Tech's Answer to Curing Rare Diseases

Speaker:

Deanna Portero, Executive Director, Fibrous Dysplasia Foundation

Interviewer:

Elyse Margolis, Managing Director, W2O Group

(Transcription by RA Fisher Ink)

Margolis: Rare disease is an important and unique public health issue positioned to benefit uniquely from innovation and healthcare. By design this group of parents, advocates, doctors, and academics working in this world, defined as small, have really profoundly changed the trajectory of some of these diseases, driven simply by the fact that there was no other choice.

This community has taken pages from every book, data analytics, genomic sequencing, AI, and today, the picture is changing as we continue to see the successful merging of healthcare technology in everything, from the way we conduct and recruit for clinical trials, to personalized medicine, to disease progression mapping, and much more. Our challenge, and what we're going to be discussing today, is how do we merge tech and healthcare innovation to drive change?

This is really a tipping point moment in rare disease. As visionary leaders like Deanna Portero, who is the executive director of the Fibrous Dysplasia Foundation, and working on the front lines catalyzing change and pushing therapeutic innovators forward, are really changing what the future of these diseases will eventually look like.

Like many, Deanna was introduced to the world of rare disease by a friend whose little sister was fighting against a rare and deadly skin disease. She unfortunately passed away at the age of 24 in 2015, but it didn't take long for Deanna to develop the point of view that barriers to progress for all 7,000 rare diseases are extremely similar and in principal surmountable.

One of Deanna's first accomplishments at the foundation was the launch of their patient registry, which is a formal research study which to date has collected almost twice as much data as the largest clinical study in this disease. So welcome, Deanna.

I want to start with just a little primer on fibrous dysplasia, if you can tell us a little bit about the family journey.

Portero: Sure. So fibrous dysplasia and McCune-Albright syndrome are related rare bone diseases caused by random genetic mutation. So that means it's not mom eating sushi, it's not dad smoking, it's not second cousins that got married somewhere up your family tree. This is a disease that occurs randomly by chance somewhere post-zygote, but pre-fetus. So this mutated cell that occurs, ultimately, all of its descendants, as they become bone, as they become different glands, they wreak havoc. And one of the most important symptoms is the effect that it has on bone. We call the bone areas that are created with these mutated cells fibrous dysplasia and it's, essentially, really a type of tumor.

And who you're looking at right now, his name is Liam. He lives in a suburb of Chicago. He's going on five. And I'll tell you how McCune-Albright syndrome and fibrous dysplasia actually affect Liam. He has these fibrous dysplasia tumors all over his body, so that obviously impacts his mobility due to involvement in his hip. These tumors are not really great for being weight bearing in general. They're extremely susceptible to fracture. So Liam, actually, the longest he's ever gone in his life without a fracture is about six months. And that's actually how Sarah Healy's son, Tyler, was diagnosed, when he took a random a fall, you know, just a normal kid playing soccer and the strongest bone in his body, his femur, just broke in half, and the surgeon later compared it to a kid who'd fallen out of a two or three-story window. So very, very weak bone.

In the case of Liam, as well as a lot of patients with fibrous dysplasia, because so many patients have involvement in their skull, as this tumor grows, it can compress your optic nerve, it can warp your temporal bone, and what the results in is blindness. It can result in loss of hearing. So it's like all of the 7,000 rare diseases, these are typically very serious diseases, and while each community small—you know, 200,000 people or under—when taken together; this is an issue that effects over 30 million Americans, according to the NIH.

Margolis: So Deanna, one of the most daunting challenges in rare diseases, given the small number of Ns, is our ability to really quickly and efficiently synthesize data and collect data. What do you think are solutions there? What's the holy grail in terms of using technology to help harness the data from the corners, from where it comes?

Portero: Well, there's a huge opportunity being squandered when it comes to clinical data. So clinical data, just the data that is generated in the course of your medical care is not being appropriately leveraged for medical research. So practices collect this data. If you see more than was practitioner, you know that this doctor has this electronic medical record, this doctor has this electronic medical record. You don't have the ability to combine this. You don't have the ability to transmit this is in a machine-readable format to researchers that can help you with your disease. And this is something that bedevils not just fibrous dysplasia/rare diseases but even just common diseases. There's huge opportunities just being squandered there because that data is not being leveraged through big data analytics, through machine learning, to find, to explore patterns that predict better outcomes for patients. So I think that that is one of the biggest opportunities that is really being squandered. I think that's a step one.

The holy grail I think also integrates in the power of patient reported outcome data, like what we're collecting in our registry, as well as genomic data, which has collapsed in pricing. It's so cheap and it's just getting cheaper every year, and it's so powerful. So I think once you are incorporating all those different datasets, if you're able to link those, you finally have the kind of data set that big data tools can realize potential for kids like Liam, kids like Tyler, on the time horizon that could make a difference in their lives.

I think there's that urgency there too knowing that this data is there, this data exists. It's just siloed, unconnected, and not being analyzed, and having massive consequences in the lives of 30 million Americans and 350 million worldwide.

Margolis: Yes, yes. From the data side to the human side of technology, social media has really transformed the way that the rare disease community has come together, has been able to mobilize in very important ways globally, and from a simple standpoint, it's really helped people just find each other and then get on the right path. You know, the statistic is that it takes seven physicians and five years on average for patients to find the right physician and the right diagnosis. Talk to us a little bit about how social media and how technology has really helped this community, and more importantly, what's next? What do you the next couple of years have in store?

Portero: Sure. So I think social media probably has the biggest impact post-diagnosis. Once a patient has their diagnosis, they're able to transition from a newly diagnosed patient to an activated patient, a patient who understands who the domestic experts are, who is well informed on treatment options, who understands what research needs to happen next, who is able to advocate in the clinical environment as well as in the social sphere. I think that social media has had a dramatic impact. In fact, when we look at progress in general in the sector, there's been a big role for public policy, so things like the Orphan Drug Act that ultimately created better incentives for biopharma to serve smaller populations. But I think if you really look at the acceleration that's been happening 2000 onward, I think a lot of that can be credited to what happens on social media when change makers are able to meet each other quicker, when the passionate patient, when the John Crowley of Amicus Therapeutics, when the Preston Campbell can evangelize directly to the patients via newsletter of the Cystic Fibrosis Foundation and engage in venture philanthropy. I think that that's really where social media is making a huge difference.

Margolis: Great. And in our last couple of seconds**,** fast forward 20 years, a prediction or a hope, where do you see rare disease going, from a tech standpoint, what is your call to action?

Portero: I think one of the biggest things that we need to see there is EMRs being smarter, EMRs—and that's electronic medical records—being more active in diagnosing. I think that the potential there to increase the speed of diagnosis based on basic clinical information is enormous. I hope to see more of a role for AI in drug discovery. I think that there is an enormous opportunity there because there is a gulf between information about the assets on

the private sector side and information about the targets on the academic side and those things are not communicating and I think AI could really bridge some of those gaps there.

And I think also just the clinical trial paradigm, there's a lot of room for improvement on that end. Especially if we also think about the fact that we want to bring precision medicine to the masses. At that point, every disease, frankly, every disease become rare. When you want to be treated for your breast cancer based on your mutations based on your medical history, you need to have sightless clinical trials, you need to have wearables, you need to have telemedicine allowing for more data to be interpreted in clinical trials. So those are some hopes—and I think realistic, because all that tech exists, it's just a matter of are we applying it towards this issue. So I think there's reason to be hopeful if we move together with urgency.

Margolis: Yes.

Portero: Because there's real human tragedies when we don't.

Margolis: Indeed. Thank you Deanna.