RARE IMPACT ★ AWARDS®

OCT 9th 2020

NORD®
National Organization for Rare Disorders

Alone we are rare. Together we are strong.®

NORD was founded over 37 years ago by a group of parents advocating for their children struggling with rare diseases. We fight to improve the lives of over 25 million Americans impacted by rare diseases.

NORD advocates for policy changes to improve the lives of Americans impacted by rare diseases at the federal and state levels.

Our research programs underscore NORD's commitment to the identification, treatment and cure of rare diseases.

Raising awareness of issues facing rare disease patients and families and providing resources for the community are at the heart of our mission.

Visit **rarediseases.org** to learn more about NORD's information and resources for rare patients, caregivers, clinicians and researchers.

NORD®
National Organization for Rare Disorders

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WELCOMES YOU TO THE 2020 RARE IMPACT AWARDS OCT 9TH, 2020

PROGRAM FOR THE EVENING

WELCOME
John Whyte, MD, MPH,
Chief Medical Officer, WebMD

OPENING REMARKS
Peter L. Saltonstall,
President and CEO, NORD

PRESENTATION OF AWARDS

MUSICAL GUEST
The cast of "Hemophilia:
The Zoomsical!"

PRESENTATION OF AWARDS

DEAR FRIENDS

Thank you for joining us for the 2020 Rare Impact Awards, which is being presented for the first time ever as a streamed event. Celebrating those making a difference in the lives of millions of Americans affected by rare disease is more important than ever for us to do together, albeit virtually, right now.

At the National Organization for Rare Disorders and as a community, the new normal in 2020 has presented many challenges for us to meet, but we've been busier than ever working to move the needle on what matters most to rare disease patients and caregivers. We now have over 50 volunteer ambassadors and community engagement liaisons across 40 states as part of our Rare Action Network (RAN), and more than 5,000 people have made their voices heard with their state or federal legislators on issues impacting rare patients and families. We've been working to protect the Affordable Care Act and ensure that rare disease patients have access to affordable coverage and treatments. Through the formation of Rare Disease Advisory Councils, we're expanding representation of our community at the state level.

Despite the pandemic, our efforts to help speed access to new rare disease treatments continue, through initiatives like the Rare Disease Cures Accelerator-Data and Analytics Platform and support of critical research and clinical trials through our grant programs. Most importantly, every day we hear from patients and caregivers across the country who need help. We are grateful that we can be there for them in these uncertain times with our COVID-19 emergency relief funds and other patient assistance programs.

We do not do these things alone. You will hear NORD's motto echoed throughout the program—Alone we are rare. Together we are strong. We'd like to take a moment to thank the tireless rare disease advocates who have created change for the entire community, congratulate the companies that had a therapy approved this year, and acknowledge the hard work and dedication of the leaders at the US Food and Drug Administration (FDA) and National Institutes of Health (NIH), who help make this progress possible.

Our sincere thanks for joining us and congratulations to our Rare Impact Award and Industry Innovation Award honorees.

Warmest regards,

Peter L. Saltonstall
PRESIDENT AND CEO

Marshall L. Summar, M.D.
CHAIRMAN, BOARD OF DIRECTORS

EMCEE FOR THE EVENING

John Whyte, MD, MPH

Dr. John Whyte, MD, MPH is a board-certified practicing physician who has been communicating to the public as well as private sectors on health and health policy issues for nearly 25 years. As someone who has been a regulator, researcher, educator, and media executive, Dr. Whyte brings a unique perspective. He is passionate about changing how we think about health.

He is currently the Chief Medical Officer, WebMD. In this role, Dr. Whyte leads efforts to develop and expand strategic partnerships that create meaningful change around important and timely health issues. He is particularly interested in evaluating consumer trends in digital health and how innovations – especially in technology – change the way health care is delivered.

Before joining WebMD, Dr. Whyte served as the Director of Professional Affairs and Stakeholder Engagement at the Center for Drugs Evaluation and Research. Dr. Whyte worked with health care professionals, patients and patient advocates, providing them with a focal point for advocacy, enhanced two-way communication and collaboration. He helped them navigate the regulatory process in a way that was more accessible. In addition, he launched the Drug Trials Snapshot program that details the participation in clinical trials for all new drug approvals – the first in a series of efforts by the FDA to help improve diverse representation and streamline the clinical trial process.

Prior to FDA, Dr. Whyte worked for nearly a decade as the Chief Medical Expert and Vice President, Health and Medical Education at Discovery Channel, the leading non-fiction television network. In this role, Dr. Whyte developed, designed, and delivered educational programming that appealed to both a medical and lay audience. This included television shows as well as online content that won over 50 awards, including numerous Tellys, CINE Golden Eagles and Freddies.

Dr. Whyte is a frequent commenter on health care topics and has written extensively, including two best-selling books, Is This Normal: The Essential Guide to Middle Age and Beyond and AARP New American Diet: Lose Weight, Live Longer. He has also edited a book on medical device regulation and authored numerous articles on drug development and personalized medicine. He writes a monthly column for WebMD magazine, and hosts a podcast that often talks about the latest trends in medical innovation.

The cast of "Hemophilia: The Zoomsical"

Art has the power to change lives, which is why Believe Limited launched Breaking Through! and "Hemophilia: The Zoomsical," a theater workshop and art therapy program built for and created in partnership with teenagers from the bleeding disorders community. First launched in 2018 at New World Stages in New York City then executed regionally at patient education events throughout the US, the 2020 program was originally scheduled to take place in Los Angeles and Chicago with two separate casts. Once circumstances changed, the creatives, production team and casts quickly pivoted and threw their energy behind what became one big, unified "Hemophilia: The Zoomsical!"

2020 HONOREES

RARE IMPACT ★ AWARDS® 2020

LIFETIME ACHIEVEMENT AWARD

Janet Woodcock, MD

DIRECTOR, CENTER FOR DRUG EVALUATION AND RESEARCH, US FOOD & DRUG ADMINISTRATION

Janet Woodcock, MD, has been an ardent advocate for the rare disease patient community for several decades. As Director of the Center for Drug Evaluation and Research (CDER) at the US Food and Drug Administration (FDA), she has provided active and successful leadership that has enabled hundreds of new therapies for rare diseases to be developed. She is currently serving in a special role in Operation Warp Speed, which is dedicated to overseeing the rapid development of therapeutics and vaccines for COVID-19.

Dr. Woodcock received her MD from Northwestern Medical School and held teaching appointments at the Pennsylvania State University and the University of California in San Francisco. She joined the FDA in 1986, initially serving as Director of the Office of Therapeutics Research and Review and Acting Deputy Director in the Center for Biologic Evaluation and Research (CBER). In 1994 she joined CDER and has served there since, other than three years (2005-2008) as Deputy Commissioner for Operations and Chief Operating Officer for all of the FDA.

She is recognized as one of the most innovative and creative managers and leaders ever to serve at the FDA. Dr. Woodcock has conceptualized many of the future-looking programs that have been adopted in the drug development and oversight processes at the FDA. She is known for assuring that all drugs approved meet the highest standards for safety, efficacy and quality, and has streamlined FDA and CDER processes so that new drugs can be developed without unnecessary delays, and are monitored during actual use to enhance patient safety.

Some of the initiatives that Dr. Woodcock has overseen include the development and implementation of the Sentinel system, which monitors drugs on a continuing basis for safety; the modernization of the national adverse drug reaction reporting system; the risk management (REMS) system which has enhanced the safe use of new drugs; and the Rare Disease Cures Accelerator initiative, which is intended to advance the framework for rare disease drug development.

In recent years, more than a third of the new drugs approved by CDER have been orphan drugs, and the percentage continues to increase. The investment community has favored the development of orphan drugs because they know that drugs for rare diseases will receive attention by the leadership at CDER and, thanks to Dr. Woodcock, at the other FDA medical centers as well.

Dr. Woodcock's personal commitment to rare diseases is reflected in her creation of offices within CDER that focus exclusively on rare diseases and orphan drugs. She has created internal training programs within CDER for medical reviewers on the special needs of patients with rare diseases and the factors that need to be considered in designing and reviewing clinical trials of orphan drugs. She has actively supported the concept that due to the small patient populations for many rare diseases, CDER must exercise flexibility in helping researchers design clinical development programs and studies for therapies for patients with rare diseases. She has been an active supporter of programs, many designed in conjunction with and implemented by NORD, that seek to gain a better understanding of the natural histories of rare diseases and to establish patient registries that can help expedite the recruitment of subjects into studies of orphan drugs.

Dr. Woodcock is a passionate advocate for incorporating the patient voice and experience into the drug development process. Under her leadership, the door to CDER has been open to patient organizations seeking to educate FDA medical staff about the patient experience with rare diseases, which has led to the initiation of more insightful and patient-friendly clinical programs. Patient groups know that Dr. Woodcock is one of their most committed advocates and that because of her dedication, every patient with a rare disease has benefited.

NORD is honored to present Dr. Janet Woodcock with a Lifetime Achievement Award.

THE ABBEY S. MEYERS LEADERSHIP AWARD

NORD® RARE CANCER COALITION

NORD has been addressing the challenges that are consistent across all rare diseases since its inception in 1983. However, within the subgroup of rare cancers, there are unique issues that patients consistently face. The Rare Cancer Coalition (RCC) was created so the extensive resources and programs of NORD could be specifically applied to rare cancers. In addition, NORD has many member advocacy organizations focused on specific rare cancers. The RCC provides an opportunity for them to come together, network and share information. Prior to its formation, there was no other unified group of rare cancer advocacy organizations in the United States; by joining as a coalition, the RCC provides the strongest voice possible to raise awareness and advocate on behalf of rare cancer patients and researchers.

When the US Food and Drug Administration's Oncology Center of Excellence leadership sought to engage the rare cancer community on COVID-19's impact on research and to hear from rare cancer patients on how the pandemic has affected their participation in clinical trials, the RCC identified patients and caregivers who shared their questions and experiences on a special listening session. When the Department of Defense launched a new Rare Cancers Research Program through their Congressionally Directed Medical Research Programs, the RCC provided patient advocates and medical research speakers at special stakeholder meetings to represent the rare cancer community.

Last year, the coalition organized the first-ever global awareness campaign for all rare cancers, reaching patients, caregivers, medical professionals, industry and government stakeholders around the world. In 2020, #RareCancerDay focused on the possibilities of genomic testing, with informational videos, patient stories, and an educational webinar featuring experts from the Pediatric Oncology Branch of the National Institutes of Health and the Dana Farber Cancer Institute.

The world of rare cancers has been changing significantly in recent years. Government organizations as well as biotech and pharmaceutical companies are demonstrating specific interest in rare cancers. As many people living with rare cancers have similar needs and challenges, creating a more cohesive representation of these cancers allows for more visibility and participation. The RCC brings forward opportunities for collaboration across research, awareness and education, impacting critical areas such as access to care and new clinical trials.

This is an exciting time in the cancer research world, as the promise of advances like precision medicine and immunotherapy are improving outcomes for patients in real time. The Rare Cancer Coalition will continue its work to raise awareness of both the unique challenges faced by patients facing rare cancers, as well as the need for greater research funding.

NORD is honored to present the Rare Cancer Coalition with a 2020 Abbey S. Meyers Leadership Award.

2020 RARE IMPACT AWARD / HONOREE

Leadership Team | Clinic for Special Children

Pennsylvania's Clinic for Special Children has provided a medical home for patients with rare genetic diseases for over 30 years. By researching rare genetic mutations within the Lancaster County Amish and Mennonite communities, the Clinic bridges the gap between genomic research and the application of practical, patient-focused medicine. It is dedicated to the idea that advanced molecular genetic techniques can be integrated into local medical care to reduce childhood death, disability, and chronic illness.

From participating in clinical trials for gene therapy to innovating unique metabolic formulas that reduce brain damage in patients with disorders like glutaric aciduria type 1, the Clinic has impacted thousands of lives. Its work in preventing the suffering of vulnerable children and adults has had countless ripple effects for the entire community, beyond the Amish and Mennonite populations it serves. The power of genetic and biochemical knowledge is used to predict and prevent disabilities in children, and the integration of its clinical and laboratory services has generated many opportunities for research and scholarly work, including natural history studies, clinical trials and gene variant identification.

In the future, translational research and innovation will be crucial to advancements in treatments for rare disease. The Clinic has started new programs like the Plain Insight Panel™, a next generation sequencing assay that tests for over 1,300 genetic mutations in a single test, to ensure that practical, patient-focused medicine is applied using cutting-edge technologies to provide the best care possible for patients. Each year at the Clinic, new variants are discovered that cause disease in the populations it serves, demonstrating the ever-changing landscape of rare disease.

The Clinic's leadership team is grateful for being honored with a Rare Impact Award. "As a dedicated team of pediatricians, clinicians, nurses, scientific researchers, administrative staff and patient advocates, we share NORD's mission and focus on the identification, treatment and cure of rare diseases. We'd like to thank NORD for this honor, and we are so grateful to the staff, board, patients and supporters of the Clinic for Special Children."

NORD is honored to present the leadership team at the Clinic for Special Children with a 2020 Rare Impact Award.

2020 RARE IMPACT AWARD / HONOREE

Charlene York

For Charlene York, rare disease is truly a family affair; both her husband and 28-year-old daughter are living with congenital muscular dystrophy (CMD). CMD is a general term for a group of genetic muscle diseases that occur at birth or early during infancy and are characterized by diminished muscle tone, progressive muscle weakness and degeneration and other symptoms. Determined to make a difference for her loved ones and others living with CMD, she began volunteering with Cure CMD in 2008.

According to Charlene, when she attended the NORD Rare Diseases and Orphan Products Breakthrough Summit for the first time in 2009, it broadened her view of the critical need for education, research and advocacy for all rare diseases. After successfully getting the last day of February permanently declared Rare Disease Day in Ohio, she decided to become more involved with the Ohio Rare Action Network (RAN) in order to raise awareness regarding rare diseases at the state and local levels.

In the CMD world, the avenues of both genetic therapy and drug development are currently being explored. For Charlene, it is exciting to see how researchers, patients and families are coming together to find a cure or treatment. It is also gratifying to know that even while a cure is being sought, living a happy, fulfilling life is possible. Both her husband and daughter are doing well. Their rare disease is part of their lives, but it doesn't define either of them.

When asked about the impact she is proudest of making for people living with rare diseases, Charlene mentions being able to bring people together and helping them to make connections as Ohio RAN Volunteer State Ambassador.

"I remember at one of the first Rare Disease Day events that I organized here in Ohio, I witnessed a woman meeting someone else with her disease for the very first time. She had lived so long with the disease and was finally able to connect with someone who knew exactly what she was going through. We were all in tears," Charlene says. "As for myself, I have always liked doing things for others and enjoy meeting new people. Making connections and seeing those connections take root is so satisfying."

NORD is honored to present Charlene York with a 2020 Rare Impact Award.

2020 RARE IMPACT AWARD / HONOREE

Erica Barnes

Erica Barnes lost her young daughter Chloe to a rare and aggressive degenerative disease, metachromatic leukodystrophy (MLD). "I am inspired by Chloe's courage and also the bravery of the families I have met along the way," Erica says. "I have a vision that I can rewrite the ending for future children with rare diseases so that they have a happy ending."

Determined to make a difference in her home state of Minnesota, Erica has made addressing the needs of the rare disease community a top priority. As she tells it, her work with the Rare Action Network in Minnesota has made her grief in losing Chloe a point of connection with others who have also suffered such a loss. Through her advocacy work, Erica has represented NORD as a Minnesota Rare Action Network Volunteer State Ambassador and provided public testimony at the federal level in support of newborn screening. She frequently speaks at national conferences and contributes to publications focused on rare diseases.

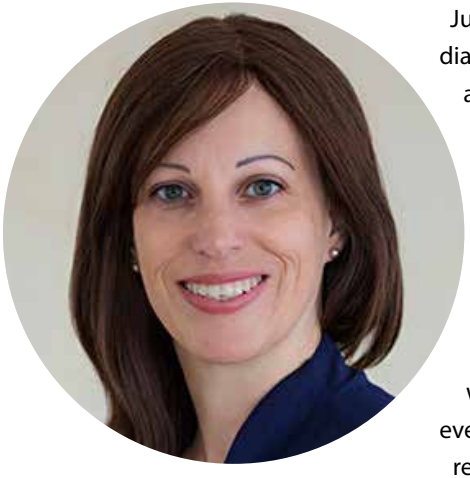
Thanks to a bill she fought long and hard for that bears Chloe's name, Minnesota is now among a handful of states that have created an advisory council to advocate for the research and treatment of rare diseases. Erica is the council administrator of the Chloe Barnes Rare Disease Advisory Council and a member of the University of Minnesota Rare Disease Day planning committee. The vision of the Council is a Minnesota where every patient diagnosed with a rare disease has access to a timely diagnosis, appropriate care and an effective treatment.

"Awards are meaningful to the degree that the organization awarding them is meaningful. To be honored by an organization as respected and dedicated to patients and families as NORD is incredibly humbling and validating," Erica says. "I don't love having personal attention on me, but I am proud to accept this Rare Impact Award on behalf of all the amazing Minnesota advocates I work alongside, and in honor of all the patients and families I have encountered along the way."

NORD is honored to present Erica Barnes with a 2020 Rare Impact Award.

2020 RARE IMPACT AWARD / HONOREE

Julia Vitarello and Timothy Yu, MD, PhD



Julia Vitarello, Founder and CEO of Mila's Miracle Foundation and Dr. Timothy Yu, a clinical neurologist and neurogeneticist at Boston Children's Hospital, have worked closely to pioneer the first genetic treatment customized for a single patient. Together, they are turning a long-held dream into reality.

Julia's daughter Mila was six years-old when diagnosed with a rare variant of Batten disease, and the prognosis was grim. The active little girl was destined to lose her vision, her ability to talk, walk, swallow and eventually her life. But Mila's diagnosis was missing one mutation. No lab could find it. Dr. Timothy Yu committed to the challenge. He investigated Mila's genome, spending evenings and weekends in his lab until he and his team eventually identified a very unusual retrotransposon—or "jumping gene."

In Mila's case, the inserted DNA was an unwanted guest that created what the cell thought was an exon, or protein-coding gene. It was disrupting assembly of the CLN7 gene, in turn compromising production of the CLN7 protein.

Dr. Yu then thought of how he might be able to give Mila a chance at life. In less than a year, he and his team designed, received approval for and delivered a genetically tailored antisense oligonucleotide (ASO) drug to Mila. The world's first FDA-approved personalized medicine was affectionately named "Milasen." Mila's treatment would not have been possible without the tireless efforts of Dr. Yu and the determination of Julia, who worked with him to make crucial decisions while raising most of the funds necessary to make her daughter's treatment possible.

The Milasen experience demonstrates the feasibility, safety and efficacy of rapid-turnaround design and delivery of ASO treatments. Mila became the first person ever to receive an FDA-approved drug created to treat just one individual, and her story has made its way into the mainstream media. Dr. Yu and Julia are now working to create a new personalized, rapid-turnaround treatment path for children across many life-threatening neurological diseases.

Scientists around the world are working to replicate Mila's treatment approach, giving hope to more children across several other rare and fatal diseases like Batten.

NORD is honored to present Julia Vitarello and Dr. Timothy Yu with a 2020 Rare Impact Award.

2020 RARE IMPACT AWARD / HONOREE

Norman Winnerman

Norman Winnerman became involved in the world of rare disease after the birth of the second of his three granddaughters. Alison Wells was born with a rare genetic birth defect diagnosed as Cornelia de Lange syndrome (CdLS). As Norm would soon learn, CdLS is named after Dr. Cornelia de Lange of the Netherlands, who in 1933 observed two children with similar physical features and medical problems in common and is now credited with describing the collection of symptoms composing the syndrome that bears her name.

A Connecticut resident, Norm knew about NORD and reached out to the organization immediately after Alison's diagnosis. When he learned about the CdLS Foundation in Collinsville, CT, he met with the foundation's Executive Director, who explained all she could about the rare disease and its prognosis. Norm volunteered to help organize their 1992 Boston Conference, and soon began working on the CdLS Foundation's National Family Conferences on an ongoing basis. He has has done so ever since and has also served a three-year term on the CdLS Board of Directors.

Shortly after retiring from a career in public education, Norm was offered a part-time job that wound up lasting 20 years, serving as Commissioner of the South-West Conference, a high school athletic league in Southwestern Connecticut. When he retired for the second time, he walked into the NORD office in Danbury and offered to help. Much to the delight of everyone at NORD since then, he's been a regular visitor.

Sadly, Alison died suddenly at the age of four and a half, the result of a medical complication associated with the syndrome. Norm's role as a volunteer with the CdLS Foundation and NORD has enabled him to work behind the scenes to lighten the load and help the staff members of both organizations do their core jobs: serving patients and families.

Being recognized for the volunteer work he's done over the past 30 years is, in Norm's words, "A great honor not only for me, but also for my wife and my entire family. I know that I would not be here with all of you were it not for a tiny little girl named Alison, who touched my heart and introduced me to a world I would never have known. My life has been enriched by the experience."

NORD is honored to present Norman Winnerman with a 2020 Rare Impact Award.

2020 RARE IMPACT AWARD / HONOREE

Patrick James Lynch

Hemophilia has long been part of Patrick James Lynch's life; he lives with the rare bleeding disorder, lost his younger brother to it, and has spent much of his professional career dedicated to improving the lives of hemophilia patients and families. Unlike most rare diseases, there are numerous treatment options for hemophilia A, with many more in the works. Patrick notes that it is vital for people with hemophilia to understand the treatment landscape in order to make fully empowered decisions about management options and risk tolerance.

Media and entertainment have the power to spark imagination, decrease loneliness, increase connectivity and empower young people to see themselves in new ways. Patrick's company, Believe Limited, creates educational, awareness-building, and fundraising content for leading national and international patient advocacy groups. Since 2015, Believe Limited has honored over 350 teens affected by hemophilia and other rare blood disorders at the largest annual patient conference for hemophilia, and has built a theater and arts therapy program for teens impacted by the disorder. The educational web series Patrick created has reached thousands of people online and lead to features in BuzzFeed, Fox News, PBS and the Huffington Post. His documentary on the first hemophiliac to attempt to climb Everest and the global disparities in hemophilia care has been seen by people around the world.

Patrick and Believe Limited have helped to raise funds toward assisting communities in developing countries and support millions of dollars' worth of treatment product being donated to hemophiliacs abroad via humanitarian aid programs. He is extremely proud of the countless lives Believe Limited's work has touched.

"The Rare Impact Award tells me that NORD recognizes the importance of supporting strong patient voices who are using media and entertainment to make a difference in the lives of patients and families, and to influence the public's perception of what it means to live with a rare disease. I am truly grateful for this honor," says Patrick.

NORD is honored to present Patrick James Lynch with a 2020 Rare Impact Award.

2020 RARE IMPACT AWARD / HONOREE

The Honorable Senator Sherrod Brown

Senator Sherrod Brown has represented Ohio in the US Senate since 2007. Prior to serving in the Senate, he served as a United States Representative for the 13th District for 14 years. He also previously served as Ohio's Secretary of State, a member of the Ohio General Assembly, and has taught in Ohio's public schools and at The Ohio State University.

Throughout his time in public office, Senator Brown has been a champion of causes that benefit the rare disease community. He has been the lead Senate sponsor of the Rare Disease Day resolution for many years and is a co-sponsor of several bills in the Senate that are beneficial to the rare disease community. He played a key role in helping to pass the Affordable Care Act, which has made health insurance more affordable and accessible for American families. This law also provides strong consumer protections that will prevent insurers from placing limits on the care patients receive and discriminating against consumers based on age or pre-existing health conditions. He has continued to support legislation to bolster patient protections and make quality, comprehensive health care more affordable for patients. Senator Brown has cosponsored legislation to protect patients from surprise medical bills, to limit access to "junk" insurance products that don't cover essential medical services, and to support non-emergency medical transportation for patients with Medicaid. The Senator has led efforts to improve mental health care options for Medicare beneficiaries, and introduced legislation to help low-income women and children access primary and preventative care.

He is married to Pulitzer Prize-winning columnist Connie Schultz. They reside in Cleveland, Ohio, and have three daughters, a son, a daughter-in-law, three sons-in-law, and seven grandchildren.

NORD is honored to present Senator Sherrod Brown with a 2020 Rare Impact Award.

2020 RARE IMPACT AWARD / HONOREE

The Honorable Representative Randi Clites

State Representative Randi Clites (D-Ravenna) currently represents Ohio's 75th House District, which includes much of central and southern Portage County.

A grassroots advocate at heart, Representative Clites began her activism when her son was diagnosed with a rare genetic bleeding disorder, then leukemia, at a young age. She has since dedicated herself to giving back to the community that has given her so much, fighting to increase access to quality, affordable health care and improve community services.

Representative Clites has worked as Associate Director of the Northern Ohio Hemophilia Foundation, Advocacy Coordinator for the Ohio Bleeding Disorders Council and has represented Ohio through the Association of Maternal and Child Health Family Scholar Program.

She served on the 2006 Legislative Task Force on the Future Funding of the state's Title V program, where she fought for Ohio's most vulnerable children. She has served as Chair of the Parent Advisory Council and is a past recipient of the Advocate of the Year award from the National Hemophilia Foundation for her continued work to increase awareness at the local, state and national levels.

In addition, she has been an active volunteer in her community, working with Akron Children's Hospital, Portage County Big Brothers and Sisters and serving as a long-time softball coach in her community.

NORD is honored to present Representative Randi Clites with a 2020 Rare Impact Award.

2020 RARE IMPACT AWARD / HONOREE

Peter Frampton

Frampton Comes Alive! is Peter Frampton's breakout album, which established him as a superstar. It's among the best-selling live albums of all time, selling more than 17 million albums worldwide, producing hit singles that have never gone out of rotation on radio, and has become a coming-of-age totem for generations of fans who grew up in the '70s and '80s.

Following a hiatus in the 1990s, Peter Frampton has remained active and ambitious in his playing. On 2006's Fingerprints, he teamed up with an all-star backing band—including members of the Rolling Stones, Pearl Jam and Frampton's own guitar hero, Hank Marvin and the Shadows—for an inventive set of originals and covers that won a Grammy Award for Best Instrumental Pop Album.

In many ways he remains a fan at heart, awed by the talents and techniques of his peers. That's what motivated him in 2013 to launch the Guitar Circus, a touring revue featuring instrumentalists from various genres and generations. Each night featured a different lineup, from Robert Cray to the late, great B.B. King.

That experience informed his latest album, All Blues, which debuted at the top of the Billboard blues chart. The blues is about connection and commiseration, but also celebration; in declaring their pain, blues artists are also proclaiming that they are alive. That's especially personal to Frampton, who was diagnosed with inclusion body myositis (IBM), a rare inflammatory disease that weakens and atrophies the muscles in the arms, hands and legs. "I'll always be able to sing, but I don't know how much longer I'll be able to play guitar. Fortunately, I have a bunch of friends who can play guitar and help me out. Some of them are pretty good, too!"

Frampton's sense of humor remains intact, as does his determination to combat inclusion body myositis and other rare diseases. "My main thing outside music is the fundraising I'm doing for myositis research at Johns Hopkins. I hope to become the face of IBM and to advocate for it and all autoimmune diseases. My work is shouting loudly and raising money for our research," he says. "I'm honored to receive the Rare Impact Award. It lifts my spirit to know that my voice can help raise awareness for funding research needed for rare diseases."

NORD is honored to present Peter Frampton with a 2020 Rare Impact Award.

Alnylam Pharmaceuticals for Givlaari™

On November 20, 2019 the FDA granted approval to Givlaari (givosiran) for the treatment of adult patients with acute hepatic porphyria, a genetic disorder resulting in the buildup of toxic porphyrin molecules that are formed during the production of heme (which helps bind oxygen in the blood).

"This buildup can cause acute attacks, known as porphyria attacks, which can lead to severe pain and paralysis, respiratory failure, seizures and mental status changes. These attacks occur suddenly and can produce permanent neurological damage and death," said Richard Pazdur, MD, Director of the FDA's Oncology Center of Excellence and acting Director of the Office of Oncologic Diseases in the FDA's Center for Drug Evaluation and Research. "Prior to today's approval, treatment options have only provided partial relief from the intense unremitting pain that characterizes these attacks. The drug approved today can treat this disease by helping to reduce the number of attacks that disrupt the lives of patients."

The approval of Givlaari was based on the results of a clinical trial of 94 patients with acute hepatic porphyria. Patients received a placebo or Givlaari. Givlaari's performance was measured by the rate of porphyria attacks that required hospitalizations, urgent health care visits or intravenous infusion of hemin at home. Patients who received Givlaari experienced 70% fewer porphyria attacks compared to patients receiving a placebo.

NORD is proud to present Alnylam Pharmaceuticals with a 2020 Industry Innovation Award for bringing this important new treatment to patients.

**FDA press release announcing the approval of the treatment.*

AveXis, a Novartis Company for Zolgensma®

On May 24, 2019, the FDA approved Zolgensma (onasemnogene abeparvovec-xioi), as the first gene therapy to treat children less than two years of age with spinal muscular atrophy (SMA), the leading genetic cause of infant mortality. Zolgensma is a one-time treatment designed to address the genetic root cause of the disease by replacing the function of the missing or nonworking SMN1 gene. Administered during a single, intravenous infusion, Zolgensma delivers a new, working copy of the SMN1 gene into a patient's cells, halting disease progression.

At the time of approval, acting FDA Commissioner Ned Sharpless, MD, said "The potential for gene therapy products to change the lives of those patients who may have faced a terminal condition, or worse, death, provides hope for the future."

SMA is a rare genetic disease caused by a mutation in the survival motor neuron 1 (SMN1) gene. The gene encodes the survival motor neuron (SMN) protein – a protein found throughout the body, which is critical for the maintenance and function of specialized nerve cells, called motor neurons. Motor neurons in the brain and spinal cord control muscle movement throughout the body. If there is not enough functional SMN protein, then the motor neurons die, leading to debilitating and often fatal muscle weakness. SMA caused by mutations in the SMN1 gene is generally classified into several subtypes, based on the age of onset and severity; infantile-onset SMA is the most severe and most common subtype. Children with this condition have problems holding their head up, swallowing and breathing. These symptoms may be present at birth or may present by the age of 6 months.

Zolgensma is indicated for the treatment of children less than two years of age with SMA. The product is an adeno-associated virus vector-based gene therapy that targets the cause of SMA. The vector delivers a fully functional copy of human SMN gene into the target motor neuron cells. A one-time intravenous administration of Zolgensma results in expression of the SMN protein in a child's motor neurons, which improves muscle movement and function, and survival of a child with SMA. Dosing is determined based on the weight of the patient.

AveXis, a Novartis company, is dedicated to developing and commercializing gene therapies for patients and families devastated by rare and life-threatening neurological diseases.

NORD is proud to present AveXis, a Novartis company, with a 2020 Industry Innovation Award for bringing this important new treatment to patients.

Daiichi-Sankyo for Turalio™

On August 02, 2019, the FDA granted approval to Turalio (pexidartinib) capsules for the treatment of adult patients with symptomatic tenosynovial giant cell tumor (TGCT) associated with severe morbidity or functional limitations and not responsive to improvement with surgery.

"TGCT can cause debilitating symptoms for patients such as pain, stiffness and limitation of movement. The tumor can significantly affect a patient's quality of life and cause severe disability," said Richard Pazdur, MD, Director of the FDA's Oncology Center of Excellence and acting Director of the Office of Hematology and Oncology Products in the FDA's Center for Drug Evaluation and Research. "Surgery is the primary treatment option, but some patients are not eligible for surgery, and tumors can recur, even after the procedure. Today's approval is the first FDA-approved therapy to treat this rare disease."

TGCT is a rare tumor that affects the synovium (thin layer of tissue that covers the surfaces of the joint spaces) and tendon sheaths (layer of membrane that covers tendons, which are fibrous tissue that connect muscle to bone). The tumor is rarely malignant but causes the synovium and tendon sheaths to thicken and overgrow, causing damage to surrounding tissue.

NORD is proud to present Daiichi Sankyo with a 2020 Industry Innovation Award for bringing this important new treatment to patients.

Daiichi-Sankyo

Genentech for Rozlytrek™

On August 15, 2019, the FDA granted accelerated approval to entrectinib (ROZLYTREK, Genentech Inc.) for adults and pediatric patients 12 years of age and older with solid tumors that have a neurotrophic tyrosine receptor kinase (NTRK) gene fusion without a known acquired resistance mutation, are metastatic or where surgical resection is likely to result in severe morbidity, and have progressed following treatment or have no satisfactory standard therapy.

FDA also approved entrectinib for adults with metastatic non-small cell lung cancer (NSCLC) whose tumors are ROS1-positive.

Genentech

Efficacy in NTRK-positive tumors was investigated in 54 adult patients who received entrectinib at various doses and schedules in one of three multicenter, single-arm, clinical trials: ALKA, STARTRK-1 (NCT02097810) and STARTRK-2 (NCT02568267); 94% received entrectinib 600 mg orally once daily. Identification of positive NTRK gene fusion status was determined in local laboratories or a central laboratory using nucleic acid-based tests prior to enrollment.

Among 54 adult patients, the overall response rate as determined by independent review was 57% (95% CI: 43, 71). Response duration was 6 months or longer for 68% of patients and 12 months or longer for 45% of patients. The most common cancers were sarcoma, NSCLC, mammary analogue secretory carcinoma, breast, thyroid, and colorectal.

Efficacy in ROS1-positive metastatic NSCLC was investigated in 51 adult patients who received entrectinib at various doses and schedules in the same three trials; 90% received entrectinib 600 mg orally once daily. The overall response rate was 78% (95% CI: 65, 89) and response duration was 12 months or longer for 55% of patients.

NORD is proud to present Genentech with a 2020 Industry Innovation Award for bringing this important new treatment to patients.

Global Blood Therapeutics for Oxbryta™

On November 25, 2019, the FDA granted accelerated approval to voxelotor (Oxbryta, Global Blood Therapeutics) for adults and pediatric patients 12 years of age and older with sickle cell disease (SCD).

SCD is a rare genetic blood disorder caused by abnormal hemoglobin, the protein in red blood cells that delivers oxygen throughout the body. SCD affects approximately 100,000 people in the United States and millions worldwide. It disproportionately affects those of African descent, along with people of Hispanic, South Asian, Southern European, and Middle Eastern ancestry.

Oxbryta is the first and only FDA-approved therapy that directly inhibits hemoglobin polymerization, the root cause of the sickling and destruction of red blood cells that occurs in all people with SCD. Oxbryta works by binding to hemoglobin, thereby increasing the red blood cell's affinity for oxygen. This keeps the red blood cell in an oxygenated state and prevents it from sickling.

Oxbryta was evaluated in more than 700 trial participants as part of a robust clinical program. Data from the Phase 3 HOPE Study, which were published in The New England Journal of Medicine in June 2019, showed that patients on Oxbryta achieved a rapid, robust and sustained improvement in hemoglobin and reduced red blood cell destruction (hemolysis), and that Oxbryta has an acceptable safety profile. Based on these data, the FDA granted accelerated approval of Oxbryta three months before the Priority Review action date and less than 5 years after initial studies in humans.

The approval of Oxbryta represents an important shift in the SCD treatment paradigm. It moves the field away from managing symptoms of SCD to addressing the root cause of the disease and potentially preventing the long-term morbidity and mortality associated with it.

NORD is proud to present Global Blood Therapeutics with an Industry Innovation Award during the 2020 Rare Impact Awards for bringing this important new treatment to patients.

2020 INDUSTRY INNOVATION AWARD / HONOREE

Vertex Pharmaceuticals for Trikafta®

On October 21, 2019 the FDA approved TRIKAFTA® (elexacaftor/tezacaftor/ivacaftor and ivacaftor), the first triple combination therapy available to treat patients with the most common cystic fibrosis mutation. TRIKAFTA is approved for patients 12 years and older with cystic fibrosis in the United States who have at least one F508del mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, which is estimated to represent up to 90% of the cystic fibrosis population.

On the day of the approval, acting FDA commissioner Ned Sharpless, MD said, "At the FDA, we're consistently looking for ways to help speed the development of new therapies for complex diseases, while maintaining our high standards of review. Today's landmark approval is a testament to these efforts, making a novel treatment available to most cystic fibrosis patients, including adolescents, who previously had no options and giving others in the cystic fibrosis community access to an additional effective therapy. In the past few years, we have seen remarkable breakthroughs in therapies to treat cystic fibrosis and improve patients' quality of life, yet many subgroups of cystic fibrosis patients did not have approved treatment options. That's why we used all available programs, including Priority Review, Fast Track, Breakthrough Therapy, and orphan drug designation, to help advance today's approval in the most efficient manner possible, while also adhering to our high standards."

Cystic fibrosis, a rare, progressive, life-shortening disease, results in the formation of thick mucus that builds up in the lungs, digestive tract and other parts of the body. It leads to severe respiratory and digestive problems as well as other complications such as infections and diabetes.

NORD is proud to present Vertex Pharmaceuticals Incorporated with an Industry Innovation Award for bringing this important new treatment to patients.

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We're proud to be recognized as one of six honorees for NORD's (National Organization for Rare Disorders) 2020 Industry Innovation Award. Through continued conversations and collaborations, we can all make a meaningful difference for rare disease communities.

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Daiichi-Sankyo

PP-US-ON-1122 08/20

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RareLaunch®

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It is estimated that more than 50% of rare diseases don't have organized representation or support, which leaves patients searching for resources and connections. NORD's **RareLaunch® Forming a Foundation** program provides education, training and support for patients to start nonprofits; for those already involved in nonprofits, the program also builds capacity and implements good governance practices to ensure their development is sustainable.

Topics addressed as part of this program include:

- Establishing a 501c(3) tax-exempt organization
- Leadership/Executive Director training
- Resources for recruiting a Board of Directors and Scientific and/or Medical Advisory Committee(s)
- Promotion of the newly established organization to continue to find and connect with members of the community through a targeted communications approach
- Development of social media platforms and strategies for continued engagement

Visit **rarediseases.org/nord-rarelaunch**
to learn more about NORD's RareLaunch® program

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The National Organization for Rare Disorders (NORD) is leading the fight to improve the lives of patients with rare diseases. We do this by supporting patients and organizations, accelerating research, providing education, disseminating information, raising public awareness and driving public policy.

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