that function, the couple decided to start their own foundation to raise money for research. “We could have more functions and raise more money,” Cristy Spooner said.

They held the second special needs fashion show in March, 2016, and raised $20,000, all of it donated to the UC-Irvine Hospital. Most of the supporters were friends and family. Now, the Spooners’ goal is to prove a point and attract funding from other foundations and companies.

Cristy Spooner’s goal is to raise $60,000 for a mouse model for the disease—research thus far, she said, has been stem cell research. A mouse model, she said, holds more promise for discovering treatments and, possibly, a cure.

Researchers are making progress, she said. They’ve discovered that the NUBPL gene is also associated with Parkinson’s disease. Cristy Spooner hopes that connection will spur more interest in research funding and a better understanding of rare diseases.

“The fact that we have to go to our friends and family and raise money on our own, the pressure that we’re really running the show is really hard,” said Cristy Spooner. “We need the support and the research and the doctors need the funding to be able to support us.”
care physicians and four specialists before getting a correct diagnosis, and patients were misdiagnosed two to three times before getting a correct diagnosis. xxii

That happened to Eddie and Stephanie Greenhill of Pikeville, Ky. Their son Noah started having problems at 3 months old. He was experiencing projectile vomiting and couldn’t eat. “We didn’t know what was wrong with him until he was 5 years old,” Stephanie Greenhill said. “He failed to thrive for five years. He was basically the disease. We just didn’t have the name for it and no diagnosis.”xxi

Just by chance, another child in Noah’s daycare was experiencing the same symptoms and had been diagnosed with eosinophilic esophagitis, a very rare condition that attacked his organs and ultimately required him to have a feeding tube installed. The Greenhills had been taking Noah to the University of Cincinnati Children’s and asked for a different doctor and convinced him to perform the necessary tests to finally diagnose the problem. xxxii (See story on page 5.)

They are not alone in that quest. In the Shire survey, 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. xxiv Most rare diseases are traced back to mutation in a single gene xxvi and many are often very complex. xxvii

Because genetics play a role in rare diseases, and many children are born with the genetic mutation, newborn screening is important in early diagnosis. Global Genes, a leading rare disease patient advocacy organization, believes more robust newborn screening efforts across the states could detect many more rare diseases. xxviii

“For many of the disorders, the earlier you know about it, the earlier a doctor knows about it, the earlier interventions can take place,” Tim Boyd, associate director of state policy for NORD, said. xxviii For that reason, NORD supports well-funded newborn screening programs in every state.

In its first State Policy Progress Report, covering 2015, NORD scored states on implementation of screening for the core and secondary conditions on the Uniform Newborn Screening Panel, which it encourages every state to adopt. Nearly all states cover the full panel of recommended core conditions, but far fewer cover the secondary conditions. Only Minnesota and New Jersey received perfect scores for the newborn screening section of NORD’s report, while Arkansas, Kansas and Louisiana were rated as insufficient in this area. xxix

The Advisory Committee on Heritable Disorders in Newborns and Children for the Secretary of Health and Human Services regularly reviews the Recommended Universal Screening Panel to guide states on diseases to include in their newborn screening. The panel includes 32 core conditions and 26 secondary conditions in those tests. xxx Many states include more than the recommended conditions, while nine states—Arizona, Arkansas, Georgia, Kansas, Montana, Nebraska, Rhode Island, Virginia and Washington—including less than the recommended core conditions before the 2016 legislative session. Mississippi includes the most conditions for screening, covering 59 conditions, while Georgia and Kansas include the least—29. xxxi

Several states have seen action with regard to improving their newborn screening efforts over the past few years. This year, California Sen. Richard Pan, MD, authored Senate Bill 1095 to expand newborn screening to include MPS 1 and Pompe, two life-threatening diseases for which treatments are available, and to add more rare diseases as they are added to the federal Recommended Uniform Screening Panel. xxxi That would change the current situation in California, which requires new legislation every time a disease is added to the federal screening panel.

“Rare diseases often are difficult to diagnose in time before permanent damage is done. SB 1095 ensures California

### Number of conditions included in newborn screenings, by US state

<table>
<thead>
<tr>
<th>State</th>
<th>Conditions Included</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alabama</td>
<td>46</td>
</tr>
<tr>
<td>Alaska</td>
<td>46</td>
</tr>
<tr>
<td>Arizona</td>
<td>30</td>
</tr>
<tr>
<td>Arkansas</td>
<td>31</td>
</tr>
<tr>
<td>California</td>
<td>57</td>
</tr>
<tr>
<td>Colorado</td>
<td>35</td>
</tr>
<tr>
<td>Connecticut</td>
<td>43</td>
</tr>
<tr>
<td>Delaware</td>
<td>43</td>
</tr>
<tr>
<td>Florida</td>
<td>36</td>
</tr>
<tr>
<td>Georgia</td>
<td>29</td>
</tr>
<tr>
<td>Hawaii</td>
<td>48</td>
</tr>
<tr>
<td>Idaho</td>
<td>46</td>
</tr>
<tr>
<td>Illinois</td>
<td>58</td>
</tr>
<tr>
<td>Indiana</td>
<td>50</td>
</tr>
<tr>
<td>Iowa</td>
<td>46</td>
</tr>
<tr>
<td>Kansas</td>
<td>29</td>
</tr>
<tr>
<td>Kentucky</td>
<td>52</td>
</tr>
<tr>
<td>Louisiana</td>
<td>33</td>
</tr>
<tr>
<td>Maine</td>
<td>38</td>
</tr>
<tr>
<td>Maryland</td>
<td>56</td>
</tr>
<tr>
<td>Massachusetts</td>
<td>53</td>
</tr>
<tr>
<td>Michigan</td>
<td>55</td>
</tr>
<tr>
<td>Minnesota</td>
<td>56</td>
</tr>
<tr>
<td>Missouri</td>
<td>58</td>
</tr>
<tr>
<td>Montana</td>
<td>30</td>
</tr>
<tr>
<td>Nebraska</td>
<td>31</td>
</tr>
<tr>
<td>Nevada</td>
<td>45</td>
</tr>
<tr>
<td>New Hampshire</td>
<td>35</td>
</tr>
<tr>
<td>New Jersey</td>
<td>57</td>
</tr>
<tr>
<td>New Mexico</td>
<td>49</td>
</tr>
<tr>
<td>New York</td>
<td>56</td>
</tr>
<tr>
<td>North Carolina</td>
<td>37</td>
</tr>
<tr>
<td>North Dakota</td>
<td>49</td>
</tr>
<tr>
<td>Ohio</td>
<td>43</td>
</tr>
<tr>
<td>Oklahoma</td>
<td>53</td>
</tr>
<tr>
<td>Oregon</td>
<td>45</td>
</tr>
<tr>
<td>Pennsylvania</td>
<td>33</td>
</tr>
<tr>
<td>Rhode Island</td>
<td>30</td>
</tr>
<tr>
<td>South Carolina</td>
<td>53</td>
</tr>
<tr>
<td>Tennessee</td>
<td>57</td>
</tr>
<tr>
<td>Texas</td>
<td>53</td>
</tr>
<tr>
<td>Utah</td>
<td>44</td>
</tr>
<tr>
<td>Vermont</td>
<td>42</td>
</tr>
<tr>
<td>Virginia</td>
<td>31</td>
</tr>
<tr>
<td>Washington</td>
<td>32</td>
</tr>
<tr>
<td>West Virginia</td>
<td>38</td>
</tr>
<tr>
<td>Wisconsin</td>
<td>48</td>
</tr>
<tr>
<td>Wyoming</td>
<td>38</td>
</tr>
</tbody>
</table>

Advisory Committee on Heritable Disorders in Newborns and Children for the Secretary of Health and Human Services
Relief of Diagnosis Opens Doors to More Frustrations for Coverage of Treatment

When Noah Greenhill started projectile vomiting when he was only 3 months old, his parents, Eddie and Stephanie Greenhill of Pikeville, Ky., began a quest for an answer that would last nearly five years.

Nothing seemed to help and doctors couldn’t give them a definitive diagnosis. He couldn’t gain weight past about 20 pounds. He finally crossed that hurdle at age 2. Then came the stalling at 25 pounds. By age 4, he weighed a mere 28 pounds.

“It was a never-ending battle” trying to get his weight up, said Eddie Greenhill.

The couple had been taking him regularly to the University of Cincinnati Children’s Hospital with no luck. Until, that is, they by chance met a couple whose child was experiencing the same symptoms and was attending Noah’s day care center.

So the next time they went back to UC Children’s Hospital, the Greenhills asked for a different doctor and asked for a test for eosinophilic esophagitis, a condition that attacked his organs.

“By the time we got a diagnosis, he was malnourished. His hair was falling out,” said Stephanie Greenhill. “He didn’t have long to live.”

But the relief at getting a diagnosis was short-lived.

“We were told as soon as he was diagnosed that there was no treatment, no medication, no cure,” said Eddie Greenhill. “Even though you have a diagnosis and you feel a little bit relieved that you know what is going on, there is nowhere to go and nothing you can do.”

Physicians started Noah on a regimen of food elimination, starting with the top eight food groups. The family waited three months then returned to UC Children’s Hospital for another endoscopy. There was no improvement.

“All of this experimental,” said Eddie Greenhill. “It’s not treating the disease. It’s not doing anything for it but trying to evaluate or decrease the severity of the disease because it is an allergy to food.”

The next step was a feeding tube. After three months with the feeding tube, Eddie Greenhill said, “Noah was starting to get some color back. His hair was coming back. He was just starting to look healthier.”

Armed with the diagnosis and the knowledge of some treatment that would help Noah, the Greenhills faced another major battle—getting insurance coverage for the amino acid-based formula that Noah needs to survive—at $40 a day.

The Greenhills filled out reams of paperwork, going back and forth between their doctor and the insurance company. It took about a month to line things up and all was well.

Until it wasn’t. When their insurance policies changed at the end of a year, the Greenhills had to go through the paperwork once again.

“It was just a battle year after year,” said Eddie Greenhill. “We had gotten to a point that we just looked at each other and said there’s no reason why we should have to fight an insurance company every time you turn around for something our son needs to live. It’s no different than an insurance company paying to put in an insulin pump and then paying for the insulin.”

So the Greenhills took a more deliberative route through the state legislature. Eddie Greenhill had known his state senator, Ray Jones, of Pikeville for years and talked with him about Noah’s situation. Then Sen. Ralph Alvarado, a medical doctor, got involved. He believed other children across Kentucky could benefit from changing the law requiring coverage for the formula—either through a feeding tube or by mouth—and expanded the legislation to cover both.

With the treatment, Noah is thriving. He plays baseball and sings in church.

“He’s very social,” said Stephanie Greenhill. “He doesn’t know a stranger. It’s not stopped him.”

But, Eddie Greenhill said, there are constant worries because someone at the school he attends must know how to feed him, what he can and cannot eat.

“It’s a constant worry that somebody will goof up somewhere through no fault of their own,” he said. One relief is that Noah, now 10, knows what he can and cannot eat.

Like many of the nearly 7,000 rare diseases that patients across the country and around the globe are facing, the awareness level is very low. Stephanie Greenhill is working with various organizations in an effort to raise awareness of her son’s disease. Awareness, she said, may help raise money for research, which she said is promising.
implements recommended newborn screening when an early
diagnosis and treatment can prevent disability and save lives,”
Dr. Pan, a pediatrician, said in introducing the bill.

**Treatments and Access**

Progress has been made in treatments and therapies for some
rare diseases over the past 33 years since the Orphan Drug
Act was approved. But that progress is limited to a fraction of
the 7,000 rare diseases people face. Even those diseases that
have gained public attention for decades have no effective

treatment. ALS, or Lou Gehrig’s disease, affects as many as
20,000 people at any given time in the U.S. xxxv Not only does
ALS have the public face of the 1930s baseball great, but it
also gained attention a few years ago with the “Ice Bucket
Challenge.” But there’s little that can be done for patients.

Stephen Finger, PhD, an economics professor at the Uni-
versity of South Carolina, didn’t flinch when an orthopedist
first mentioned ALS as a possible diagnosis for his condition.
"I assumed it was a treatable disease and I was facing some
sort of unpleasant treatment but that I was eventually going
to come out on the other side,” Dr. Finger said. "I think the
general public loses sight of the idea that here is something
where you currently don’t come out on the other side of it.” xxxv

And while no cure exists for ALS, there are some things
that can make life easier. Finger said as the disease progress-
es, patients rely more and more on equipment and tech-
nology to keep them connected to their families and to the
outside world. States could help people with ALS specifically
by ensuring they have access to the equipment and technol-
gy they need to help them continue to be a part of life and
be connected. “You keep someone connected to the outside
world and they still have a will and a drive to survive. With-
out that, they don’t,” he said. xxxvi

Those medical devices, medical foods and even the drugs
needed to treat people often are inaccessible to people with
rare diseases. Getting those treatments to the people who
need them often requires state legislatures to act on individu-
ral rare diseases.

In Kentucky, for instance, Rep. Rita Smart of Richmond,
a state director for Women in Government, this year filed a
floor amendment to an unrelated bill to require insurers to
cover treatment for a young girl in her district, Katherine
Belle, who suffers from a rare form of Mitochon-
drial Complex 1 Deficiency. Her neurologist
prescribed a compounded mixture of vitamins
and supplements. This “Mito cocktail,” which costs
around $250 a month, won’t cure the disease, but
it can help to increase muscle tone, stamina and
stability, and help to decrease intention tremor. xxxvii

But the family's health insurance wouldn't cover it. Rep. Smart didn't know much about the disease
Katherine Belle was fighting, but was happy to
work with another representative, House Majority
Caucus Chair Sannie Overly, to ensure the treat-
ment is covered. "The insurance companies fight
it tooth and nail," Smart said. She said legislators
could use more education about rare diseases and
the needs families face. "I know, from my point
of view, (this legislation) was under Banking and
Insurance and I’m not on that committee,” she said.
"Unless somebody in your district actually peti-
tions you, then it sort of gets lost in the shuffle.” xxxviii

Because there are so many rare diseases, and many have
very specific needs, it’s difficult to comprehensively address
those needs, Rep. Smart said. The NORD State Policy Prog-
ress Report, found scattered coverage for the treatments and
therapies patients with rare diseases need. It found:

- Only half the states include some mandate for private
  insurers to cover the special nutrition items patients with
  rare diseases need. In those states without the mandate,
  patients need help paying for 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 the conditions covered under the law, which
makes it difficult to ensure coverage for new disorders that
are added to the federal newborn screening panel.
- Some insurers place therapies for patients with rare disease
  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. xxxix

(From our perspective, we work really hard to make sure once
the research is done that people have access to drugs that will
help them. Cost-sharing within health plans can be a major
barrier to people accessing treatment. Once cost-sharing gets
above a certain level, you start to see fewer people accessing
treatment.”

—Tim Boyd
National Organization for Rare Disorders

xxxv, xxxvi, xxxvii, xxxviii, xxxix
Stephen Finger, PhD, had just become a father in 2010 with the birth of his daughter and again in 2012 with the birth of his son. Then little things started to crop up. He noticed some weakness and stiffness in his hands. He was having a bit of trouble getting his wallet out of his pocket. And buttoning his daughter’s buttons. Undoing the buckle on the car seats. Changing diapers.

Those minor annoyances continued for about a year. So in 2013, Finger, an economics professor at the University of South Carolina, decided to go to an orthopedist. He thought he had carpal tunnel or some minor muscular condition affecting his hands and dexterity. The orthopedist quickly referred him to a neurologist in Columbia, S.C., who referred him to a neurologist at Duke University Hospital.

The diagnosis—ALS, amyotrophic lateral sclerosis, Lou Gehrig’s disease.

“In a matter of a couple of months I went from thinking I had a pretty minor hand problem to being told I had a neurodegenerative disease with no effective treatment and no known cure and a life expectancy of two to five years,” said Dr. Finger. “Given where we were in our lives and in our family’s lives, it was pretty horrifying diagnosis.”

While ALS has been in the spotlight since the Yankees’ first baseman Lou Gehrig was diagnosed and made his famous speech on the field, there are still many questions but no cure. Progression of the disease varies from patient to patient.

“For us, the part we latched onto was that we really didn’t have any idea of how fast or how slow my progression was going to be and, to some extent, that liberated us to sort of focus on living each day to the fullest because we didn’t want to wake up two years later thinking that there was this ticking time bomb and wonder why we had wasted the time we had had,” said Dr. Finger.

Over the past three years, the progression of his disease has been relatively slow compared to many of his friends. His hands and arms are very weak; he can still walk around his home and office a little, but he has to be careful; falls are a major concern. ALS affects all the voluntary muscles in your body, which include the muscles in the diaphragm. As those muscle atrophy, you lose the ability to breathe. That’s how patients eventually die from the disease, Dr. Finger said.

Part of Dr. Finger’s effort to focus on living each day to the fullest is connecting with other ALS patients. He publishes a blog, writes for the Huffington Post, and has active Facebook and Twitter pages all in an effort to raise awareness about ALS and the challenges patients like him face. He was diagnosed with ALS at age 39, young for such a diagnosis, and needed to feel part of the broader community to not feel so alone.

“One of the motivations for the blog is to share my story with other people going through the disease,” said Dr. Finger, “to share my perspective on what I am going through, not to say that it’s the right perspective or the wrong perspective. It is my perspective.”

“It’s also a way to share his story with his circle of friends and to the outside world of what someone with ALS faces in fighting the disease, “what’s really on the line when we’re talking about getting funding for this disease, getting the appropriate advocacy work with this disease.”

He also hopes it helps people to understand the impact on patients and the families, “who, on a daily, on an hourly, on a minute basis, are forced to deal with something that they never would have otherwise dreamed of dealing with.”

He speaks out, he said, because of the need to educate people on the realities of the disease.

“I think because the disease is so debilitating physically, oftentimes you don’t see patients publicly who are really living through the hardships of the disease,” he said.

Those life stories are important tools to share with state policymakers to illustrate how access to technology can help ALS patients continue to live productive lives and to help families keep their heads above water.

“Without that outside access to technology to be able to communicate with the outside world it becomes harder and harder for these people to continue to be productive to continue to have the will to fight the disease,” Dr. Finger said.

State leaders can play a role in helping to ease the burden on families. Some states provide funding for ALS clinics, which are staffed with a neurologist and social workers, respiratory therapists, nutritionists and other health care pro-
can't have access to them,” Tim Boyd of NORD said. “From our perspective, we work really hard to make sure once the research is done that people have access to drugs that will help them. Cost-sharing within health plans can be a major barrier to people accessing treatment. Once cost-sharing gets above a certain level, you start to see fewer people accessing treatment.”

NORD strives to keep cost-sharing for patients below $100 per month to ensure people can access the medicine, Boyd said. The 2015 NORD State Policy Progress Report found that while the utilization of special tiers for prescriptions is staggering, some states have enacted policies to help make the drugs patients with rare diseases need more accessible, although they are in the minority. A few states have mandated caps on out-of-pocket expenses for specialty medications or capped co-pays on a per-drug, per-month basis, while some have mandated total caps for all drug cost sharing. For instance, Maine has set an annual cap of $3,500 on prescriptions, with deductibles applied, while New York sets a $70 per month cap per prescriptions following standard benefit design and prohibits cost-sharing in excess of cost-sharing, deductibles or co-insurance for non-preferred drugs or their equivalent. Vermont sets an annual cap of $1,300 per individual or $2,600 per family and the cap is linked to IRS inflation adjustments.

Shining a Spotlight

The challenges rare disease patients face in getting coverage are just a part of a long line of challenges to gain attention for their disease and the impediments patients of all rare diseases face. Each year since 2008, World Rare Disease Day draws attention to rare diseases and the needs of patients and families. Tim Boyd of NORD said the passage of the Orphan Drug Act in 1983 helped to fuel the conversation about rare diseases and that interest continues. “The awareness around rare disease is growing and, in particular, the awareness of common challenges is growing,” he said. “In the past, people would typically think about these diseases in the context of a single condition or a single disease.”

NORD, Global Genes and other patient advocacy groups focused on rare diseases as a whole have joined forces, as have other advocacy groups focused on specific rare diseases. NORD, for one, is building a Rare Action Network with a presence in every state aimed at support and education for local grassroots communities. Boyd said it’s important for state legislators to hear from their constituents who are fighting rare diseases. While NORD’s overall advocacy aims to give voice to those people, “that grassroots relationship and grassroots network is critical to us to address these things on a state by state basis. Our ability to operate in every state and effect change in every state depends on us having an advocacy network to actually carry things out,” Boyd said.

That means patients and their families are still the best advocates for their disease, and social media is making it much easier.

Consider the Kentucky case of Katherine Belle, the young girl with the rare form of Mitochondrial Complex 1 Deficiency. Rep. Smart said Senate Bill 18 covering insurance contracting for providers was passed by the Senate and moved to the House, where she attached the floor amendment to ensure Katherine Belle and 600 other children throughout the commonwealth would have access to the Mito Cocktail. Rep. Smart believed insurance companies lobbied the Senate to reject that amendment. That created a firestorm on social media. “They (families of children with the disorder) started contacting their local senators and representatives and talking about it on social media,” Rep. Smart said. “I think social media is going to be the trick to getting these types of bills passed. I do think that social media and technology will help these families lobby to find out who the families are and they can join together and organize groups that can do more lobbying work than they could before.”

Social media is doing much more than that. The ALS Ice Bucket Challenge in 2014, for example, raised $115 million for research, patient and community service, public and professional education and fundraising. But Stephen Finger, the University of South Carolina professor who suffers from ALS, said it did something else that was keenly important—it updated the face of the disease and highlighted the fact that there is still no cure or effective treatment.

“One of the things that was almost working against the
community until the ice bucket challenge was the idea that Lou Gehrig is the still the face of the disease,” Finger said. “He’s an incredible face of the disease for many reasons, but he also passed away 75 years ago. For people who weren’t intimately aware of what ALS was, you would think that something we knew about 75 years ago and had a very public profile would have been cured or at least treated in the interim.”

Finger said the Ice Bucket Challenge, which was largely driven by and through social media, brought attention to the patients who are fighting the disease today. He and others like him strive to do that as well. He publishes a blog, writes for the Huffington Post, and has active Facebook and Twitter pages all in an effort to raise awareness about ALS and the challenges patients like him face. He was diagnosed with ALS at age 39, young for such a diagnosis, and needed to feel part of the broader community to not feel so alone.

“One thing I have found is that with social media, it’s really allowed me and my family to connect with people in similar situations to ours,” he said. “By definition, if you have a rare disease, you don’t have many peers and, given that I’m a bit on the younger side of the distribution in terms of the average ALS patient, I don’t have many peers where I live who are in similar family situation.”

Filmmaker Patrick O’Bryan was diagnosed with ALS in his early 30s. He filmed his journey through diagnosis to his current paralysis in the documentary, “Transfatty Lives.” In his filmmaker’s notes, O’Bryan, aka DJ Transfatty, explained what prodded him over the course of a decade as he filmed the documentary. “As I progressively became more and more paralyzed I assembled a great group of friends and cameramen and I launched a website to fundraise and keep my documentary film afloat. The messages from others across the country and the world, also facing immense difficulties, helped fuel my mission to make my film. It started to feel like more than just my story, but also my duty to tell the story that others are facing in their own adversities.”

ALS is a rarity in the rare disease world—the attention to the disease has been steady and widespread. That doesn’t always happen. Of the 7,000 rare diseases, only a few—like ALS, Parkinson’s and cystic fibrosis—are well-known and have a very public face; the large majority of rare diseases do not. That’s why drawing attention to rare diseases as a whole is important to get laws passed, raise money for research and patient care, and prod along the efforts to find a cure for the patients whose voice isn’t as loud as patients with more common diseases.

NORD says one in 10 people are suffering from a rare disease of some sort whether they know it or not. Dr. Virginia Kimonis, the UC-Irvine rare disease researcher, said if you combine all the rare diseases, they become a common disorder. That knowledge could help amplify the individual voices by joining them together.

Looking Ahead
While many of the research dollars are awarded at the federal level, state policymakers are important to the process in many areas. Tim Boyd of NORD said policymakers can use the 2015 State Policy Progress Report—and the planned subsequent reports—to gauge how well their state is doing with regard to rare diseases and how they can do better. “First and foremost, they should try to engage with these families … listen to what these families are saying about their experience in accessing health care and accessing the health care system,” Boyd said. He said it’s important for state policymakers to understand that many health care decisions are made with the broader population in mind that don’t always work well for people with rare diseases.

Some states are working to address that gap. Illinois in 2016 considered legislation—House Bill 4576—to establish a Rare Disease Commission to provide education for state policymakers on the diseases, as well as access and coverage issues. Similar legislation was filed to establish the West Virginia Advisory Council on Rare Diseases to advise state agencies on research, diagnosis, treatment and education relating to rare diseases. Connecticut in 2015 established a task force to examine research, diagnoses, treatment and education related to rare diseases and to ultimately make recommendations for a group of experts to advise the Department of Public Health on Rare Diseases.

“They (families of children with the disorder) started contacting their local senators and representatives and talking about it on social media. I think social media is going to be the trick to getting these types of bills passed. I do think that social media and technology will help these families lobby to find out who the families are and they can join together and organize groups that can do more lobbying work than they could before.”

—Rep. Rita Smart
Kentucky House of Representatives
also established an Advisory Council on Rare Diseases in 2015 that would include advocates for rare diseases, including health care professionals, academic researchers, and a rare disease patient and foundation representative.\(^{vi}\)

Dr. Finger, who has battled ALS for nearly four years, hopes state leaders will consider what they can do to help ease the burden on families. The returns are massive because of the impact those services have, he said. "For a disease without a cure, without an effective treatment, those types of services make an enormous impact on the lives both in terms of the quality of life and the longevity of the lives of patients," he said.\(^{lii}\)


\(^{iii}\)(FAQ About Rare Diseases, 2016)

\(^{iv}\)NORD History of Leadership. (2016, April 22). Retrieved from National Organization of Rare Disorders: http://rarediseases.org/about/what-we-do/history-leadership/


\(^{vi}\)Kimonis, D. V. (2016, April 22). Author interview with Dr. Kimonis. (M. Branham, Interviewer)

\(^{vii}\)Spoonser, C. (2016, April 25). Author interview with Cristy Spoonser. (M. Branham, Interviewer)


\(^{xi}\)Kesselheim, E. and S. (2016, April 20). Author interview with Greenhill. (M. Branham, Interviewer)


\(^{xiii}\)(Stokowski, 2014)

\(^{xiv}\)Developing Products for Rare Diseases and Conditions. (2016, April 22). Retrieved from U.S. Food and Drug Administration: http://www.fda.gov/ForIndustry/DevelopingProductsforRareDiseasesConditions/ucm2005525.htm

\(^{xv}\)Kimonis, (2016)


\(^{xvii}\)(Rare Disease Impact Report, 2016)

\(^{xviii}\)Greenhill, E. and S. (2016, April 20). Author Interview with Greenhill family. (M. Branham, Interviewer)

\(^{xix}\)(Greenhill, 2016)

\(^{xx}\)(Rare Disease Impact Report, 2016)


\(^{xxiv}\)(Boyd, 2016)


\(^{xxvi}\)The Recommended Uniform Screening Panel. (2016). Retrieved April 22, 2016, from baby’s first test: http://www.baby’sfirsttest.org/newborn-screening/ the-recommended-uniform-screening-panel


\(^{xxxiii}\)(Finger, 2016)


\(^{xxxvii}\)(Boyd, 2016)


\(^{xxxix}\)(Boyd, 2016)

\(^{xl}\)(Boyd, 2016)

\(^{xli}\)(Smart, 2016)


\(^{xliii}\)(Finger, 2016)

\(^{xiv}\)(Finger, 2016)


\(^{xlvi}\)(Boyd, 2016)

\(^{xlvii}\)(Kimonis, 2016)

\(^{xlviii}\}(Boyd, 2016)


\(^{lxxiii}\)(Finger, 2016)
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.¹

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.² 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.³

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 gene⁴ and many are often very complex.⁵ 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.⁶

Even when they do get a correct diagnosis, many patients with rare diseases have either no or ineffective treatment options available to them.⁷ 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.⁸ Since then, the U.S. Food and Drug Administration has approved treatments for more than 360 rare diseases.⁹
- 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.¹⁰

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.¹¹

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.¹
The National Organization for Rare Disorders—or NORD—has 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.\textsuperscript{xii}
US S2188

**Rare Disease Innovation Act**

- This bill amends the Federal Food, Drug, and Cosmetic Act to expand the humanitarian device exemption to authorize the Food and Drug Administration (FDA) to exempt from effectiveness requirements certain medical devices intended to benefit fewer than 8,000 individuals. Currently, the FDA may exempt devices intended to benefit fewer than 4,000 individuals. Within 18 months of enactment of this Act, the FDA must define “probable benefit” for these devices. The FDA must report every five years on the effect of this expansion of the humanitarian device exception.

- **Primary Sponsor**
  - US Senator Corey Gardner (R-CO)

- **Co-Sponsor**
  - US Senator Joe Donnelly (D-IN)

- **Latest Action**
  - Oct. 2015 referred to Committee on Health, Education, Labor, and Pensions

US S 2030

**Advancing Targeted Therapies for Rare Diseases Act of 2015**

- This bill would allow the sponsor of an application for the approval of a targeted drug to rely upon data and information with respect to such sponsor’s previously approved targeted drugs.

- **Primary Sponsor**
  - US Senator Michael Bennet (D)

- **Co-Sponsors**
  - US Senator Richard Burr (R)
  - US Senator Orrin Hatch (R)
  - US Senator Elizabeth Warren (D)
  - US Senator Mike Enzi (R)

- **Latest Action**
  - Committee on Health, Education, Labor, and Pensions ordered to report bill with an amendment (substitute favorably)

CA SCR 108

**Relative to Rare Disease Day**

- This measure would proclaim February 29, 2016, as Rare Disease Day in California and provide for the recognition of Rare Disease Day in California in subsequent years, as specified.

- **Primary Sponsor**
  - Senator Bill Monning (D)

- **Co-Sponsors**
  - Senator Joel Anderson (R)
  - Senator Jean Fuller (R)
  - Senator Steve Glazer (D)
  - Senator Isadore Hall (D)
  - Senator Robert Hertzberg (D)
  - Senator Mark Leno (D)
  - Senator Andy Vidak (R)
  - Senator Robert Wieckowski (D)

- **Latest Action**
  - Passed in the Senate; waiting on action in the Assembly

IL HB 4576

**Rare Disease Commission**

- Creates the Rare Disease Commission Act. Provides for the creation of the Rare Disease Commission. Defines terms. Provides that initial appointments shall be made by February 1, 2017. Provides required criteria and considerations for appointees and nominations to the Commission. Includes provisions regarding the terms, vacancies, and compensation for the Commission’s membership. Requires that the Commission meet at least quarterly and submit an annual report due no later than December 31 of every year.
to the General Assembly. Provides that the Department of Public Health shall provide administrative and other support to the Commission. Effective January 1, 2017

Primary Sponsor
- Rep. Sonya Harper (D)

Co-Sponsors
- Rep. Mary Flowers (D)
- Rep. Litesa Wallace (D)
- Rep. Monique Davis (D)
- Rep. John Anthony (R)
- Rep. Stephanie Kifowit (D)
- Rep. Robert Martwick (D)
- Rep. Kathleen Willis (D)
- Rep. Michael Zalewski (D)
- Rep. Al Riley (D)

Last Action
- March 2016; Currently adding co-sponsors to the bill

WV HB 4526

Establishing an Advisory Council on Rare Diseases

Establishes a Rare Disease Advisory Council to advise state agencies on research, diagnosis, treatment and education relating to rare diseases. The council will be made up of 10 members, which include: the Secretary of the Dept. of Health and Human Resources, or a designee; 9 members shall be appointed by the Governor, including: 3 physicians with experience in rare disease, 3 people over the age of 18 who have a rare disease or are a family member of a person with a rare disease; a registered nurse or APRN with experience treating rare diseases; 1 person with an advanced degree in public health or a health-related field who has experience in medical research; a representative from a patient-based organization/advocacy group; the President of the Senate and the Speaker of the House will each appoint one senator and delegate. Duties of the council include: coordinate statewide efforts for the study of the incidence of rare disease within the state; research and identify priorities relating to the quality of and access to treatment and services provided to persons with rare diseases; develop policy recommendations relating to the quality of and access to treatment and services provided to persons with rare diseases.

Primary Sponsor
- Rep. Matthew Rohrbach (R)

Co-Sponsors
- Rep. Christopher Stansbury (R)
- Rep. Larry Faircloth (R)

Rep. Bradley White (R)
- Rep. Theresa Waxman (R)
- Rep. Denise Campbell (D)
- Rep. Amy Summers (R)
- Rep. Joe Ellington (R)

Latest Action
- The bill was sent to the House Finance Committee

Similar Bills
- WV SB 606- Senate version of the above bill

KY SCR 109

Designate February 29, 2016 as Rare Disease Day:
This measure would proclaim February 29, 2016, as Rare Disease Day in Kentucky

Primary Sponsor
- Senator Ray Jones (D)

Latest Action
- Bill sent to senate committee on state & local government committee

MA H 1977

An Act To Create A Massachusetts Rare Disease Advisory Council

The Commissioner of the Dept. of Public Health shall appoint a rare disease advisory council to coordinate statewide effort for the study of the incidence of rare disease with in Massachusetts and the status of rare disease community.

Primary Sponsor
- Representative Paul Heroux (D)

Co-Sponsors
- Rep. James Cantwell (D)
- Rep. Josh Cutler (D)
- Rep. Viriato DeMacedo (R)
- Rep. Geoffrey Diehl (R)
- Rep. Benjamin Downing (D)
- Rep. Gloria Fox (D)
- Rep. Denise Garlick (D)
- Rep. Joseph McKenna (R)
- Rep. Paul Murfley (D)
- Rep. Michael Moore (D)
- Rep. Tom Sannicandro (D)
- Rep. Frank Smizik (D)
- Rep. Chris Walsh (D)

Latest Action
- March 2016; Senate Concurred
**NJ A 3137**

Establishes the New Jersey Rare Disease Advisory Council

- Established a 16 member advisory council in the Dept. of Health that will assist the Commissioner of Health in establishing and implementing a rare disease registry program; conduct thorough and comprehensive study of all issues relating to the quality and cost-effective of, and access to, treatment and services provided to persons with rare diseases in NJ and to develop policy recommendations from these studies.

- **Primary Sponsor**
  - Rep. Ronald Dancer (R)

- **Latest Action**
  - Introduced and referred to the Asm. Health and Senior Services Committee

- **Similar Bills**
  - NJ S 914
  - NJ S959

**TN SB 2084**

Access to Pediatric Rare Disease Treatment Information Act

- Provides for sharing of essential treatment information for children with cancer among certain health care institutions.

- **Primary Sponsor**
  - Sen. Doug Overbey (R)

- **Latest Action**
  - March 2016; Assigned to General Subcommittee of Senate Government Operations Committee

**RI SB 236**

An Act Relating To Human Services—Rare Disease Community Support, Resource Coordination, And Quality Of Life Act Of 2015

- Establishes the Rhode Island Rare Disease Community Advisory Council which would ensure that state-of-the-art information on rare disease education, treatment, and access to care is available to health care providers and survivors. This advisory council will serve as a consensus group designed to coordinate efforts in state resources, private entities, and social services, including bringing additional monies to the state to fund improvements in the treatment of rare diseases.

- **Primary Sponsor**
  - Sen. James Doyle (D)
  - Sen. V. Susan Sosnowski (D)
  - Sen. Joshua Miller (D)
  - Sen. Donna Nesselbush (D)

- **Latest Action**
  - May 2015; committee recommended measure he held for further study
FOR IMMEDIATE RELEASE

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

[CITY], [STATE]—[DATE]—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.

###

Media contacts:

[If applicable, insert your organization’s media officer with contact information]
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 __________________________, 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 @Global Genes

Doctors need more office visits to adequately address a rare disease patient’s symptoms. #WRDD2016 #CareAboutRare @Global Genes

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

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

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

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

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

Rare diseases require more resources from doctors, their staffs. #WRDD2016 #CareAboutRare @Global Genes

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

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