

# NEWS FROM NORD

AN UPDATE FOR OUR MEMBERS AND FRIENDS

FALL 2011



## ALONE WE ARE RARE. TOGETHER WE ARE STRONG.

NORD is people ... patients, their families, their caregivers, and the dedicated researchers seeking to improve their lives. Through the stories of real people, we present the everyday realities and challenges of living with rare diseases.

Our annual report for 2010 features photos of some of the 1 in 10 Americans who have rare diseases. There are certain themes their stories have in common. While the journey of each individual and family affected by a rare disease is unique, there are many intersecting points at which the 30 million Americans with rare diseases, and millions more around the world, share common goals, challenges and life experiences.

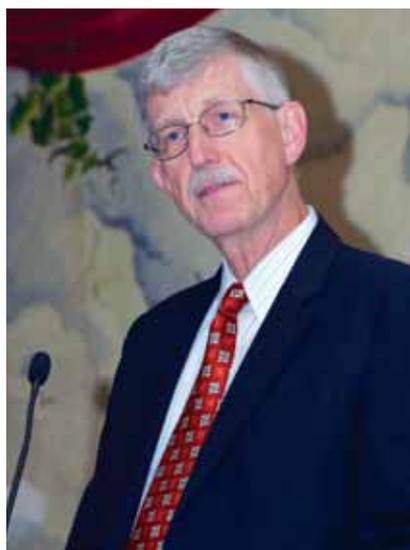
On page 4, you'll find brief stories behind the photos in the NORD annual report. Stories and videos are also posted on the NORD website ([www.rarediseases.org](http://www.rarediseases.org)).



## NORD CONFERENCE JUDGED A MAJOR SUCCESS



Conference Participants



NIH Director Francis Collins, MD, PhD

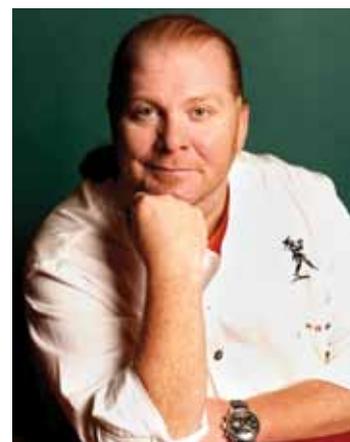
Thought leaders from patient organizations, government, industry, and the research and investment communities exchanged views and shared their expertise at the first-ever U.S. Conference on Rare Diseases and Orphan Products, held October 11-13 in Washington DC. The event drew nearly 400 participants.

"I've attended many conferences over the years but this is the first time I came away feeling that something really significant and good might come of it," one participant noted afterward. That was a sentiment expressed by many. With nearly 80 speakers, including some of the nation's leading experts on the science, economics, and ethics of rare diseases and orphan product development, there was something for everyone.

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## MARIO BATALI FOUNDATION ESTABLISHES RESEARCH FUND WITH NORD

Widely acclaimed chef, restaurateur, and author Mario Batali is well known for his charity work on behalf of children and, in particular, to ensure that children are "well read, well fed and well cared for." Now, the Mario Batali Foundation has established a research fund at NORD to help children who have rare diseases.



**"I feel particularly passionate about protecting and helping these children, whose entire future rests on the vital research and awareness of these rare diseases. Every child deserves a fair shot at a healthy and long life."**

The fund, to be known as the "Mario Batali Foundation Pediatric Rare Disease Research Fund", will raise funds for research and further awareness of rare disorders affecting children. The first Request for Proposals will be announced soon on the NORD website.

"Sadly, children are the most frequent victims of rare diseases," said NORD President and CEO Peter L. Saltonstall, "and many of these diseases are serious and life-altering. NORD is extremely grateful to the Mario Batali Foundation for its desire to help these children and their families."

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# CAPITOL HILL STAFF BRIEFED ON NEED FOR RARE DISEASE CAUCUS



Moderator Timothy Coté, MD, MPH, with parent panelists



Catherine Collins describes her family's experience with Duchenne muscular dystrophy.

A Capitol Hill briefing for congressional staff members on the purpose of the Rare Disease Congressional Caucus drew a standing-room-only crowd in the Rayburn House Office Building on November 9. The event, hosted by the caucus co-chairs with help from NORD and the Rare Disease Legislative Advocates, was to educate legislative staff members so they could encourage their bosses – members of Congress – to support the caucus.

Moderated by NORD's Chief Medical Officer, Timothy Coté, MD, MPH, the briefing featured short presentations by Anne Pariser, MD, of the Food and Drug Administration (FDA), and Stephen C. Groft, PharmD, of the National Institutes of Health (NIH), followed by several descriptions by parents of the devastating effects specific rare diseases have had on their children's lives.

The staff briefing was just the latest development in the caucus's one-and-a-half year history. There are more than 300 caucuses (also known as Congressional Membership Organizations) in the U.S. Congress. They focus on issues ranging from protecting America's fisheries to exploring space. Each must be renewed for each new two-year session of Congress.

Earlier this year, at the start of the 112th Congress, the Rare Disease Caucus was renewed and, at the same time Rep. Fred Upton (R-MI) stepped down from his role as the Republican co-chair of the caucus because he was assuming chairmanship of the House Committee on Energy and Commerce.

Rep. Upton passed the torch to Rep. Leonard Lance (R-NJ) to serve as the GOP co-chair of the renewed "Rare Disease Caucus." Rep. Joseph Crowley (D-NY) resumed his role as the caucus's co-chair for the Democratic Party. Known as the Rare and Neglected Diseases Caucus when it was first established, the caucus was renewed as simply the Rare Disease Caucus.

As this article went to press, the Rare Disease Caucus stood at a strong 32 members of the House of Representatives. Additionally, four senators have made a commitment to the rare disease community. Each member joins the caucus with a willingness to be engaged and search for legislative solutions to the problems facing patients and their caregivers.

A list of caucus members is posted on the NORD website. (Click on Advocacy at the top of the home page.) NORD encourages its members and friends to check this list periodically and, if their U.S. Representative is not on it, to write to him or her about the importance of joining the caucus. Tips for contacting your members of Congress can also be found on the NORD website.

## GEARING UP FOR RARE DISEASE DAY 2012



Rare Disease Day

Rare Disease Day 2012 will mark the 5th global observance of this special day and will also fall on February 29th – the ultimate rare day! As the national sponsor for Rare Disease Day, NORD is planning an observance that will be bigger and better than ever.

Rare Disease Day was launched in Europe in 2008 by EURORDIS. The following year, EURORDIS asked

NORD to sponsor this event in the U.S., and since then, it has spread around the world. In 2011, more than 60 nations observed Rare Disease Day.

Highlights of Rare Disease Day 2011 included a resolution by the U.S. Senate and proclamations or resolutions by governors or legislatures of all 50 states. More than 550 patient organizations, government agencies, health-related institutions, and companies developing treatments for rare diseases signed on as "Rare Disease Day Partners", indicating their support of the day. Individuals who did not have an organization signed on as Rare Disease Day Ambassadors.

The official U.S. website for Rare Disease Day ([www.rarediseaseday.us](http://www.rarediseaseday.us)) will launch very soon with the new campaign for RDD 2012. Activities and events planned for this year will include:

- A media blitz of patient stories, photos, interviews, and videos to raise awareness of what it's like to live with a rare disease
- A "Handprints Across America" campaign in which supporters will be encouraged to download the Rare Disease Day logo, which resembles a hand, and submit photos of themselves holding or wearing the logo at locations around the U.S. for a photo gallery to be posted on the RDD website
- Educational curricula and resources for teachers of high school, middle school, and elementary school students
- A press kit and other tools to help patient organization leaders and others share the global themes of Rare Disease Day
- A three-day event, to be announced soon, in Washington DC for patient organization representatives and others that will include special programs at NIH and FDA

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President . . . . . Peter L. Saltonstall

Vice President for Communications . . . . . Mary Dunkle

Art Direction & Design. . . . . SandorMax Marketing

For more information, contact us at: 203.744.0100 / [orphan@rarediseases.org](mailto:orphan@rarediseases.org)



**NORD**  
National Organization for Rare Disorders

# NORD CONFERENCE JUDGED A MAJOR SUCCESS

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## Speakers Include NIH Director and Social Security Commissioner

Keynoters such as National Institutes of Health (NIH) Director Francis Collins, MD, PhD, and Social Security Commissioner Michael Astrue addressed general sessions. In addition, there were breakout sessions in three tracks:

- Patient Organizations
- Industry/Investors
- Rare Disease Researchers

Tracks 1 and 2 focused on topics of particular interest to patient advocates and those involved in developing orphan products or investing in the life sciences. Track 3 was a training course for rare disease investigators developed last year by Anne Pariser, MD, of the Food and Drug Administration (FDA). Dr. Pariser is Associate Director for Rare Diseases in FDA's Center for Drug Evaluation and Research (CDER) Office of New Drugs.

## Major Study Released at Conference



Frank Sasinowski

In the opening session, NORD Board Chair Frank Sasinowski presented findings from a major new study conducted by NORD to determine whether, and in what ways, FDA may have applied flexibility in its review of non-cancer orphan drugs approved since the 1983 Orphan Drug Act. (See related story on page 5.) Later, Sasinowski further discussed the study in a press briefing.

Another highlight of the opening session was the introduction of the newly appointed Acting Director of FDA's Office of Orphan Products Development, Gayatri Rao, MD, JD, who spent her first days on the job at the conference. Dr.

Rao told the audience she could think of no better orientation than to participate in such an event drawing together the various stakeholder groups.

In addition to Dr. Collins and Commissioner Astrue, general session speakers included:

- **Fred Hassan**, Chairman of the Board, Bausch & Lomb
- **Janet Woodcock**, MD, Director of FDA CDER
- **Jeffrey Shuren**, MD, JD, Director of FDA's Center for Devices and Regiological Health
- **Mark McClellan**, MD, Director of the Engelberg Center for Health Care Reform at the Brookings Institution
- **John Crowley**, Chairman and CEO of Amicus Therapeutics whose family was the subject of the movie *Extraordinary Measures*
- **Christopher Austin**, MD, PhD, Scientific Director, NIH Center for Translational Therapeutics

## Building Strong Patient Leadership A Highlight

A workshop for patient representatives on the second day focused on building strong patient organizations and helping patients participate more fully in the search for treatments. Day two also featured a session for industry representatives on "How to Select/Vet Candidate Diseases" and one on "International Markets and Product Development".

Speakers in sessions of particular interest to patients included:

- **Kari Luther Rosbeck**, President and CEO, Tuberous Sclerosis Alliance
- **Ronald J. Bartek**, President and Co-Founder, Friedreich's Ataxia Research Alliance
- **Ann Brazeau**, VP of Development, MPN Research Foundation
- **Stephen C. Groft**, Director, NIH Office of Rare Diseases Research
- **Margaret Anderson**, Executive Director, FasterCures

“Drug development for rare diseases is different and requires flexible regulatory standards. We need uniform articulation and application of these policies.”

– Janet Woodcock, MD,  
Director of FDA Center for Drug Evaluation and Research

The Rare Disease Investigators Training Course was taught largely by team and division leaders from FDA. In addition to many others, speakers included:

- **Robert Temple**, MD, Deputy Center Director for Clinical Sciences in FDA CDER
- **Marc Walton**, MD, PhD, Associate Director for Translational Sciences, FDA CDER
- **Thomas Fleming**, PhD, Professor of Biostatistics, University of Washington

## Social Security Commissioner Makes Surprise Announcement

On the third and final day of the conference, Social Security Commissioner Michael Astrue announced that 13 additional conditions – all of them rare – were being added to the Compassionate Allowances list. This is a list of diagnoses so disabling that patients who have them qualify for expedited review of applications for Social Security disability benefits. NORD and its medical advisors have assisted in the creation of this list, as have rare disease experts at NIH and others. (See related story on page 7)

## Related Conference to Take Place in Europe in May



Yann Le Cam

Yann Le Cam, CEO of EURORDIS, announced that a similar conference will take place in Europe in the spring as part of the partnership between EURORDIS and NORD. The two conferences are expected to become annual events, one in the U.S. in the fall and one in Europe in the spring. In 2012, the European conference will take place in May in Brussels.

The U.S. Conference on Rare Diseases and Orphan Products was co-sponsored by NORD and DIA (Drug Information Association), a nonprofit global professional society, with collaboration from FDA, NIH, EURORDIS and the Duke University School of Medicine.

# ALONE WE ARE RARE. TOGETHER WE ARE STRONG.

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## I AM LULU I HAVE PARANEOPLASTIC SYNDROME (PNS)



*"My husband, Robert, calls my rare disease a 'brutal and unkind disease.' But it all began with very mild symptoms – numbness in three of my toes."*

Lulu was walking the family's dog in the backyard of her suburban home when she noticed her first symptoms: numbness in the three smallest toes on her left foot. She was 44 and had never experienced any significant health problems. Over time, Lulu developed increasing

numbness, stiffness and pain, and after much searching she was given a diagnosis of "stiff person syndrome."

Later, it was determined that she had breast cancer and that her neurological symptoms actually resulted from her body's response to the presence of cancer. While her cancer treatment appears to have been successful, she has lost her hearing and mobility as a result of PNS. Lulu's journey with this rare syndrome has been fraught with difficulty, but Robert has been her constant advocate. Everyone who meets this remarkable couple is struck by their devotion to each other.

## I AM IAN I HAVE LANGERHANS CELL HISTIOCYTOSIS



*"I'm doing well now but my mother says our family has been 'forever changed' by my rare disease."*

Ian was diagnosed at age 15 months with Langerhans cell histiocytosis, in which too many of this type of white blood cell are produced and build up in various parts of the body. The disease impacts about 1 in 200,000 people each year.

He is now 6. Since his original diagnosis, he has endured 129 IV chemotherapy treatments, 440

doses of oral chemotherapy, 1,252 doses of steroids, 315 x-rays, 10 MRIs, 6 CT scans, 5 PET scans, 7 bone scans, 2 bone marrow biopsies, 22 emergency room visits, and three hospital stays.

"Through it all," his mother says, "our family has been fortunate to find a caring and supportive community through the Histiocytosis Association of America, a NORD member. The histio family is small but active in raising awareness and raising money for research to find a cure for all of our histio warriors!"

## I AM BEN I HAVE CONGENITAL HYPERINSULINISM



*"In the first days of my life, my parents worried about the fact that, even for a newborn, I seemed unusually sleepy and lethargic."*

"My pregnancy with Ben was perfect and perfectly unremarkable," his mother Julie notes. "And Ben seemed healthy at birth, with double nines on his Apgars." But before they left the hospital, Ben became irritable and constantly hungry. At home, he slipped into a lethargic state.

Twenty-four hours after bringing Ben home from the hospital, the family was headed to their pediatrician's office and, from there, to the neonatal intensive care unit. Ben's blood sugar was so low it didn't even register.

After learning that both parents had Eastern European ancestry, an endocrinologist told them about a condition known as congenital hyperinsulinism. Ben is now 15 and has many interests. His mother is the executive director of an organization called Congenital Hyperinsulinism International, founded in 2005. She advises other parents of children with rare diseases to connect with each other and with medical experts. "You don't have to do it all alone," she says.

## I AM GABY I HAVE APS TYPE I



*"There are only about 150 people in the U.S. who have my rare disease. It took 10 years to get a diagnosis."*

Gabriella's family attended their first NORD patient/family conference in 2006, soon after her rare disease was diagnosed. "We learned invaluable lessons from those who had walked the path before us," her father, Todd, says. "You'd be surprised what you're capable of with a little direction and caring family and friends."

APS is a very rare and complex disorder of the immune system that causes multiple endocrine glands to malfunction. Gaby's family adopted a motto: "If it is to be, it's up to me!" And they have lived up to it, creating a website, connecting with medical experts, and raising nearly \$200,000 for research on APS type I with golf tournaments and other events supported by their relatives, co-workers and friends. They have funded three grants – with another to come soon – through the NORD Research Grant Program.

**LEARN MORE AND WATCH PATIENT VIDEOS IN OUR 2010 ANNUAL REPORT BY SCANNING THIS QR CODE, OR BY VISITING OUR WEBSITE.**



# I AM RAYVEN I HAVE CORNELIA DE LANGE SYNDROME



"I have fun playing with the other kids at events hosted by the Cornelia de Lange Syndrome Foundation."

Rayven is nine years old and was born with a genetic condition known as Cornelia de Lange syndrome. It is named for a Dutch pediatrician.

This syndrome is present at birth but not always diagnosed since its effects may be subtle. In Rayven's case, her pediatrician noticed that the 5th digit on her hands and feet was small, a

subtle sign that helped lead to a diagnosis. This condition affects both girls and boys of all races and ethnic backgrounds. It can result in a wide range of physical, cognitive and medical challenges.

Rayven's family is active in the Cornelia de Lange Syndrome Foundation, which is a member of NORD. "Every other year, the foundation has a conference where the children can play with other children who have the same syndrome," her mother says. "Parents can meet medical experts and get advice from special education experts."

# I AM JANELLE I HAVE DEVIC'S DISEASE



"I majored in theatre in college and was on a national Sesame Street tour. But in a matter of months, I went from rehearsals to rehab."

Devic's disease, also known as neuromyelitis optica, is often mistaken for multiple sclerosis. This is a problem because it can result in patients receiving inappropriate treatment.

In this very rare disease, the protective outer covering of the nerves is lost. Signs and symptoms include blindness in one or

both eyes, weakness in the legs or arms, and painful muscle spasms. Janelle's symptoms began as simple fatigue and a tingling in her toes. Eventually, she had a few falls, including one that unnerved her fellow cast members in a Sesame Street road show. She was sent to the clinical center at the National Institutes of Health (NIH), sometimes called "America's clinical research hospital."

Janelle's very supportive parents and her naturally buoyant personality have seen her through some difficult times with this disease that tends to recur in clusters of periodic attacks.

## NORD STUDY CONCLUDES FDA IS FLEXIBLE IN REVIEWING THERAPIES FOR RARE DISEASES



### Study Catalogues Flexibility in Orphan Drugs Approved Since 1983

NORD released a landmark report documenting flexibility in the FDA review of potential treatments for patients with rare diseases, in the opening session of the U.S. Conference on Rare Diseases and Orphan Products. The report examined the basis for FDA's approval of 135 non-cancer "orphan drugs" – those for rare diseases – since the Orphan Drug Act was enacted in 1983 to provide incentives to encourage development of treatments for rare diseases.

This is the first study of its kind ever conducted and the first time there has been a systematic examination of the basis for approval for any category of drug products over such a long period of time. The study concluded that flexibility had been applied in two-thirds of the cases.

"We are gratified that this study on behalf of the entire rare disease community found that there is supportable evidence to document flexibility by FDA medical reviewers," said NORD President and CEO Peter L. Saltonstall. "This study should provide an extra level of confidence for investigators, companies and investors who are considering developing new drugs for rare diseases."

Sandy Walsh, of the FDA Office of Public Affairs, responded to media inquiries about the study with the following comments: "We think the report released by NORD shows FDA's sensitivity to meeting unmet medical needs. Our record, as evidenced in the report, shows that

we are exercising judgment to address the unmet medical needs of patients with rare diseases. The cases looked at in the report were overwhelmingly one-of-a-kind treatments where no treatment existed. Development of treatments for rare diseases presents special challenges to researchers. We work every day to help make new therapies a reality."

The issue of flexibility is important to the patients and families NORD serves, Saltonstall said, because the patient population available for testing of orphan drugs is, by definition, more limited than for drugs for more prevalent diseases.

For each of the 135 non-cancer drugs approved as orphans since 1983, NORD sought to access documentation such as FDA approval letters, labeling at the time of approval, decision memoranda, and reviews. In most cases, the documents were retrievable.

Those documents were then divided into three categories: those that, in NORD's judgment, would have met the traditional data requirements for effectiveness; those whose approval was based on flexibility applied as a result of some documented FDA system for flexibility; and those whose approval appeared to reflect case-by-case flexibility.

NORD Board Chair Frank Sasinowski, who authored the report and presented the study at the conference, said the study showed that FDA recognizes the importance of therapies for persons with rare disorders. "It would be helpful for such flexibility and importance to be recognized in a formal FDA policy," he said, "and for FDA officials to incorporate and recognize that flexibility in a systematic way in their evaluations of each new therapy in development and under FDA review for Americans with any rare disease."

The entire report is posted on the NORD website under News & Events/NORD Publications. NORD's press release is located under News & Events/News.

# MARIO BATALI FOUNDATION ESTABLISHES RESEARCH FUND WITH NORD

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NORD's research program provides seed-money grants, mostly to researchers at university campuses and teaching hospitals, in the hope that promising studies will attract additional support later. At least one has resulted in development of a treatment approved by the Food and Drug Administration (FDA), a device known as the titanium rib for children with thoracic insufficiency syndrome.

Widely known for his television appearances on shows such as the PBS series "Spain ... on the Road Again" with actress Gwyneth Paltrow, Mr. Batali also co-owns numerous restaurants and is an expert on the history and culture of Italian cuisine. He established the foundation after realizing that, while he actively supported many charitable causes over the years, his chief interest was in improving the health and education of children. "I feel particularly passionate about protecting and helping these children, whose entire future rests on the vital research and awareness of these rare diseases," Mr. Batali said. "Every child deserves a fair shot at a healthy and long life."

## PATIENT ADVOCATES ASK FDA TO LET THEM HELP DEFINE RISK

Several weeks ago, NORD and approximately 30 of its members and advocacy partners sent letters to key FDA officials seeking more frequent and regular opportunities to provide input on decisions related to relative risks and potential benefit for new drugs and medical devices.

The letters were a follow-up to meetings the patient advocates had earlier with senior FDA officials. They included specific proposals for enhanced communication between the patient community and FDA to ensure that the voices of patients with chronic and rare diseases are heard in risk-benefit determinations and related policy decisions.

Now FDA has responded to the letters by inviting NORD and its partners to a meeting to further discuss the topic. Those present at the meeting included key representatives from key FDA divisions, such as the Center for Drug Evaluation and Research (CDER), Center for Biologics Evaluation and Research (CBER), and Center for Devices and Radiological Health (CDRH).

"We're very happy with FDA's response on this topic," said NORD Vice President of Public Policy Diane Dorman. "We feel the agency is hearing our concerns and wants to work with the patient community to ensure that patient voices are heard."

The letters were sent to Dr. Theresa Mullin, Director of FDA's Department of Planning & Informatics, and Dr. Jeffrey Shuren, Director of CDRH. Copies were sent to Health and Human Services Secretary Kathleen Sebelius, FDA Commissioner Dr. Margaret Hamburg, and Dr. Janet Woodcock, Director of CDER.

"Patients need to have opportunities to communicate with FDA medical reviewers on the risks they are willing to run in exchange for a potential though unproven benefit," said Peter L. Saltonstall, NORD President and CEO. "For example, a patient with a serious disease and no approved therapy may have a perception of risk that is very different from that of someone who has treatment alternatives. We believe that FDA can make more informed decisions about investigational products and about which products to approve if they hear directly from patients."

In addition to laying out specific suggestions as to how and when patient input might be submitted, the letters proposed that FDA create a portal on its website where it can post specific requests for patient contributions to certain issues.

# CONNECTING PATIENTS AND CAREGIVERS AROUND THE WORLD



NORD and EURORDIS recently announced the launch of the 14th global online patient community. This one is for patients and families affected by Waldenstrom macroglobulinemia.

RareConnect, launched as a joint initiative of EURORDIS and NORD last year, provides a safe

platform for rare disease patient organizations to develop online communities across continents and languages.

The driving force behind the project is the desire to help patients and caregivers share experiences and increase their access to quality information. Patient organizations in the U.S. and Europe collaborate with NORD and EURORDIS on this project. Disease-specific organizations play a key role in governing, moderating, and providing content for the communities.

The online communities are divided into three sections: What, Meet, and Learn. The "What" section features patient stories and blog-style updates from patients and patient organizations. The "Meet" section is a forum, moderated by volunteers and offering human translation services across five languages: English, French, Spanish, Italian, and German. The "Learn" section provides information in the form of frequently asked questions, documents, recently published news and scientific articles, upcoming events, and helpful resources.

To learn more about RareConnect, and the 14 disease-specific communities active at this time, go to: [www.rarediseasecommunities.org](http://www.rarediseasecommunities.org).



## AN UPDATE ON THE NORD RESEARCH PROGRAM

Through the NORD Research Program, nine Requests for Proposals (RFPs) have been posted for 2011. These RFPs are for seed-money grants funded largely by patients, their families and friends, and patient organizations.

In the spring, RFPs were posted for seven grants for the study of six diseases. An additional set of RFPs for three more diseases was posted in the fall. Currently, the NORD Medical Advisory Committee is reviewing full proposals for the RFPs posted in the spring and abstracts and letters of intent for those posted in the fall.

The 2011 RFPs are for study of the following rare diseases:

- Adenoid Cystic Carcinoma (ACC)
- Corticobasal Degeneration
- Diffuse Scleroderma (Systemic Sclerosis)
- Primary Orthostatic Tremor (POT)
- Pseudomyxoma Peritonei (PMP)
- Tarlov Cysts
- Trimethylaminuria (TMAU)
- Congenital Skeletal Abnormalities Associated with Cat Eye Syndrome or Other Rare Chromosomal Disorders

Decisions on award recipients for the first set of RFPs will be made by NORD's medical advisors in late November. Grant recipients for the second set of RFPs will be announced in March 2012.

For more information on NORD's research grant program, write to the program coordinator, Stefanie Putkowski RN ([research@rarediseases.org](mailto:research@rarediseases.org)) or go to the NORD website and click on "Medical Professionals" at the top of the home page.

## KEEPING OUR MEMBERS ENGAGED AND ACTIVE!

At NORD, keeping our members engaged and active is more than just a goal. It resonates throughout our organization as an essential part of what NORD is all about. We understand that our members are a key part of the solution, and we value their support and collaboration in our advocacy, awareness, and educational programs.

Since NORD's inception in 1983, its core values have been shaped and articulated by patient organization leaders who identified common challenges and recognized the importance of working together. Today, NORD's membership community consists of more than 160 patient advocacy organizations located throughout the U.S. and overseas.

### Strengthening and Growing NORD's Membership Community

Currently, a major initiative is underway to:

- strengthen the two-way communication between NORD and its members
- provide guidance and leadership to help NORD members build the capacity of their own organizations and adopt best practices
- increase our membership to enhance opportunities for collaboration among rare disease patients and families

A grant from the Medtronic Foundation has made possible three regional meetings for NORD member organizations as one step in this process. The first of these meetings took place in Chicago in September. The second is being organized for the New York City area in December, and a third is planned for March 2012 in San Francisco.

These one-day meetings provide a wonderful opportunity for our members to share ideas and acquired wisdom in areas such as advocacy, organizational and board development, education and awareness, research programs, social media and websites, and fundraising. In addition, NORD senior staff share updates about current public policy initiatives and how to work more closely with NORD in advocacy and through national campaigns such as Rare Disease Day.

### Watch For Additional Opportunities to Collaborate

In 2012, NORD will continue to expand its services targeted to all levels of patient organization needs. We look forward to growing the collective voice and continuing to facilitate engagement with the broader rare disease community. NORD members recognize the power of speaking together to promote their shared interests, even though their disease-specific missions may be somewhat different.

For more information about NORD's dynamic membership community, contact Ana Maria Vallarino, Director of Membership (amvallarino@rarediseases.org) or click on "For Patient Organizations" on the NORD home page (rarediseases.org).

## SSA COMMISSIONER ANNOUNCES 13 COMPASSIONATE ALLOWANCES



Michael Astrue

At the recent NORD conference, Social Security Commissioner Michael Astrue announced that 13 additional rare medical diagnoses are being added to the Compassionate Allowances list. This is a list of diseases so disabling they qualify those affected for expedited review of applications for disability assistance.

NORD and its Medical Advisory Committee have provided input to the Social Security Administration (SSA) for the list over the past few years. In July, NORD President and CEO Peter L.

Saltonstall spoke at a Capitol Hill event commemorating the fact that the list at that time included 100 diseases.

The 13 diagnoses added to the list in this most recent expansion are: malignant multiple sclerosis, paraneoplastic pemphigus, multicentric Castleman disease, pulmonary Kaposi sarcoma, primary central nervous system lymphoma, primary effusion lymphoma, Angelman syndrome, Lewy body dementia, Lowe syndrome, corticobasal degeneration, multiple system atrophy, progressive supranuclear palsy, and the ALS/Parkinsonism dementia complex.

SSA's press release is available online at: <http://www.ssa.gov/pressoffice/pr/ss-expands-compassionate-allowances.html>

View the complete list of Compassionate Allowances at: <http://www.socialsecurity.gov/compassionateallowances/>

## NORD VP PRESENTS INVITED TESTIMONY ON PDUFA

Vice President of Public Policy Diane Dorman spoke as an invited patient panelist at the final public hearing for the reauthorization of the Prescription Drug User Fee Act (PDUFA). This Act, which provides important funding to FDA, must be reauthorized periodically. The current process is known as PDUFA V.

Dorman spoke in support of provisions related to rare diseases in the current draft of PDUFA. She also focused on two topics of particular interest to rare disease patients and families at this time: risk tolerance and conflict of interest.

Regarding conflict of interest, she noted that NORD has joined with approximately 50 other organizations deeply concerned about the possibility that, with rare diseases, qualified and knowledgeable medical experts and patients may not have an opportunity to make their voices heard in the review process since the pool of available experts may be small and it may be difficult to find individuals not considered "conflicted" under the current provisions.

The hearing on October 24 was the final opportunity for public input on the current PDUFA draft. The draft will soon go to the Energy and Commerce Committee in the House and the HELP (Health, Education, Labor & Pensions) Committee in the Senate.

## NORD SUBMITS CITIZEN'S PETITION TO FDA

In September, NORD submitted a Citizen's Petition to FDA requesting that a documented policy be established regarding the review of potential treatments for people with rare diseases. The request was made in conjunction with a report submitted by an FDA committee to Congress on June 27, 2011: Improving the Prevention, Diagnosis and Treatment of Rare and Neglected Diseases.

Citizen's Petitions may be submitted to FDA by individuals or organizations requesting specific actions to issue, amend, or revoke regulations. In this case, NORD is asking that a specific policy related to orphan drugs be included in a guidance document that FDA must issue no later than December 27, 2011, following the June report to Congress.

While expressing concern that currently no policy specific to orphan drugs exists, NORD also praised FDA's history of flexibility in the

review of orphan products. The Citizen's Petition specifically asks for FDA documentation to guide future drug reviews and to:

- Acknowledge that clinical trials for most orphan drugs are fundamentally different from trials for drugs for common conditions
- Acknowledge that FDA review of orphan drugs is therefore different from the review of other products
- Document that, while the need to demonstrate both effectiveness and safety remains unchanged, it will now be official FDA policy to apply special flexibility in the review of orphan drugs.

NORD's press release with more information on this topic is available on NORD's website under News & Events/News.

# MEET NORD'S SENIOR STAFF



**PETER L. SALTONSTALL**  
PRESIDENT AND CEO

"Before joining NORD, I was very involved in issues related to patient safety. My commitment has always been to patients, and at NORD I saw an opportunity to make a difference on a broader level."



**TIMOTHY COTÉ, MD**  
CHIEF MEDICAL OFFICER

"I joined NORD because of the great opportunity to 'keep wearing the white hat,' forgoing the riches of industry for a mission even more treasured."



**PAMELA GAVIN**  
SENIOR VICE PRESIDENT

"Working at NORD enables me to be part of a team dedicated to bringing forth positive changes for a community I care so much about."



**MARY COBB**  
SENIOR VICE PRESIDENT OF  
MEMBERSHIP AND ORGANIZATIONAL  
STRATEGY

"Patients are at the forefront and heart of what we do. I joined NORD for the opportunity to use my experience in organizational growth and development to help patients in a meaningful way."



**MERIDITH BOLADO**  
VICE PRESIDENT OF DEVELOPMENT

"I have a close personal friend whose daughter has an undiagnosed rare disease. I come to work every day with a passion for helping others struggling with those types of issues."



**DIANE DORMAN**  
VICE PRESIDENT OF PUBLIC POLICY

"We watched my Dad die of a rare disease for four years. I hope I can make a difference for those unable to advocate for themselves."



**MARY DUNKLE**  
VICE PRESIDENT OF  
COMMUNICATIONS

"I've been with NORD for 12 years. It's a unique and wonderful organization that provides a voice for people who are quietly doing heroic things."



**MARY ORCUTT**  
VICE PRESIDENT OF FINANCE

"I joined NORD because I saw the organization as a great opportunity to use my skills to make a positive impact on the lives of patients."



**NORD**  
National Organization for Rare Disorders

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.

**Connecticut Office**  
55 Kenosia Avenue • PO Box 1968  
Danbury, CT 06813-1968  
203.744.0100  
Fax 203.798.2291

**Washington DC Office**  
1779 Massachusetts Avenue NW, Suite 500  
Washington, DC 20036  
202.588.5700  
Fax 202.588.5701

[www.rarediseases.org](http://www.rarediseases.org)