

RARE IMPACT AWARDS®

JUNE 28 2021

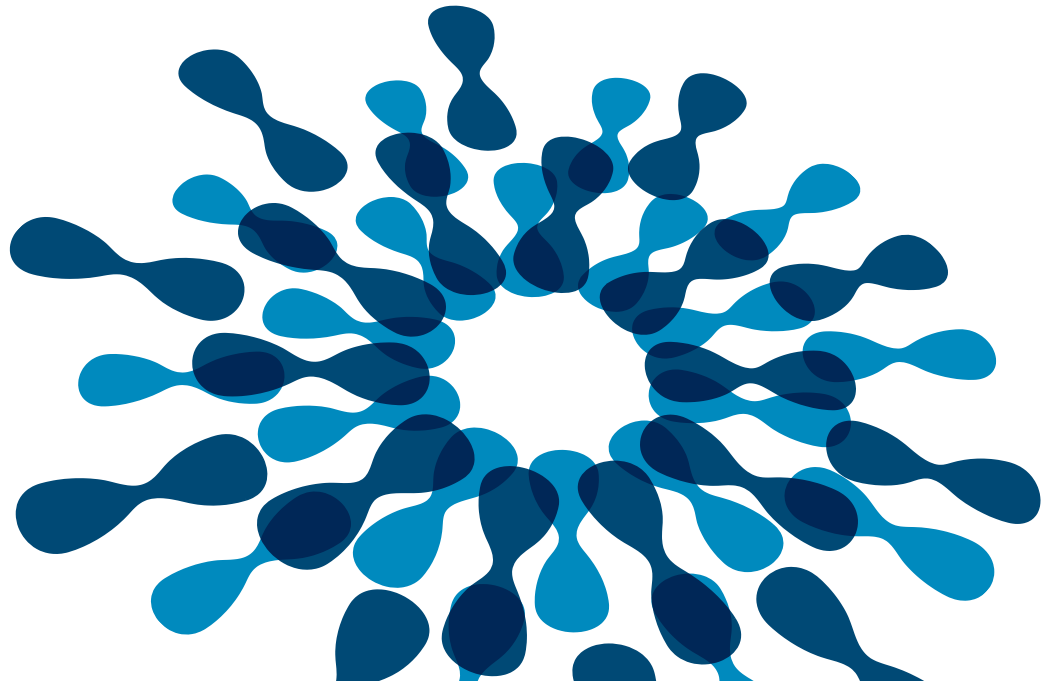


NORD®
National Organization
for Rare Disorders



NORD[®]
National Organization
for Rare Disorders

WELCOMES YOU



TO THE 2021 RARE IMPACT AWARDS®



DTRF's co-founders,
Marlene Portnoy and
Jeanne Whiting

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

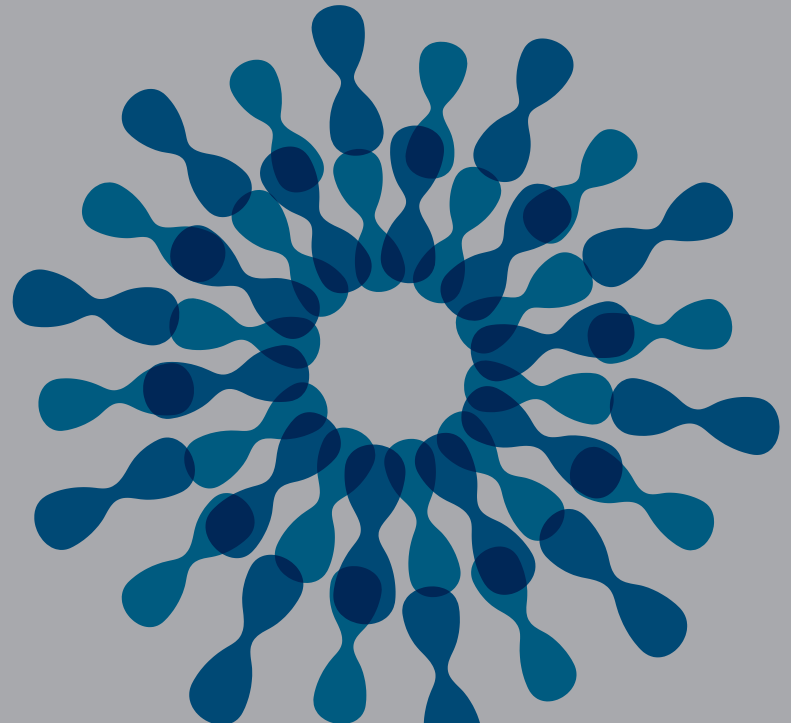
Ali Stroker

PRESENTATION OF AWARDS

MUSICAL GUEST

Ali Stroker

PRESENTATION OF AWARDS



DEAR FRIENDS



Thank you for joining us for the 2021 Rare Impact Awards program, presented as a virtual event as we continue to do our part to keep our community connected but safe during the COVID-19 pandemic. Despite the enormous challenges that we have faced during the pandemic, there are heroes to be found among us from whom we can draw inspiration and motivation to keep moving forward.

At the National Organization for Rare Disorders, we are proud to honor these people, groups and companies for their achievements. We're humbled to work alongside them and appreciate their tenacity and commitment to creating a better future for our community.

Last year it was hard to anticipate what 2021 might bring. For many, it's been hopeful as the rollout of vaccines puts the worst of the pandemic behind us. But there is still much to be done to deliver on the promise of better standards-of-care and more accessible healthcare, faster and more accurate diagnosis for patients, and greater treatment options for all rare diseases.

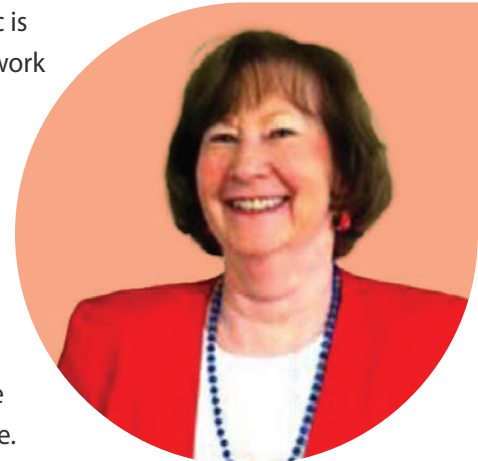
One lesson from the pandemic is clear – we are stronger when we work together, unified in our cause. It's how we started this community as patients and caregivers advocating for the Orphan Drug Act back in the early 1980s, and it's how we will continue to make an impact for the over 25 million Americans and 300 million people worldwide affected by rare disease.

We'd like to take a moment to thank the tireless rare disease advocates who continue to drive change that benefits the entire community, congratulate the companies that had a therapy approved this past year, and acknowledge the hard work and dedication of the leaders at the US Food and Drug Administration and the National Institutes of Health, 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 & CEO



KAY HOLCOMBE
CHAIR, BOARD
OF DIRECTORS



EMCEE FOR THE EVENING

JOHN WHYTE MD, MPH

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 for 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 they could comprehend "to" 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.

Dr. Whyte is also a bestselling author and regular contributor to WebMD magazine.

MUSICAL PERFORMANCE

ALI STROKER



Paralyzed from the chest down since age two, Ali Stroker has faced overwhelming adversity. Despite often appearing on stages with limited accessibility, she is a trailblazer. Ali won a 2019 Tony Award for her role as Ado Annie in *Oklahoma!*, making her the first actress who uses a wheelchair for mobility to do so.

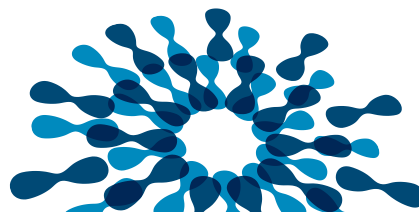
It wasn't the first time Ali made history. In 2015 she was the first actress in a wheelchair to appear on Broadway when she originated the

role of Anna in Deaf West's acclaimed revival of *Spring Awakening*. She is also the first actress in a wheelchair to graduate from the NYU Tisch drama program.

She has also appeared on several popular television shows including, *The Glee Project*, *Glee*, *Lethal Weapon*, and Comedy Central's *Drunk History*. She's performed solo at the Kennedy Center in Washington, DC, New York's Town Hall, Lincoln Center for the Performing Arts and Carnegie Hall.

A humanitarian and advocate, Ali is a co-chair of Women Who Care, which supports United Cerebral Palsy of New York City. She's a founding member of Be More Heroic, an anti-bullying campaign that tours the country and connects with thousands of students each year. Ali's remarkable ability to improve

the lives of others through the arts, whether disabled or not, is captured in her motto: "Making your limitations your opportunities."



2021 HONOREES



2021
RARE
IMPACT
AWARDS
HONOREES

2021 PUBLIC HEALTH LEADERSHIP AWARD

PETER MARKS

MD,
PHD
DIRECTOR, CENTER FOR
BIOLOGICS EVALUATION
AND RESEARCH, US
FOOD AND DRUG
ADMINISTRATION



Peter Marks, MD, PhD has been a leader in the fight against COVID-19. As the Director of the Center for Biologics Evaluation and Research (CBER) at the US Food and Drug Administration (FDA), he has been part of reviewing and authorizing vaccines and therapies to combat the pandemic, while also continuing to champion the critical work of advancing

treatment options for rare diseases.

CBER is responsible

for assuring the safety and effectiveness of biological products, including vaccines, allergenic products, blood, and blood products, and cellular, tissue, and gene therapies. Dr. Marks, together with his team at the FDA, review and provide advice during biological product development; evaluate applications and make approval decisions based on safety and effectiveness data; monitor the safety of biological products, and conduct research that supports future discoveries.

Dr. Marks received his

graduate degree in cell and molecular biology and his medical degree at New York University. He completed an internal medicine residency and hematology/medical oncology fellowship at Brigham and Women's Hospital in Boston, where he subsequently joined the attending staff as a clinician-scientist and eventually served as Clinical Director of Hematology.

After spending several years in the pharmaceutical industry, Dr. Marks subsequently returned to academic medicine at Yale University where he led the adult leukemia service and served as Chief Clinical Officer of Smilow Cancer Hospital. He joined the FDA in 2012 as Deputy Center Director for CBER, becoming the Director in 2016. Dr. Marks is board certified in internal medicine, hematology and medical oncology, and is a Fellow of the American College of Physicians.

NORD is honored to present Dr. Peter Marks with a 2021 Public Health Leadership Award.



THE ABBEY S. MEYERS LEADERSHIP AWARD



THE DESMOID TUMOR RESEARCH FOUNDATION

In 2005, Marlene Portnoy and Jeanne Whiting co-founded the Desmoid Tumor Research Foundation (DTRF). It started at a kitchen table with an audacious goal to transform their community and the outlook for those fighting desmoid tumors, benign soft tissue tumors also known as aggressive fibromatoses. DTRF's path to meeting that goal is through driving research. Desmoid tumors, while not life-threatening themselves, can cause life-threatening problems if they compress critical internal organs, blood vessels or nerves.

DTRF initiated a groundbreaking project to develop the first validated

Patient Reported Outcome tool in desmoid tumors and created the first ever US patient registry specifically for desmoid tumor patients. Data from that registry, created in partnership with NORD, is now part of another landmark global initiative, the Rare Disease Cures Accelerator – Data and Analytics Platform (RDCA-DAP). RDCA-DAP seeks to use data across the rare landscape to speed clinical development of new treatments and drive innovation.

DTRF has raised millions of dollars to fund cutting-edge research that is developing treatment options

and uncovering new disease insights. In the community of rare diseases and cancer, DTRF has become known as a model of how patient advocates can power research through collaboration, making a significant impact across diseases and setting the standard for how the drug development process can be made better through direct and robust patient engagement at every level.

DTRF initiated a major multi-institution collaborative drug screening project that yielded important results.

Five promising drugs were identified as inhibiting desmoid tumor cell growth without affecting normal

fibroblast cells. Drug screen projects are typically funded by pharmaceutical companies, which all too often means that rare diseases aren't included. DTRF's initiation and funding of such a large collaborative project for uncovering new possible treatment options for desmoid tumors is now a model in the rare disease community.

NORD is honored to present the Desmoid Tumor Research Foundation with the 2021 Abbey S. Meyers Leadership Award.



JADE DAY



Jade Day knows first-hand the challenges of raising a child with several rare diseases, one of which is FG Syndrome Type 1. She refused to accept that her son Gavin's quality of life, even if he lived, would be exceptionally low. It took over seven years for Gavin to get a diagnosis, but with Jade as his champion, he continues to thrive in spite of the odds.

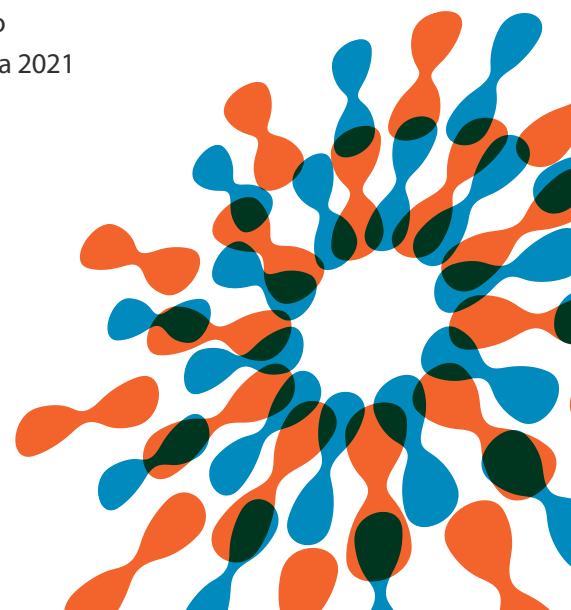
Outside her home life of being a mother to her three boys, Jade is Vice President of A Twist of Fate – Arterial Tortuosity Syndrome. Jade also helped form the first Rare Disorders Workgroup at the

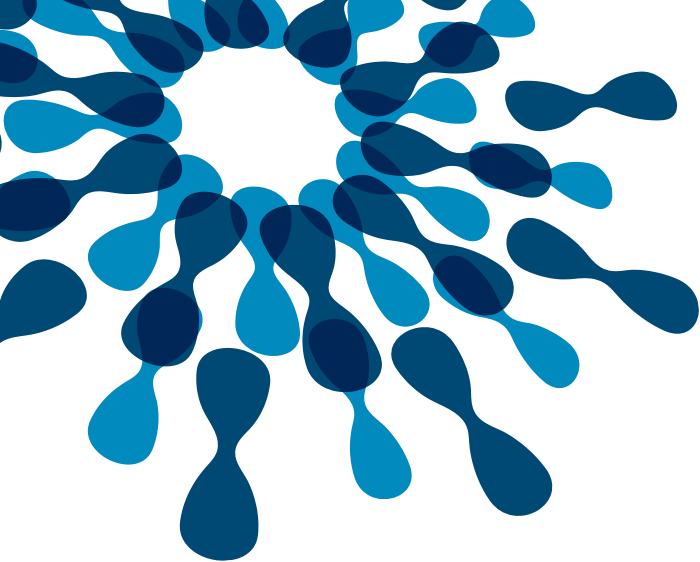
Indian Healthcare Services and lobbied nationally for rare disease causes. As a member of the Cherokee Nation, Jade has worked actively to bring awareness to the existence and prevalence of rare diseases within the Native American community.

Jade's efforts for her son have resulted in significant contributions to the broader rare disease landscape. She has served in a leadership capacity as part of NORD's Rare Action Network in Oklahoma. In July of 2016, Jade and her three sons traveled to the Primary Children's Hospital at the University of Utah in Salt Lake City to meet with Dr. John Opitz, the geneticist who

discovered FG1 Syndrome. For one week, Jade and Gavin were tested and interviewed for research purposes on both X-linked carriers and FG1 Syndrome. This research will impact genetics research for decades to come and will hopefully lead to a discovery that will improve the lives of millions.

NORD is honored to present Jade Day with a 2021 Rare Impact Award.





2021 RARE IMPACT AWARD ✨ HONOREE

DANIELA DELGADO



Thirteen-year-old Daniela Delgado lives with severe Von Willebrand (VWD) Type 1C and Ehlers-Danlos Syndrome (EDS), but that has not slowed down her efforts to help her community. She realized early on that kids with life-threatening illnesses and disabilities needed something to smile about. Coming from a family of avid bakers, Daniela decided that creating special customized cakes for these children was the best way to accomplish her dream of more smiles – and that is how her non-profit organization, Daniela's Little Wish was born.

Daniela posts regularly on social media to help educate others and drive rare disease awareness. Pre-pandemic, she traveled around the country giving motivational speeches to children and their families in the bleeding disorders community. Daniela has been a guest speaker at the NORD Rare Disease Day event in Connecticut, advocating for children, like herself, living with chronic conditions.

At her young age, Daniela's work in advocating, educating, and raising awareness for all those facing bleeding disorders gives hope and inspiration to others living with painful and debilitating diseases. Her efforts help empower other young people to look beyond their diseases to find that their illness does not need to define them or limit their purpose in life. At 13 years of age, she exemplifies what it means to be an individual successfully rising above to create a way to be of service to others.

Through Baking Smiles for Kids, her baking classes for people of all ages, and her efforts to educate and increase bleeding disorder awareness, Daniela has touched and inspired many with her story and her passion.

NORD is honored to present Daniela Delgado with a 2021 Rare Impact Award.

2021 RARE IMPACT AWARD ✨ HONOREE

SUSAN

FERNBACH RN, BSN



Susan Fernbach's passion is providing genetic education for patients, families and clinicians. She created the Evenings with Genetics seminar series aimed at demystifying the diagnosis, care and research for genetic disorders, with each month featuring a different topic with genetics faculty and family speakers.

With Department of State Health Services funding, Susan has led seminars and resource fairs for underserved areas across Texas, many featuring simultaneous Spanish translation. In partnership with The Arc of Greater Houston, Susan

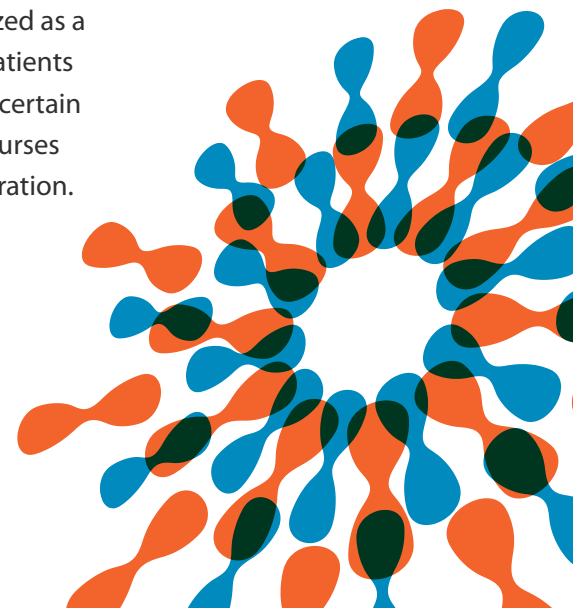
has planned six annual conferences, provided in Spanish, for families in the Houston area dealing with rare and genetic disorders.

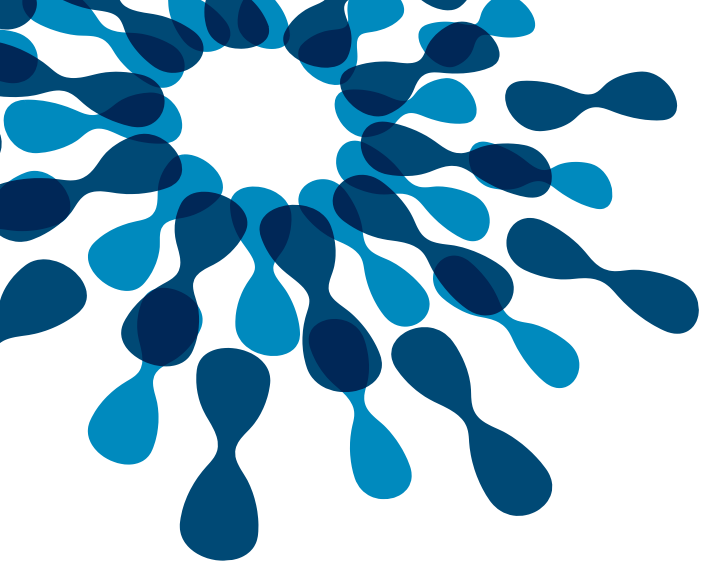
In 2016, Susan planned the first annual Houston Rare Disease Day event with over 35 rare disease support organizations represented. This effort has grown tremendously and now includes a committee of faculty, staff, and leaders of rare disease groups from across the state. She reaches out to the wider medical educational community, and in turn has been a speaker at events hosted by school districts as well as at nursing

organizations and schools of nursing. She is honored to do her part in raising rare disease awareness and inviting others to engage with and join our broader rare community.

Susan's work has brought together a network of organizations and advocacy groups to provide education and resources for families. She has been recognized as a champion for those patients and families facing uncertain futures by the Texas Nurses Annual Nursing Celebration.

NORD is honored to present Susan Fernbach with a 2021 Rare Impact Award.





2021 RARE IMPACT AWARD 🌟 HONOREE

THE HONORABLE
REPRESENTATIVE
HANNAH KANE



Massachusetts' 11th House District Representative Hannah Kane is a tremendous advocate for the rare disease community. She was the lead legislative sponsor for the Rare Disease Advisory Council (RDAC) bill for two years as a member of the Joint Committee on Ways and Means and the ranking minority member of the Joint Committee on Public Health.

As the co-sponsor for the effort to establish an RDAC, which began in 2015, Representative Kane went above and beyond. She argued that this council would help give a stronger voice to rare disease patients and caregivers. She worked closely with both advocates and NORD staff, and ultimately secured the bill into a larger health care package that was signed into law in January 2021.

Representative Kane is a graduate of Boston University's School of Management. A Republican representing the Shrewsbury and Westborough, she was recently sworn in

for her fourth term in the Massachusetts House of Representatives. She serves as the ranking minority member of both the Joint Committee on Health Care Financing and the Joint Committee on Public Health and she is also a member of the Joint Committee on Mental Health, Substance Use and Recovery and the new Joint Committee on Racial Equity, Civil Rights and Inclusion.

She is deeply committed to public service, serving as an elected Town Meeting member for 18 years, an appointed member of the Shrewsbury Finance Committee, President of the Shrewsbury Public Schools Foundation and a member of the Shrewsbury Coalition for Addiction Prevention and Education.

NORD is honored to present Representative Hannah Kane with a 2021 Rare Impact Award.

2021 RARE IMPACT AWARD ✨ HONOREE

ROBERT LONG



Nearly a decade ago, Robert Long's life was turned upside down when he was diagnosed with anaplastic astrocytoma, a rare and aggressive form of brain cancer. An All-American punter at Syracuse, Rob was on a path to the NFL when he was diagnosed. His prognosis was less than encouraging. His surgery, recovery and treatment took 16 months.

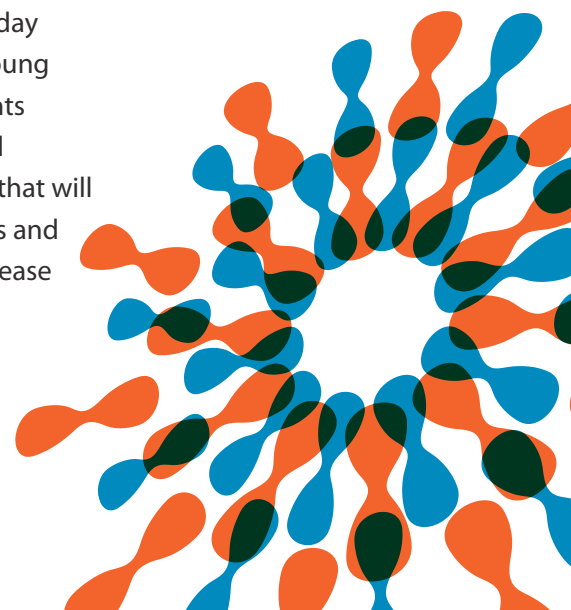
An experimental treatment was the key to his survival. Although Rob had to give up his dream job of playing in the NFL, he was inspired to give back to the rare disease community as

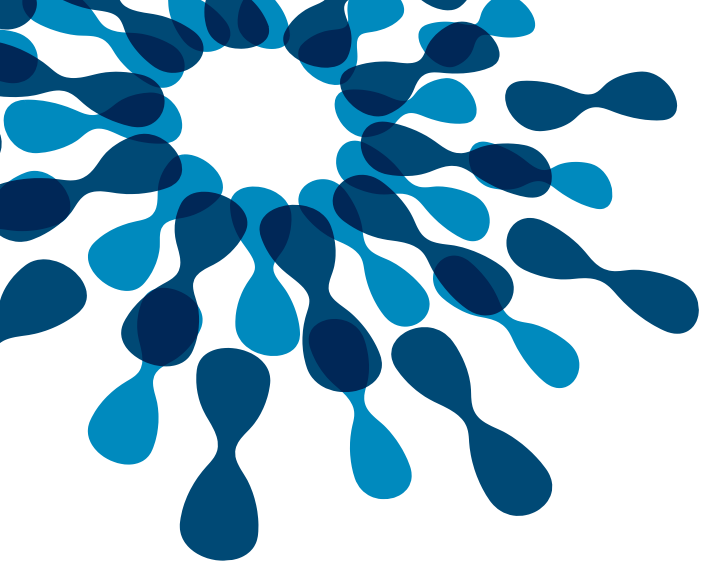
part of his new future. After nearly two years serving as Uplifting Athletes' Director of Rare Disease Engagement, Rob became its second Executive Director at the end of 2018. As the Executive Director of Uplifting Athletes, he is not only connecting college athletes to the work of fighting rare diseases, but is also completely reimagining how we fund, elevate and celebrate rare disease researchers through the Young Investigator Draft.

Moving the Young Investigator Draft from concept to reality was one of Rob's many achievements

that is moving the needle in the fight against rare disease. The Draft treats researchers much in the same way we treat football stars entering the NFL. The selected researchers receive the funding, resources and recognition they need to be the champions our community needs on day one of their service. Young Investigator Draft grants fund collaborative and translational research that will uncover the next cures and treatments for rare disease patients.

NORD is honored to present Robert Long with a 2021 Rare Impact Award.





2021 RARE IMPACT AWARD ✨ HONOREE

CATHLEEN
LUTZ PHD, MBA



Dr. Cathleen Lutz, Senior Director, Mouse Repository and In Vivo Pharmacology at the Jackson Laboratory (JAX) has dedicated years contributing to life-saving research into rare diseases.

Dr. Lutz personally has oversight of a growing collection of more than 12,000 unique strains, including over 1,700 live colonies for distribution to the scientific community for the development, characterization and validation of mouse models that support their research and drug discovery goals. When she becomes aware of a specific rare disorder, she will identify what mouse model, if any, may help with research. She provides advice and support to research teams around the world as the model is utilized, in the hope that researchers and clinicians will develop therapeutics and cures.

Dr. Lutz' team within JAX's Rare and Orphan Disease Center have worked with over 30 rare disease organizations. Through these collaborations the team has created a systematic approach for rare disease drug discovery. Efforts that once took 10-15 years to create a mouse model, test for drug efficacy, and bring studies to clinical trials can now take less than two years. Dr. Lutz's personal conviction and JAX's nonprofit mission align to ensure the best models are always available to patients. If there is no model available, she works directly with the families to develop one. Often this is provided at no cost.

Dr. Lutz's contribution is more than groundbreaking science – she truly understands the importance of showing families there's someone in their corner.

NORD is honored to present Dr. Cathleen Lutz with a 2021 Rare Impact Award.

2021 RARE IMPACT AWARD ✨ HONOREE

THE HONORABLE
US SENATOR PATTY

MURRAY



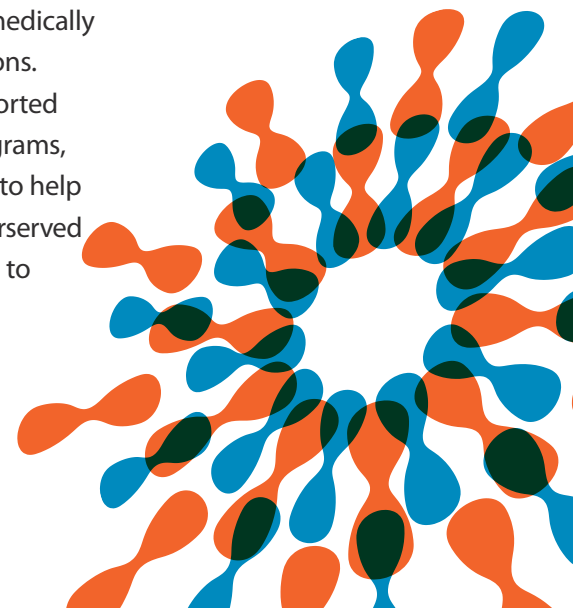
Senator Patty Murray is Chair of the United States Senate Committee on Health, Education, Labor and Pensions (HELP) and a member of the Appropriations Committee. A retired educator and longtime advocate, she is Washington state's first female US Senator.

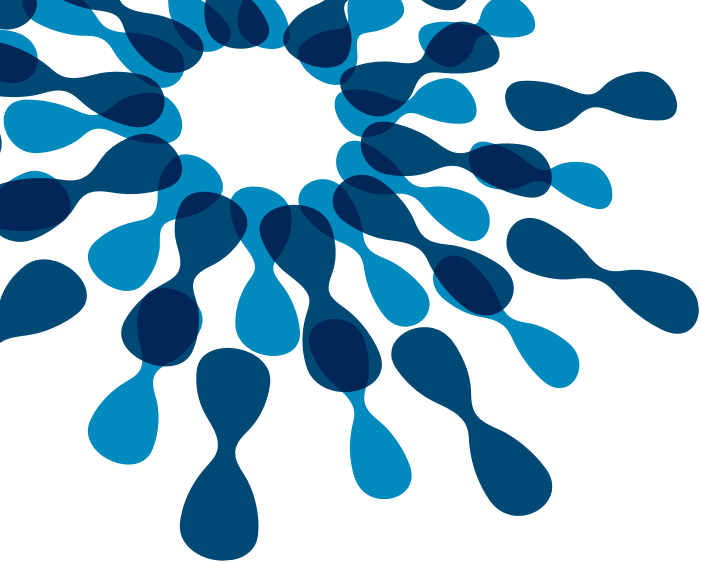
Senator Murray has championed many health priorities, among them expanding access to quality health care while working to rein in costs and ensure access to mental health coverage and services. Senator Murray also supports prevention efforts to not only decrease long-term care costs, but also promote wellness, prevent disease, and protect against public health emergencies.

Her accomplishments relating to health legislation are significant. Senator Murray helped pass healthcare reform legislation that will ensure Americans have access to quality, affordable healthcare and will significantly reduce long-term costs. This law will also ensure that insurance companies can never again refuse coverage for preexisting conditions or cut off care when they decide it is too expensive. She has worked with Senate colleagues to pass the FDA Safety and Innovation Act which will provide the FDA with the necessary tools to support innovation and provide access to life-saving medications.

Senator Murray has championed legislation to reauthorize the Children's Health Insurance Program and increase the number of children covered to almost 10 million. She has also fought for increased funding for community health centers and the National Health Service Corps, which work to improve health care access to medically underserved populations. Additionally, she supported federal telehealth programs, which use technology to help people in rural or underserved areas get better access to healthcare services.

NORD is honored to present Senator Patty Murray with a 2021 Rare Impact Award.





2021 RARE IMPACT AWARD ✨ HONOREE

KAM

REDLAWSK



Kam Redlawsk is an industrial designer, artist, and writer living with GNE Myopathy (GNEM), a rare and progressive genetic muscle debilitating disorder. This disease has led her to being a wheelchair user for the past six years and eventually will result in quadriplegia. She has dedicated the past 10 years to raising awareness and, despite chronic pain and debilitating fatigue, she pushes forward using her experiences to further her message that life is what we make it and together, we must live more.

When Kam came to California in 2007 for her design career, she met two brothers who shared her diagnosis and had started their own non-profit for GNEM research. True to her personality, she saw help was needed and volunteered; her pro bono design work for the nonprofit was key in raising awareness for this rare disease. As the first GNEM patient blogger, her design campaign for the nonprofit was about broadening the audience by encouraging other patients

to share their stories. Kam has helped spread the word and today there are GNEM groups worldwide including in Korea, Japan, India, and the UK, with patients representing all ethnicities engaged and sharing their stories.

Her impact goes beyond the GNEM community. She has shared her struggles and triumphs generously and honestly with her followers. From the cover of the LA Times to art galleries featuring her work and international television; her work and advocacy have had an impact. Kam shares her wheelchair travels to inspire both able and disabled individuals to understand that there are ways to creatively overcome our everyday challenges.

NORD is honored to present Kam Redlawsk with a 2021 Rare Impact Award.

2021 RARE IMPACT AWARD ✨ HONOREE

NATASHA SHUR, MD



Dr. Natasha Shur has been a clinical geneticist for over a decade. She leads the Telemedicine Genetics Program under the Rare Disease Institute at Children's National Hospital.

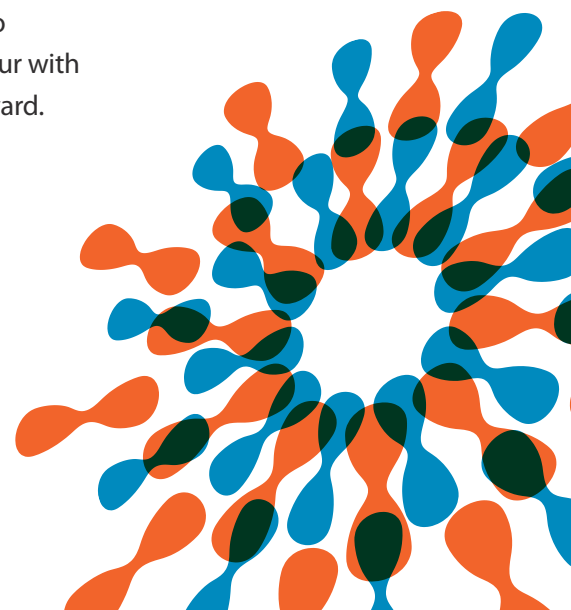
With the onset of the pandemic, medical professionals had to move swiftly to virtual models to continue providing care to their patients. Dr. Shur was instrumental in converting the genetics clinical unit to 100% telemedicine over a few days in early March 2020. She built out the systems and support needed for an active in-home telemedicine program, an innovation that is helping many rare disease families.

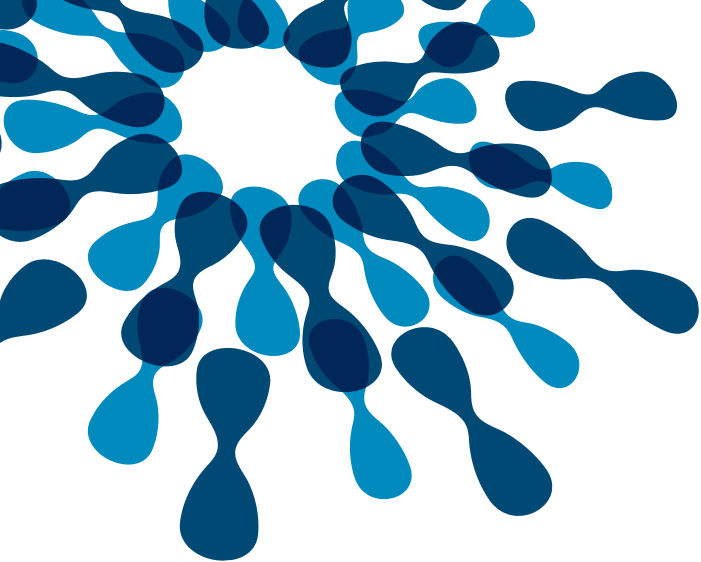
With virtual care, wait times for visits are reduced. No-show rates are lower as families can easily make and keep at-home appointments, patient satisfaction has increased; there are fewer admissions and patients are not at risk of contracting COVID-19. Telehealth also provides care to patients in underserved rare disease communities who often lack easy access to services.

Dr. Shur has also been an expert voice within the rare community, helping us better understand the shift to telehealth. She lent her expertise to NORD to create a series of informational videos answering critical questions

during the early days of the pandemic about telehealth. Dr. Shur's innovative, successful, and empathetic approach to providing care in these unprecedented times has proved the effectiveness of telemedicine as another tool for clinicians to best care for their rare disease patients.

NORD is honored to present Dr. Natasha Shur with a 2021 Rare Impact Award.





2021 RARE IMPACT AWARD 🌟 HONOREE

JASON SICKLICK, MD, FACS



Dr. Jason Sicklick is making significant contributions to Gastrointestinal Stromal Tumor (GIST) patients and to the rare disease community as a whole. In the United States, there are approximately 4,000-6,000 cases of GIST diagnosed each year, with 150-200 of these cases diagnosed as the hereditary subset known as SDH-deficient GIST that primarily affects adolescents and young adults. As one of

the founding members of the Life Raft Group's SDH Consortium, he is part of a group striving to find a cure for this disease through global collaboration.

Dr. Sicklick is a Professor of Surgery at University of California San Diego's Moores Cancer Center. He possesses a unique combination of expertise as a skilled surgeon, inquisitive translational scientist, and compassionate clinician. He specializes in detection, diagnosis, treatment and care for patients with GIST. One of

the major road blocks to SDH-deficient GIST research was that previously there were no reliable human cell lines or mouse models. However, his team has since been able to successfully grow SDH-deficient GIST tumor cells from patients that accurately model the disease. He has tested thousands of existing FDA approved drugs and novel compounds on these cells in the race to find an effective treatment.

Dr. Sicklick is also the co-principal investigator of a large scale clinical trial driven by genomics. The Investigation of Profile Related Evidence Determining Cancer Therapy (I-PREDICT) study has applied precision medicine to cancer treatment, using genomics to uncover what combination of available therapies may provide the best patient outcomes. The findings of this study may positively impact innovation for all genetic rare diseases.

NORD is honored to present Dr. Jason Sicklick with a 2021 Rare Impact Award.

2021 RARE IMPACT AWARD ✨ HONOREE

DANYELLE SUN



Danyelle Sun joined the Wisconsin Rare Action Network® in May 2018 as the Volunteer State Ambassador. Prior to that she had served Cure SMA as a board member and Wisconsin chapter officer, building community and championing a better future for children living with spinal muscular atrophy (SMA), a disease she knew well as the mother of two children affected by the disorder. She currently serves on the staff of Cure SMA as the Social Work Manager.

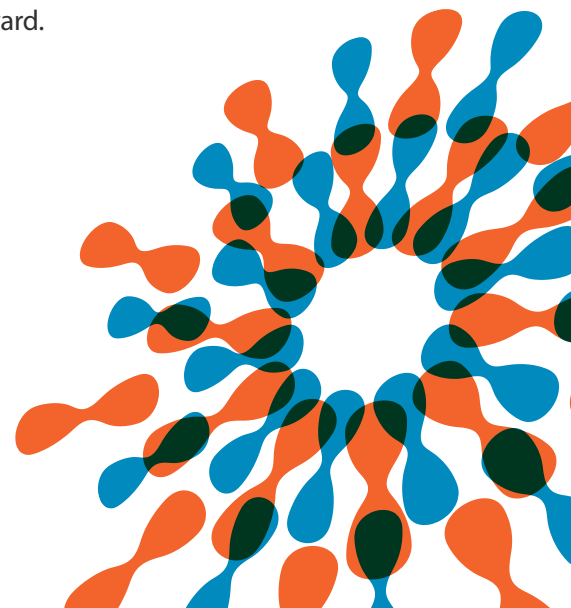
Through her incredible advocacy work on newborn screening and educating of elected officials and their staff

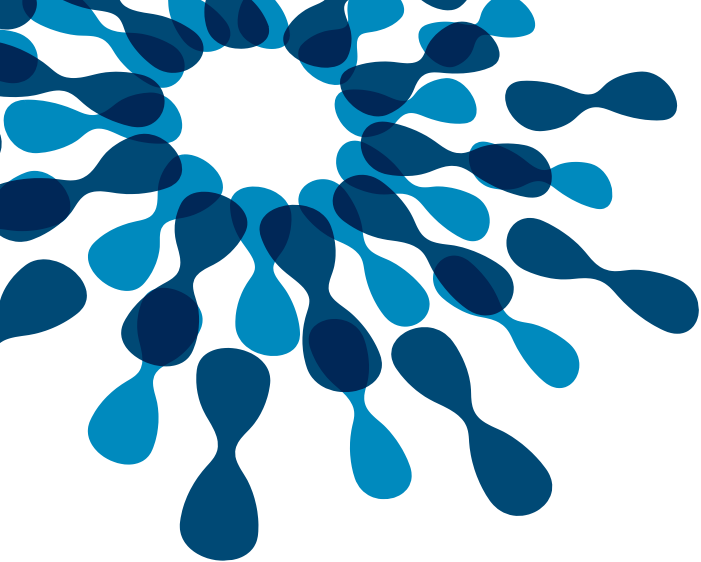
on rare diseases, Danyelle has successfully built a strong network of over 280 advocates throughout Wisconsin. She is leading the Wisconsin Rare Action Network in building a coalition of groups to propose legislation for a rare disease advisory council in the state of Wisconsin. She has worked tirelessly with state legislators and their staff on numerous policy initiatives affecting the rare disease community.

Danyelle is a true inspiration to all who meet her. She is quick to say that her children's disabilities don't change their desires and goals; it just makes the "how" they make those dreams a reality

a bit different. She is always willing and ready to share her story, host events and mentor others as they grow as advocates. The rare disease community is fortunate to have Danyelle leading the way in Wisconsin.

NORD is honored to present Danyelle Sun with a 2021 Rare Impact Award.





2021 RARE IMPACT AWARD 🌟 HONOREE

MARY WOOTTEN



Mary Wootten, mother of three, joined the Rare Action Network in October of 2018. She is also a Program Associate at the Oley Foundation, a non-profit organization that provides information, peer support, networking opportunities and more for individuals who depend on home intravenous and/or tube-fed nutrition and their families. Since her appointment as the Volunteer State Ambassador for New York, she has helped spearhead the growth of rare disease advocacy in her home state and driven positive policy change on behalf of the entire rare disease community.

Mary has actively garnered support from elected officials at the state capitol on step therapy and rare disease advisory council legislation, the latter of which was passed into law in 2020. She has convened in-person advocacy events in Albany and elsewhere throughout

the state. As a result of her efforts, the New York Rare Action Network has grown to more than 1,085 individual advocates, many of whom she connects with daily helping them get to know their legislators and identify resources that may help them in their day-to-day rare disease fight.

Mary's hard work and dedication have made the many victories benefiting rare disease patients and families in New York possible. It is NORD's hope for all of our ambassadors to mirror the same successes of the New York Rare Action Network that we have had under Mary's leadership.

NORD is honored to present Mary Wootten with a 2021 Rare Impact Award.

SPECIAL RECOGNITION

CHRIS AUSTIN, MD



After nearly two decades at the NIH and almost a decade of leadership as the founding director of the National Center for Advancing Translational Sciences (NCATS), Dr. Austin is embarking on a new chapter as CEO-Partner at Flagship Pioneering.

While at NCATS, Dr. Austin has been an innovator, a collaborator, a problem solver, and a challenger of the status quo. These characteristics have been crucial to his work in overcoming long-standing scientific and operational issues that can hamper the

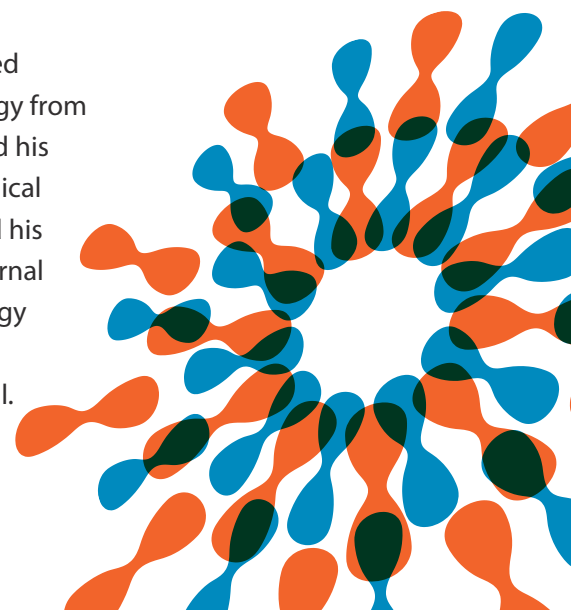
process of turning promising biomedical advances into new diagnostics and health innovations. His drive to improve the translational process has been fueled by his experience as a geneticist, drug developer, neuroscientist, and clinician.

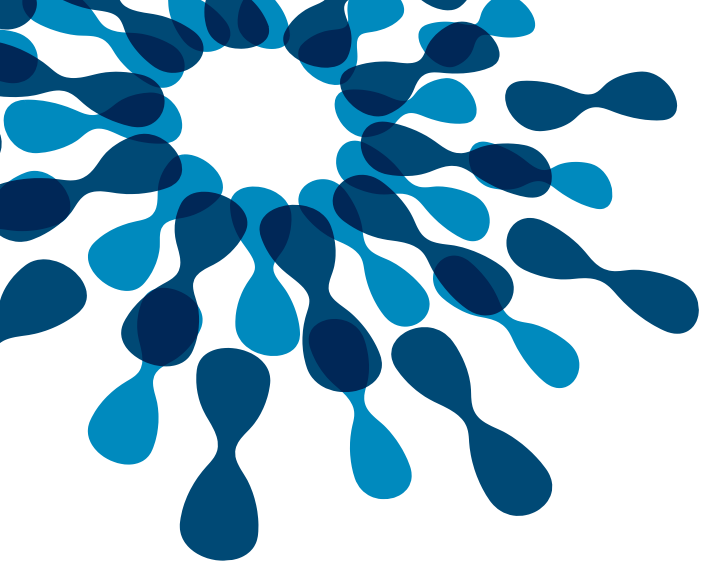
Under Dr. Austin's leadership, the Center has made great strides in its ambitious vision to transform translation from an empirical process into a predictive science. As part of this work, Dr. Austin supported rare diseases research nationally and internationally to meet

the need for timely diagnosis and effective treatments for more than 300 million people globally who are affected by about 7,000 different rare diseases. He also served as chair of the International Rare Diseases Research Consortium (IRDiC).

Dr. Austin graduated with a degree in biology from Princeton and received his MD from Harvard Medical School. He completed his clinical training in internal medicine and neurology at world-renowned Massachusetts General.

NORD is honored to present Dr. Chris Austin with special recognition for his decades of work in support of the rare disease community.





2021 INDUSTRY INNOVATION AWARD  HONOREE

ALNYLAM PHARMACEUTICALS FOR OXLUMO™



On November 24, 2020, the FDA approved OXLUMO™ (lumasiran) injection for subcutaneous use, the first-ever therapy available for the treatment of primary hyperoxaluria type 1 (PH1) to lower urinary oxalate levels in pediatric and adult patients. PH1 is a rare genetic disease characterized by oxalate overproduction. The excess production results in the deposition of calcium oxalate crystals in the kidneys and urinary tract and can lead to the formation of painful and recurrent kidney stones, nephrocalcinosis, progression to kidney failure, and systemic organ dysfunction.

“The approval of Oxlumo represents a great triumph of community involvement to address a rare disease. It is a result of input from patients, treating physicians, experts and sponsors at a patient-focused drug development meeting and through other collaborative efforts,” said Norman Stockbridge, MD, PhD, Director of the Division of Cardiology and Nephrology in the FDA’s Center for Drug Evaluation and Research.

Oxlumo was evaluated in two studies in patients with PH1. First in a randomized, placebo-controlled trial in patients six years and older, the Oxlumo group patients had a 65% average reduction of oxalate in the urine, compared to an average 12% reduction in the placebo group. By the sixth month of the study, 52% of patients treated with Oxlumo reached a normal 24-hour urinary oxalate level; no patients treated with the placebo did. In the second study, an open-label study in patients younger than six years, 16 patients all received Oxlumo. The study showed, on average, a 71% decrease in urinary oxalate by the sixth month.

NORD is proud to present Alnylam Pharmaceuticals with a 2021 Industry Innovation Award for bringing this important new treatment to patients.

ASTRAZENECA FOR KOSELUGO™

On April 10, 2020, the FDA approved Koselugo (selumetinib) for the treatment of pediatric patients, two years of age and older, with neurofibromatosis type 1 (NF1), a genetic disorder of the nervous system that causes tumors to grow on nerves. Koselugo is the first drug approved by the FDA to treat this debilitating, progressive and often disfiguring rare disease that typically begins early in life.

“Everyone’s daily lives have been disrupted during the COVID-19 pandemic, and in this critical time we want patients to know that the FDA remains committed to making patients with rare tumors and life threatening diseases, and their unique needs, a top priority. We continue to expedite product development for these patients,” said Richard Pazdur, MD, Director of the FDA’s

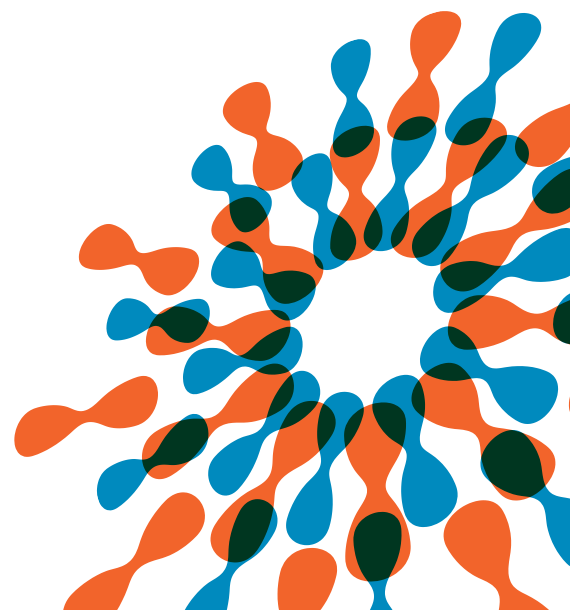
Oncology Center of Excellence and Acting Director of the Office of Oncologic Diseases in the FDA’s Center for Drug Evaluation and Research.

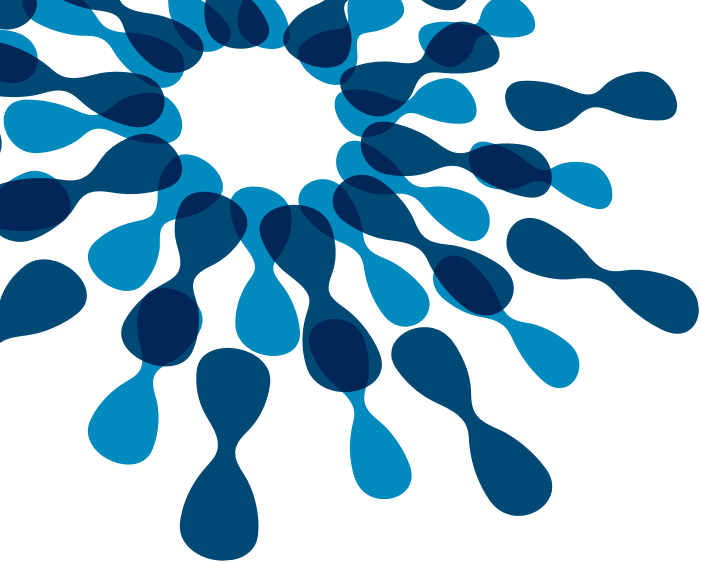
Koselugo is approved for patients who have symptomatic, inoperable plexiform neurofibromas (PN), which are tumors involving the nerve sheaths and can grow anywhere in the body, including the face, extremities, areas around the spine and deep in the body where they may affect organs. NF1 is usually diagnosed in early childhood and appears in an estimated 1 out of every 3,000 infants.

As a kinase inhibitor, Koselugo works by blocking a key enzyme, which results in helping stop tumor cell growth. The FDA approved Koselugo based on a clinical trial conducted by the National Cancer Institute of

pediatric patients who had NF1 and inoperable PN.

NORD is proud to present AstraZeneca with a 2021 Industry Innovation Award for bringing this important new treatment to patients.





2021 INDUSTRY INNOVATION AWARD  HONOREE

BLUEPRINT MEDICINES FOR AYVAKIT™



On January 09, 2020, the FDA approved Ayvakit (avapritinib) for the treatment of adults with unresectable or metastatic gastrointestinal stromal tumor (GIST) – a type of tumor that occurs in the gastrointestinal tract, most commonly in the stomach or small intestine – harboring a platelet-derived growth factor receptor alpha (PDGFRA) exon 18 mutation. Ayvakit is a kinase inhibitor, meaning it blocks a type of enzyme thus preventing cancer cell growth.

“GIST harboring a PDGFRA exon 18 mutation do not respond to standard therapies for GIST. However, today’s approval provides patients with the first drug specifically approved for GIST harboring this mutation,” 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. “Clinical trials showed a high response rate with almost 85% of patients experiencing tumor shrinkage with this targeted drug.”

The FDA approved Ayvakit based on the results of a clinical trial involving 43 patients with GIST harboring a PDGFRA exon 18 mutation, including 38 patients with PDGFRA D842V mutation. The trial measured how many patients experienced complete or partial shrinkage of their tumors during treatment. For patients harboring a PDGFRA exon 18 mutation, the overall response rate was 84%, with 7% having a complete response and 77% having a partial response. For the subgroup of patients with PDGFRA D842V mutations, the overall response rate was 89%, with 8% having a complete response and 82% having a partial response.

NORD is proud to present Blueprint Medicines with a 2021 Industry Innovation Award for bringing this important new treatment to patients.

EIGER BIOPHARMACEUTICALS, INC. FOR ZOKINVY™

On November 20, 2020, the FDA approved Zokinvy (lonafarnib) capsules to reduce the risk of death due to Hutchinson-Gilford progeria syndrome and for the treatment of certain processing-deficient progeroid laminopathies in patients one year of age and older.

"Hutchinson-Gilford progeria syndrome and progeroid laminopathies are rare genetic diseases that cause premature aging and death and have a debilitating effect on people's lives," said Hylton V. Joffe, MD, MMSc, Director of the Office of Rare Diseases, Pediatrics, Urologic and Reproductive Medicine in the FDA's Center for Drug Evaluation and Research. "With today's approval, Zokinvy is the first FDA-approved medication for these devastating diseases. The FDA will continue to work with stakeholders to advance

the development of additional new, effective and safe therapies for these patients."

Patients with Hutchinson-Gilford progeria syndrome and progeroid laminopathies experience accelerated cardiovascular disease from the buildup of defective progerin or progerin-like protein in cells. Most patients die before the age of 15 years from heart failure, heart attack or stroke.

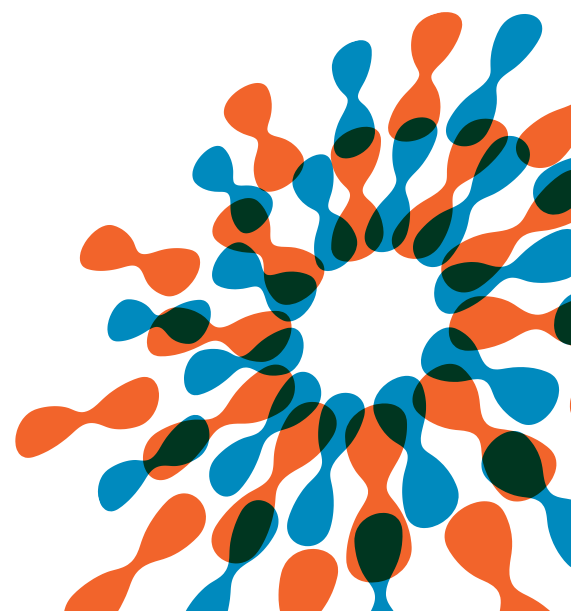
Zokinvy, a farnesyltransferase inhibitor, is an oral medication that helps prevent the buildup of defective progerin or progerin-like protein. The effectiveness of Zokinvy for the treatment of Hutchinson-Gilford progeria syndrome was demonstrated in 62 patients from two single-arm trials that were compared to matched, untreated patients from a separate natural history study. Compared to untreated patients, the lifespan of Hutchinson-Gilford progeria

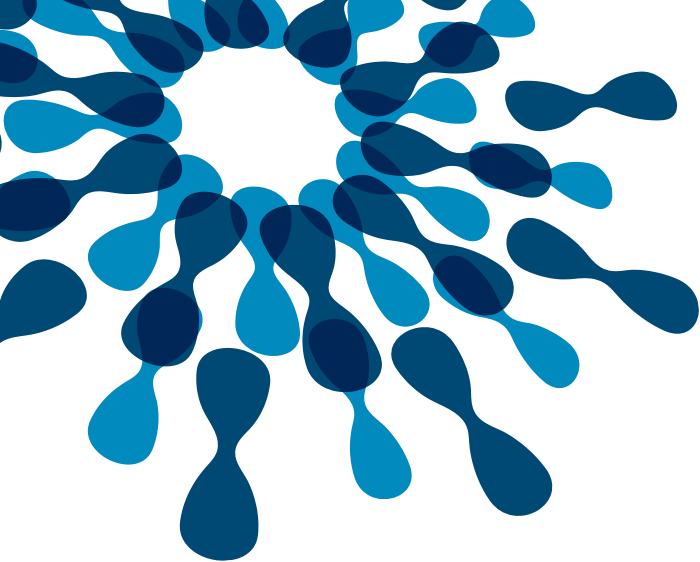
syndrome patients treated with Zokinvy increased by an average of three months through the first three years of treatment and by an average of 2.5 years through the maximum follow-up time of 11 years.

NORD is proud to present Eiger Biopharmaceuticals, Inc. with a 2021 Industry Innovation Award for bringing this important new treatment to patients.



EIGER
BIOPHARMACEUTICALS





2021 INDUSTRY INNOVATION AWARD  HONOREE

**HORIZON
THERAPEUTICS
FOR TEPEZZA™**



HORIZON

On January 21, 2020, the FDA approved Tepezza (teprotumumab-trbw) for the treatment of adults with thyroid eye disease, a rare condition where the muscles and fatty tissues behind the eye become inflamed, causing the eyes to be pushed forward and bulge outwards. The disease can be incapacitating with patients experiencing a variety of symptoms including, eye pain, double vision, light sensitivity and difficulty closing the eye.

“Currently, there are very limited treatment options for this potentially debilitating disease. This treatment has the potential to alter the course of the disease, potentially sparing patients from needing multiple invasive surgeries by providing an alternative, non-surgical treatment option,” said Wiley Chambers, MD, Deputy Director of the Division of Transplant and Ophthalmology Products in the FDA’s Center for Drug Evaluation and Research. “Additionally, thyroid eye disease is a rare disease that impacts a small percentage of the population, and for a variety of reasons, treatments

for rare diseases are often unavailable. This approval represents important progress in the approval of effective treatments for rare diseases, such as thyroid eye disease.”

Tepezza was approved based on the results of two studies consisting of a total of 170 patients with active thyroid eye disease who were randomized to either receive Tepezza or a placebo. Of the patients who received Tepezza a significant number experienced a reduction in eye protrusion (71% in the first study and 83% in the second) compared to the placebo (20% and 10% respectively in the two studies).

NORD is proud to present Horizon Therapeutics with a 2021 Industry Innovation Award for bringing this important new treatment to patients.

INCYTE FOR PEMAZYRE™

On April 17, 2020, the FDA granted accelerated approval to Pemazyre (pemigatinib), the first treatment approved for adults with certain types of previously treated, advanced cholangiocarcinoma. Cholangiocarcinoma is a rare form of cancer that forms in bile ducts, which are slender tubes that carry the digestive fluid bile from the liver to gallbladder and small intestine.

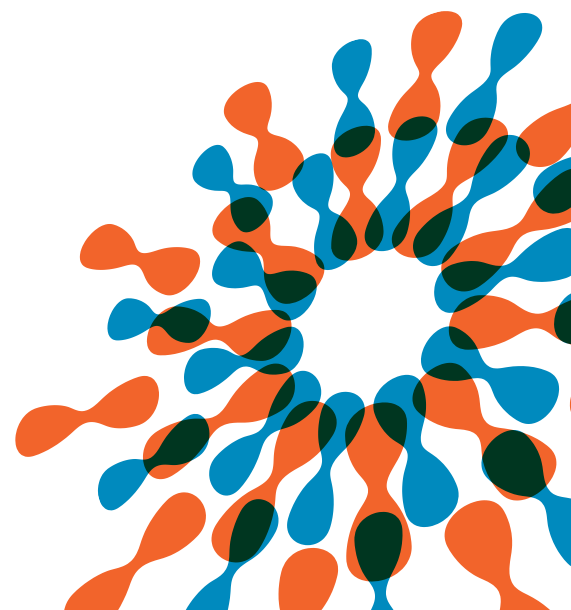
“This approval demonstrates that while we continue to focus our efforts on addressing the COVID-19 pandemic, the FDA remains committed to the important work of reviewing treatments for patients with cancer and other serious conditions,” said Richard Pazdur, MD, Director of the FDA’s Oncology Center of Excellence and Acting Director of the Office of Oncologic Diseases in the FDA’s Center for Drug Evaluation and Research. “With Pemazyre, we considered the observed

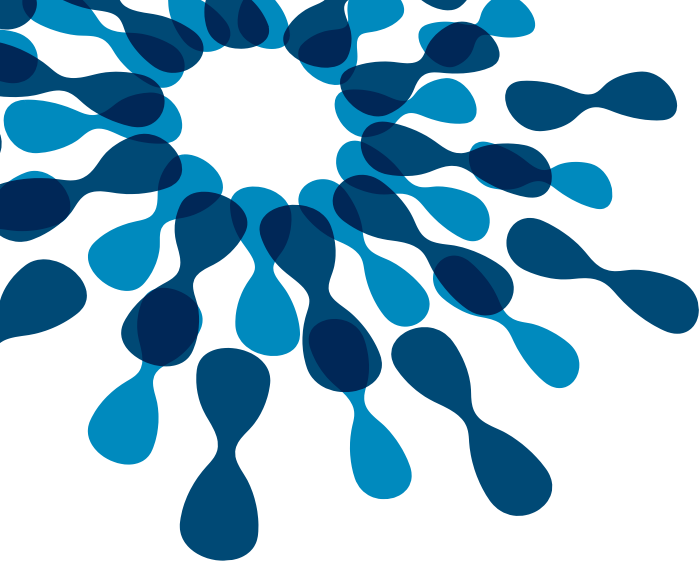
efficacy results to be clinically meaningful and the overall risk to benefit assessment for patients with tumors harboring FGFR2 gene fusions and other rearrangements to be favorable, particularly when we considered that these patients have no other good options following first line treatment with chemotherapy.”

At diagnosis, a majority of patients with cholangiocarcinoma have advanced disease, meaning that the disease is no longer treatable with surgery. For these patients, there have been no FDA-approved therapies before Pemazyre’s approval; a combination of chemotherapy drugs has been the standard initial treatment. FGFR2 fusions have been found in the tumors of approximately 9% to 14% of patients with cholangiocarcinoma. Pemazyre is a tablet that works by blocking FGFR2 in tumor

cells to prevent them from growing and spreading.

NORD is proud to present Incyte with a 2021 Industry Innovation Award for bringing this important new treatment to patients.





2021 INDUSTRY INNOVATION AWARD  HONOREE

LILLY ONCOLOGY
FOR RETEVMO™

Lilly | ONCOLOGY

On May 8, 2020, the FDA approved Retevmo (selpercatinib) capsules to treat three types of tumors – non-small cell lung cancer, medullary thyroid cancer and other types of thyroid cancers – in patients whose tumors were caused by an abnormal RET gene. Genomic alterations in the RET kinase lead to overactive RET signaling and uncontrolled cell growth. Retevmo is the first therapy approved specifically for lung and thyroid cancer patients with the RET gene alterations.

“Innovations in gene-specific therapies continue to advance the practice of medicine at a rapid pace and offer options to patients who previously had few,” 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. “The FDA is committed to reviewing treatments like Retevmo that are targeted to specific subsets of patients with cancer.”

The FDA approved Retevmo on the results of a single-arm, multi-center, open-label, multi-cohort clinical trial involving patients with each of the three types of tumors with RET alterations. Retevmo is a kinase inhibitor, meaning it blocks a type of enzyme to help prevent the cancer cells from growing. Before beginning treatment, the identification of a RET gene alteration must be determined using laboratory testing.

NORD is proud to present Lilly Oncology, with a 2021 Industry Innovation Award for bringing this important new treatment to patients.

REGENERON FOR INMAZEB™

On October 14, 2020, the FDA approved Inmazeb® (atoltivimab, maftivimab and odesivimab-ebgn), a mixture of three monoclonal antibodies, for the treatment of infection caused by Zaire ebolavirus in adult and pediatric patients. Zaire ebolavirus is one of four Ebolavirus species that can cause a potentially fatal human disease.

“Today’s action demonstrates the FDA’s ongoing commitment to responding to public health threats—both domestically and abroad—on the basis of science and data,” said then-FDA Commissioner Stephen M. Hahn, MD, “This approval was made possible because of our steadfast dedication to facilitate the development of safe and effective treatments for infectious diseases as part of our vital public health mission.”

Inmazeb targets the glycoprotein that is on the surface of Ebola virus. Glycoprotein attaches to the cell receptor and fuses the viral and host cell membranes allowing the virus to enter the cell. The three antibodies that make up Inmazeb can bind to this glycoprotein simultaneously and block attachment and entry of the virus.

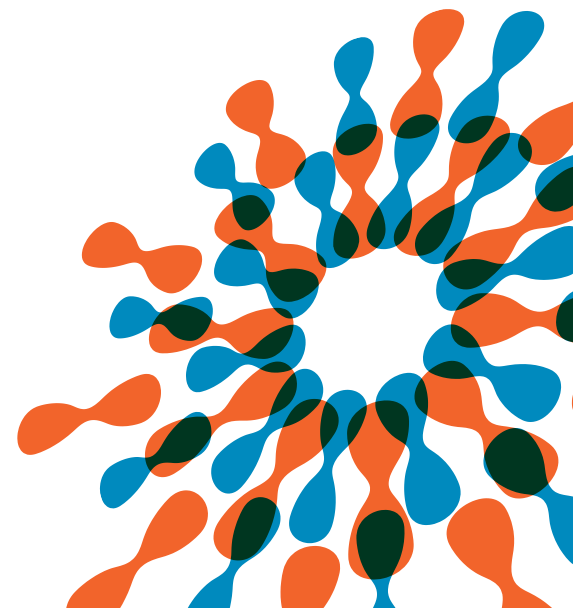
The safety and efficacy of Inmazeb was evaluated in 382 adult and pediatric patients with confirmed Zaire ebolavirus infection in one clinical trial (the PALM trial) and as part of an expanded access program conducted in the Democratic Republic of the Congo during an Ebola virus outbreak in 2018-2019.

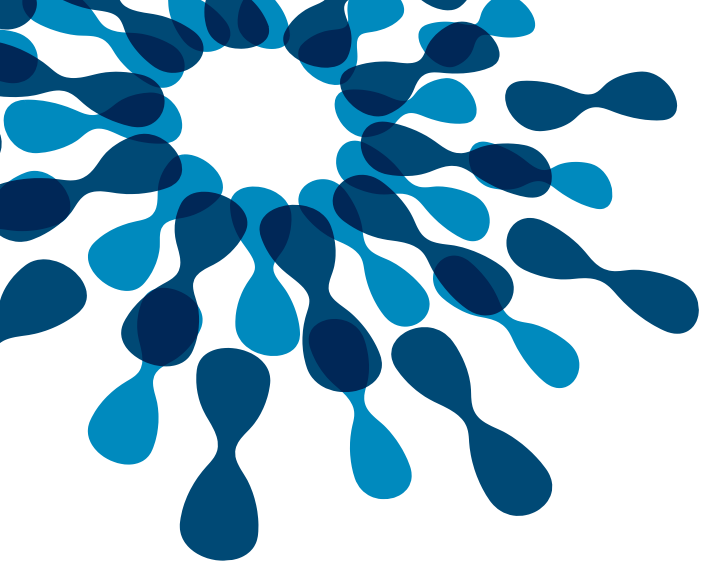
As part of an agreement announced in July 2020, Regeneron will deliver an established number of Inmazeb treatment doses over the course of six years

to the Biomedical Advanced Research and Development Authority to build national preparedness for public health emergencies.

NORD is proud to present Regeneron with a 2021 Industry Innovation Award for bringing this important new treatment to patients.

REGENERON
SCIENCE TO MEDICINE®





2021 INDUSTRY INNOVATION AWARD  HONOREE

RIDGEBACK BIOTHERAPEUTICS FOR EBANGA™



RIDGEBACKBIO

On December 22, 2020, the FDA approved Ebanga™ (Ansuvimab-zykl), a human monoclonal antibody, for the treatment of Zaire ebolavirus in adult and pediatric patients. Zaire ebolavirus is one of four Ebolavirus species that can cause a potentially fatal human disease. It is transmitted through blood, body fluids, and tissues of infected people or wild animals, and through surfaces and materials, such as bedding and clothing, contaminated with these fluids. Ebanga prevents the virus from entering human cells by inhibiting receptor binding.

“The devoted Ridgeback team embarked on this mission with one goal in mind – to stop the spread of Ebola and stop the devastation created by this aggressive disease,” said Wendy Holman, CEO and co-founder of Ridgeback Biotherapeutics LP. “The Ridgeback team has been tireless in their efforts, united in their mission and focused on bettering the lives of the patients we impact. I am so thankful to this committed

team, and I look forward to how we can further our mission of helping people by serving unmet needs for those with grievous illness.”

The efforts of the PALM study team conclusively demonstrated Ebanga’s safety and efficacy in a randomized controlled trial conducted during the second largest and longest outbreak in the history of the Democratic Republic of Congo. The study showed patients on Ebanga were more likely to survive than those in the control arm. Of the 174 patients who received Ebanga, 35% died after 28 days, compared with 49% of the 168 control patients. The study team’s efforts represent a landmark achievement in the development of medical countermeasures for emerging infectious diseases.

NORD is proud to present Ridgeback Biotherapeutics LP with a 2021 Industry Innovation Award for bringing this important new treatment to patients.

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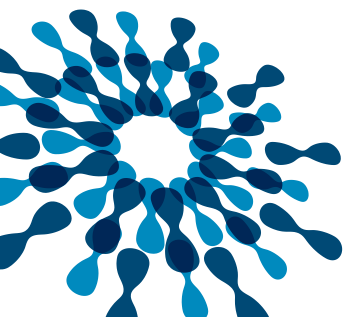
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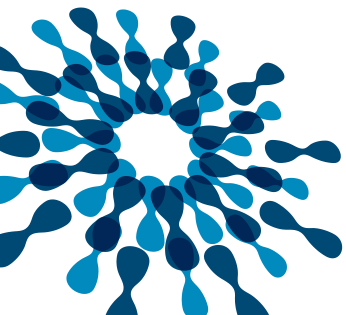
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Congratulations to all the 2021 Rare Impact Award Honorees.



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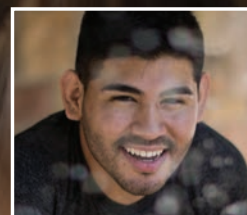
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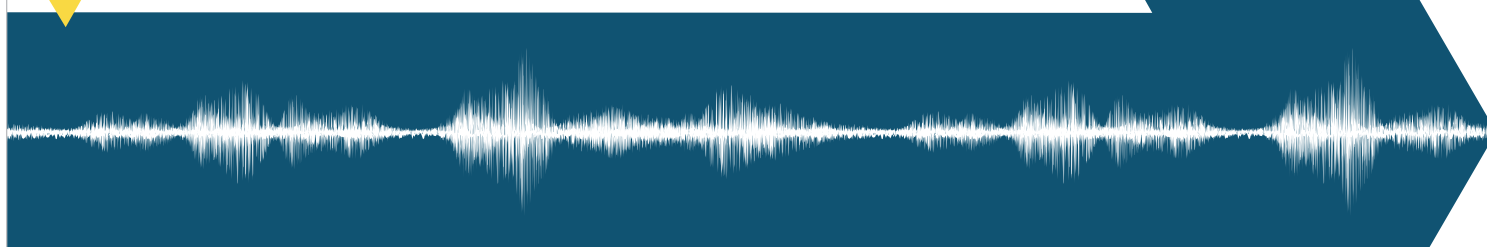


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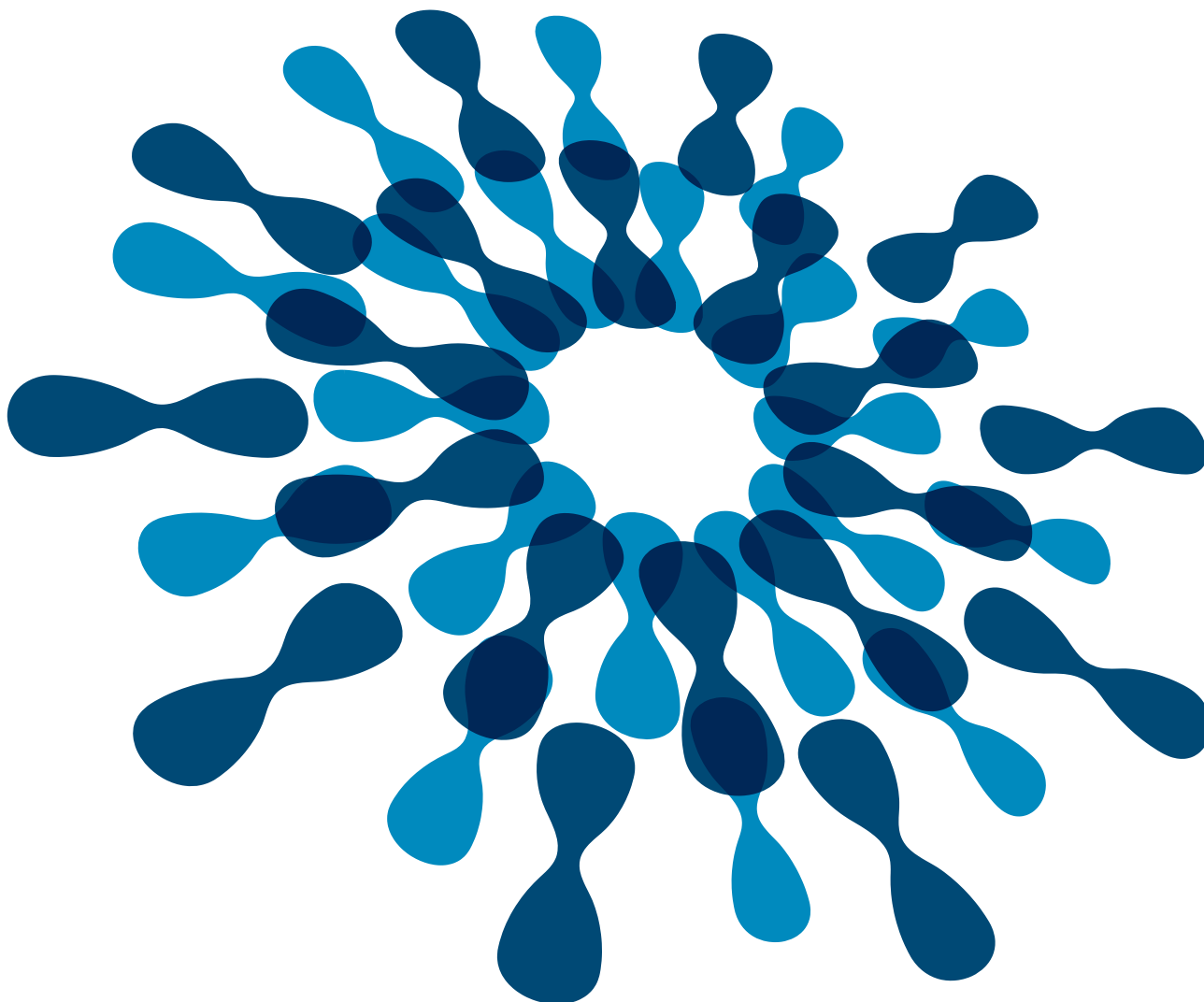
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