NEWS FROM NORD

AN UPDATE FOR OUR MEMBERS AND FRIENDS — SUMMER 2013

CELEBRATING 30 YEARS: EMPOWERING THE RARE DISEASE COMMUNITY!

Saluting the heroes of 1983, recognizing major milestones, and looking to the future.

DRUGS OF "LIMITED COMMERCIAL VALUE"

On June 29, 1979, a task force chaired by Marion Finkel, MD, of the Food and Drug Administration (FDA), published a report calling for measures to address what it perceived as a significant public health issue. The issue was that drugs "of limited commercial value" – for small patient populations – were not being developed.

The members of the task force came mostly from FDA and the National Institutes of Health (NIH). All served as volunteers. The report they produced called for directing more resources toward the research, development, and distribution of drugs for people with rare diseases. This helped set the stage for events that ultimately resulted in the *Orphan Drug Act*.

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Abbey Meyers (seated center) with Sen. Orrin Hatch and Rep. Henry Waxman

RARE DISEASE DAY 2013 WRAP-UP

Blair Van Brunt and Sarah MacDonald at Massachusetts RDD 2013 State House event

Gracie Van Brunt playing at RDD 2013

A media blitz, events for legislators in five states, a photo gallery, and a research "Hall of Fame" were among the highlights of Rare Disease Day 2013. This year's campaign also included free resources for teachers, awareness activities at hospitals and universities, messages of support from several Members of Congress, and a letter from President Obama.

Rare Disease Day was launched in Europe in 2008 by EURORDIS. The following year, NORD brought it to the U.S. Since then, the observance, always on the last day of February, has grown each year.

More than 70 nations participated in 2013, each using the same theme: Rare Disorders Without Borders. More than 700 organizations, companies, and institutions signed up as Rare Disease Day Partners with NORD.

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JPA AND NORD TO COLLABORATE

The Japan Patients Association (JPA) and NORD have signed a "Memorandum of Understanding" to explore ways to work together to the benefit of rare disease patients in Japan and the U.S. The partnership was formally established with a signing ceremony at the NORD offices in Washington DC.

> "Our organizations have a shared mission and goals, we would like to make use of our valuable knowledge and experiences for the mutual benefit of patients in both countries."
>
> *– JPA President Tateo Ito*

Seated, from left: NORD President Peter L. Saltonstall and Japan Patients Association President Tateo Itoh; Standing, from left: Mary Dunkle, Pamela Gavin, Yukiko Nishimura, Jason Barron and Casey Morris

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SSA REACHES MILESTONE; PRAISES NORD FOR HELP

Peter L. Saltonstall (left) with Michael Astrue at the Capitol Hill event where the expansion of the CA Program was announced.

At a Capitol Hill event in December 2012, Michael J. Astrue – who was completing his term as Social Security Commissioner – announced the addition of 35 diseases to the Compassionate Allowances list for expedited disability review. Mr. Astrue also praised NORD for supporting the program and presented a Commissioner's Award to Diane Dorman, NORD Vice President for Public Policy.

NORD President and CEO Peter L. Saltonstall spoke at the event, thanking Mr. Astrue and his staff for the CA Program, which he said "demonstrates true compassion for Americans with seriously disabling rare diseases."

Mr. Astrue established the program soon after being appointed Commissioner by President Barack Obama. The purpose is to identify diseases that meet Social Security disability standards so that patients with those diagnoses may receive their benefit decision within days rather than months or years. With the 35 added diseases, there are now 200 on the list, many of which are rare.

In his remarks, Mr. Saltonstall said that Americans living with rare diseases often struggle with "overwhelming medical and financial challenges." He told the story of a young wife and mother whose 33-year-old husband died of frontotemporal dementia, a progressive neurological disorder, and said it illustrated the plight of many families.

"She wrote to us about caring for her 5-year-old son while also managing her husband's medical needs and the family finances," he said. "She described how she had to quit her job and let the family's three-bedroom house go into foreclosure." The CA Program, he added, made it possible for her to focus on the things that really mattered during a very difficult period.

JPA AND NORD TO COLLABORATE

Continued from Cover

NORD President and CEO Peter L. Saltonstall said the partnership reflects growing awareness of the importance of global collaboration in the rare disease community. Such collaboration, he said, "will drive progress toward new treatments, improved public policies and better lives for people with rare diseases."

In 2009, NORD entered into a strategic partnership with EURORDIS (the European Organisation for Rare Diseases). The two organizations work together on initiatives such as Rare Disease Day to promote global awareness of the challenges that people around the world with rare diseases share.

The JPA is a nonprofit umbrella organization established in 2005. It focuses on rare and intractable diseases and has a membership of approximately 300,000 from 72 organizations, including individual patient groups and regional centers. JPA provides patient/family services and advocacy on public policies. Its website is at www.nanbyo.jp.

News from NORD is published by the National Organization for Rare Disorders

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FDA, MEDICAL DEVICE INDUSTRY AND NORD ANNOUNCE NEW PUBLIC-PRIVATE PARTNERSHIP

Food and Drug Commissioner Margaret Hamburg, MD, in December announced the launch of the first-ever public-private partnership (PPP) to focus exclusively on promoting the development of innovative medical devices for patients. NORD is a founding member and the only patient organization in the PPP.

Other founding members, in addition to the FDA, are LifeScience Alley, a biomedical science trade association; and several companies involved in developing medical devices. The partnership will be called the Medical Device Innovation Consortium (MDIC).

In remarks made at the launch, NORD President and CEO Peter L. Saltonstall said: "Over the past several years, we have sought to forge a productive relationship with the FDA and the medical devices industry because we believe that progress can be made only if all segments of the medical system work together. We hope this public-private partnership will serve as a model for additional collaboration."

The mission of the MDIC will be to foster innovation, increase the tools available to evaluate new medical devices to assure that they are safe and effective, and ensure that innovative devices get to patients as quickly as possible while still protecting public health.

Senators Amy Klobuchar and Al Franken participated in the launch announcement, as did Commissioner Hamburg and FDA Center for Devices and Radiological Health Director Jeffrey Shuren, MD, JD. Mr. Saltonstall said that NORD's role will be to provide the patient perspective in the new partnership.

30 YEARS: NORD AND RARE DISEASE MILESTONES

1979

FDA/NIH Task Force Issues Report
Calling for measures to address the need for more resources to be directed toward drugs for small patient populations.

1979 – 1980

Patient Advocates Form Ad Hoc Coalition
Leaders of rare disease patient organizations form a coalition to provide advocacy together on behalf of legislation to encourage the development of treatments for people with rare diseases.

1980

Pre-Orphan Drug Act
Only 10 new drugs were developed solely by industry for rare diseases in the decade before 1983.

RARE DISEASE DAY 2013 WRAP–UP

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FIVE STATE HOUSE EVENTS

In California, Connecticut, Massachusetts, New Jersey, and Texas, patient advocates and supporters hosted events to educate state legislators about the challenges of living with rare diseases. NORD worked with Blair Van Brunt of the Shwachman Diamond Syndrome Foundation and Sarah MacDonald of MassBIO, organizers of the Massachusetts event, to develop tools and resources for others.

MEDIA OUTREACH

Rare Disease Day 2013 featured a blizzard of patient stories, photos and videos in the social media, TV, radio and newspapers. Throughout the day and the weeks leading up to it, there was a steady drumroll of stories to enhance public awareness and understanding. Good Morning America host Robin Roberts talked about Rare Disease Day. The Today Show welcomed mothers of children with a rare syndrome known as CDKL5. ABCNews.com, CNN.com, and Huffington Post all featured stories.

Move YOUR Way for Rare Disease Day; Texas State House event; and CSL Behring support for Rare Disease Day

HANDPRINTS ACROSS AMERICA

Supporters submitted photos of themselves holding the Rare Disease Day logo. CBSNews.com included 16 of these photos in a major spread headlined "Rare Disease Day Puts Spotlight on 30 Million Americans."

RARE DISEASE RESEARCH HALL OF FAME

The rare disease community was invited to post photos and brief tributes to researchers. These will be available for viewing throughout the year.

NIH AND OTHER EVENTS

NIH hosted a symposium for patient advocates and researchers. Notre Dame University students wrote a special version of their "fight song". Several research institutes hosted conferences. Many schools provided special activities for students, and teachers could download a free curriculum from NORD's Rare Disease Day US website.

To read about Rare Disease Day in the U.S., go to www.RareDiseaseDay.US. To reach about Rare Disease Day around the world, go to www.RareDiseaseDay.org.

Mary Caruso and her daughters, Sam and Alex, with Representative Vincent Candelora at the Connecticut State House event.

Brooke Foster telling New Jersey State Legislators about the rare disease of particular interest to her, mastocytosis.

A proclamation was displayed at the California State House event.

State legislators participating in the Connecticut event listen to Steve Bajardi of the NORD board and the International Rett Syndrome Foundation.

Orphan Drug Act Passed

1983

NORD Founded

NORD is incorporated to represent the shared interests and goals of all Americans affected by rare diseases.

1983

First Orphan Drug Approved

1983

CELEBRATING 30 YEARS: EMPOWERING THE RARE DISEASE

Continued from Cover

Real-life rare disease patients served as extras in the seond *Quincy* episode

ON CAPITOL HILL

At about the same time, certain legislators were becoming aware of this issue. In the late 1970s and early '80s, the Subcommittee on Health and the Environment of the House Energy and Commerce Committee, chaired by Representative Henry Waxman (CA), held hearings and gathered evidence on the problem. Rep. Waxman ultimately drafted legislation creating financial incentives to encourage the development of treatments for people with rare diseases.

Rep. Waxman served as the bill's primary sponsor in the House. Senator Nancy Landon Kassebaum (KS) introduced it in the Senate.

> "For those with rare diseases, this is a tragic, and sometimes hopeless, situation. For the rest of us who believe that people with rare diseases suffer no less and are no less deserving because they are so few, this situation is intolerable. It must be changed."
>
> *– Rep. Henry A. Waxman, opening statement for Orphan Drug Act hearings, March 8, 1982*

AD HOC PATIENT COALITION

The patient voice was added to this conversation with the formation of an ad hoc coalition of patient advocates that included representatives of organizations for people with Huntington's disease, Marfan syndrome, epidermolysis bullosa, and other rare diseases. Abbey Meyers, who was affiliated with the Tourette Syndrome Foundation, played a key role in organizing this coalition.

Abbey and others testified before a 1980 hearing of Rep. Waxman's subcommittee. The room was nearly empty but there was one reporter taking notes who posted a brief story in the LA Times.

Jack Klugman as *Quincy*

That story was seen by Maurice Klugman, a writer and producer for a popular television show, Quincy, ME. Klugman's brother, Jack, was the star of the show. Maurice Klugman later called Abbey Meyers, told her that he had a rare disease, and asked how he could help the patient coalition.

The result was an episode of Quincy that sparked national interest. Viewers sent letters to Klugman that he forwarded to Meyers. That was the beginning of the ad hoc coalition's mailing list.

Later, Klugman was invited by Rep. Waxman to testify at a 1980 hearing on the topic. While it's not uncommon today for Hollywood celebrities to appear on Capitol Hill, it was highly unusual in that day. Klugman's testimony was widely reported in newspapers, including on the front page of the New York Times.

> "On the evening before the hearings … Dr. Van Woert and I … called Jack at his hotel because we knew that he was uncertain about his speech and needed advice. Jack expressed a deep feeling that he was just an actor, and he couldn't understand why he would have an impact on the Congress. He said he had read our speeches from last year's testimony and had cried. He wondered if anything he could say to Congress could be more meaningful than what we had already said.
>
> Dr. Van Woert and I explained that our speeches were given to a half-empty hearing room….We told him that the great difference between last year and this would be the fact that people would listen and the press would report his testimony."
>
> *– Abbey Meyers in a report to the Tourette Syndrome Foundation Board, March 9, 1981*

When the bill later appeared to have become stalled in Congress, Klugman devoted a second Quincy episode to the topic. This time, many real-life rare disease patients served as extras, staging a march on Capitol Hill in a scene filmed in California.

Ultimately, the Orphan Drug Act was approved by the House of Representatives on Dec. 14, 1982, and by the Senate three days later. President Ronald Reagan signed the bill on January 4, 1983.

1987 — **NORD Establishes First-ever Patient Assistance Program**

1989 — **National Commission Issues Report**
The National Commission on Orphan Diseases, chaired by Jess Thoene, M.D., board chair and medical advisor to NORD, conducts a major study and issues a report on the experiences of patients and families affected by rare diseases.

1995 — **NORD Establishes Research Program**
Patients and patient organizations may provide grants for the study of diseases with limited or no other source of funding.

COMMUNITY!

AD HOC PATIENT COALITION BECOMES NORD

Just before the bill was enacted, the members of the patient coalition decided to formalize as an umbrella organization representing all people with rare diseases. They realized, as NORD's slogan states today: Alone we are rare. Together we are strong.

On May 4, 1983, the incorporation of the National Organization for Rare Disorders (NORD) was completed. Initially, Jack Klugman served as NORD's honorary chairman. Rare disease medical experts including Jess Thoene, MD, and Melvin Van Woert, MD, provided key leadership during the formation of NORD.

Over the years, many patient advocates from NORD Member Organizations have played key roles in charting NORD's course and its advocacy positions. Today, NORD represents approximately 200 Member Organizations and partners with many other stakeholders in the rare disease community.

NORD AND THE ORPHAN DRUG ACT

Since 1983, the Orphan Drug Act has been amended on three occasions – in 1984, 1985 and 1988. NORD has opposed other efforts to revisit the law out of concern that changes might weaken it. The law is considered by most to have been highly successful. Since 1983, more than 400 drugs and biologics have been approved as orphans. Hundreds more are in the research pipeline.

> "We have come here today to express our support for Mr. Waxman's bill. For, if this bill does not become law, generations of people with hundreds of rare diseases will have no future."
>
> *– Abbey Meyers at a press conference on Dec. 16, 1981*

Photo taken just before Jack Klugman testified before Waxman Subcommittee in 1981: Congressman Ted Weiss; Judy Wertheim (tourette syndrome); Abbey Meyers; Jack Klugman; Melvin Van Woert, MD; Adam Seligman; Seated: Betty Teltsher (tourette); Sharon Dobkin (myoclonus); Niss Ryan (narcolepsy); Marjorie Guthrie, widow of folksinger Woody, (Huntington's disease).

NATIONAL COMMISSION ON ORPHAN DISEASES

In the late 1980s, Congress established a National Commission on Orphan Diseases. The Commission's report issued in 1989 has become a classic document in the history of the U.S. rare disease community.

In the prologue, Dr. Thoene, who chaired the commission, wrote: "Hidden among the many strident voices clamoring to be heard by Congress and to receive a share of our national resources is the persistent murmur of those left outside the current health care system. Lacking adequate health insurance, a correct diagnosis, and any real hope of effective therapy, persons with rare diseases are truly medically disenfranchised."

Stephen C. Groft, PharmD, and other NIH staff played key roles in the commission's study and report. Over the years, NORD has continued to work closely with the NIH, FDA and other government entities.

MILESTONES ALONG THE WAY

Since 1983, there have been many key milestones achieved by the rare disease community with NORD leading the way. Among others, these include:

- Establishment of the ClinicalTrials.gov website
- The Rare Diseases Act of 2002, which led to establishment of the NIH Rare Diseases Clinical Research Network (RDCRN)
- Publication of two editions of The Physician's Guide to Rare Diseases and a 2003 edition of The NORD Guide to Rare Disorders for physicians
- Establishment of an Associate Director for Rare Diseases position in FDA's Center for Drug Evaluation and Research
- Establishment and later expansion of the Compassionate Allowances Program within the Social Security Administration
- Expansion of FDA's Rare Disease Program and other major rare disease provisions in the FDA Safety and Innovation Act of 2012

See the 30th Anniversary timeline on NORD's website (www.rarediseases.org) for additional milestones.

LOOKING TO THE FUTURE

Today, NORD partners with all stakeholders in the rare disease community:

- To promote timely diagnosis for people with rare diseases
- To address the medical and financial challenges of living with rare diseases
- To promote faster, less costly development of treatments
- To assure that all patients have access to these treatments

Established 30 years ago by the members of the ad hoc patient coalition, NORD remains the only national organization representing all Americans with rare diseases with programs of advocacy, education, research and patient/family services.

Rare Diseases Act Signed into Law
Providing for the establishment of the NIH Rare Diseases Clinical Research Network.

2002

NORD Publishes Guide to Rare Disorders

2003

NIH Establishes Undiagnosed Diseases Program

2008

Guests arriving at 2012 NORD Celebration, which was held at Union Station.

U.S. Representatives Ed Towns (NY-10) and Cliff Stearns (FL-6) with emcees Patricia Richardson and Michael Scott

Alexion representatives and the Ziegler family representing the aHUS community at the 2012 NORD gala

Corporate awards honor companies for innovative therapies approved during the previous year.

30th Anniversary Celebration

14 MAY TH 2013
Tuesday

Mellon Auditorium
1301 CONSTITUTION AVE NORTHWEST
WASHINGTON, DC 20240

RECEPTION
6:30 PM
DINNER
7:30 PM
Black tie optional

NORD TO HOST 30TH ANNIVERSARY CELEBRATION ON MAY 14

NORD will honor heroes of the past and present – and renew its commitment to innovation for the future – at a special 30th Anniversary Celebration on May 14 at the Mellon Auditorium in Washington DC.

Individuals who played key roles in the history of the Orphan Drug Act and of NORD will be honored. Also, awards will be presented to four companies for orphan products approved by FDA in 2012.

A highlight and unique feature of the evening will be the creation of a 30th Anniversary video to be shown at future events and posted online. Interviews will be conducted for the video at the May 14 event. In addition, a new initiative to be launched by NORD and the Genzyme Boston Marathon team will be announced.

The evening will be dedicated to the memory of actor Jack Klugman for his role related to the Orphan Drug Act. Honorees will include:

For leadership related to the Orphan Drug Act:

- Representative Henry Waxman (CA)
- Senator (retired) Nancy Kassebaum (KS)
- William Corr, Deputy Secretary of Health & Human Services and former Congressional aide to Rep. Waxman

For leadership on behalf of patients and patient organizations:

- Abbey S. Meyers
- Jess Thoene, MD
- Marlene Haffner, MD
- Stephen C. Groft, PharmD
- Lars Uno-Larsson

For orphan products approved by FDA in 2012:

- Aegerion Pharmaceuticals, Inc.
- NPS Pharmaceuticals, Inc.
- Onyx Pharmaceuticals, Inc.
- Sigma-Tau Pharmaceuticals, Inc.

To attend the NORD 30th Event:
Registration is now open at rarediseases.org

Member of EURORDIS Rare Diseases Europe

NORD and EURORDIS Partnership
2009

Rare Disease Day

NORD Sponsors first US Rare Disease Day
2009

Rare Disease Position Established in CDER
For the first time, FDA CDER has an Associate Director for Rare Diseases.
2010

NORD MEMBERSHIP COMMUNITY GROWS AND SHARES EXPERIENCES

With 10 new Member Organizations having joined the NORD family since October, and regional member meetings taking place in Orlando and Chicago, the NORD membership community continues to grow and strengthen its bonds through shared experiences.

"In December, our members came together in Orlando to share thoughts regarding capacity-building and how to build critical mass to achieve organizational goals," says Mary Cobb, Senior Vice President of Membership and Organizational Strategy. "Topics ranged from board-staff interaction to implementing scientific meetings, among others."

Planning is now underway for a regional meeting for NORD Member Organizations in Chicago. The agenda will focus on the role of patient organizations in encouraging and accelerating research on rare diseases.

NORD Member Organizations played a significant role in Rare Disease Day 2013 activities. In particular, several were involved in helping organize State House events.

At a regional member meeting in Orlando in December, participants took a minute to show their support for Rare Disease Day.

NEW MEMBERS OF NORD SINCE OCTOBER 2012 ARE:

- Basal Cell Carcinoma Nevus
- CMTC/OVM-USA
- Cloves Syndrome Community
- Council for Bile Acid Deficiency
- Dup15q Alliance
- Hageman Foundation
- International Pemphigus and Pemphigoid Foundation
- International WAGR Syndrome Association
- Lymphangiomatosis & Gorham's Disease Alliance
- Turner Syndrome Society of the US

HAVE YOU VISITED NORD'S BLOG YET?

Over the past six months, posts on NORD's blog have covered a broad range of topics, from the outlook for investment in orphan product R & D to a patient's determination to run a 5K race despite having a serious rare disease. If you haven't done so yet, we recommend that you make a visit to: http://blog.rarediseases.org … and we hope you'll join the conversation. Your thoughts are important to us!

NORD POSTS RFPS FOR 2013 RESEARCH GRANTS

Requests for Proposals for seven 2013 NORD Research Grants have been posted on the website. The deadline for submitting abstracts and letters of intent is May 15, 2013. All information is at www.rarediseases.org, and questions may be directed to research@rarediseases.org.

SCAN CODE TO VIEW NORD'S COMPLETE TIMELINE

Affordable Care Act Signed

Provides reforms, such as ending annual and lifetime insurance caps and eliminating discrimination based on pre-existingconditions.

2011

First Annual NORD / DIA Conference

2011

FDA Safety and Innovation Act Approved by Congress and signed by President Obama.

2012

NORD

30 YEARS | EMPOWERING THE RARE DISEASE COMMUNITY

DONATE

Be part of NORD's future.

Donate online, by phone, or by mail today to support NORD programs for the rare disease community.

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.

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