Center for Healthcare Innovation

Equitable Access to Rare Disease Therapies Workshop Executive Summary

Table of Contents

Introduction

On May 23, 2019, the Center for Healthcare Innovation (CHI) held an Equitable Access to Rare Disease Therapies Workshop which took place in Washington D.C.

The workshop focused on identifying solutions to the challenges preventing equitable access to rare disease therapies in the United States. This workshop brought together patients, families, patient advocacy groups, biopharmaceutical firms, providers, policymakers, and other NGOs to discuss how to achieve equitable access to care. The panels focused on the latest developments and prominent challenges in accessing novel rare disease treatments, as well as encouraging and accelerating innovation and utilization. In addition, we also discussed the current barriers to care, including delayed access to treatments and drug pricing.

The workshop featured a Distinguished Keynote Address by Dr. Marshall Summar, Chairman of the Board of National Organization for Rare Disorders. Panel discussions included industry, provider, and patient focuses to facilitate dialogue around the challenges and opportunities for each group.

This executive summary aims to:

- Define the challenges for patients, providers, and other professionals in the rare disease community
- Summarize the innovative ideas and potential solutions on innovation and access
- Discuss best practices to better serve rare disease patients both in current and future state

The paradigm shift towards patient-centered care and increasing emphasis on rare diseases highlights the need for innovative treatments and technologies. This executive summary aims to serve as a guide of the ideas and insights regarding the challenges and calls to action in the rare disease community.

Background

With just 5% of the over 7,000 rare diseases addressed by some form of drug treatment, 95% of rare diseases remain without a therapeutic option. Rare diseases affect 1 in 10 Americans, yet many of these debilitating conditions impact a very small number of patients. The biopharmaceutical industry is delivering tremendous innovation, from disease-modifying medications to curative gene therapies, to address previously unmet medical needs. It is imperative that these innovations be accessible to those who need them. This workshop brought together patients, families, patient advocacy groups, biopharmaceutical firms, providers, policymakers, and other NGOs to discuss how to achieve equitable access to care for rare diseases. Topics included current barriers to care, ensuring earlier access, and drug pricing. Our goal is to empower patients and families who are affected by rare diseases.

7,000

Rare diseases exist and new ones are discovered each year

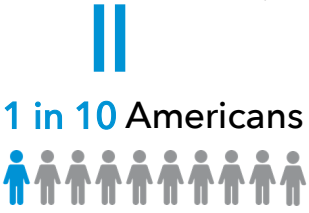
5% of rare diseases have FDA-approved treatments

80% of rare diseases are inherited

Rare disease affects...

30 million people in the United States

1 in 10 Americans



30 million people in the European Union

350 million people worldwide

The vast majority of rare disease patients are **CHILDREN**

SOURCE: https://globalgenes.org/

I. Industry Panel

MODERATOR: Dr. Chitra Edwin, PhD, RAC, is a regulatory affairs and compliance professional with over two decades of experience in the biotechnology, and diagnostics private sector. She has been a key team leader in the product development of approved, infectious diseases, oncology and cardiology products. She has held leadership positions in the biopharmaceutical, and diagnostic industry including Spotlight Innovation, the Cleveland Heart Lab (Quest Diagnostics), Mass Biologics, and Chiron Corporation (Novartis). She is an Adjunct Associate Professor of Pharmaceutical Sciences, and was the Director of the Capstone Project, Master's in Drug Development Program, College of Pharmacy, University of Cincinnati. Dr. Edwin earned her Ph.D. in Medical Microbiology and Immunology from the University of Minnesota, and then trained at the Harvard Medical School as a Post-doctoral Research Fellow at the Brigham and Women's Hospital.

Mr. Matt Hall is the CEO at Human Care Systems (HCS). Matt founded HCS in 2008 to deliver systematic, evidence-driven solutions to improve real world health outcomes. HCS provides complete therapy support solutions to pharmaceutical companies globally. Prior to founding HCS, Matt co-founded the Kerdan Group, a biopharma and life science consulting firm. Matt has also run a healthcare information business, worked in venture capital and as a consultant at Bain & Company. He has lived and worked in Europe, Japan, South Africa and South Korea. Matt holds a bachelor's degree with honors in Political Economy from Williams College.

Mrs. Jamie Van Iderstine, Vice President of Client Engagement, sits on the leadership team of Cyan Health, where she develops comprehensive and high-impact market access strategies for a spectrum of biopharma clients, large and small, across many categories, including multiple rare diseases. Prior to her tenure at Cyan, Jamie cultivated her deep market access expertise at Ogilvy Commonhealth, where she led market access launches in complex disease states, includingpulmonary hypertension and cystic fibrosis. With experience navigating all points in the product life cycle and addressing a wide range of market scenarios, Jamie specializes in connecting the dots of opportunity required to maximize access potential for pharmaceutical brands. Challenged daily to overcome the obstacles that stand between patients and the therapies they need, Jamie has developed a passion for principles of patient access and affordability.

Ms. Crystal Vanuch is President and CEO of Capitol Hill Solutions located in Washington DC. With over 12 years of experience leading large and small Public Relations, Government Affairs, Policy and Advocacy teams for Healthcare companies such as Pfizer, Vivus, Amylin, and Avanir Crystal has led major patient access and reimbursement initiatives at the state and federal level. Founding Capitol Hill Solutions 2 years ago, she works with clients in the healthcare industry to identify regulatory and legislative challenges that could impede best practice patient care. With a robust background in establishing relationships with Congressional, Agency and Advocacy stakeholders, Crystal has experience in passing tough legislation at the state and federal level that has major impacts to clinical research, development, and commercialization of pharmaceutical products.

Dr. Sharad Verma, PhD, is Director of Research and Development for the Neurofibromatosis Therapy Acceleration Program (NTAP), and an Assistant Professor in the Department of Neurology, at The Johns Hopkins University School of Medicine. In these roles, Dr. Verma oversees basic science and translational initiatives as they pertain to the discovery and development of therapeutics for the treatment of plexiform and cutaneous neurofibromas. Before joining Johns Hopkins University, Dr. Verma was in the pharmaceutical industry for over 14 years at Bayer and GlaxoSmithKline, serving in various R&D leadership roles in the discovery and clinical development of oncology therapeutics. Dr. Verma was a member of multiple project teams spanning the areas of signal transduction, cancer epigenetics, and immuno-oncology, which delivered a total of 6 clinical development candidates (including one that is now FDA approved).

The Orphan Drug Act, passed in 1983, served to establish incentives for the development of orphan drugs. The 1984 amendment defines orphan drugs as any disease or condition that affects less than 200,000 individuals in the United States.[1] This effort has been primarily considered to be a success in its goal of encouraging the development and marketing of orphan drugs as the number of drugs receiving the necessary designation has been increasing (Figure 1). However, despite these considerable improvements, the majority of rare diseases remain without treatment. With approximately 250 new rare diseases and conditions discovered each year, that majority is continually increasing.[1]

Additionally, costs for medical care for diseases outside of the more common rare diseases remain concerningly high. The median annual price for an orphan drug in 2017 was over $46,800, with a mean cost of $87,319.[2] Conversely, the median yearly cost of the top ten rare disease therapies, determined by the number of patients, in that same year was $1,216 with a mean of $9,676 (Figure 2).[2] The inverse relationship

Figure 1

Number of Orphan Indications Approved Since 1983

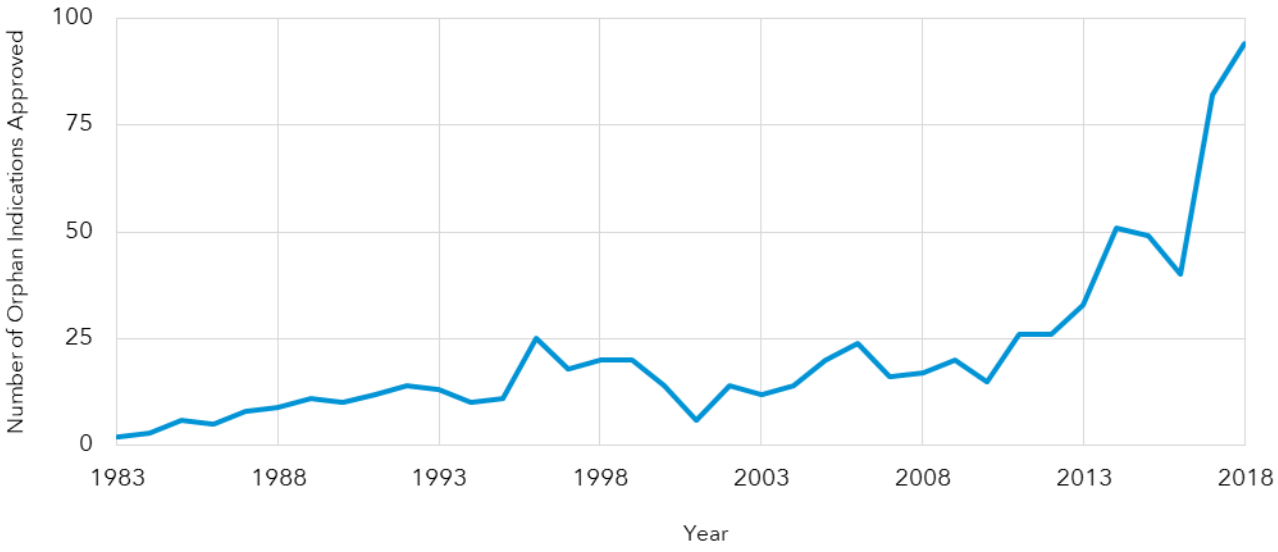
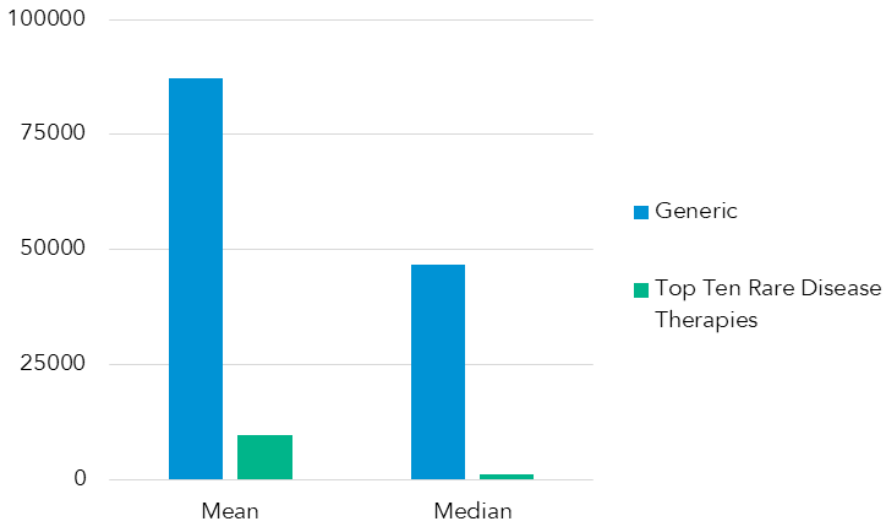


Figure 2

Average Cost of Therapies Per Year Per Patient



between costs and the number of patients can be extremely beneficial to those with the most common rare diseases. It is problematic as it leaves a patient population with high-cost barriers that have the potential to inhibit a patient's ability to receive proper treatment. The average patient population per orphan therapy is 5,730 people. Given this number and the number of rare diseases known, there is a sizeable population without affordable treatment options.

Given these successes and challenges, more incentives and motivators are necessary to help bridge the gap between the rates of approving treatments and the discovery of new conditions. This need is because in addition to barriers in access to care that rare disease patients experience, fewer than 10% receive disease-specific treatment.[3] Many panelists emphasized the opportunity to take more advantage of incentives that the FDA has already allowed for, in some cases suggesting expanding on current programs and frameworks. An example of this is priority review vouchers, which would permit for "passes" to expedite FDA approval. A

company that receives one of these can either use it themselves or sell them to other corporations as a means of generating funds for further research and projects. Panelists called for those to apply pressure to expand this current system and to allow for the voucher to be repurposed to advance areas such as genetics and pediatrics. In addition to this, panelists recommended that patent protections are extended to incentivize innovation further. There is the potential to apply known drugs and treatments to broader patient populations. As a panelist put it, "when we learn about genotype, we learn about phenotype." New knowledge and innovation can lead to opportunities for repositioning drugs, which can save valuable time, given that the traditional drug discovery process takes on average 10-12 years. There have been successes with this idea as well: of the 51 new medicines that got to market for the first time in 2009, 30% of them were repositioned drugs.[4]

"New knowledge and innovation can lead to opportunities for repositioning drugs, which can save valuable time, given that the traditional drug discovery process takes on average 10-12 years."

One of the panelists stressed that we need dialogue between academics and private companies. Furthermore, advocacy groups need to be brought in to the process to provide patient and business perspectives. Having these partnerships can help in facets like navigating market dynamics, such as with competing generic drugs. The orphan drug designation was revisited and emphasized as a priority of the utmost importance. A panelist stressed that the FDA needs to realize and prioritize based on the resources companies are putting into developing these drugs. A proposed additional incentive could be having a higher priority for review, which would reduce wait times. Additionally, extended patent protections for specific indications can further incentivize innovation.

Given the small base of patients with rare diseases, there can be issues with finding patients for clinical trials and having those trials be feasible for a patient to participate. Given feasibility, the panel discussed that a company needs to have a certain level of awareness about the realities of living with a particular disease and take these into account when deciding how to run clinical trials (Table 1). These issues also compound themselves with funding. This is both on the part of those running the trial and with patients regarding compliance. To begin to address these issues, companies need to prioritize early and consistent dialogue with advocacy organizations. More specifically, these partnerships should be developed early on in the design process, so there can be constant feedback to relay to the FDA.

Table 1

Clinical Trial Participant Experience by Age Groups

	18-34 yr.	35-44 yr.	45-54 yr.	55-64 yr.	≥65 yr.
Somewhat or difficult to understand consent form	20.9%	16.2%	7.7%	8.1%	5.7%
Somewhat or very disruptive on daily routine	47.8%	35.8%	19.8%	15.7%	11.8%
Overall time commitment too much	15.9%	11.6%	9.0%	8.4%	5.5%
Missing too much work	13.9%	9.3%	5.9%	5.5%	1.4%
Study procedures at home too cumbersome	10.9%	8.7%	5.9%	5.4%	3.9%
Childcare or other family care cost of much	10.0%	6.4%	2.2%	1.4%	0.9%
Slow reimbursement for out-of-pocket expenses	9.5%	7.5%	5.6%	3.0%	2.9%

Regarding the experiences of individual patients, there are cases where there must be a proxy for efficacy, which can at points be problematic. Additionally, it may be hard to communicate the actual value of the therapy, especially when working with a strict amount of material. The most prominent proposed solution was the utilization of virtual models, which may help alleviate some of the burdens on patients. Virtual models can be better tailored to fit around the experience a particular patient is having, making clinical trials far more feasible. Patients can be better supported within the comfort of their homes for some visits, greatly easing the burdens placed on them.

There is the overarching "time to diagnosis" issue that leaves patients without proper treatment or access to clinical trials for years. The average time it takes for rare disease patients to receive an accurate diagnosis is eight years.[5] Furthermore, patients visit, on average, 7.3 physicians, before being given a diagnosis,[6] further complicating the path to treatment. One panelist provided the statistic that for those who are born with their diseases displaying symptoms, treatment is usually not obtained

until they are six years old. The timing is crucial as some diseases could be cured or have their symptoms severely reduced if treated within the first six months. Within this, misdiagnoses lead to delays in receiving proper care, as is the case for over 40% of rare disease patients.[7] A potential solution to these issues, Universal Screening Panels, is developed and recommended federally. Addressing this discrepancy is critical: 3 out of 10 children with a rare disease will not live to see their 5th birthday,[5] so by delaying care, lives are in danger.

Financial illiteracy is an extremely prevalent problem for patients as it can cause insurmountable damage for themselves and their families. One of the panelists provided the statistic that approximately four out of five patients do not accurately calculate out of pocket costs for a regular doctor visit. Additionally, according to a 2016 survey, people are generally twice as uncomfortable figuring out how to pay for healthcare than technology.[8] These groups in health finance have an opportunity as well as an obligation to help with this issue. Studies report that 86% of clinicians, clinical leaders, and executives believe that physicians do not have the proper training to help patients navigate the financial burdens of treatment. However given this issue, there is a chance for interest groups to help as well as an opportunity for advocacy groups to get involved.

The panel brought up the fact that at the core of many of these present issues, we must remember that companies need to and act in the interest of turning profits. They need to consider questions such as investment

8 YEARS

Average time it takes for rare disease patients to receive an accurate diagnosis

7.3 PHYSICIANS

are visited before rare disease patients being given a diagnosis

3 OUT OF 10

children with a rare disease will not live to see their 5th birthday

returns, whether the disease can be diagnosed quickly, how reimbursement for doctors work, and at what costs will it be for the payers. A panelist mentioned that there is an opportunity in this as the drive for profit can push pharma companies to develop drugs in the largest unmet areas. If a company can develop a drug and the market is big enough, they make a profit while being able to provide treatment for a group that previously had none available. Where this logic does not work, the incentives previously mentioned can play a role in creating the opportunities that drive innovation in a capitalist sector. There are also considerations for upward investment within companies and the drugs they already develop. A greater understanding of genetics allows for the correct application of available options. In this expanding field, it is becoming evident that gene therapies are the future of this growth.

"A greater understanding of genetics allows for the correct application of available options."

The most emphasized point on this panel was that more collaborative efforts with the government and advocacy groups are needed to address many of these issues further. As a panelist stated, one of the most prominent misunderstandings is that the FDA and pharma have a contentious relationship; two bodies that need to work in conjunction with one another. Patients need support that is specific to how they are suffering rather than blanket solutions. Advocacy is an essential part of these efforts, and those who fund these efforts are the drivers behind the agendas. It is important to note that much of the work addressing these issues starts in Congress. Many of these advocacy groups do not have enough donors behind them to afford lobbyists. This problem brings in the inevitable question of how to address this gap in monetary capital for different interest groups in this debate.

"Policy makers may also consider epidemiologic information on prevalence and disease burden - in combination with scientific, political, economic, ethical, and other factors - in making decisions about the allocation of resources for biomedical research. Decisions about research spending, for example, sometimes favor the relatively more common rare conditions such as ovarian cancer, neurofibromatosis, and sickle cell disease, but decision makers have also directed resources to extremely rare diseases, constituent with the value judgements underlying the adoption of special politicizes to encourage research on rare diseases."[9]

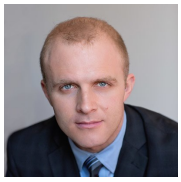
II. Provider Panel

Mr. Joseph Gaspero is the CEO and Co-Founder of CHI. He is a healthcare executive, strategist, and researcher. He co-founded CHI in 2009 to be an independent, objective, and interdisciplinary research and education institute for healthcare. Joseph leads CHI's research and education initiatives focusing on including patient-driven healthcare, patient engagement, clinical trials, drug pricing, and other pressing healthcare issues. His leadership stems from a wide array of experiences, including founding and operating several non-profit and for-profit organizations, serving in the U.S. Air Force in support of 2 foreign wars, and deriving expertise from time spent in industries such as healthcare, financial services, and marketing. Joseph's skills include strategy, management, entrepreneurship, healthcare, clinical trials, diversity & inclusion, life sciences, research, marketing, and finance.

Dr. Sameer Ather, MD, PhD, is a Cardiologist and Staff Physician at Birmingham VA Medical Center. He is also is the President and CEO of XpertDox, a platform for rare disease patients to find personalized recommendations for their condition including specific information about their disease, list of expert doctors and hospitals and peer to peer support. Dr. Ather did his medical residency and PhD at Baylor College of Medicine, Houston, TX, and completed a cardiology fellowship at the University of Alabama at Birmingham. During the course of his clinical practice, Dr. Ather saw patients struggling to navigate the complex world of health care. XpertDox was built to help improve patient access to doctors, hospitals, clinical trials and peer to support by leveraging big data analytics.

Dr. Suvankar Majumdar, MD, is Chief, Division of Hematology at Children's National Health System. Dr. Majumdar was born in Zambia, attended the University of Zimbabwe College of Health Sciences and conducted his postdoctoral medical education at the University of Mississippi. Dr. Majumdar served as the director of the Comprehensive Pediatric Sickle Cell Program at the University of Mississippi Medical Center. He previously directed the Mississippi Hemophilia Treatment Center and is a recognized leader in hematology and sickle cell disease. In addition to his broad clinical expertise, Dr. Majumdar is an accomplished researcher and a principal investigator of NIH-funded studies.

Ms. Rosemarie Truman is an entrepreneur, growth strategist, distinguished corporate executive, angel investor and prolific startup catalyst. Rosemarie's specialty is growth strategy, having led strategies for 50 of the global Fortune 100 companies in over 15 countries. Rosemarie led strategies that resulted in $300+ billion in yearly revenue and 190+ new products. Currently, Rosemarie is the Founder and CEO of the Center for Advancing Innovation (CAI), a 501c3 non-profit which has been coined "Tinder for Startups" and "Shark Tank on Steroids." CAI partners with 80 organizations across the US to gain access to 170,000 inventions and has launched 280+ startups over the last 4 years; created 3500 knowledge-based jobs.



Dr. Kent Werner, MD, PhD, is Co-founder and CEO of Cogentis Therapeutics. Dr. Werner is a board-certified neurologist and neuroscientist. He holds Assistant Professor appointments in the Departments of Neurology at the Uniformed Services University and adjunct at Johns Hopkins University. He has 14 years of experience in biomedical research and seven years in clinical training. Kent received his MD and PhD in Cellular and Molecular Medicine in the department of Neuroscience in 2012 at the Johns Hopkins University under Dr. Solomon Snyder. His areas of research include neurodegenerative conditions to include dementia and traumatic brain injury and their relationship to sleep.

One of the many challenges that underserved groups face is the lack of knowledge and opportunity surroundings clinical trials. A panelist expressed that many people, especially those in these underserved groups, do not have the opportunity to find clinical trials, nor are they recruited to be in one. A 2001 survey found that 85% of patients were initially unaware or unsure that participation in a clinical trial was an option for them. It also found that 75% of those patients would have been willing to participate.[10] A 2011 survey also found that negative attitudes about participation significantly decreased after learning about clinical trials.[10] Furthermore, this problem of lack of knowledge and awareness of clinical trials expands to practicing physicians as well. Even clinicians do not often know which or in some cases, even what type of doctor treats certain rare diseases. This concern is an area of note as 77% of patients in clinical trials first learn about them from their healthcare providers.[10] The problem grows exponentially as with many rare diseases, there are only one or two doctors in the country that have experience with patients with that particular disease. These problems, as well as social barriers, including race, ethnicity, and literacy, create an environment that places the responsibility of seeking care and clinical trials on the patient and their families. An issue surrounding this is compliancy and the burden of getting patients to their visits. Pharma often does not pick people from underserved groups due to issues such as these. As one panelist expressed, "we are expecting people to find their way and also find all of their effort to be worthwhile."

"85% of patients were initially unaware or unsure that participation in a clinical trial was an option for them."

With regards to life-threatening cases, clinical trials may be the only chance a patient has. The communication and knowledge barriers between clinicians and researchers need to disappear to serve patients better and potentially save the lives of patients with rare diseases. On average, about 7% of patients are referred to a clinical trial by their healthcare provider.[12] This lack of communication and knowledge does not match the growing opportunities that clinical trials are providing. A massive development for trials has been the emergence of gene therapies. The goal of finding a "champion" physician and the promise of new treatments has led to more patients actively wanting to be involved in research studies. Their participation is contingent upon knowing that the trial exists in the first place, which is why the discrepancy needs to be addressed.

Over time diagnosing a patient with a rare disease has become quicker and less expensive. The same is not correct for receiving treatment. While improvements to genetic test pricing have occurred as general pricing has gone down, there is still much room for improvement. As one panelist noted, much of this testing is even denied by a patient's insurance company; this issue in itself takes valuable time to work through.

Additionally, the panel addressed the overarching question of why rare diseases are so hard to treat. An issue that was raised by one of the panelists was that pharmaceutical companies tend to shelve 7 out of 10 drugs. Of those that survive, about 86.2% fail to win approval, significantly reducing potential treatment options.[11] An issue, referred to as the "invisible hand syndrome," highlights that we do not know what shelved drugs can be harnessed to be successfully commercialized. Another panelist highlighted that the gap between discovery and clinical trials is unfathomably wide. This is especially problematic for small companies that may not have the resources to sustain throughout this gap of time. Given that pharma relies on these smaller companies for innovation, we are potentially losing out on huge advancements.

"An issue, referred to as the "invisible hand syndrome," highlights that we do not know what shelved drugs can be harnessed to be successfully commercialized."

The example of Sickle Cell disease, in which the disease was discovered 110 years ago but only has two known treatments currently, was brought up for discussion with the panel. In the last 30 years, we have developed a better understanding of the mechanism of the disease, which has lead to targeted treatments. Despite these improvements in our knowledge, the common issues of lack of communication and coordination have slowed progress. More people are successfully recruited to participate in trials, but the approach has admittedly not been very coordinated. Without coordination and the availability of information, progress has undoubtedly been stifled.

One question raised once again brought up the topic of technology in reference to how it can be used to link people in clinical trials as well as facilitate engagement. A panelist further emphasized the importance of technology going forward in this, adding that we cannot count on humans to connect promptly in areas where the connections need to be made. The technology could aid with this issue, especially with the maintenance of care issues such as follow-up appointments and primary care. Examples of this include sensors on beds, doors, and medication containers to collect valuable data.[13]

III. Distinguished Keynote Address

Dr. Marshall Summar, MD
Director, Rare Disease Institute at Children's National Health System
Chairman of the Board of National Organization for Rare Disorders

Dr. Summar is well-known for his pioneering work in caring for children diagnosed with rare diseases. He joined Children's National in 2010 where he leads the Division of Genetics and Metabolism. He founded and Directs the Rare Disease Institute in 2017, the first dedicated home for the clinical care of patients with genetic rare diseases and The National Organization for Rare Disorders first designated Rare Disease Clinical Center of Excellence. Dr. Summar's laboratory works on both devices and treatments for patients with genetic rare conditions and adapting knowledge from rare diseases to mainstream medicine. His work has resulted in new drugs in FDA clinical trials for patients with congenital heart disease and premature birth. He has over 30 patents and patent applications. His laboratory is best known for its work in the rare diseases affecting nitrogen, ammonia, and amino acid metabolism. Dr. Summar has also organized and led a number of international work groups to develop and publish standards of care and treatment for rare diseases resulting in significant improvements in outcomes. He has built remote/telemedicine programs to reach patients currently without genetic care access. Dr. Summar is board-certified in Pediatrics, Clinical Genetics, and Biochemical Genetics and has been listed with Best Doctor's in America since 2004. He serves as President of the Board of Directors of the National Organization for Rare Disorders and is the past president of the Society for Inherited Metabolic Disorders. At NORD he is spearheading an effort that has created digital registries for families to collect long -term information about poorly understood diseases. He is very active in newborn screening policy issues and in developing testing and follow-up systems.

III. Distinguished Keynote Address

The keynote address was presented by Dr. Marshall Summar, Chairman of the Board of National Organizers for Rare Disorders, who has been working in his chosen field since 1985. He is known for his groundbreaking work regarding care for children with rare diseases and holds board certifications in both pediatrics and clinical genetics and biochemical genetics. He currently leads the Division of Genetics and Metabolism at Children's National, seeing over 8,000 rare disease patients each year. In his keynote address, Dr. Summar began by introducing rare diseases and the highly specialized field. Rare diseases affects approximately 1 in 10 Americans and only 5% of rare diseases have FDA-approved treatments. Additionally, the vast majority of rare disease patients are children, making work in pediatrics all the more essential.

Due to the chronic nature of these diseases, Dr. Summar focused on the unique patient experience of those living with rare diseases. Given the small percentage of affected patients, there is limited data, preventing those with these diseases from obtaining answers and predictability, which is of particular importance to patients. Due to the low prevalence of these conditions and limited clinical expertise, diagnosis and treatments often require the coordination of multiple specialists. These limitations also force patients and their families to become knowledgeable, or experts, in their own disease states. Due to this shift in the traditional patient-physician relationship, incorporating patients in the decision-making process and providing comprehensive information is critical to the success of treatment and care.

"Given the small percentage of affected patients, there is limited data, preventing those with these diseases from obtaining answers and predictability, which is of particular importance to patients."

Dr. Summar attributes much of the advancement in rare disease care to collaboration regarding both standards of care and the sharing of information to fuel new developments and opportunities. However, there is still a tremendous amount of progress to be made. With new research, the number of rare diseases with a clear molecular diagnosis is "exploding," yet in 2017, there were only approximately 600 diseases with available therapies. Even for those with treatment options, they often suffer debilitating financial costs. For example, with pediatric hospitals, 34% of children have a clear genetic condition, yet they account for 51% of all hospital bills.[14]

Another challenge and opportunity highlighted was the lack of medical geneticists available to advance care in this field. There has been an increase in wait times and average new patient caseloads, but not an increase in the already small number of geneticists.[15] There is an opportunity to incentivize young people to go into this field to meet the demands of the patients. Overall, Dr. Summar's address sought to educate on the current issues his organization has identified as well as inspire participants to work to address these issues.

IV. Patient Panel

MODERATOR: Ms. Ricki Fairley, MBA, is a the Founder, President and Thought Leader of DOVE Marketing, a marketing agency with a mission to deliver iconic thinking, strategic problem solving and creative genius to clients seeking profitable business results. Fairley is a seasoned marketer with over 30 years of marketing experience including 20 years in brand management at Johnson & Johnson, Nabisco, Reckitt & Colman and The Coca-Cola Company, and over ten years in agency leadership encompassing strategic planning and consulting for numerous Fortune 500 companies. Ricki is Chair of the Board of Trustees and chairs the Marketing Committee for the Triple Negative Breast Cancer Foundation. As a Stage 3A Triple Negative Breast Cancer Survivor/Thriver, she speaks regularly to advocate for awareness of the disease.

Ms. Christina Hartman, MPH, is Senior Director of Policy and Advocacy at EveryLife Foundation for Rare Diseases. Christina is experienced in elevating the voices of patients, parents, scientists and clinicians to have a positive impact on health and nutrition policy. She worked with staff and member leadership at the American College of Cardiology to develop an agenda of improving cardiovascular health outcomes. At the Pew Charitable Trusts, she worked with a broad range of partners to advance legislative goals that incentivize the development of new antibiotic drugs. Prior to that, she served as an analyst at the Centers for Disease Control and Prevention and in the Office of the Secretary for the the U.S. Department of Health and Human Services (HHS).

Ms. Saira Sultan, JD, is Executive Director of Haystack Project and President & CEO of Connect 4 Strategies, LLC. Saira has represented corporate, nonprofit, and government interests in the legislative and regulatory health policy arena for more than 20 years in Washington, D.C. Saira brings a decade of experience working with market access, health outcomes, and commercial teams in pharmaceutical companies, including Pfizer and Sanofi. Focusing in areas such as oncology, rare and extremely rare therapies, vaccines, and specialty products in sickle cell, pain, addiction, cell and gene therapy and more, Saira has worked extensively with CMS, FDA and Capitol Hill. Saira's leadership at the Association of Community Cancer Centers (ACCC) allowed her to build strong relationships with the oncology community and gain insights into the evolution of oncology care.

Mr. Jim Cavan, MS, is President and CEO at Backpack Health. Jim has great passion for helping healthcare companies tackle systemic problems, and 20 years of executive health research and startup leadership experience. The development of Backpack Health is the result of several key goals of his, including improved access to and control of medical information, and less obtrusive collection of research data. Along with his wife, Julia, he is an avid traveler and amateur chef. When not working, he can be found fly fishing or deep in a book. He lives with Julia and son, Hunter, in Dover, Massachusetts.

Ms. Lydia L. Seiders is a passionate advocate, driven to provoke positive changes globally. Her background includes roughly 20 years of advocacy and 15 years of policy work. She serves as an adviser globally for grassroots efforts, with work published on national and international platforms. In 2016, her oldest daughter was diagnosed with a rare form of bone marrow failure. At her daughter's request, she worked endlessly to create a cause within her daughter's fight and has been successful on an international scale. Worldwide, she partners with bone marrow registries and foundations to ensure patients have accurate information pertaining treatment options. As a result of this work, she was appointed as the first Volunteer Maryland State Ambassador for the NORD's Rare Action Network in 2017.

Ms. Wendy White is Chief Patient Officer at Vitrisa Therapeutics. Wendy is an innovative leader working for over 20 years in Rare Disease at the intersection of advocacy, technology and business. She founded, grew and transitioned Siren Interactive Corporation to Dohmen Life Science Services as part of a long-term vision to integrate patient-level insights into the drug delivery continuum. Wendy was recognized by PharmaVOICE magazine in 2015 and 2012 as one of the 100 most inspiring people in the pharmaceutical industry and by MedAd News with a Manny Award for her work in rare disease. She currently serves as Chair of Global Genes, Chair Emeritus for the Healthcare Businesswomen's Association (HBA) and an Executive Advisor for the Chicago Life Science Consortium.

IV. Patient Panel

The patient panel concentrated on bringing together different perspectives from experts, innovators, and advocates to address the challenges that face rare disease patients on an individual level. The first portion of the panel discussion focused on the impact rare diseases have on patients. One of the complications discussed was that the economic environment surrounding medical care places burdens on providers to charge more in a commercial setting. This factor can lead to systematic discrimination against people with rare diseases. As discussed previously, there is a concerningly large discrepancy in treatment costs between those with more common rare diseases and those with smaller patient populations, creating additional barriers to proper care. One of the proposed mediums of this necessary collaboration is establishing registries for the less developed or known rare disease communities. An effort such as this has many potential benefits, such as connecting patients, their families, and professionals to better share information and work towards comprehensive care and treatment.

"There is a concerningly large discrepancy in treatment costs between those with more common rare diseases and those with smaller patient populations, creating additional barriers to proper care."

An important feature and benefit that a panelist noted was that having a registry allows for patients to have control over the access and therefore their own information. Registries can also allow for the more efficient accumulation of data both for research purposes as well as lobbying for more government resources. There is a strong demand for other forms of information technology with self-service programs, which have seen successes (Table 2) such as web messaging, with a potential to increase process efficiency. Yet, there are also concerns that the increased workload for physicians and costs may outweigh the benefits.[16]

Regarding this overarching issue, one of the most common aspects brought up in the panel was the difficulty many experienced in sharing and coordinating information. Given that a patient on average will see 7.3 doctors before receiving a diagnosis,[17] valuable time can be wasted trying to transfer records to all of the doctors. This also adds to the point of ownership of medical information and history: by streamlining this process to the control and benefit of the patient and their family, there is a returned sense of ownership over their own records which can provide a sense of control. A panelist shared her own story in seeking a clinical trial for her child; the panelist described having to argue with a doctor in order to get her child admitted into a clinical trial that ended up saving their life. They stressed that the Orphan Drug Act cannot go away as it would take away the opportunity for the creation of new treatments that could go onto trials.

Table 2

Impact of Self-Management Programs on Service Use

Health Problem	Impact of Self-Management Programs on Service Use
Chronic Pain, Arthritis	Up to 80% Reduction in Visits to Health Professionals
Arthritis, Insomnia, Asthma	Up to 44% Reduction in Visits to General Practitioners
Sickle Cell Disease, Asthma	Reduction in Hospitalizations and Length of Stay
Insomnia	Up to 15% Reduction in Visits to Specialists
Sickle Cell Disease, Asthma	Up to 39% Reduction in Emergency Room Visits

Quality of life for patients and their caregivers was also emphasized by the panelists. Fatigue for both is common, especially with registries as there is currently little coordination between them. Overall, industry in general needs to be more concerned with quality of life. Ways that this could be accomplished may be in the simplification or automation of processes that can be cumbersome for patients as well as data DIY programs that can ease burdens.

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Tuesday, November 5, 2019 | Atlanta, GA

This symposium brings together best-in-class entrepreneurs, executives, investors, policymakers, researchers, scientists, and technologists drawn from the Atlanta metro region and across the United States. The symposium will focus on the interesting intersection of business, management, strategy, healthcare, and medicine. The emphasis will be on compelling, cross-functional, and inter-disciplinary themes. We will discuss the business case for how diversity, inclusion, and equity --in addition to being beneficial for humanity and society-- are catalytic for decreasing costs, increasing revenues, and ultimately driving higher profits at the organizational level. The event will punctuate the crucial importance of diversity, inclusion, and engagement for modern organizations. Most importantly, we will examine the "why" and "how" D&I are good for patients and their families. This will be an engaging, highly interactive exchange of ideas and information, with the hope and expectation that multiple subsequent collaborations between participants will emerge in the wake of the event.

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Developing the Next Generation of Culturally Competent Healthcare Leaders Workshop

Friday, February 7, 2020 | Austin, TX

CHI's workshop brings together healthcare leaders, medical school academics and leaders, providers, patients, pharma, and other stakeholder groups for an innovative and collaborative discussion on how to best equip our future healthcare leaders with the knowledge, skills, and attitudes that allow them to adequately treat all patients and communicate with their families. Medical school and other healthcare training curriculums must be designed to appropriately train cultural competence. By proactively beginning to think about culturally competent healthcare leaders of tomorrow, we can begin to make steps towards addressing the broader healthcare disparities and heath inequities that afflict the U.S. healthcare system. The workshop will bring together an intimate group of interdisciplinary healthcare thought-leaders, executives, academics, and industry leaders for action for a day of idea sharing, brainstorming, networking, and co-learning.

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