Turning Patient Engagement into Scientific Disruption

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The phenomenon of patients fundraising and then sponsoring scientific research has grown. These organizations range from the country's major disease foundations (such as the AMERICAN HEART ASSOCIATION and others) to much smaller patient advocacy groups (PAGs) for rare diseases. In many ways, the NATIONAL INSTITUTES OF HEALTH has embraced PAGs through its Rare Diseases Clinical Research Network. Turns out, these organizations do more than raise awareness and money—they change how research is done.

In this issue of *Healthcare Transformation*, we focus on three individuals who have turned their skill sets, their fear, even their anger, into advocacy. Each offers a model for thinking about science as a networked community inspired by engagement with patients who want to help others.

EMILY'S ENTOURAGE: Emily Kramer-Golinkoff and drug development

Emily notes that her very rare “nonsense” mutations are actually the cause of a third of all genetic diseases. That means the science that helps her can be scaled up to a wide variety of illnesses. But today, she fights for the development of drugs and targeted therapeutics with a pace not normally seen in the award-and-publication world of traditional academics.

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BRINGING SCIENCE HOME: Nicole Johnson and social support

Nicole focused her research sponsorship on living optimistically with a chronic illness, assessing the needs of patients, and understanding the role of happiness in survival rates. She created communities, online and in-person, for young adults with type 1 diabetes (T1D), who too often are “lost to care” as they move away from home.

RARE CANCER RESEARCH FOUNDATION: Mark Laabs and strategic crowd sourcing

Mark understood the danger of silos—divisions between disease-specific research and institutions. He’s focused on speeding work on rare cancers by breaking down barriers to sharing cell lines, animal models, and patient experiences. He believes the many foundations can sustain their work by “inventing one wheel and using it together,” rather than inventing the wheel for each disease.

Courtesy of Cheyenne Gil Photography.

Emily Kramer-Golinkoff: EMILY’S ENTOURAGE

Emily Kramer-Golinkoff’s lung function is 35% on her best days. Until now, exciting new therapeutics for cystic fibrosis (CF) did not target her genetic form—a group of rare “nonsense” mutations.

But here’s what she found: there isn’t a comprehensive strategic plan in action to create breakthroughs methodically in every rare disease.

That’s why she took it upon herself to find a cure. First out of desperation—to her, it is do or die. But also, in part, because once she started, she found that scientists are not computers; they are humans with big hearts who respond to personal relationships and want to help.

And those relationships became the EMILY’S ENTOURAGE model for disruption of major science.

Her story illustrates the power of patients who form scientific foundations, select advisory boards, lobby, build awareness, and fund research. They put the ultimate goal up front—to save lives, including their own. And they create new networks and ways of operating that push against institutional and organizational barriers.

“He believes the many foundations can sustain their work by ‘inventing one wheel and using it together,’ rather than inventing the wheel for each disease.”

The intensity of PAGs such as **Emily's** have indeed reshaped the nation's research agenda. In fact, at several major institutions, there are cell models derived from **Emily Kramer-Golinkoff's** own cells—proudly labeled with her name—to test novel therapies.

In response to the crisis of rare and orphan diseases, the national RARE DISEASES CLINICAL RESEARCH NETWORK has sped the development of clinical consortia in more than 200 rare diseases overcoming a series of hurdles for multi-site clinical research, and providing central data coordination at the UNIVERSITY OF SOUTH FLORIDA.¹

We talk often about disrupting the delivery of care by engaging patients.

But we do not plan enough to disrupt our science through patient-centered design thinking.

At its core, design thinking begins with empathy. *Healthcare Transformation* had the chance to interview **Emily Kramer-Golinkoff** in between her launch of a revised scientific advisory board, speaking engagements, award acceptances, and her relentless drive to accelerate science.

“Our lives imploded when we realized that the rate of progression of my disease was most likely going to exceed the rate of advances in science.”

Michael Hoad: Thank you for joining us. The challenge of being optimistic and fighting for change is a key one for us as we publish this journal. The core question that I want to ask you is about the idea of a patient setting up a foundation and sponsoring significant trials that we might otherwise expect to be funded by the NATIONAL INSTITUTES OF HEALTH. How did you come to that decision?

Emily Kramer-Golinkoff: Raising money has never been our goal. It's a means to achieve our actual goal, which is creating faster breakthroughs to treat nonsense mutations of CF like mine.

And it all started because, in 2011, I was at a point in my disease where I was seeing its progression and was getting sicker and sicker, and we were looking at the drug development pipeline, and we were seeing all of these targeted therapeutics coming. But as we looked a little deeper, we could see they were targeted to particular mutations, none of which I had. I have two copies of a rare nonsense mutation. And because we are a small community of folks with nonsense mutations of CF, there wasn't a lot of work being done for us.

1. Further reading includes: "The partnership of patient advocacy groups and clinical investigators in the rare diseases clinical research network," *Orphanet Journal of Rare Diseases*, 2016;11; which concludes: "Patient Advocacy Groups ... help ensure the feasibility and success of research protocols by assisting with study design and patient recruitment, and support training programs."

In a sense, our lives imploded when we realized that the rate of progression of my disease was most likely going to exceed the rate of advances in science. It was really an awful realization. We couldn't resign ourselves to that. We really had a "do or die" moment, where we were, like, there isn't a lot going on that we feel we can cling to.

Being totally naïve, the one thing we knew that we believed in was story-telling. We filmed a homemade video that told the story, and we used the channels we were familiar with, which were e-mail and social media, and we sent it to everyone we knew.

And we asked them to donate, but more important than that, to pass it on. And even beyond that, to believe that we could do something better. It was to our complete shock that people jumped on board. In the first week, we raised \$40,000.

I wish I could tell you we had some great vision and a grand plan. But we really didn't. We sort of dipped our toe in the water in a moment of desperation. And we kept being shocked by the response and the impact we were able to have.

In each moment of the evolution of EMILY's ENTOURAGE, we've grown beyond what we could have imagined, and then we try to create the infrastructure to support it.

But what we had was laser focus on what we wanted to achieve. And that was we wanted to fast-track research and drug development for nonsense mutations of CF.

And we quickly learned that the thing that was missing in the world wasn't just money. The biggest thing that we found missing was this strategic, coordinated, and focused approach to accelerating drug development. We want drugs to save lives for me and other people like me, who don't have time to wait for some of the other really exciting things happening in genetics. We don't have time for that.

That is what led us to create an alliance, a collaborative group of people who believe in having all stakeholders at the table. We believe that when we can get people to connect to the humanity of what we're doing, and we can gather them together in one room with collisions of ideas and technologies and data-sharing in real time, that's the way we can shatter some of the traditional silos, get people to work faster and smarter, and hopefully get breakthroughs in record time.

“The biggest thing that we found missing was this strategic, coordinated, and focused approach to accelerating drug development.”

We want to clear the way. We don't want organizational and operational barriers slowing the progress. We're not skilled enough to be the scientists who are actually doing the drug development in the labs, but what we can do is troubleshoot so that the only thing that stands between us and breakthroughs is science. And we found that a lot of the time, that's not the case.

Michael: What are the three pieces of advice you would give another patient starting out to do this?

Emily: You don't realize the value of your story until you start telling it. You think your story and your experiences are just in some small corner of the earth, and you think they are just anecdotal. But they actually carry tremendous power to touch people's hearts and to create action. And so I would encourage that even when it's uncomfortable and even when it goes against your nature, as it does for me, start telling your story.

Second, just show up—to the lab, to the companies. I think what's been shocking for me to find is how much relationships matter. Even for the most academic scientists and the companies out there that are so corporate you don't even think there are humans behind it. Make it so that when they're working on your cells, or your mutation of the disease, it is not about a digital code, it's about Emily, or Bill, or Susan, or whoever it is—a person with a story and possibility and big things to do in the world.

And then third would be that you don't have to accept the status quo. It's easy to think these big institutions have it all figured out, and it's a well-oiled machine, and that everything is being done in this super thorough, comprehensive way. When you start peeling away the layers, you realize that's not always the case. When you're fueled by desperation, it spurs incredible amounts of innovation and creativity and urgency that can be disruptive in a productive way and can do things differently.

I have a friend who always says, "Systems are perfectly designed to produce exactly what they produce." And if you're not happy with what they're producing, you have to shake things up and do things differently.

So that desperation that patients and families feel can be a huge asset if you think of it as a propeller rather than a handicap.

Michael: Aside from medicine, what helps you live with a chronic, progressive illness? What helps you get through each day, when you have to take hours of breathing treatments, etc.?

Emily: I spend three to four hours a day on treatments. I take 30 pills. I take four insulin shots. That's my healthy baseline. When I get sick, I go in the hospital and step everything up. The truth is that I do everything I can to slow the progression of my disease, and I always have.

“It's easy to think these big institutions have it all figured out, and it's a well-oiled machine.”

If I could keep up my daily routine and stay right where I currently am health-wise, that would be a dream come true. But unfortunately, that's not the case. No amount of hard work, love, or passion can effectively stop the inevitable progression of this fatal disease.

What sustains me is I have an incredible support network, without which I couldn't do any of what I do, and frankly wouldn't want to. In addition to my parents, my siblings, and my close friends of course, I have an amazing online community of incredibly close friends with CF.

I don't know if you know, but people with CF can't be near each other. We can pass dangerous bacteria only to one another. So the online community has been a real lifeline. There are people where I know some of the most intimate details of their lives, but I'll never be able to meet them in person. So those people are essential not only for support and camaraderie, but they are an incredible source of information. We crowdsource information all the time, and they have led me to treatments that have been incredibly helpful. When I get bad news or bad test results, I have an amazing doctor who I work closely with, but they are the first people I go to because they can help me crowdsource the latest information on my extremely specific issue from all around the world in minutes.

EMILY'S ENTOURAGE is also sustaining in that it's giving me hope. There are all these people, including some who I didn't know before we started this, who have rallied behind us and become our core team. They dedicate hours upon hours, not to mention heart and soul, to this endeavor and to this dream of a better future. So the ENTOURAGE, the amazing folks that make it up, have become an incredible source of motivation.

Michael: How did you pick the group's scientific advisory board?

Emily: We have an amazing chairman of our board, **Kevin Foscett** (UNIVERSITY OF PENNSYLVANIA). His daughter was a close friend of mine in high school. I heard he worked on CF in the past, but never really understood the depth of his work with CF until a couple of years ago when my parents and I just decided to knock on their door. We asked if we could just pick his brain and talk to him for just a few minutes. We like to say we haven't left.

He's allowed us to tap into this tight-knit scientific community and brought a tremendous amount of knowledge, expertise, and guidance. Since then, we've had two scientific symposia, and then we had an in-person meeting with our board. So we've gotten to be close with a lot of scientists in the field, and we've been so shocked and uplifted by how eager they are to work with us. So we thought about who would be our dream team, and everyone we asked kept saying yes. So it was really a humbling experience for us. We were methodical in how we picked—we picked a lot of people specializing in CF, nonsense mutations and nonsense-mediated decay, and also in translational research and drug development because we're not here to publish papers—we're here to get life-saving treatments to patients quickly.

Michael: So scientists are human too.

Emily: Yes. And they have big hearts. At this point, we've become really close to many of the scientists. They are brilliant when it comes to their scientific work, but their hearts are deeply invested too. And that's affirming for us. That these busy people with lots of demands on them have chosen to devote their time and talent to what we're doing, it's a really powerful recognition that we're doing promising work.

Michael: Can you talk about your gala on December 3rd?

Emily: Yes, we have an annual gala that is the launch of our annual campaign. This year, we anticipate more than 500 people will attend. There will be dinner, drinks, a silent auction, musical entertainment, and an inspiring program where I will speak, and we are so thrilled to have **Marta** and **Bob Adelson** as our honorary chairs. We started EMILY'S ENTOURAGE with a video, and so every year, we come out with a new video that tells my story and the EMILY'S ENTOURAGE story in a format that is shareable. We tell everyone at the gala that on the following Monday morning, they will receive that video embedded in an e-mail. So we ask them to be ambassadors and when they get that e-mail to pass it on their network and encourage people to keep spreading. That's how we raise our money each year. There's a unique level of devotion and energy.

Michael: Is there anything you want to add about the push that comes from patients who fight for treatments for their illnesses?

Emily: One other thing I didn't mention: we really are in a race against time. So I have late-stage disease. I have about 35% lung function when I'm at my best. We are painfully aware that is right above the threshold for end-stage disease, where the only treatment option is lung transplant, and that is a very bumpy road with dismal outcomes at best. And so all it takes is one small cold, one little blip, to be the downfall. We feel the crunch of time all the time.

“They are brilliant when it comes to their scientific work, but their hearts are deeply invested too.”

We're racing, and we're scrappy and resourceful. It's personal and it's painful and it's desperate. I think it's easy in these stories to think it's exhilarating from the outside. But when it's your life that dangles in the balance, there's this huge additional component and urgency.

There have been major breakthroughs for CF in the past few years that treat the root cause of CF rather than just the symptoms. They have given people real, tangible hope for a future they never thought they would have, but unfortunately they don't help those of us with nonsense mutations like me. It's a bittersweet experience watching my friends get their life-saver—my heart is happy for them, but desperate too: desperate to join them on the other side of possibility.

Courtesy of Eric Youngghans, USF Health.

Nicole Johnson: **BRINGING SCIENCE HOME**

In 2010, USF HEALTH in Tampa announced a \$5.6 million grant from the PATTERSON FOUNDATION of Sarasota to build a new resource for living optimistically with chronic illness.

The initiative was led by **Nicole Johnson**, DPH, diagnosed with T1D early in her college career. At the time, T1D was still seen as a limiting illness—she was told she would be unlikely to finish college, should not engage in stressful activities, and probably wouldn't survive a pregnancy.

Nicole converted her anger about the diagnosis into action. She not only finished college, but she earned a doctorate in public health. She not only engaged in stressful activities, but she won the Miss America competition in 1999, competing to reduce the stigma of T1D. And she couldn't be more proud of her daughter.

The first research sponsored by BRINGING SCIENCE HOME targeted the parents of children with T1D. She tapped into a reservoir of chronic grief by parents who felt guilty for passing on an inherited disease and had to adjust to parenting as “insulin Mom,” nagging their children to test blood glucose levels.

This focus on human communities of care and support led to studies of the emotions of living with an illness.

As she says, “In a system built on trust, I would reveal more. Everything feels judgmental—that I’ve done something wrong. If authentically cared about, I would reveal more of the actual struggles that exist. We fill up the time before we get to heavy things—questions about mortality and depression are easy to mask.”

Nicole Johnson in her words:

My role is to be the voice of the forgotten patient. Healthcare is so fragmented and difficult to understand; we need facilitators to help and advocate for their needs.

What would make it ideal is if I were a valued partner with my physician. At times, I’m left feeling like I drain time from making money with the next patient. Nothing is easy and seamless. So nothing feels safe in the day-to-day. There’s no warm blanket of support.

Patient guilt is at times debilitating. For example, I have a blood draw tomorrow, but my blood sugar went up 300, so my average is now blown, so I feel like a disappointment to the doctor and to myself.

I need a beautiful orchestration of mind, body, and spirit to deal with my chronic disease. I’m grateful for the wisdom and the advances of science. But there is incomplete science: The science about how my mind works and plays into my illness is not rich enough and not recognized by my provider.

In the future, I dream of emotional intelligence and sensitivity being seen as the pillars of understanding.

“In a system built on trust, I would reveal more. Everything feels judgmental—that I’ve done something wrong.”

Courtesy of Mark Laabs.

Mark Laabs: RARE CANCER RESEARCH FOUNDATION

Mark Laabs abruptly lost the sight in his right eye during a meeting in Beijing in 2012. After talking with four doctors, he found the Head of Ophthalmology at one of China's best medical schools—she told **Mark** it looked like ocular melanoma, but she had never seen one. **Mark** started Googling “ocular melanoma” in the taxi on the way to the airport.

Although it's the most common eye cancer, a melanoma in the eye is rare. **Mark** jumped on a plane to Philadelphia where he found one of the few major centers for cancers of the eye. Next step: WILLS EYE HOSPITAL for radiation of the tumor, and oncology follow-up at JEFFERSON HEALTH.

But as **Mark** investigated rare cancers, he found a common theme—from a cancer his father had and another that struck a friend.

Mark saw the commonalities that **Emily Kramer-Golinkoff** found. Rare disease research is plagued by a lack of resources, not a lack of scientific ideas. Researchers spend their time keeping lights on, developing generations of animal models, finding or buying cell lines, sequencing genes, and struggling to access large databases of the clinical experiences of patients.

Mark explained, “These organizations were spending their money on infrastructure, not science. And they were inventing the wheel over and over again for things we call utilities.”

Mark decided to apply his talents as a social entrepreneur and formed the RARE CANCER RESEARCH FOUNDATION in 2013. His question: what would a high functioning foundation look like?

His first step: a survey of 20 foundations asking them, first, to identify their top-three problems, and, second, what they were good at doing well. He then matched the list of problems against the list of solutions. Elegant.

He then created price-based or prize-based incentives for sharing cell lines, then animal models, then software for patients to contribute their own clinical data. The first success was developing cell lines.

“Rare disease research is plagued by a lack of resources, not a lack of scientific ideas.”

“I’m especially proud that researchers have access to cell lines contributed by patients that are now in the public domain,” **Mark** says. “Where a dozen organizations had access to those cell lines, now a couple hundred can work with them.”

“My pitch was: ‘collaborate on inventing the wheel once, and then we can use the wheel together.’”

Inventing “one wheel” wasn’t a new thought for **Mark**, who had worked in the social sector around the globe. But in each nonprofit project, he worried about sustainability and scalability. Now, he brought the same concern to organizations working on rare cancers.

“This is my personal north star: how can I have the maximum impact over my lifespan? That’s what led me to what’s called social enterprise or social business, where it starts with questions of maximizing social impact.”

“The same is true of rare disease foundations. We need to build organizations where scalability and sustainability are directly tied to the good they are trying to do in the world.”

The key to scalability: even though **Mark** is interested in ocular melanoma, by speeding organizational solutions, he accelerates work on all rare cancers. And for patients, he makes it easier to contribute their own cells and their own experiences.

As a result, we see the power of engaging patients, the partnerships with scientists, and, as **Emily** calls it, the opportunity for positive disruption. ■■■

“We need to build organizations where scalability and sustainability are directly tied to the good they are trying to do in the world.”