


# Coming Full Circle: Reflections and Inspirations from a Cystic Fibrosis Patient Scientist Panel

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## Abstract

Care for many progressive chronic diseases continues to improve, allowing patients to survive and thrive for longer periods of time<sup>1</sup>. People living with such conditions may now find themselves able to achieve long-term goals in education and career development<sup>2</sup>. Many people now occupy the dual roles of scientist and patient<sup>3</sup>. This commentary article synthesizes experiences of scientists and advocates with the progressive genetic disease cystic fibrosis (CF) who collaborated on a career development session for the Cystic Fibrosis Foundation’s inaugural ResearchCon event in 2019. It explores how such collaborations affirm and transform individual perspectives on patient science and its importance in broader scientific research agenda setting. We first share our own individual insights about the experience and impact of the ResearchCon panel session before progressing to discussion and future directions centering the shared insights from one another’s reflections.

## Keywords

cystic fibrosis, scientific research, career development, patient voices, professional collaboration

## Highlights

### What do We Already Know About This Topic?

Disparities persist in chronic disease research and care that inclusion of patient scientists can help to mitigate.

### How Does Your Research Contribute to the Field?

Our work outlines specific opportunities for engagement of scientists with direct lived experience of CF in research efforts serving our community.

### What Are Your Research’s Implications Towards Theory, Practice, or Policy?

Scientists with CF should be actively centered in related research initiatives to promote both innovation in scholarship and justice in care.

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## Introduction

Care for many progressive chronic diseases continues to improve, allowing people to survive and thrive for longer periods of time.<sup>1</sup> Individuals living with such conditions may now find themselves able to achieve long-term goals in education and career development that previously would have seemed unrealistic.<sup>2</sup> Populations impacted by these conditions thus increasingly count among their number many people who occupy the dual roles of scientist and patient.<sup>3</sup> This unique constellation of experiences and knowledges positions patient scientists as a specific population and chronic disease researchers as a general group to better understand and respond to progressive illness.<sup>4</sup> It also helps people with CF both within and beyond the scientific community achieve empowerment.<sup>5</sup> And contributing data in research can instill a tremendous sense of purpose and meaning in our lives with this shared disease.<sup>6</sup>

The above discourse concisely summarizes current research on patient science in the progressive disease landscape. It also tells—albeit in miniature—the story of our own careers.<sup>7</sup> And like most people with progressive diseases who find ourselves able to fulfill long-term career horizons, we seldom forget that our journeys to this point have not been smooth or simple.<sup>8</sup> Rather, our discovery of what patient science means in our own lives and how best we can translate those interests into impact has taken many twists and turns.<sup>9</sup> But it has also involved “coming full circle” with respect to both our work and the underlying health condition that inspired it.

## Cystic Fibrosis 101

Cystic fibrosis (CF) is a progressive genetic disease in which the mucous membranes produce thick and sticky mucus like rubber cement instead of thin and slippery liquid secretions. It results from defects in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, which codes a chloride/bicarbonate channel protein key to regulating fluid transport across the mucous membranes.<sup>10</sup> The “classic” autosomal recessive presentation of CF includes lung disease caused by a cycle of mucus obstruction, infection, and dysregulated inflammation<sup>11</sup> and gastrointestinal manifestations including malabsorption and pancreatic insufficiency, impaired biliary and liver function, or dysmotility and intestinal obstruction.<sup>12</sup> Per this abbreviated summary and the “classic” qualifier, individual people with CF may experience different facets of the disease with wildly disparate outcomes. Indeed, cystic fibrosis exists across a disease spectrum: CFTR genotypes are a poor predictor of disease severity and manifestation, which suggests complex interactions between environment and non-CFTR genetic background.<sup>13</sup> Additionally, mild or non-symptomatic presentations of CFTR dysfunction are increasingly recognized.<sup>14</sup>

CF is a rare disease impacting approximately 30,000 people in the United States, just over half of whom are adults.<sup>15</sup> Unlike most other rare diseases, CF requires stringent infection control protocols to prevent people from passing virulent respiratory bacteria to one another that can cause permanent harm to those with the disease.<sup>16</sup> People with CF are thus recommended not to interact with one another in person, and to avoid crowded and poorly ventilated spaces even outside the context of viral pandemics. However, technology facilitating realtime online interaction via videoconference has proliferated since the introduction of current CF infection control guidelines in the late 1990s. This has produced increasing collaboration among scientists living with CF, as has growing longevity within the community due to improved and diversifying treatment options. Collaboration efforts between different scientists with CF have often begun at community conferences using videoconference technology.

These include the BreatheCon and ResearchCon conferences now offered annually by the national Foundation. BreatheCon is exclusively for people with CF. It aims primarily to help those living with the disease connect with and support one another. ResearchCon is for all in the CF scientific community, including and beyond those living with the disease. It aims to highlight current advancements in CF research and care, and invites feedback on new scientific directions for the Foundation itself.<sup>17</sup>

## Notes from the First Author

I was tentatively but inconclusively diagnosed with cystic fibrosis as a young child. When I was first given a diagnostic workup for CF, the gene controlling the function of the protein that does not work properly in people who have the disease had not been discovered yet. Only a couple years later, that gene—called the CF transmembrane conductance regulator (CFTR)—was discovered. But it would take a long time before the scientific community would understand the vast diversity of issues with this gene that can cause people to have CF symptoms.<sup>18</sup> As a result, it would take over 25 more years before my own diagnosis was conclusively confirmed.<sup>19</sup> With a progressive disease whose median life expectancy in 2019 was only 47 years, that is a long time to wait for answers.

So long before my diagnosis was ever confirmed, I began the process of searching for answers on my own—and helping others in the chronic disease community find theirs. I became a scientist because I knew my own experiences gave me unique perspectives on chronic illness, and what was missing in health care for people like me. I also became a scientist because I was born as the result of a science experiment—I was conceived through a clinical trial of artificial insemination—and raised in my parents’ neuroembryology lab at a medical school. I thus came full circle in my adult life to both my professional background and the original diagnosis I started out with. Along

the way, I joined a panel of other patient scientists living with CF for a new type of conference: the inaugural ResearchCon event described above. Appropriately, the panel was titled “Coming Full Circle.”

Here we offer a narrative review and synthesis of our own reflections and takeaways from the 2019 ResearchCon event and the collaborations that emerged from it. We first provide an overview of the panel and our process for working together before transitioning into individual reflections on each of our experiences as planners and speakers. We then summarize and explore common threads from these reflections, focusing primarily on the process of working together and how it has shaped our perspectives as practicing scientists living with a common disease. Those of us who have worked in program evaluation and related areas of science continue to incorporate formative assessment robustly in our daily work. So to conclude we offer recommendations for future research on CF, both within and beyond the patient community. And to provide context for how each of our individual journeys with CF compare to the broader population of our peers with the same disease, we each provide a positionality statement as a supplement to the main text. One of our peer reviewers suggested this approach to enhance the impact of the article on social justice in health science, which we greatly appreciate!

### *Overview of the Panel*

Before I joined the planning team for the inaugural ResearchCon event, I only knew one other health scientist with cystic fibrosis personally. I had a colleague in public health at another university who had been diagnosed at birth; we had begun collaborating on CF-related research the previous summer. I had made that connection through a mutual friend several months after having my own diagnosis confirmed. It felt very affirming to meet and begin working with someone whose health experiences paralleled mine so closely despite their having received a conclusive diagnosis much earlier in life, compared to my own tentative diagnosis at age 5 that was not confirmed until I was 32. I found it comforting on a personal level to know that despite decades of poor management, I still had a fighting chance at better health going forwards.<sup>20</sup> But perhaps even more exciting to me was the prospect of sharing research experiences and building kinship with someone who understood me so well.

Until late 2018 though, I had thought myself lucky to find another health scientist with CF at all, let alone one so close to my own age and career stage. Both of us were university professors in similar ranked positions who held public health degrees and worked with major medical centers. We had both commented on the pervasive messaging surrounding CF and health sciences careers while we were growing up—specifically, the idea that these careers were not safe or otherwise feasible for us. As individuals, we enjoyed proving people wrong about what we could safely do and the impact we could make in the process.<sup>20</sup> But we also spoke of a

tremendous sense of isolation and loneliness—of being the only ones like us. I remember telling my colleague, “I knew there must be others like me out there, but unless our friend had introduced us I don’t know that I would ever have found you.”

My world was about to get much bigger very quickly. Toward the end of 2018, I participated in the Cystic Fibrosis Foundation’s annual BreatheCon event for adults with CF. During one of the keynote sessions, I wound up dialoguing in the participant chat stream with a biocomputing specialist from Alabama who was studying CF genetics. I thought it would be pretty exciting to connect him with some of my own students, especially those in the Biomedical Sciences PhD program with which I worked collaboratively. I figured that I would open up an opportunity for our many students interested in biocomputing, several of whom worked with the geneticist whom I had consulted for information about my own rare CFTR mutation, to meet an active scientist in that field who lived with CF. Our students have usually found it quite exciting to meet a social and behavioral medical scientist with a rare disease when working with me, so I thought correctly that they would jump at the chance to interact with my colleague in affirmation of their own interests in biocomputing and genetics.

However, this connection was just the beginning. I had included my father, who had served for nearly a decade as chair of that department, on the email thread with our colleague who had so many advisees interested in genetics applications of biocomputing. He suggested that we bring my colleague in for a department seminar. But having learned something over many years of hearing me say “nothing about us without us” in response to the notion of people without chronic conditions attempting to speak for those with them, he insisted that I be the one to host. This initiated a cascade of emails about technology support requirements for hosting the seminar, which spurred my colleague to mention that perhaps this would be the first of many collaborations that we could achieve through distance technology. Given that we finished the draft of this manuscript during the peak of the global COVID-19 pandemic, we note that the use of such technologies has expanded greatly both within and beyond the CF community. We see these changes as a boon for scientific collaboration in CF research specifically as well as more generally. It amazes us now to reflect on how far our community has come since the event that first brought us together.

At BreatheCon 2018, we had just learned that the CFF would be offering its first ever ResearchCon event at the end of February 2019. Unbeknownst to me, my colleague had jumped onto the planning team for ResearchCon just as I had done at first opportunity—and had agreed to participate in a panel of scientists with CF doing research on CF. With a few keystrokes, I had suddenly become part of that group, and entered a much larger world of scientists with life histories very similar to my own. In the space of a year and a half, I had gone from being an island to finding a couple of friends to joining an entire club—one I never knew existed until that

moment where I agreed to serve on the panel. Suddenly I was immersed in group chats over the Slack collaboration platform about the panel as my colleague and I planned our seminar. I thus quickly felt like part of something much bigger, in a way that my general participation in various aspects of CF community and advocacy had never achieved. I had stopped wondering if there were others like myself out there, and instead begun collaborating with some of those individuals as part of a dynamic team.

### *Reflections from Participants*

After the panel had concluded and our group had debriefed the experience via videoconference, we discussed the idea of developing a manuscript to share our takeaways from this collaboration with the broader chronic disease community. We found powerful resonance in the idea of building togetherness in the isolation of rare disease, and bridging that more universal experience of social progress with the more niche experience of patient science. Because each of us came from a different background scientifically, being patient scientists meant different things to each of us when we began working together. Yet we also found consonance in our sense of priorities for CF research in the present and future. The “Coming Full Circle” title for our panel thus proved especially apt, as the experience both affirmed and transformed our individual perspectives on patient science and its importance in broader CF research agenda setting. So we tasked ourselves with each writing about 500 words summarizing our experiences and what we learned. Below we share our own individual reflections before progressing to discussion and future directions.

*Author 1.* Our first planning call for the CF scientist panel felt like the proverbial breath of fresh air. The five of us excitedly shared our personal stories and our passions for our respective areas of science with no coaxing whatsoever from CFF staff, instead easily exceeding the original time window for that first meeting. I had thought that perhaps my easy rapport with James, my colleague who had invited me to serve on the panel, had been another stroke of luck just like my collaboration with my only prior colleague who had CF. But by the time that initial meeting concluded, I felt a distinct sense of having known my new colleagues for much longer than a couple of hours.

I felt energized and impressed by the unique specializations, accomplishments, goals, and histories of each of my fellow panelists. We had a diverse array between us of genetic profiles, diagnosis journeys, and illness histories just as we had a wide range of educational backgrounds, professional competencies, and scientific interests. Our enthusiasm for learning from one another emerged quickly and became cause for shared celebration as we continued working together to plan the panel.

Perhaps my favorite activity that we did in support of our planning process—other than the calls themselves—was

generating questions in a shared document and then answering each other’s inquiries about our scientific careers. I have always gravitated toward written forms of expression as a means of getting to know people, as I love the reflection and introspection that writing about one’s own experiences involves. Getting to read about my colleagues’ journeys in their own words—and seeing the evolution in each of their thinking as they continued to adjust their responses based on our shared discussion—felt incredibly special and intimate. It also inspired me, as did reflecting on my own experiences.

People talk a lot about “inspiration porn” involving people with challenging health conditions, and rightly so. What I found so refreshing about reading my colleagues’ narratives and allowing them to shape my own thinking was the balance their words struck between candor about the hardships of living with CF and confidence in their own abilities to negotiate these challenges in their continued scientific work. As a professor who had regularly and unflinchingly told stories about throwing up quarts of black slime when dealing with a bowel obstruction, I relished the dark humor and biting wit in many of our narratives, as well as the steely strength these anecdotes communicated. But I found myself thinking anew about the more delicate side of being a health scientist with CF: the constant immersion in both abject fear and ebullient hope.

This feeling seemed readily understood and shared by our group. Moreover, my colleagues clearly understood and beautifully articulated how holding these two experiences in mind simultaneously made all of us better and more effective scientists. In helping one another to bring these stories to life in ways that would resonate with future generations of health scientists living with CF, we thus also supported one another in finding greater peace and affirmation in our own journeys—both the challenges and the triumphs.

For me, an amazing part of participating in this panel for ResearchCon was how a shared mission for a single event blossomed into a shared sense of expansive possibilities for the future. But equally powerful were also all the precious tiny moments of deep kinship, like when we would make fun of Tré for drinking so much LaCroix, or when Miriam would anticipate one of my terrible puns and start laughing preemptively, or when we would pump our fists with joy in hearing about Ella’s success with phage therapy. None of us felt sure when we had made the transition from colleagues to friends, but it had certainly happened by the time we went live for our panel at ResearchCon.

*Author 2.* I was asked to be co-chair of ResearchCon a few months prior to the event, along with a physician at the CF Foundation. It was the first ever research-focused virtual event for the CF community. It was also the first time I had ever come to the realization that I could have some reputation and influence as a person with a scientific voice in the CF community to have been asked to lead such an event. In the last few years, since graduating from college with a biology

degree, my work in an environmental microbiology lab testing bacteria in water systems has had an intersection with CF, as some of the same bacteria are commonly found in lungs affected by CF. Having this background knowledge has created an interest in me and allowed me to explore and understand CF research and be involved in various research committees tasked in providing patient perspectives on research questions and methods. Providing this scientific voice to the CF community has also burgeoned from a desire to connect with others with CF, share ideas, and learn from each other to gain the best health outcomes for ourselves.

Needless to say, I excitedly agreed and spent the following months meeting with groups of doctors, researchers, and fellow community members, some of which were fellow patient scientists, picking topics and forming the content for various sessions within the conference. The event came to fruition on Rare Disease Day, February 28th. Only after the event did I reflect on the whole process from start to end, and grasped what an incredible event it was. The collaboration that took place between those of us with CF and the doctors and researchers caring for our health really makes me proud to be a part of a community that is striving to understand the science of research and having a hand in advancing our own health outcomes. Through meetings with doctors, researchers, and those interested in understanding the patient experience, I have also cultivated an interest in science communications.

From this event, which logged nearly 700 attendees, I was amazed by the intellect of the CF community in understanding biological mechanisms and familiarity with prior research, as was evidenced by the questions posed to the researchers and speakers. I recall on more than one occasion, a doctor remarked about the insightful nature of the question asked. It is clear that those of us with CF and our families are thinking critically about treatments for CF and are not content with a layman's grasp.

Our desire to gain the knowledge and tools we need to understand the science, to seek clinical trial opportunities, and to suggest new avenues of research is rapidly growing. As the lives of more people with CF reach well into adulthood, the importance we place on understanding our health and our futures leads us to seek information and the best possible care for ourselves. The rapid advancements in research in recent years parallel our interest in being active participants and collaborators in research and care. We are actively empowering ourselves to better our lives, and the value of our roles in communicating and creating the future of CF treatment with our doctors is immeasurable.

I have thought about it plenty time myself but also hearing echoing thoughts from this group of fellow CF scientists reinforces the fact that there needs to be more direct patient involvement in the development of research questions and methods. Day to day, we live with the effects of CF, and we pay attention to symptoms and treatment regimens closely. We track changes in our body's reactions and form hypotheses for

these changes. We have ideas for what could make our lives better and what questions we need answers to. We positively contribute to collaborations with research teams, but I hope that researchers could tell from this event that we are capable of more! I hope there will be more opportunities for forging collaboration and patient involvement in future research development because there are still a lot of unknowns in CF care. By joining our voices as we did in this ResearchCon and encouraging others with CF to pursue careers in science fields, to building confidence to collaborate with and ask questions of their care teams to create joint plans for optimal care, we are shaping the future of research and treatments for CF.

As someone whose life is significantly affected day to day by having advanced lung disease and the increasing hardship and burden, having the opportunities to bridge my education with my personal experience of dealing with a chronic illness and being able to share these emotions with my fellow CF scientists and project our voices to the larger CF community encouraging others to assist in their own empowerment makes me feel an immeasurable sense of accomplishment and hope our futures.

**Author 3.** As adults with CF we all have been through suffering because of this disease. We cannot change this, but we can persevere in life despite the challenges and keep the flame of what inspires us in the first place!

Perseverance for me means embracing challenges and becoming involved in interesting life experiences such as the ResearchCon meeting that we all participated in as invited speakers. ResearchCon was the first online conference organized by the CF Foundation. I would like to take everyone with me into this unique journey.

Imagine you have a rare genetic disease such as cystic fibrosis and that the only people you cannot meet in person are others with the same disease. That is due to the cross-infection that happens between people with CF. Growing up with that reality before the boom of the internet era was one of toughest things to deal with as a teenager who is constantly going through medical treatments, and would very appreciate the support of others undergoing similar experiences. I am old enough now to be able to reflect on earlier days of CF treatment, and to have experienced this sense of isolation firsthand. Being "old enough" with CF means you are an adult despite having a so called, "childhood disease."

Imagine that you grew up with CF and decided to pursue a career in science. Now imagine that after so many years one day you would meet in a video chat not only 1 but 4 others that like you had CF and were health *scientists* as well!

That sunny day after work, I entered the video chat not knowing what to expect. After seeing 4 new faces, I immediately felt I was very close to them. I felt surrounded by others who despite being different from me were also so similar. The hour we were talking for the first time felt like 5 minutes. That is the type of experience only us with this isolating disease can share!

I felt energized by these connections and brainstormed about what we had discussed on my way home. After the first talk we all agreed to generate a document telling our stories, struggles and motivations behind our career path.

I was impressed by how committed we all were about spreading knowledge related to CF. After knowing them a little more I could so easily recognize their talents. Ella and Tré who besides their job in science also are amazing writers for the CF community. James and Xan who both have so much talent with oral communication were perfect for managing the questions during our meeting with the public.

When the day of our talk to the public arrived, I was honestly very nervous. We entered the video chat and right before going live, we had a quick excited chat between us, where everyone was supportive and reassuring. We went live! We asked each other key questions about our life with CF and our work in our field. Once again, we had such a nice conversation but this time with many others watching it. I felt incredibly empowered by hearing their stories and sharing mine at the same time as we were interacting with the public!

After that experience I am hoping for many other events like this! I appreciated that this event was also made by and for the CF community. I am looking forward to connecting with more people and through these interactions we can slowly change something so important! We can change the isolating nature of this disease! Let us use the technology in our favor and connect even more, because connections mean knowledge and knowledge mean power!

*Author 4.* The ResearchCon scientists' committee was a pioneering enterprise: It was the first ever joint panel I know of comprising adults with CF who work in science.

For decades, cystic fibrosis has been a child-facing disease so, naturally, advocacy had been child-led or at least child-focused. In recent years, with improving medications, treatments, communication channels, and technology, the CF community has seen a trend towards a larger focus of adults leading the conversation. This is great for the community at large: Adults with CF perceive life differently, have nuanced opinions and critiques of existing systems, and can communicate in a more effective manner than children or parents. Adults with CF have the lived experience of growing up with CF combined with the wisdom that comes with adulthood. This blend of personal experience and emotional intelligence allows for adults with CF to better convey the best way to provide a child with CF the world they deserve. In science and medicine, just as any other long-existing infrastructure, there exist many problems.

Our panel was led and operated by all adults with CF that work in science. It was full of interesting discourse surrounding our lives with CF. We worked together to discuss what would have been valuable for us as young kids and young students. What sort of message would have been good for us to see? Our hope was to be a panel of people with widely varied experiences communication in the shared

language of CF and science. The goals were multifold: We provided commentary on how science and medicine can be improved for a niche community like CF and we also provided insight to parents of kids with CF and young people with CF about what a life fully lived can look like for people with CF.

It is critical for the people most directly affected by policies and decisions to be at the vanguard of the conversation. That includes science and medicine. Until recent years, it was not necessarily possible for a group of scientists with CF to be at the forefront—whether that was due to the lack of scientists with CF, technological restraints, or whatever else. Today, that is possible, and I firmly believe our panel set a strong precedent for how these conversations can happen and how they can make an influence.

It was also critical on a personal level. In life, we look for the people that share in our experiences. It is a beautiful testament to the world we live in that there was a panel of scientists with CF that I could talk to about my experiences. Science has a reputation of grinding its practitioners to a fine dust and that they can never accomplish enough. When you are somebody that is already having to work against a chronically ill body, the nose-to-the-grindstone culture is not well-suited. It can be difficult to discuss this with mentors or colleagues who do not truly understand what living with CF is like.

Our panel provided me with newfound friends that have the shared experiences with me that will hopefully lead to life-long friendships. I look forward to staying in contact with them and working to improve the world for a very long time.

*Author 5.* Like a number of events and connections in my life, I could not have anticipated that the small, seemingly incidental choice I made to reconnect with the CF community would lead to finding a like-minded community of scientists and friends, nor that it would lead to a handful of opportunities to enrich my own career as a scientist.

My connection to the community of people with CF and their families began in my teenage years, when I joined the email discussion list Cystic-L, as well as other discussion groups on early social media platforms, such as LiveJournal. As a young teenager, this connection to a broader community catalyzed my taking ownership of my CF care and my desire to learn more about how the disease and its treatments worked. As I entered adulthood and college, a combination of changing priorities and social circles and a few deaths left me with fewer personal connections to others with CF. However, in the last two years, I rekindled my connection to the CF community, which ultimately led me to my ResearchCon compatriots.

The first return step was joining the CF Foundation (CFF) Community Voice program, which is a network for people with CF and our families to share their input and experiences with the CFF via surveys, focus groups, and other opportunities. Through Community Voice, I learned about

positions available on the CFF Adult Advisory Council (AAC), and decided to apply. I had recently completed my master's degree, which included a computational thesis project focused on using genomic and biochemical data to better predict the regulation of CFTR (the cystic fibrosis gene) across different types of cells. At the same time, I was beginning to feel settled into my new career as a computational biologist supporting a lab using genome sequencing for rare disease diagnosis; together, these made me feel like I had "made it," to an extent, and that perhaps my perspective as both a patient and professional scientist could be valuable to the community and provide a new role for me to explore. When I was offered a position on the AAC, I was suddenly thrust into a new community of fellow CF adults.

In turn, this led to my participation in BreatheCon (a virtual event where adults with CF can interact without risk of cross-infection), where I met Xan amid a conversation of science careers, and immediately noticed their passion as a mentor and health sciences professor—so, I decided to introduce myself and explain a bit about my line of work in bioinformatics and genomics. Meanwhile, when the idea of ResearchCon was pitched to the AAC, I immediately wanted to be involved: a virtual event to connect the CF community to ongoing research could not have aligned better with my interests. From the early planning calls, I met fellow patient scientists Ella and Tré, and not long after, our CFF facilitators suggested that we work together to organize a panel discussion about science and research careers and CF. Immediately, I thought of Xan, and Ella brought Miriam on board.

Our first calls with the group were invigorating—the five of us had an immediate rapport, enhanced by our shared experiences as health scientists living with CF. We eagerly engaged in discussing each other's career paths and research areas. It was immediately apparent that our challenge would be curtailing ourselves to an hour panel discussion. In the others' career paths, I saw reflections of my own experiences: the ways my education enriched my self-knowledge about CF and vice versa; how we wrestled with doubts about the long educational and career paths encountered in academia and medicine; the strong doctor-patient relationships enabled by our comfort with biomedical jargon; and the specter (whether far-off or near) of changing health status and concomitant career changes. Despite our differing health histories, we all understood the daily struggle to balance our own self-care with our desire to advance our fields and serve our peers and students, and the challenges of navigating a professional career amid the unpredictability of a chronic illness. We learned from each other: for example, I hope that I am many years off from having to contemplate a change or reduction in my professional role as a result of cystic fibrosis, but if that occurs in the future, I also hope that I can learn from Ella's example of how to approach that transition with grace and optimism, by seizing opportunities like ResearchCon to remain connected to a community in need of my passion and skill.

Apart from inspiration and camaraderie, the connections I have made to these fellow scientists through BreatheCon and ResearchCon have been directly beneficial to my own career; this manuscript is one such opportunity. A second: Xan recently arranged for me to come to their institution as a seminar speaker and share my work with students and faculty. This was notable not only as my first seminar talk and first opportunity to represent my institution in this way, but also because this opportunity is not often extended to junior staff scientists. Many of our ResearchCon discussions helped me conceptualize and frame how I presented my professional role in genome sequencing to Xan and their university colleagues: discussing our desires to include voices from lived experience in the research world helped me create a more impactful and memorable presentation by directly using own voices to highlight the impact of the work. Finally, my new role in the CF community inspired me to seek out more professional development and leadership opportunities at work, resulting in my nomination to the Alabama Leadership Initiative, a statewide training program for emerging leaders.

In total, my ResearchCon experience became a template to help me integrate my "paid career" as a researcher and my "unpaid career" of CF management and advocacy into a more unified whole. Our discussions helped me see how the roles of patient and scientist can build off of and support each other, and I count myself lucky to have met such wonderful colleagues.

### *Shared Themes from Participant Narratives*

Our experiences as panelists clustered broadly around two interrelated themes: realizing we were not alone; and seeing the power in coming together. Each of us discovered the depth and breadth of CF patient science community in our own individual ways via collaboration for this panel. We also realized the tremendous impact potential of our collective voices and actions, informed by both our lived experiences with CF and our training as scientists.

Within these general thematic areas, we also identified multiple subthemes. These included science as a distinct mode of relating to CF community, challenging notions of what success in science means, debunking myths about what people with CF can achieve, the importance of role models in visioning the future, combining our strengths to translate a broad array of scientific work for community audiences, overlap between doctor/patient and scientist/scientist collaboration, and how research improves our ability to advocate for each other and ourselves.

Our reflections showed a consciousness of how scientific careers offer us a different route to supporting and engaging others in the CF community. Even those of us who were heavily involved in CF spaces continuously cited feeling the need to "reconnect" after receiving our scientific training and beginning our professional practice. These feelings often intensified as our scientific practice became more focused on

CF specifically, as we found ourselves connecting to CF experiences and voices in new ways.

Perhaps because of this, we also found ourselves challenging notions of what success in science means. As researchers with CF, we have had to discover for ourselves what it means to succeed in a field that often does not intuitively accommodate the dynamic physical experiences of living with a disease that affects so many different aspects of life. From safety to stamina and so much more, we are constantly learning and reflecting on what our bodies can do—and how to balance those considerations with what our minds long to contribute. Yet in most scientific careers, this is readily done with a bit of ingenuity—one of the same qualities that drew us to science in the first place.

So, we often find ourselves debunking myths about what is possible for people with CF. This phenomenon likewise appeared as a prominent theme in our individual reflections, and was mirrored in our collective discussions before and after the ResearchCon event itself. Our scientific careers have shown us the diversity of what we can achieve while living with different arrays of complications from the progression of our CF, and also the creativity we can develop in adapting our work around these challenges. Being able to dialogue with other scientists has absolutely heightened both our ability to make these adjustments and our sense of self-empowerment in doing so.

It thus proves unsurprising that our individual reflections all highlight the importance of role models for our own ability to envision an impactful future in science and in advocacy—as well as in life overall. Being able to see our own successes through the eyes of others, and share lessons learned from our tribulations within and outside of the professional research sphere, helps us think through how we ourselves will handle challenges with our CF management that impact our ability to do certain types of research tasks. Looking to peer mentors inspires creativity and also comfort in planning for our own futures in the scientific community. It also heightens the triumphant feeling of breaking down barriers when we can commune about those experiences with colleagues who truly understand those struggles firsthand.

This kinship also catalyzes professional innovation, as reflected by our shared insights about combining our powers to translate a broad array of scientific work for community audiences. As scientists both managing and studying CF, we speak in two languages that meet in the middle through our professional practice. Dialoguing actively with other scientists with CF across different disciplines helps us develop vocabulary that enhances the ability of all people living with CF to become more agentic in our own medical care and life planning. Our reflections on the ResearchCon experience also show how the audience for our translation of scientific findings into community impact prominently includes ourselves as well as others.

Interplay between doctor/patient and scientist/scientist relationships absolutely constitutes a central theme of our

insights about the ResearchCon experience. All of us were struck in one way or another by how the dialogue we share with other scientists as colleagues mirrors many facets of positive interaction between us as people with CF and our clinicians. In our own unique ways, each of us has found ways to educate our care professionals and build spaces that invite people with CF more actively into the process of reflecting on opportunities for improvement in our care. We have also found ample support for these activities in the credibility our scientific credentials afford, as well as the vocabulary our training for our own professional practice has given us.

Improved advocacy for ourselves as people managing CF thus constitutes a shared output of advocating for one another in research. We have all shared insights in our individual reflections and collective discussions about how using our scientific voices to bring experiences, preferences, insights, and values more to the forefront has shaped our own ability to care for ourselves as well. This is perhaps the most critical lesson from our experience in coming together for scientific collaboration in support of ResearchCon: that just as our personal biographies give us unique value as scientists, so too does our scientific practice offer unique value for our self-management of CF.

## Conclusions and Future Directions

We offer a few concluding thoughts, beginning with the fact that patient science remains nascent in the CF community. This likely owes partly to the simple fact that living well into adulthood with CF is a relatively new phenomenon. Many science careers in our community have ended before they truly began—much to the detriment of our collective well-being and empowerment in the present, as well as our hope for the future. Yet aging with CF is becoming more of a widespread phenomenon these days; hope is more often blossoming into action by result. And action in research and professional development requires action in community infrastructure. Our recommendations thus follow in this spirit.

The US CF science community is absolutely undergoing transformational changes in who engages with research content. People with CF are now heavily involved not only in contributing data, but also in collecting it. We are also becoming more activated in translating conceptual outputs from research to concrete changes in our clinical care and self-management. Yet the ease with which we can access research outputs remains inconsistent.

Likewise, limitations on our access to CF research spaces and resources come from different places. Content in academic journals often hides behind paywalls.<sup>21</sup> For people like our panelists, this may not present a challenge. But CF community members who do not actively practice science in their career journeys may still wish to engage with research content—and should have robust opportunities to do so.



More concerning is how CF science spaces beyond the printed page often prove inaccessible to those of us living with the disease directly.<sup>22</sup> Content for the North American Cystic Fibrosis Conference (NACFC) is not consistently livestreamed for community members to view in real time. We do often get to access recorded sessions, but this does not give us the opportunity to participate in dialogue through the event chat—to offer our perspectives on science about us. As a result of these kinds of lags in scientific spaces catching up to progress in our patient communities, far too much science about us still happens without us in a general sense.<sup>23</sup>

Our positionality statements accompanying this article also reflect specific persistent gaps in CF research and implementation of findings. Subsequent ResearchCon events have consciously centered issues of racial/ethnic/cultural<sup>24</sup> and sex/gender/sexuality justice<sup>25</sup> in science. Deep disparities persist with respect to these characteristics not only in care access<sup>26</sup> and survival outcomes<sup>27</sup> among people living with CF, but also in research participation.<sup>28</sup> Each of us has focused our own activities with the national Foundation since that initial 2019 panel on amplifying the voices and contributions of scientists and advocates from intersectionally marginalized backgrounds who are living with CF.

We thus collectively recommend creative thinking and bold innovation in scientific spaces for the community to ensure that those living with CF ourselves can participate fully—regardless of our individual professional credentials. Videoconference technology offers a vital tool for amplifying community voices robustly and consistently in CF care planning and delivery.<sup>29</sup> We also applaud advancements in the recent past that have opened up scientific education and practice discourse more to those of us who occupy both worlds.<sup>30</sup> ResearchCon offers one example, but certainly is neither an exhaustive resource nor a substitute for full inclusion in existing spaces.

Indeed, our own experiences as patient science panelists for ResearchCon showed us both the excitement and the potential in creative approaches to research engagement for people living and aging with CF. We echo calls from other scholars for the direct impact of videoconference technology innovations on care delivery and outcomes<sup>31,32</sup> and advocate for the extension of these practices to clinical research. As the landscape of adult life and professional development with CF changes rapidly in response to innovations in scientific research and evidence-based care, so too should those areas evolve swiftly in response to our experiences and perspectives. In the process, the experiences of scientist advocates with direct lived experience of both disease management and intersectional oppression should be centered in planning future directions for CF research and care alike.

### *Statement of Informed Consent*

This manuscript describes an oral history project that was not required to go through human subjects' protection review

because it did not meet the criteria for human subjects research at our respective institutions.

### *Positionality Statements*

*Author 1.* My own lived experience with CF has been shaped by intersecting forms of privilege and disadvantage. I have consistently had health insurance and full-time employment, as have my parents. I even grew up in their research laboratory at a medical school that had an affiliated CF care center. Yet my family and I still struggled to get an accurate diagnosis and proper care. Being racially white meant that doctors at least considered the possibility that I had CF and initially tested me for it at age 4; being multiethnic and of multiracial lineage may help to explain why I have only rare CFTR mutations and was not diagnosed conclusively until age 32. Likewise, my experiences as a bisexual and agender member of the CF community give me unique insight into unmet needs in science and care alike. My work as a CF researcher, educator, and advocate also draws on my neurodivergence and my history of domestic abuse and PTSD.

*Author 2.* Being of Eastern European descent, I have an uncommon class 1 CFTR mutation that has been a factor in my severe disease progression since childhood. I was diagnosed at 18 months old due to having constant pulmonary and digestive ailments causing my mother to be in the pediatrician's office with me very frequently. Being a Caucasian female, my diagnosis although was not immediate, was suspected due to my consistent symptoms. However, there is no history of CF in my family lineage and having two older healthy siblings, the possibility of CF was not among the physician's first thoughts either. Having lived with only a third of my potential overall lung capacity for the last 8 to 10 years, I have been physically limited in the level of exertion and activities in which I can partake. I have strived to not let this affect my social, romantic, and familial connections, but there are the effects of feeling left behind, different, and inadequate. Nonetheless, my life outlook is that cystic fibrosis does not define me, but it has shaped my life. Earning a bachelor's in biology, and subsequently working in a lab, allowed me to explore and partake in research design and communications with researchers working in CF. From there, participating in research committees and advisory boards providing the patient experience with the science understanding, has expanded my understanding of healthcare from the research pipeline to marketing. I now work with healthcare companies to improve their relation to community populations and make the industry more person centered, improving health outcomes in the process.

*Author 3.* I am originally from Brazil and have spent the last four years living and working in the US as a biomedical postdoctoral CF researcher. In addition, I have rare mutations of the CF gene, which happens more frequently among

community members with diverse backgrounds like mine. My main struggle growing up with CF in Brazil was accessing critical newly developed CF therapies. Unfortunately, this problem happens due to many complex reasons such as high drug prices, or market and government interest, making Brazil and other Latin American countries experience, in some cases, a decade of delay in approving new medications compared to the US.

Despite the challenges I face, I still consider myself fortunate, as due to my work in science, I could find a pathway to relocate to the US to work, allowing me to access new treatments. In addition, in early 2021, a medication that treats the underlying cause of CF was approved for one of my rare mutations. My recent access to this medication has profoundly alleviated my symptoms and harsh experiences with this disease. Nevertheless, many patients with other rare mutations are still not eligible for this type of drug, a reality that I am all too familiar with. With my dual background as a CF researcher and person with CF, I have continually worked to advocate directly with company representatives and fellow scientists about these issues. I approach people by telling my own story and highlighting the importance of expanding new medicines to all CF mutations and the need to increase access to different parts of the world. Unfortunately, not all patients with CF benefit from the same breakthrough drug simply due to a genetic lottery or a geographic location. My goal is to keep advocating to change this reality for patients and have a more just and inclusive world.

**Author 4.** I am a scientist passionate about science and its utility in building a better world for all. As a white, cis-gendered, heterosexual man, I am privileged in many ways; but, as a disabled person living in the United States, I am also disadvantaged. Yet as my disability is mostly invisible, my disadvantage—and the prejudice I face—is not as overt as somebody who might be more visibly disabled. Ultimately, these pieces of my identity dovetail together to drive my belief that the privileged should fight for a better world for everybody. This is especially true in science, which, in my opinion, has an inherent mechanism (the scientific method) capable of driving inequities into the past. I consider it a moral responsibility to use these parts of my background to learn and contribute to building the better world we all deserve.

**Author 5.** My personal history with CF has significantly impacted my identity and career as a researcher, and has been at times both typical and unusual. I was diagnosed with CF around eighteen months of age following failure to thrive symptoms. I was later found to have the homozygous F508del CFTR variant, which is the most common and well-known genetic cause of CF among white people in the US who have northern European ancestry. (Parenthetically, I think it is interesting to note that I have realized while writing this introduction how much that, as a white, cisgender bisexual man

living in the US, our society gives me a huge amount of latitude regarding how much—or how little—I want to focus my narrative on these aspects of my identity.) Today, the common and well-understood genetics underlying my CF allow me access to disease-modifying CFTR modulator therapy. Unusually, I have experienced fewer lung infections than is common in CF, leaving me with a high lung function and a lower burden of hospitalization than many of my peers. Fortunately, I have had relatively easy access to CF-specific care throughout my life: as a child, my family lived in Baltimore, Maryland, near Johns Hopkins—one of the pioneer CF centers; as an adult living in northern Alabama, I am within a two-hour drive of the University of Alabama in Birmingham, which also hosts a cutting-edge and research-focused CF center. Also fortunately, my health allows full-time employment, which gives me access to both the health insurance and the paid time off necessary to access this care with a minimum of financial strain. In the workplace, I feel able to be very open about CF, due to their health focus and a forward-thinking hiring policy of non-discrimination based on genetic information. Because I have been able to see the positive impact of having a genetic diagnosis in my own life, these experiences have motivated my current career: working in bio-informatics to support the genetic diagnosis of rare diseases. (Without the underlying knowledge linking the CF phenotype to the CFTR gene and its molecular function, few of the advancements in CF care realized during my lifetime would have been possible.) Similarly, one of my favorite roles within the CF community has been to help educate the community about research and the science of CF, because my education in science has given me an important lens to better manage my own health.


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#### References

1. Nowakowski ACH. Cystic Fibrosis Kidney Disease: 10 Tips for Clinicians. *Frontiers in medicine* 2018a;5:242.
2. Lynn J, Adamson DM. *Living Well at the End of Life. Adapting Health Care to Serious Chronic Illness in Old Age*. Rand Corp Santa Monica CA; 2003 January 1.

3. Nowakowski ACH., Sumerau JE, Mathers LAB. Building a Bright Future for Diabetes Care and Management by Writing Where It Hurts. *PLAID: People Living with And Inspired by Diabetes* 2016;2(1).
4. Dudley L, Gamble C, Preston J, et al. What difference does patient and public involvement make and what are its pathways to impact? qualitative study of patients and researchers from a cohort of randomised clinical trials. *PLoS One*. 2015 June 8;10(6):e0128817.
5. Donley AM, Sumerau JE. *Living with Diabetes in a Historically Black Community: Insights from Residents* PLAID: *People Living with and Inspired by Diabetes*; 2019:36-44.
6. Damschroder LJ, Pritts JL, Neblo MA, Kalarickal RJ, Creswell JW, Hayward RA. Patients, privacy and trust: Patients' willingness to allow researchers to access their medical records. *Soc Sci Med*. 2007 January 1;64(1):223-235.
7. Nowakowski ACH., Sumerau JE (Eds.). (2017). *Negotiating the Emotional Challenges of Conducting Deeply Personal Research in Health*. New York, NY: Routledge.
8. Nowakowski ACH. Hope is a four-letter word: riding the emotional rollercoaster of illness management. *Sociology of Health & Illness* 2016;38(6):899-915.
9. Nowakowski ACH, Sumerau JE. Reframing health and illness: a collaborative autoethnography on the experience of health and illness transformations in the life course. *Sociology of health & illness* 2019;41(4):723-739.
10. Harris WT, Kirk KL. *CFTR and Cystic Fibrosis. In Ion Channels and Transporters of Epithelia in Health and Disease*. Springer; 2016:519-552.
11. Birket SE, Rowe SM. Revealing the molecular signaling pathways of mucus stasis in cystic fibrosis. *J Clin Invest*. 2019 October 1;129(10):4089-4090.
12. Sathe MN, Freeman AJ. Gastrointestinal, pancreatic, and hepatobiliary manifestations of cystic fibrosis. *Pediatr Clin*. 2016 August 1;63(4):679-698.
13. Drumm ML, Ziady AG, Davis PB. Genetic variation and clinical heterogeneity in cystic fibrosis. *Annu Rev Pathol*. 2012 February 28;7:267-282.
14. Zielenski J. Genotype and phenotype in cystic fibrosis. *Respiration*. 2000;67(2):117-133.
15. Corriveau S, Sykes J, Stephenson AL. Cystic fibrosis survival. *Curr Opin Pulm Med*. 2018 November 1;24(6):574-578.
16. Saiman L, Siegel JD, LiPuma JJ, et al. Infection prevention and control guideline for cystic fibrosis: 2013 update. *Infect Control Hosp Epidemiol*. 2014 August;35(S1):s1-s67.
17. Borowitz D, Dulcan E, Cipriani D. Creating a virtual environment to enable two-way learning about the relevance of clinical research. *Annals of the American Thoracic Society*. 2020 March;17(3):271-273.
18. Clancy J, Benitez DS, Fajac I, Jain M, Rowe SM, Sawicki G. Applying recent advances in the science of CFTR-based therapeutics to improve outcomes in patients with cystic fibrosis. *J Cyst Fibros*. 2017 May 1;16(3):e9-e10.
19. Nowakowski ACH. Neverland: A Critical Autoethnography of Aging with Cystic Fibrosis. *The Qualitative Report* 2019;24(6): 1338-1360.
20. Nowakowski ACH. Original parts: Aging and reckoning with cystic fibrosis related kidney disease. *Patient Experience Journal* 2018b;5(1):7-10.
21. Willinsky J. *The Access Principle: The Case for Open Access to Research and Scholarship*. : MIT Press; 2006.
22. McDonagh JE, Bateman B. 'Nothing about us without us': considerations for research involving young people. *Archives of disease in childhood - Education & practice edition*. 2012 April 1;97(2):55-60.
23. Chu LF, Utengen A, Kadry B, et al. "Nothing about us without us"-patient partnership in medical conferences". *BMJ*. 2016; 354:i3883.
24. McGarry ME, , Gibb ER, , Oates GR, and Schechter MS. Left behind: the potential impact of CFTR modulators on racial and ethnic disparities in cystic fibrosis. *Paediatric Respiratory Reviews* 2021.
25. Kidd KM, Sequeira GM, Voss RV, et al. Caring for gender diverse youth with cystic fibrosis. *J Cyst Fibros*. 2020 November 1;19(6):1018-1020.
26. DiMango E, Simpson K, Menten E, Keating C, Fan W, Leu CS. Health Disparities among adults cared for at an urban cystic fibrosis program. *Orphanet J Rare Dis*. 2021 December;16(1): 332-335.
27. McGarry ME, Williams WA, McColley SA. The demographics of adverse outcomes in cystic fibrosis. *Pediatr Pulmonol*. 2019 November;54 Suppl 3:S74-S83.
28. West NE, Kazmerski TM, Taylor-Cousar JL, et al. Optimizing sexual and reproductive health across the lifespan in people with cystic fibrosis. *Pediatr Pulmonol* 2021. S89-S100.
29. Rowbotham NJ, Smith SJ, Elliott ZC, et al. Adapting the James Lind Alliance priority setting process to better support patient participation: an example from cystic fibrosis. *Research Involvement and Engagement*. 2019 December;5(1): 24-28.
30. Nowakowski ACH, Sumerau JE. Should we talk about the pain? Personalizing sociology in the medical sociology classroom. *Teaching Sociology* 2015;43(4):290-300.
31. Calthorpe RJ, Smith S, Gathercole K, Smyth AR. Using digital technology for home monitoring, adherence and self-management in cystic fibrosis: a state-of-the-art review. *Thorax*. 2020 January 1;75(1):72-77.
32. Wood J. *Telehealth in Cystic Fibrosis: Does the Integration of Modern Technology with Traditional Care Improve Health Related Outcomes?* Doctoral dissertation, Curtin University of Technology.