Find out how Chan Zuckerberg is helping advance rare disease research

Learn about the exciting new developments helping treat and diagnose rare diseases

Kristin Chenoweth
Discover how this Broadway star balances a thriving career with Meniere’s Disease

De-risk drug development for epilepsy and tumor growth in tuberous sclerosis complex by partnering with our Preclinical Consortium. Learn more at tsalliance.org/preclinical today.
Emily V. Gordon on Living with “The Big Sick”

Writer and real-life subject of “The Big Sick” discusses the big-screen adaptation of her own rare illness and the importance of self-advocacy in health.

Having an unknown and undiagnosed disease can be extremely frustrating and stressful, as it can take years, as it did in your case, to come up with a diagnosis. How was the process for you?

What eventually helped me was applying for life insurance and getting rejected because I was deemed too unhealthy to cover. This led me to start asking questions to my doctor that he couldn’t answer or help with. My new doctor helped me find a new treatment for myself that has made me healthier than I’ve been in years.

What moment did you decide to write “The Big Sick”?

There was no moment, really. It is just a story that no one could make up that we felt like, under the tutelage of Judd Apatow, was a story that other people might want to see. And we were the only ones to tell it.

What was that process like, writing about a subject so close to you?

It’s an interesting thing to figure out how you show illness in a movie. If we kept true to exactly what my illness looks like and how it manifests, it would be an accurate movie but not a very good movie.

And in writing with my husband, we got to relive and restructure one of the most harrowing experiences of our lives and turn it into something sweet and meaningful, and somehow less scary. I highly recommend it.

Supporting People Living With Rare Diseases and Their Dedicated Caregivers

People living with rare diseases and their families sometimes need help in accessing vital services. NORD® is there to lend a hand.

With rare diseases affecting more than 25 million Americans and 90 percent of the over 7,000 rare diseases still without an FDA-approved treatment, the need for research and progress in drug development remains as pressing as ever. When there is no treatment available, the medical and supportive care costs can be insurmountable for a person living with a rare disease. When a treatment does exist, it may unfortunately be out of a patient’s financial reach.

A way forward

For more than 35 years, the National Organization for Rare Disorders (NORD®) has been leading the fight to improve the lives of people with rare diseases and their families. When financial restrictions make obtaining treatment difficult, an already challenging situation can become even more stressful. An integral part of NORD’s mission is to help rare disease patients access medical care and treatments that without assistance might not be possible for them.

Helping more patients get the care they need

Since 1987, NORD’s RareCARE has designed and administered patient assistance programs to help patients who are struggling to secure life-saving or life-sustaining medication and access to care they could not otherwise afford.

Providing care for caregivers

Earlier this year, NORD® launched the Rare Caregiver Respite Program, a first-of-its-kind assistance program designed for caregivers of a child or adult diagnosed with a rare disorder.

Through the Rare Caregiver Respite program, NORD® provides financial assistance to eligible caregivers so that an interim caregiver may be secured. Potential applicants can reach out to NORD® to determine if they meet eligibility requirements. The grant may be used throughout a calendar year or in a single award. The fund continues to accept additional donations to ensure this unique program serving all 7,000 rare disease states is sustained for years to come.

For more information and instructions on how to apply for the Rare Caregiver Respite program and other NORD patient assistance programs, visit rarediseases.org.
Compassion in Medicine: Why Caring Matters

The Rare and Undiagnosed Network (RUN) partnered with the University of Utah School of Medicine’s Pediatric Medicine Interest Group to discuss what it is like to live in a diagnostic odyssey with a rare or undiagnosed disease.

Ava Szajnuk, 12 years old, spoke to the students there. She has been through nine years of living in the world of the unknown. She has had four cranial surgeries and is shunt-dependent. She almost lost her vision and her life during the summer of 2013 to a subdural hygroma. “I wish that being shunt-dependent was the least of my worries; unfortunately, it is not. I live with an undiagnosed autonomic neuropathy. I suffer from fatigue and pain every day. We have been told that we are 20-plus years ahead of science. I would love for the doctors to spend two hours in my body and tell me if they would be able to handle it for 20-plus years,” she said.

We hope all medical students consider being a geneticist. You are the future for our undiagnosed community. We need your help and your expertise.

The importance of empathy-driven care

We know all medical students are aware of the Hippocratic Oath. An oath, as future physicians, you will take: “First, do no harm.” In the world of undiagnosed rare diseases, doctors can sometimes do harm with their lack of empathy, curiosity, or drive to figure out the problem patients. We truly hope the next time you meet someone that does not feel well, you will listen with compassion and you will tell them that you believe them. We hope you will say that you will do everything in your power to help them.

How you treat patients, outside of the science, is crucial to giving hope to these families living in the world of the rare and undiagnosed.

Gina Szajnuk, Co-Founder and Executive Director, Rare and Undiagnosed Network (RUN)
Worldwide, 400 million people have a rare disease, including 25 to 30 million Americans. While there are as many as 7,000 rare diseases, often, most aren’t well understood and less than 5 percent have approved treatments.

Now a movement is underway to raise awareness and improve patient outcomes. The Rare As One Project, a new science program by the Chan Zuckerberg Initiative (CZI), is dedicated to bringing rare disease communities together and accelerating research by offering capacity-building grants, tools, and training. “We want to help the rare disease community get the message out that rare is common,” says Tania Simoncelli, Rare As One Project lead and CZI’s science policy director. “Collectively, rare is common.”

Founded in 2015 by pediatrician Dr. Priscilla Chan and her husband, Facebook founder and CEO Mark Zuckerberg, CZI has three core areas of focus: science, education, and justice and opportunity. Each area is paired with engineering, grant-making, policy work, impact investing, and advocacy.

The Rare As One Project is part of CZI’s goal to cure, prevent, or manage all diseases by the end of the century. They’re confident that shared knowledge and support across different diseases can bring patients, researchers, doctors, and other key stakeholders together to advance understanding and treatments of rare diseases.

“We need to develop strategies and approaches to tackle these diseases as one community, not one disease at a time,” says Simoncelli, who has also worked at the White House Office of Science and Technology Policy, the U.S. Food and Drug Administration (FDA), the Broad Institute of MIT and Harvard and the American Civil Liberties Union.

Solving common problems
Traditionally, medicine and research are structured around supporting diseases and illnesses that affect large populations, not small groups. In rare disease, there is a numbers problem, as patient communities are often spread around the globe and each individual research institution typically has few patients.

Rare As One is looking at ways to help these rare diseases communities work together to solve common problems. Rare As One is starting by helping patients build communities, which then attracts researchers to study their respective diseases. Next, they’re leveraging technology to help patient and research communities accelerate research — such as creating templates for rare disease groups to share information, track research progress, and crowdsource research ideas.

“Patients really have to be at the center of the whole system and patients have the power to really change it,” says Simoncelli.

CZI is committed to this effort long-term. The Rare As One program will roll out in early 2020 with initial funding of $4.5 million. They’re currently reviewing applications from patient-led 501(c)(3) organizations to offer 10 rare disease groups two years of funding to develop and launch patient-led collaborative research networks working with clinicians and scientists.

CZI’s work is inspired by the stories and incredible work of countless rare disease advocates. One of those is Dr. David Fajgenbaum.

Success story
Dr. Fajgenbaum is a rare disease patient, doctor, and advocate. During his third year of medical school, he was hospitalized for five months and became so ill that he was given last rites. He was 25 when he was diagnosed with idiopathic multicentric Castleman disease (IMCD), a rare disease where the immune system attacks and shuts down the body’s vital organs, including the heart, liver, kidneys, and bone marrow.

An orphan drug helped him for about a year until he relapsed. He was discouraged because that was the only drug in development at the time for his condition.

“I promised my dad and my sisters that I would dedicate the rest of my life to try to cure this disease,” he says.

In all, Dr. Fajgenbaum
Kristen Castillo has had five dangerous relapses, but he’s currently in his longest remission ever thanks to a treatment that he identified in the lab after testing his own samples. The drug, Sirolimus, was developed for kidney transplantation. He realized it could manage his condition, too, and has been taking it for five-and-a-half years.

“This is a true success story,” says Dr. Fajgenbaum, who is the co-founder and executive director of the Castleman Disease Collaborative Network (CDCN), where he spearheads the Collaborative Network Approach. “If I hadn’t tried this already FDA-approved drug that was developed for something totally different, I most certainly would not be alive right now.”

He’s encouraged by the potential of shared knowledge and research: “How can we take what we learn from one disease and apply it to other diseases?”

**Patient-driven approach**

The status quo for research is that foundations raise money and researchers apply to have their research ideas funded. But Dr. Fajgenbaum flipped that notion by inviting patients, physicians, and researchers to proactively determine what research questions need to be answered and then proactively recruiting researchers to do the work.

His new book, “Chasing My Cure” is a blueprint for patients to follow as they look for a cure to their rare disease and inspiration for anyone facing seemingly insurmountable challenges. “It’s really a dream of mine that we can speed up the pace of progress for many diseases beyond Castleman disease,” says the assistant professor of medicine in translational medicine and human genetics at the University of Pennsylvania, and associate director of patient impact for the Penn Orphan Disease Center. “Put patients, physicians, and researchers all at the table together and really have it be a patient-driven approach.”

He says building communities can take time but it’s essential to the success of treating and potentially curing rare diseases. For example, it took a few years to build the Castleman disease community before they crowdsourced and shared data. “We always ask the question, ‘What drugs are already FDA-approved that might be able to help patients with Castleman disease tomorrow based on the research we just found?’” he says, noting there are 1,500 FDA-approved drugs. “While we’re waiting for new drugs to make it out to the market, we really need to leverage these existing drugs.”

**Patient advocate**

Heidi Bjornson-Pennell also knows what it’s like to manage a rare disease. Two of her three children were diagnosed as toddlers with primary ciliary dyskinesia (PCD), a rare, progressive genetic disorder. Nowadays, her children, a 13-year-old daughter and an 11-year-old son, are managing their disease as best as they can but they have challenges, including recurrent respiratory infections and loss of lung function.

“We are really on a hunt for a cure before their lung function declines further,” says Bjornson-Pennell.

A lawyer by trade, she’s now a patient engagement strategist at CZI, where she champions the voices of rare disease patients and supports efforts to accelerate progress against disease. “It’s been very inspiring to see how CZI is really working to elevate that voice,” she says. “The patients are the ones who have the greatest understanding of their disease. They’re the ones with the greatest sense of urgency regarding their disease and the ones most devoted to advancing treatments and finding cures.”

She feels a “great sense of optimism and hope that something can be done that will accelerate progress.”

Bjornson-Pennell says over half of rare disease communities don’t have organizations rallying for treatments and research. Rare As One is an opportunity for rare disease patients to be seen and heard. Plus, it advances medicine. “Research puts us much closer to that goal of trying to cure and prevent all diseases by the end of the century,” she says. “Progress in one, translates into progress in many.”
What is OFEV?
OFEV is a prescription medicine used to treat people with a lung disease called idiopathic pulmonary fibrosis (IPF). It is not known if OFEV is safe and effective in children.

Important Safety Information
What is the most important information I should know about OFEV (nintedanib)? OFEV can cause harm, birth defects or death to an unborn baby. Women should not become pregnant while taking OFEV. Women who are able to become pregnant should have a pregnancy test before starting treatment and should use birth control during and for at least 3 months after your last dose. If you become pregnant while taking OFEV, tell your doctor right away.

What should I tell my doctor before using OFEV?
Before you take OFEV, tell your doctor if you have:
- liver problems
- heart problems
- a history of blood clots
- a bleeding problem or a family history of a bleeding problem
- had recent surgery in your stomach (abdominal) area
- any other medical conditions.
Tell your doctor if you:
- are breastfeeding or plan to breastfeed. It is not known if OFEV passes into your breast milk. You should not breastfeed while taking OFEV.
- are a smoker. You should stop smoking prior to taking OFEV and avoid smoking during treatment.
- have liver problems.

What are the possible side effects of OFEV?
OFEV may cause serious side effects. TELL YOUR DOCTOR RIGHT AWAY if you are experiencing any side effects, including:
- Liver problems. Unexplained symptoms may include yellowing of your skin or the white part of your eyes (jaundice), dark or brown urine, pain on the right upper side of your stomach area (abdomen), bleeding or bruising more easily than normal, feeling tired, or loss of appetite. Your doctor will do blood tests regularly to check how well your liver is working during your treatment with OFEV.
- Diarrhea, nausea, and vomiting. Your doctor may recommend that you drink fluids or take medicine to treat these side effects. Tell your doctor if you have these symptoms, if they do not go away, or get worse and if you are taking over-the-counter laxatives, stool softeners, and other medicines or dietary supplements.
- Heart attack. Symptoms of a heart problem may include chest pain or pressure, pain in your arms, back, neck or jaw, or shortness of breath.
- Stroke. Symptoms of a stroke may include numbness or weakness on 1 side of your body, trouble talking, headache, or dizziness.
- Bleeding problems. OFEV may increase your chances of having bleeding problems. Tell your doctor if you have unusual bleeding, bruising, or wounds that do not heal and/or if you are taking a blood thinner, including prescription blood thinners and over-the-counter aspirin.
- Tear in your stomach or intestinal wall (perforation). OFEV may increase your chances of having a tear in your stomach or intestinal wall. Tell your doctor if you have pain or swelling in your stomach area.

The most common side effects of OFEV are diarrhea, nausea, stomach pain, vomiting, liver problems, decreased appetite, headache, weight loss, and high blood pressure.

These are not all the possible side effects of OFEV. For more information, ask your doctor or pharmacist. You are encouraged to report negative side effects of prescription drugs to the FDA. Visit www.fda.gov/medwatch or call 1-800-FDA-1088.

Please see Patient Information on the next page.

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(08/19) PC-US-110601
Read this Patient Information before you start taking OFEV and each time you get a refill. There may be new information. This information does not take the place of talking to your doctor about your medical condition or your treatment.

What is the most important information I should know about OFEV?
OFEV can cause birth defects or death to an unborn baby. Women should not become pregnant while taking OFEV. Women who are able to become pregnant should have a pregnancy test before starting treatment with OFEV. Women who are able to become pregnant should use birth control during and for at least 3 months after treatment. If you become pregnant while taking OFEV, tell your doctor right away.

What is OFEV?
• OFEV is a prescription medicine used to treat people with a lung disease called idiopathic pulmonary fibrosis (IPF).
• It is not known if OFEV is safe and effective in children.

What should I tell my doctor before taking OFEV?
Before you take OFEV, tell your doctor if you:
• have liver problems
• have heart problems
• have a history of blood clots
• have a bleeding problem or a family history of a bleeding problem
• have had recent surgery in your stomach (abdominal) area
• are a smoker
• have any other medical conditions
• are pregnant or plan to become pregnant. OFEV can harm your unborn baby. OFEV can cause birth defects or death to an unborn baby. See “What is the most important information I should know about OFEV?”
• are breastfeeding or plan to breastfeed. It is not known if OFEV passes into your breast milk. You should not breastfeed while taking OFEV.

Tell your doctor about all the medicines you take, including prescription and over-the-counter medicines, vitamins, and herbal supplements such as St. John’s wort. Keep a list of the medicines you take and show it to your doctor and pharmacist when you get a new medicine.

How should I take OFEV?
• Take OFEV exactly as your doctor tells you to take it.
• Your doctor will tell you how much OFEV (nintedanib) to take and when to take it.
• Take OFEV with food. Swallow the OFEV capsules whole with a liquid.
• Do not chew or crush OFEV capsules.
• If you miss a dose of OFEV, take your next dose at your regular time. Do not take the missed dose.
• Do not take more than 300 mg of OFEV in 1 day.
• If you take too much OFEV, call your doctor or go to the nearest hospital emergency room right away.
• Your doctor should do certain blood tests before you start taking OFEV.

What are the possible side effects of OFEV?
OFEV may cause serious side effects, including:
• See “What is the most important information I should know about OFEV?”
• liver problems. Call your doctor right away if you have unexplained symptoms such as yellowing of your skin or the white part of your eyes (jaundice), dark or brown (tea colored) urine, pain on the upper right side of your stomach area (abdomen), bleeding or bruising more easily than normal, feeling tired, or loss of appetite.
• Your doctor will do blood tests regularly to check how well your liver function is working during your treatment with OFEV.
• diarrhea, nausea, and vomiting. While you are taking OFEV, your doctor may recommend that you drink fluids or take medicine to treat these side effects. Tell your doctor if you have diarrhea, nausea, or vomiting or if these symptoms do not go away or become worse. Tell your doctor if you are taking over-the-counter laxatives, stool softeners, and other medicines or dietary supplements that can cause diarrhea.
• heart attack. Tell your doctor right away if you have symptoms of a heart problem. These symptoms may include chest pain or pressure, pain in your arms, back, neck or jaw, or shortness of breath.
• stroke. Tell your doctor right away if you have symptoms of a stroke. These symptoms may include numbness or weakness on 1 side of your body, trouble talking, headache, or dizziness.
• bleeding problems. OFEV may increase your chances of having bleeding problems. Tell your doctor if you have unusual bleeding, bruising, or wounds that do not heal. Tell your doctor if you are taking a blood thinner, including prescription blood thinners and over-the-counter aspirin.
• tear in your stomach or intestinal wall (perforation). OFEV (nintedanib) may increase your chances of having a tear in your stomach or intestinal wall. Tell your doctor if you have pain or swelling in your stomach area. The most common side effects of OFEV are diarrhea, nausea, stomach pain, vomiting, liver problems, decreased appetite, headache, and weight loss. These are not all the possible side effects of OFEV. For more information, ask your doctor or pharmacist.

How should I store OFEV?
• Store OFEV at room temperature 68°F to 77°F (20°C to 25°C).
• Keep OFEV dry and protect from high heat.

Keep OFEV and all medicines out of reach of children.

General information about the safe and effective use of OFEV.
• Medicines are sometimes prescribed for purposes other than those listed in a Patient Information leaflet. Do not use OFEV for any condition for which it was not prescribed. Do not give OFEV to other people, even if they have the same symptoms you have. It may harm them. This Patient Information leaflet summarizes the most important information about OFEV. If you would like more information, talk to your doctor. You can ask your pharmacist or doctor for information about OFEV that is written for health professionals.
• For more information, go to www.ofev.com or call Boehringer Ingelheim Pharmaceuticals, Inc. at 1-800-542-6257, or (TTY) 1-800-459-9906.

What are the ingredients in OFEV?
Active ingredient: nintedanib.

This Patient Information has been approved by the U.S. Food and Drug Administration.

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Rare Autoimmune Diseases: Individually Rare, Collectively Common

While millions of Americans are diagnosed with an autoimmune disease, many of the diseases in this category are considered extremely rare.

The American Autoimmune Related Diseases Association (AARDA) estimates that 50 million Americans have one or more autoimmune disease. In total, there are more than 100 autoimmune diseases.

The challenge of accurate diagnosis
For people with rare autoimmune disease, getting a proper diagnosis can be one of the most difficult challenges they face. AARDA conducted a survey of autoimmune disease patients and found that the majority of those eventually diagnosed with autoimmune diseases had significant problems in getting a correct diagnosis. Many were incorrectly diagnosed with a variety of conditions that have no specific blood test to confirm the diagnosis. Many were told that their symptoms were “in their heads” or that they were under too much stress. Furthermore, the survey revealed that 62 percent of autoimmune disease patients had been labeled as chronic complainers or were told that they were overly concerned with their health in the earliest stages of their illnesses. On average, autoimmune patients see four different doctors over a three-year period before an accurate diagnosis is made. Many rare autoimmune diseases have confusing and unrelated symptoms, which makes it difficult to arrive at a timely and accurate diagnosis.

Areas of improvement
Increased research funding, for both individual rare autoimmune disease and collective autoimmune disease, is necessary to streamline the diagnosis and treatment of rare autoimmune disease. In addition to research, it is important to provide autoimmune disease awareness and education for patients, practitioners, and caregivers. To learn more about the American Autoimmune Related Diseases Association’s mission to help facilitate collaboration in areas of autoimmune disease education, public awareness, research, and patient services, visit www.aarda.org.

Virginia Ladd, President and Executive Director, American Autoimmune Related Diseases Association (AARDA)

In May, Novartis’ subsidiary AveXis won U.S. Food and Drug Administration approval to market Zolgensma, its potentially curative, one-time gene therapy for a rare and fatal disease: spinal muscular atrophy (SMA) type 1.

SMA type 1 is one of thousands of rare diseases. In the United States, any condition that affects fewer than 200,000 people is considered to be a rare disease. According to the National Institutes of Health (NIH), there are an estimated 7,000 rare diseases, about 80 percent of which have an underlying genetic cause.

More than half of these conditions affect children and many of them are deadly. About 30 percent of children afflicted with a rare disease will die before the age of five.

Improving the lack of treatment options
About 95 percent of the rare diseases that have been identified to date are without an approved treatment today. About 30 million people in the United States and 400 million people globally suffer from a rare disease.

The convergence of information technology and biotechnology, the movement of low-cost sequencing into clinical use, and the incorporation of artificial intelligence throughout the rare disease continuum are accelerating improvements to research, diagnosis, and care for patients.

The proliferation of low-cost wearable sensors and cameras has transformed smartphones into ubiquitous tools that can be harnessed to monitor patients with chronic conditions, and the emergence of regenerative therapies to not only treat, but functionally cure rare genetic diseases, has fueled new hope among rare disease patients for a brighter future in which they may be free of their conditions.

The challenges ahead
While science and technology are propelling advances in the rare disease space at a rapid pace, the ability to fully capitalize on the opportunities before us will depend on how well society is able to address a range of issues outside the realm of science.

Though scientific challenges remain, there are financial, policy, and man-made barriers that arise when large numbers of organizations and individuals with competing interests try to address complex problems.

As Christopher Austin, director of the National Center for Advancing Translational Sciences at the NIH, said in the newly issued Global Genes’ report, “Next: Imagining the Future of Rare Disease,” half of the failure of translating science into treatments is science and the other half is social science, involving things such as human behavior, organizational behavior, and incentives. “We need to innovate in both,” he said.

Austin said the biggest challenge is in getting all of the players, including the patients, to realize that they may be more productive, not only for themselves but for everybody else, by focusing on the similarities among the diseases rather than the differences.

Daniel S. Levine, Journalist, Global Genes

The Future of Rare Disease Depends on More Than Just Science

Why innovations outside of science are also vital to developing comprehensive treatment and diagnoses for rare diseases.

Just Science

The Future of Rare Disease

Depends on More Than Just Science

Why innovations outside of science are also vital to developing comprehensive treatment and diagnoses for rare diseases.
Prenatal genetic screening and testing has changed dramatically in recent decades. New screening options can help expecting parents understand the possibility of certain conditions with less risk to the fetus.

Genetic screening can occur as early as the embryonic stage as well as throughout a pregnancy. Carrier screening for genetic conditions like cystic fibrosis have become part of routine prenatal care. Patients determined to be a carrier of a genetic condition generally have no signs or symptoms, but their children are at greater risk.

Patients now have access to screening tests to determine if they are a carrier for hundreds of genetic conditions, as well as testing for abnormal chromosomal number in a fetus.

New ways to stay informed
Noninvasive prenatal screening (NIPS) is the newest and most sensitive approach to Down syndrome-screening. It relies on fetal DNA fragments in the mother’s blood that are shed from the placenta.

Using NIPS can result in false positives (i.e. a positive screening result that does not mean a fetus is affected). NIPS offers the lowest false positive rate when compared to other screening methods. All positive screening test results should be confirmed using amniocentesis or chorionic villus sampling (CVS).

A closer look at health
If a patient desires more information, diagnostic testing via amniocentesis or CVS should be considered. Amniocentesis is generally performed at 16-20 weeks of pregnancy, while CVS is done at 11-13 weeks. For each procedure, a sample is sent to the laboratory for chromosome analysis. An additional test called a microarray, where smaller parts of chromosomes/DNA segments are analyzed, can also be performed to help detect hundreds of other genetic disorders. It is always the patient’s decision whether or not to opt for diagnostic testing, screening, or no screening/testing at all.

Decisions on prenatal genetic screening and testing options can be complex. Healthcare providers can use guidelines and educational materials from national organizations like the American College of Medical Genetics and Genomics to help explain these concepts. Medical genetics professionals, such as genetic counselors and board-certified medical geneticists, are available to help interpret what the tests can and cannot tell.

Susan Klugman, M.D., FACMG, FACOG, Board Director of Clinical Genetics, American College of Medical Genetics and Genomics

A Clinical Trial for Arginase 1 Deficiency

A clinical trial of a study drug is currently enrolling participants with Arginase 1 Deficiency (ARG1-D). ARG1-D is a rare inherited disease that leads to the build-up of arginine in the body. Find out if you may be eligible to participate.

www.arginase1.com
Kristin Chenoweth is one of Broadway’s most vibrant and positive stars, but many may not know that she has quietly fought a personal battle with Meniere’s disease, a disorder that affects the inner ear.

“It’s a battle, one that I’ve taken on and I’m so happy to have the opportunity to talk about it,” Chenoweth said. “A lot of people have had it and can’t describe it, and once they know and understand what it is, they can deal with it better.”

The side effects of Meniere’s can be debilitating. Tinnitus (a ringing or fluttering in the ear) and migraines are common symptoms.

“The main aspect that I suffer with is vertigo,” Chenoweth explained, “which can feel like either falling down an elevator very quickly or spinning — imagine if you were in a helicopter that was spinning.”

Balancing career and health
As a live performer, such symptoms can be at best frustrating and at worst incapacitating.

“It’s been very frustrating in a work environment to have vertigo hit me, or a migraine, or any of the various symptoms while I’m trying to work,” she said. “You look sort of like you’ve had too much to drink. You can’t quite keep your balance, or you can’t see.”

Meniere’s symptoms usually arise without warning.

“You can’t decide when it’s going to happen, so you can’t work around it. Inconvenient is a very respectful word for a very disrespectful disease,” she said.

For a singer like Chenoweth, the scariest side effect is the potential loss of hearing.

“One of the ways to help Meniere’s is to have an inner ear surgery to help to correct what I would call a piece of tape in your inner ear that holds onto the little bones that help your balance. So let’s say my piece of tape is bad and needs to be replaced, but with the replacement, you will most definitely be deaf,” she explained.

Managing a lifelong illness
When the symptoms kick in, Chenoweth said there’s not much she can do but wait for it to pass.

“When I feel it coming on — it sounds silly, but everybody has their way — I definitely pray immediately, and I ask God to just help me through it,” she said. “And I call my mom for support.”

Chenoweth said that the best resource for support is the love of friends and family. “I’m very lucky that I have a very small but tight-knit, close, loving group that surrounds me with love,” she said. “Love can get you through a lot of things.”

Taking positive steps
For those suffering from rare diseases like Meniere’s, the constant battling can feel lonely.

“For people who feel hopeless — which I’ve been there, believe me — I look for things to inspire me,” Chenoweth said. “A lot of that comes through music.” Chenoweth’s latest album, “For the Girls,” available September 27, is an ode to female musicians that have personally inspired Chenoweth, from Dolly Parton to Ariana Grande.

While Meniere’s disease has been a burden in Chenoweth’s life, she feels that sharing her story has helped her move beyond the burden by helping others understand.

“I like to do something where I can give to somebody,” she said. “I think when you give, you receive, and that’s the secret right there.”

Ross Elliott
Emerging Breakthroughs in Personalized Medicine Improves Care for Rare Disease Patients

New tests and therapies may help doctors diagnose rare disease patients a lot faster and lead to more effective treatments.

Although scientists have long understood that most rare diseases are caused by harmful genetic mutations, it often takes several years for doctors to identify which gene is causing a patient’s specific symptoms. These years take a toll on patients and their families, who watch loved ones suffer despite dozens of costly tests and visits to various doctors and specialists. Even after the disease is diagnosed, existing daily maintenance medications can sometimes prove woefully inadequate.

Fortunately, new tests and therapies emerging in an era of personalized medicine are helping physicians use diagnostics to determine which medical treatments will work best for each patient.

Genetic testing
A new technology called next-generation sequencing (NGS) can test for thousands of genetic mutations at one time. Scientists hope NGS tests may someday replace the battery of single-gene tests that doctors often use to understand the potential causes of a patient’s symptoms. The use of NGS tests may help shorten the diagnostic odysseys that many rare disease patients must endure before receiving an accurate diagnosis, thereby reducing associated expenses and improving patients’ lives.

Personalized medicine
Meanwhile, to improve the prospects for patients with rare diseases after they are diagnosed, the biopharmaceutical industry is developing an emerging group of personalized medicines known as gene therapies. Gene therapies promise to deliver lasting benefits by reversing the genetic causes of diseases. The U.S. Food and Drug Administration approved the first gene therapy, called Luxturna (voretigene neparvovec), in 2017. By revising a harmful genetic mutation, Luxturna can restore vision to patients with Leber congenital amaurosis, a rare genetic retinal disease.

Researchers studying the benefits of NGS tests, gene therapies and other personalized treatments hope that their work will help advance a new era in healthcare that quickly targets more effective treatments to patients who will benefit from them.

Edward Abrahams, President, Personalized Medicine Coalition

America’s Most Successful Public Health Program at Risk

A life-saving bill is set to expire on September 30, 2019. The Senate must act. Twelve-thousand babies and their families are depending on it.

George Fox loved growing up in Florida. When he and his high school sweetheart got married and settled down in the Florida Keys, they looked forward to a dream life. After their son Phoenix was born, George beamed when the doctor remarked on how strong his baby appeared.

Within weeks, however, little Phoenix began showing symptoms of a life-threatening heart abnormality. After months of tests, he was diagnosed with Pompe disease, an incurable muscle-wasting condition that can cause heart weakness and, if left untreated, can result in death. One in 28,000 Americans suffer from Pompe.

Screening programs save lives
Of the 4 million babies born in the United States each year, 1 in 300 are found to have a potentially devastating condition through screening. Newborn screening is the most successful public health program in the history of our country.

A bill currently before Congress, the Newborn Screening Saves Lives Reauthorization Act, or S. 2158, would continue to provide assistance to states in order to improve and expand newborn screening programs.

Time is running out
If S. 2158 does not pass, other states may not be able to implement testing for Pompe and other newly-detectable diseases. Please go to RareScreening.org to contact your Senators and ask them to co-sponsor and support S.2158.

Mark Dant, Chairman of the Board of Directors, The EveryLife Foundation for Rare Diseases

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PV, a rare chronic blood cancer, can bring a multitude of symptoms, including fatigue, night sweats and itching to name just a few. And these symptoms can change so slowly over time you might not notice. In fact, new symptoms, or changes in the frequency or severity of your PV symptoms can be a sign of disease progression. You track and discuss blood counts with your Healthcare Professional to monitor your PV and help prevent blood clots and other serious complications. But it's just as vital to be aware of—and discuss—any new or changing PV symptoms as well. For a more complete list of symptoms, and tools to help you track them, visit TakeActionPV.com