NEWS FROM NORD

AN UPDATE FOR OUR MEMBERS AND FRIENDS FALL 2010

I AM 1 IN 10

One of NORD's consistent messages is that rare diseases aren't really rare. When considered together—and they should be, because there are significant challenges that all people with rare diseases have in common—they affect nearly 30 million Americans or almost 1 in 10 of us. NORD's logo illustrates the 1 in 10 theme. So does our current annual report, which features photos of several people who have rare diseases. To read the stories behind those photos, go to page 4.

NORD TO HIGHLIGHT MEDICAL FOODS ISSUE

On February 10, 2011, NORD will host a conference in Washington, DC, to focus attention on the fact that many families are struggling to provide the medical foods and special formulas needed by children and adults with certain metabolic diseases. In many cases, without these special foods, the patients would develop serious, even life-threatening medical problems. But because these foods are not considered medicines, insurers (even Medicare and Medicaid) don't always reimburse for them. This is a serious problem. NORD plans to bring together the experts who can focus attention on it.

SOCIAL SECURITY TO ROLL OUT MORE COMPASSIONATE ALLOWANCES IN 2011

NORD hosts a monthly teleconference for its 150 Member Organizations. These conferences feature speakers on a variety of topics of interest to leaders of rare disease patient organizations. The speaker for the October teleconference was Nancy Schoenberg, Director of Compassionate Allowances and Disability Outreach for the Social Security Administration (SSA). She provided some important information for people seeking disability assistance through SSA...and a prediction that the Compassionate Allowances list will be expanded in 2011. Continued on page 7.

HELP NORD PROMOTE RARE DISEASE CAUCUS

NORD 2010 Gala

In the early months of 2010, NORD identified an unmet need for Americans with rare diseases: There did not yet exist in the U.S. Congress a specific forum for the exchange of ideas and concerns related to rare diseases and meeting the needs of patients and families. Today, such a forum exists. Together with allies in Congress as well as the broader advocacy community, NORD succeeded in creating the Rare and Neglected Diseases Caucus, and its launch was announced at the NORD Gala in May of 2010. Now, we need your help to make this caucus truly effective. Continued on page 2.

WHAT'S HAPPENING IN WASHINGTON?

BY JASON BARRON, NORD ASSOCIATE DIRECTOR OF PUBLIC POLICY

Researching a treatment for a rare disease and seeing that treatment through to market is no small effort. All therapies that successfully make their way from good ideas in a laboratory to available options for patients have been put to task many times over. The situation becomes even more challenging when patient populations are small and additional constraints are made on researchers, industry, and the regulatory agencies charged with protecting consumers.

TASK FORCE WITH NIH AND FDA

This is precisely why NORD assembled a task force with the National Institutes of Health (NIH) and the Food and Drug Administration (FDA) to review and assess the development process for orphan products, from potential target identification to post-approval marketing and consumer availability.

In early January of 2010, NORD, NIH, and FDA embarked upon a historic collaborative project to identify barriers to orphan product development and collectively explore their resolution. This task force also addresses a key recommendation of the Institute of Medicine's recent report, Rare Diseases and Orphan Products: Accelerating Research and Development, released October 4, 2010.

FOCUS GROUPS

As its first action, the task force resolved to undertake a comprehensive review of orphan product development. As part of that process, NORD commissioned a qualitative assessment of key stakeholders' views in the form of four focus groups. The constituencies represented by the focus groups were as follows:

- Academic Medical Researchers
- Patient Advocates
- Investors and Venture Capitalists
- Industry Executives from the Pharmaceutical and Biotechnology Sectors

A clear need emerged from the findings of this review. To date, there is no guide available for clinical investigators that focuses on the challenges specific to the development of orphan products. Additionally, the practical roles and resources of NIH, FDA, and patient advocacy groups in the development process for rare disease therapies are not widely known. Patients benefit when investigators and industry are well informed of what the regulatory expectations are for a proposed rare disease therapy.

The NIH, FDA, NORD task force is currently drafting a handbook that will better inform researchers. Its primary aim is illuminating how best to navigate the process of getting a new therapy to market—be it a drug, biologic, or medical device. The handbook combines the input of all three groups in the interest of being as comprehensive as possible regarding the clinical and regulatory aspects of orphan products development. While the target audience is investigators, it will likely be useful to many others who also have a stake in the development of orphan products.

EXPANDING OUR HORIZONS WITH GLOBAL COLLABORATION

Thinking globally makes good sense in many aspects of life, but perhaps nowhere is it more appropriate than with rare diseases. With few resources and patients scattered around the world, it's clearly the best way to drive progress in patient services and developing treatments.

NORD has long had a friendly, collaborative relationship with the European Organization for Rare Diseases, EURORDIS. However, in October 2009 we formalized that relationship by signing a strategic partnership to work together in key areas related to education, awareness, and advocacy.

One of the first co-sponsored projects will be online patient communities. These communities will provide safe online environments hosted by trusted partners (NORD, EURORDIS and their disease-specific partners) where patients can share information, learn from the experts, and describe their own experiences with rare diseases.

A pilot community on CAPS (cryopyrin-associated periodic syndromes) may be viewed at www.rarediseasecommunities.org. It was unveiled at the recent Health 2.0 Conference in San Francisco and received a good response. NORD and EURORDIS hope to launch communities on additional rare diseases in 2010, and many more next year.

NEW CONGRESSIONAL CAUCUS: HOW YOU CAN HELP

Working with the broader advocacy community, NORD has helped establish a Rare and Neglected Diseases Congressional Caucus. This new caucus presents a tremendous opportunity for the rare disease community. However, it does not yet have robust membership or a policy agenda that can drive positive, meaningful legislation.

NORD is committed to a caucus that has both, and accomplishing this is of the highest priority. A determined and bipartisan Rare and Neglected Diseases Caucus can result in a Congress that is better educated about this constituency and more capable of addressing the issues.

NORD is encouraging all its members and friends to ask their representatives in Congress to join the caucus. We'll post a list on NORD's website—and update it regularly—of all members of the Caucus.

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NORD
National Organization for Rare Disorders

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NORD GOES SOCIAL

SHARING OUR MESSAGE IN THE NEW SOCIAL MEDIA FORUMS

Tai Spargo
Special Projects Coordinator

A recent story on the ABC News website pointed to NORD's Facebook page as a good example of how the social media can be especially helpful to people with rare diseases. The reporter told NORD that many patients she interviewed for the story had recommended NORD's Facebook page as a good place to connect with others experiencing similar problems and challenges.

A year ago, NORD didn't have a presence on Facebook. However, we decided it was important to reach out to patients, families, and the public through these new media, while still maintaining our traditional avenues of communication. We did this in the hope of reaching a broader audience with our key messages while also helping patients network and share helpful information with each other.

Today, NORD hosts a rare disease patient community on Inspire.com. We also have Facebook and Twitter pages, both for NORD itself and for the Rare Disease Day US community. And we are creating new global online patient communities with our European partner, EURORDIS. (See related story on page 2.) NORD's social media presence is overseen by Tai Spargo, Special Projects Coordinator..

Our efforts are paying off: In October, NORD and Siren Interactive, a company that partnered with NORD in a Rare Disease Day awareness project, were honored at the prestigious MM & M (Medical Marketing and Media) awards in New York City for effective use of the social media.

In all of these efforts, our goal is the same as it was years ago when the postman arrived at the NORD offices with bags of mail each day: to connect people who feel isolated and to help patients and families find patient organizations, medical experts, clinical trials, and other resources specific to their needs.

GOT A QUESTION? WE MAY BE ABLE TO HELP

NORD has an Information Service dedicated to helping patients and families. The staff includes Information Specialists who are knowledgeable about patient organizations and a broad range of other resources. These staff members help patients and their families find clinical trials, medical experts, financial assistance to obtain needed medications, and many other resources.

They are also aware of many government resources such as the NIH Genetic and Rare Diseases Information Center (GARD), which provides excellent service to patients and families seeking information about specific diseases.

The staff includes a genetic counselor and a registered nurse, both of whom have many years of experience in their fields. Anyone with a question or concern may call toll free (800-999-NORD) or write to genetic_counselor@rarediseases.org or RN@rarediseases.org. While the Information Service staff may not be able to help in every situation, they will certainly give it their best effort.

Marsha Lanes, MS, CGC
NORD Genetic Counselor

Stefanie Putkowski, RN
NORD Nurse

HELP US TEACH OTHERS ON RARE DISEASE DAY 2011

NORD is already making plans for the national observance of Rare Disease Day 2011. Visit the Rare Disease Day US website (www.rarediseaseday.us) for frequent updates and ways you can get involved in this observance both on the actual day (Feb. 28, 2011) and in the weeks leading up to it.

Rare Disease Day 2011 activities will include:

- Creating a library of short (two minutes or less) videos about specific rare diseases that will answer three questions: What is the name of my disease? How does it affect my daily life? And what do I do to overcome it?
- A major national conference hosted by NORD on the needs of patients related to medical foods
- A survey to be done by NORD and the PEW Internet and American Life Project related to rare diseases and the Internet
- Creating a database of physicians who are experts in specific rare diseases

Rare Disease Day

Rare Disease Day provides a wonderful opportunity to teach the public—and professionals—about specific diseases and to convey that rare diseases are an important public health concern receiving too little attention around the world.

I AM 1 IN 10

YOU'LL SEE THESE FACES IN NORD'S 2009 ANNUAL REPORT. HERE ARE THE STORIES BEHIND THE PHOTOS.

I AM BRIAN DREXLER

Brian Drexler loves travel, music and sports, including volleyball, golf, soccer, and snowshoeing. He is a Special Olympics athlete who enjoys the camaraderie of events that allow him to meet many different people.

Brian has a condition known as Moebius syndrome, and he is a member of the Moebius Syndrome Foundation—one of NORD's Member Organizations. Brian and his family participate in many activities organized by that foundation.

Moebius syndrome is a rare neurological disorder that is present at birth. It primarily affects the sixth and seventh cranial nerves leaving those with the condition unable to move their faces. They can't smile, frown, or blink their eyes. They also can't move their eyes from side to side.

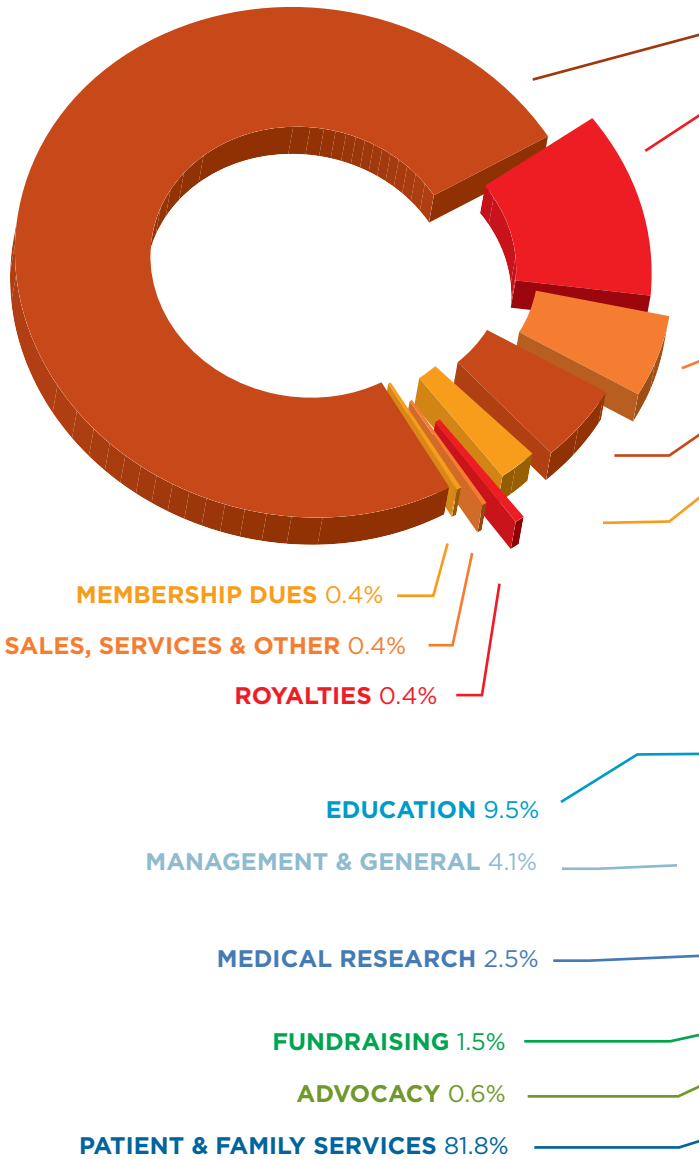
Other cranial nerves may be affected, and the condition may result in other medical problems. As children with this condition grow older, however, the lack of facial expression and inability to smile may be the primary visible symptoms.

No one knows for sure what causes Moebius syndrome, but it is thought to be genetic. Most cases appear to be sporadic, occurring once in a family. Research is being conducted around the world on the causes of this syndrome and possible treatments.

A procedure known as "smile surgery", in which a segment of muscle is removed from the thigh and implanted into the face, may help some affected children smile. In 2007, three physicians conducting research on the origin and development of Moebius syndrome were awarded the Nobel Prize in Medicine for their work.

2009 Revenues a

REVENUES



I AM BERNADETTE SCARDUZIO

Both her father and grandfather had the same rare condition that Bernadette Scarduzio has, but they were affected in a milder way. For Bernadette, the disorder progressed quickly and has significantly affected her life.

Her condition is a neurological disorder known as Charcot-Marie-Tooth (CMT) disease, named for three physicians who first identified it in 1886. This disorder is also known as hereditary motor sensory neuropathy (HMSN). Symptoms typically first appear in adolescence or early adulthood. They include pain; muscle weakness and atrophy affecting the feet, legs and hands; a distinctive gait; and frequent falls.

"I've had some tough times but it's made me stronger," says Bernadette, who serves as a national spokesperson for the Hereditary Neuropathy Foundation. With friends, she is producing a documentary to show how this condition impacts the daily lives of those affected.

CMT is caused by mutations in genes that produce proteins that help nerve cells communicate with distant muscles such as in the arms, legs, hands, and feet. There is no cure for it at this time, but treatment—including physical and occupational therapy, braces and other orthopedic devices, and orthopedic surgery—may help.

Though she is just 31 years old, Bernadette has had 20 surgeries over the years on her ankles, feet, toes, and hands. A year and a half ago, she began using a motorized scooter that gives her greater mobility.

I AM BRYN WELTON

Bryn and Pat Welton, both originally from Wales, will celebrate their 50th wedding anniversary next year. The observance will be especially poignant because the couple's loving partnership has included facing together the challenges brought on by a disabling progressive disease that Bryn was found to have when he was just 37 years old.

As a youth and young man, Bryn loved competitive sports, including rugby and squash. While playing squash one day in 1973, he went for a shot but fell to the ground when his knee mysteriously failed to support him. That was his first indication that something was wrong.

Bryn's muscle weakness grew steadily worse, and eventually he was diagnosed with primary progressive multiple sclerosis. Most people with MS have periods of remission. For the small percentage with Bryn's type, however, the disease is steadily progressive.

In spite of this, Bryn traveled the globe for many years as a trouble-shooter for his company, Perkin Elmer, fixing problems related to the chemical side of instrumentation. Pat kept things going on the home front and cared for the couple's two sons, the younger of whom was just three when Bryn first got his diagnosis.

Although he required a wheelchair, Bryn drove himself to work each day and managed all the logistics related to world travel. In fact, in many respects his career is a metaphor for his triumph over MS.

"I've always been a problem-solver," Bryn says. "If I can't do something the way I've done it in the past, I figure there must be another way."

nd Expenses

GRANTS & RESTRICTED CONTRIBUTIONS 74.7%

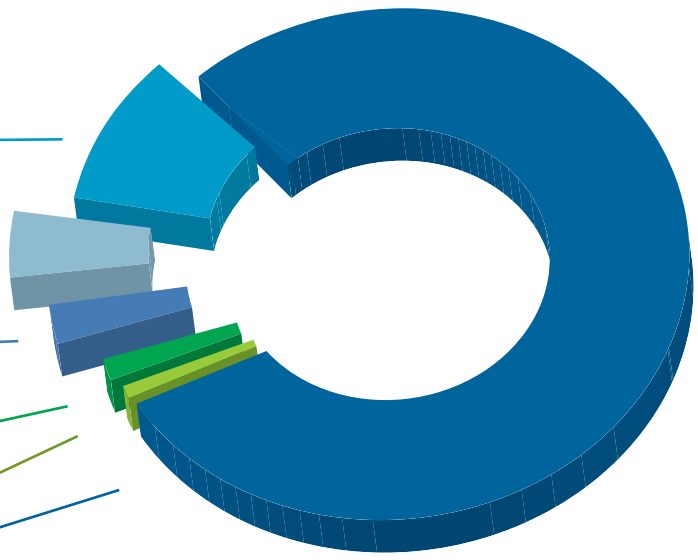
ADMINISTRATIVE FEES – PATIENT PROGRAMS 11.5%

CONTRIBUTION 5.4%

FUNDRAISING EVENTS 4.8%

INVESTMENT INCOME 2.4%

EXPENSES



I AM MARTHA BRYCE

Martha Bryce was 32 years old when she had her first-ever "grand mal seizure". "Not unusual," her doctor said. "Many people have a one-time seizure, never to be troubled again."

But then Martha developed another perplexing symptom: When eating, she would often feel faint after the first few bites, nearly black out, and then feel fine again. If Martha hadn't been a nurse—and found an organization known as STARS—she might never have gotten to the truth.

The seizure occurred while Martha was on vacation and visiting the Hoover Dam. Other symptoms, including briefly losing consciousness while eating, began soon afterward. It happened so frequently that she developed a coping mechanism. "I would lower my head as soon as I began to feel lightheaded," Martha says. "My husband and co-workers became accustomed to seeing me go through this strange ritual during meals."

Eventually, the lightheadedness led to actual loss of consciousness, and Martha had a second seizure. At that point, she was given a diagnosis of epilepsy, but the pieces of the puzzle still didn't quite seem to fit together.

One day, Martha happened upon the website of the STARS organization and that was the beginning of her journey to a correct diagnosis. Eventually, she learned that she had a condition known as "swallowing syncope" in which the act of swallowing triggers a chain of events that causes the heart to stop beating briefly.

A pacemaker stopped the symptoms completely. Martha shared her story with NORD for Rare Disease Day 2009. Since then, she has been featured in the Washington Post's Medical Mysteries column and on the Mystery Diagnosis TV show. Martha hopes that telling her story may help others obtain a correct diagnosis with less delay.

I AM PAIGE BOURHILL

When Paige Bourhill visited the NORD office in Connecticut recently with her mother and grandfather, every member of the staff wanted to meet her. That is because Paige's family's search for a diagnosis tugs at the heartstrings of all who hear about it. And, sadly, lack of a diagnosis is not an uncommon problem in the rare disease community.

When Paige was born on August 4, 2005, she was a beautiful and healthy baby. In fact, her parents, Annette and Dave, called her "super baby" because she never got sick.

That all changed on June 18, 2006. On that day, Paige developed a fever, and her parents were told by her pediatrician that she had a viral infection, should be kept hydrated, and needed to rest. For five days, she slept almost continuously. On the sixth day, she awoke with no fever but experienced a seizure. In the emergency room, a CT scan revealed lesions in her brain.

Since then, Paige has had eight MRIs, two muscle biopsies, four spinal taps, hundreds of blood tests, numerous EEGs, and many other tests. She underwent evaluation through the NIH Undiagnosed Diseases Program. But, while Paige's family now knows a little more about her specific symptoms, they don't know what transformed their healthy baby into a child with multiple medical problems.

Paige is now four years old and does not walk or talk. However, she is blessed with parents and a younger brother who will never give up on her and who will fight to help her achieve her greatest potential. Paige and her family represent a large community of Americans who are believed to have rare diseases but don't have the comfort of being able to put a definitive name on their disease.

I AM DANIEL AZIERE

Dan Aziere had his first surgery at age five, and he's had 18 additional surgical procedures since then. Dan has a rare genetic condition known as multiple hereditary exostoses (MHE), which results in the growth of benign bone tumors.

These bony growths may cause pain, limited range of motion, and pressure on nerves, blood vessels, the spinal cord, or surrounding tissues. While some people are only minimally affected, others require frequent surgeries, especially during childhood and the teen years when the bones are growing. While the condition is sometimes inherited from an affected parent, there is no family history in some cases.

As a teenager, Dan went through a brief period of depression, troubled by teasing about the "bumps" on his legs or the way he walked.

Today, however, with strong support from his family and the realization that things could always be worse, Dan finds himself in a unique position to help others put life's troubles in perspective. He works as an insurance claims adjuster and frequently meets with people who have just gone through an automobile accident.

"When we first start talking, they're upset," Dan says. "They're having a bad day. But after we talk for a bit, they usually leave feeling a lot better about their situation."

Although Dan has never met another person with MHE, he's been helped by connecting through the Internet with an organization known as MHE Coalition. More than 1,600 families in 57 countries around the world are registered with the Coalition as having MHE.

NORD TO FOCUS ON MEDICAL FOODS ISSUE

BY ANA MARIA VALLARINO, NORD NATIONAL URBAN FELLOW FOR 2010

Monaco family, with Stephen in wheelchair and Caroline behind him

Medical food treatment, food intended for specific dietary management of a disease or condition for which there are distinctive nutritional requirements, is the difference between a healthy, full-functioning child and a severely disabled child who requires a wheelchair, is unable to communicate verbally and is fed through a gastrostomy tube. This is the difference between siblings Stephen Monaco (age 13) and Caroline Monaco (age 8).

Stephen and Caroline both have an inborn error of metabolism, a disease called isovaleric acidemia. Stephen, unfortunately, was not diagnosed at birth. He was born a seemingly healthy baby and remained that way until the age of three when he had a metabolic crisis that left him physically and mentally disabled. Caroline, screened at birth, was detected to have isovaleric acidemia. Her disorder has been monitored and treated through medical foods ever since.

Newborn screenings have made progress in the last decade with various states now screening for all 29 disorders recommended by the American College of Medical Genetics. And although there is still work to be done around newborn screening, the other critical half of treatment lacks so much attention.

Treatments, such as medical foods, allow for individuals like Caroline to grow and lead fulfilling lives. However, only 28 states have successfully passed legislation mandating that medical foods be made available. Unfortunately, the cost of purchasing medical foods is extremely high, and may be prohibitive for many families.

You are invited to attend a medical foods conference that NORD will host in February 2011, in Washington, DC. This will be an exciting opportunity to interact with key stakeholders and add to the timely discussion on addressing gaps in access and reimbursements for individuals who need treatment through medical foods. Participants will include medical experts, medical food production companies, federal agencies, public and private payers on the state level, and patient advocates.

WATCH FOR NEWS ON
COMPASSIONATE ALLOWANCES

Speaking at NORD's regular monthly teleconference for its 150 Member Organizations, Nancy Schoenberg of the Social Security Administration said additional public hearings are planned for 2011 to identify more conditions to add to SSA's Compassionate Allowances list. This is a list of a diseases so disabling that those who have them qualify for expedited review when they apply for Social Security disability assistance.

Ms. Schoenberg, who oversees the Compassionate Allowances program, said that public hearings are planned on two general topics: cardiovascular diseases, including rare congenital heart defects, and autoimmune diseases.

She also assured the patient leaders on the teleconference that SSA continues to review requests to add diseases to the list on an ongoing basis. Patient organization leaders who feel their diseases may qualify for this may submit a request directly to Ms. Schoenberg (Nancy.Schoenberg@ssa.gov) or to NORD staff members, who will then forward the requests to Ms. Schoenberg.

Michael Astrue established the Compassionate Allowances program soon after he was named Commissioner of Social Security in 2007. He did this because of his awareness, through his own family's experience, of delays in the review of applications, associated with the fact that those conducting the review may not be familiar with the rare disease in question.

NORD worked closely with Mr. Astrue and his staff in the establishment of the Compassionate Allowances program, and later helped SSA expand the initial list of 50 rare diseases and cancers with an additional group of 38 rare diseases. Ms. Schoenberg said she anticipates that additional diseases may be added to the CA list in the summer of 2011. Review the current CA list at www.ssa.gov/compassionate allowances

NORD Medical Advisor Honored by U.S. House

The U.S. House of Representatives recently approved a resolution (H.Res.1499) honoring Robert M. Campbell Jr., MD, for providing children with life-saving medical care. Specifically, Dr. Campbell, who is a member of NORD's Medical Advisory Committee, was honored for his role as the inventor and developer of the expandable titanium rib. This medical device has saved the lives of hundreds of children with a rare condition known as thoracic insufficiency syndrome.

Congresswoman Debbie Wasserman Schultz (D-FL) sponsored the resolution. Co-sponsors Frank Pallone (D-NJ) and Phil Gingrey, MD (R-GA) also helped present the resolution for approval on the House floor.

Dr. Campbell is an orthopedic surgeon at Children's Hospital of Philadelphia. He has been a medical advisor to NORD for about 10 years.

PUBLIC HEARINGS
SPOTLIGHT RARE DISEASES

NORD representatives have testified at several key public hearings in recent months. The following are excerpts from news stories published following:

- a public hearing hosted by the Food and Drug Administration (FDA) to get input on FDA policies and procedures and
- a hearing hosted by the U.S. Senate HELP Committee on how to accelerate the development of treatments for rare pediatric diseases.

NORD CALLS FOR STATEMENT OF POLICY ON ORPHAN DRUG DEVELOPMENT

Noting that only about 200 of the nearly 7,000 diseases classified as rare currently have treatments, the National Organization for Rare Disorders (NORD) today called for a Food and Drug Administration (FDA) statement of policy on regulation of therapies for rare disorders.

NORD also urged FDA to work with the research community to reduce regulatory uncertainty in the development of medicines for rare disorders.

"There are still almost 6,800 rare disorders for which there are no FDA-approved therapies," said Frank J. Sasinowski, chairman of the NORD Board of Directors, in the opening address at a two-day public hearing sponsored by FDA. "Perhaps most discouraging is that many affected with these rare disorders do not even see any research being conducted on their conditions."

Referring to the Orphan Drug Act of 1983, Sasinowski said that more than 350 new treatments for rare diseases have been approved by FDA since that law was enacted. This is good progress, he said.

"But to NORD and the patient community," he added, "it seems as if the low-hanging fruit have been harvested. While much has been accomplished, much more remains to be done."

NORD TELLS SENATE: PARENTS TOO OFTEN HAVE TO FUND SEARCH FOR THEIR CHILDREN'S TREATMENTS

An advocate for people with rare diseases today told a U.S. Senate committee that the burden of funding and driving research on rare diseases too often falls upon patients and their families.

"As a society, it is wrong for us to expect people with devastating diseases to fund the search for their treatments," said Diane Dorman, vice president for public policy of the National Organization for Rare Disorders (NORD). "There are nearly 7,000 rare diseases, and only about 200 of them have treatments. Many are not being studied by any researcher in government, academic, or industry.

"Through golf tournaments, raffles...even bake sales and car washes, it's too often the patient community that funds and drives rare-disease research. We need a more significant commitment at the federal level."

Dorman said the word "rare" is misleading, since about one in 10 Americans have diseases classified as rare. While each disease is unique, there are many problems and challenges that all people with rare diseases share, she added.

Dorman told the committee that federal funding and guidelines are needed for natural history studies, patient histories and other basic tools to make clinical research possible. And, she said the Food and Drug Administration (FDA) should institute a statement of policy on rare diseases and orphan products to reduce regulatory uncertainty and encourage researchers to develop treatments for diseases that have none.

NORD is a unique federation of individuals and voluntary health organizations dedicated to helping people with rare diseases and assisting the organizations that serve them. NORD is committed to the identification, treatment and cure of rare diseases through programs of education, advocacy, research, and patient services.

To request a copy of NORD's annual report, contact: NORD Development Office, PO Box 1968, 55 Kenosia Avenue, Danbury, CT 06813, (203) 744-0100

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