

[00:00:00] **Stephen Calabria:** . From the Mount Sinai Health System in New York City, this is Road to Resilience, a podcast about facing adversity. I'm Stephen Calabria.

[00:00:10] Today on the show we welcome Dr. Ibrahim Elsharkawi. Dr. Elsharkawi is a medical biochemical geneticist and an assistant professor of genetics at the Icahn School of Medicine at Mount Sinai.

[00:00:21] Having begun his medical career in Boston, Dr. Elsharkhawi is helping spearhead the creation of a new clinic at Mount Sinai to research and take care of patients with Down syndrome. We're honored to have Dr. Elsharkhawy on the From the Doctor Ibrahim Elsharkawi, welcome to Road to Resilience.

[00:00:41] **Dr. Ibrahim Elsharkawi:** Thank you. I'm happy to be here.

[00:00:42] **Stephen Calabria:** Could you give us a bit of your background?

[00:00:44] **Dr. Ibrahim Elsharkawi:** Sure. I'm a medical biochemical geneticist. I'm board certified in pediatrics, medical genetics, and medical biochemical genetics, so I take care of people, primarily kids and adults, as well, with genetic conditions, metabolic conditions, and mitochondrial conditions that are genetic in origin.

[00:01:03] **Stephen Calabria:** One of the patient populations that you serve is people with Down syndrome. In medical and lay person's terms, what is down syndrome?

[00:01:13] **Dr. Ibrahim Elsharkawi:** I think to answer that, I would first need to provide a broad overview of genetics and chromosomes. So if you think of all of our DNA as a book, if the genes, the many, many different genes that we have that tell our bodies how to work in various ways, are words on a page, and the pages themselves are chromosomes that contain the genes.

[00:01:34] Typically we all have 23 pairs of chromosomes, so 46 chromosomes altogether. We inherit a chromosome from each parent. So that's why it's 23 pairs. In Down syndrome, people with Down syndrome have an extra copy of chromosome 21. So rather than the typical two copies of chromosome 21, they have three copies of chromosome 21.

[00:01:57] This is also known medically as trisomy 21. And there are about 200 to 300 genes on chromosome 21. And so that extra copy and that extra gene

dosage can lead to not just having Down syndrome, but being at increased risk for medical and neurobiological and developmental conditions that are unique to people with Down syndrome.

[00:02:20] **Stephen Calabria:** Now, before we continue, in prepping for today, you've made a point that it is Down syndrome and not Down's syndrome. What explains the discrepancy there?

[00:02:31] **Dr. Ibrahim Elsharkawi:** So, Down syndrome is named after the physician, Dr. John Langdon Down, who is a UK physician who first kind of classified features of Down syndrome and first recognized the characteristic facial features of people with Down syndrome in the 1800s.

[00:02:48] In the U. S., we don't say, for example, Down's Syndrome, we say Down Syndrome. Similarly, for Turner's Syndrome, a different kind of genetic condition, we don't say Turner's Syndrome.

[00:02:58] We say Turner syndrome. In the UK, they phrase it a little differently, but here in the US, that's the typical language we use.

[00:03:06] **Stephen Calabria:** Is there a verified genetic link?

[00:03:10] **Dr. Ibrahim Elsharkawi:** So it's an extra copy of chromosome 21, and it most often happens sporadically. It's not inherited. There are certain types, very rarely, certain types of Down syndrome known as translocations, chromosomal translocations that that may be inherited.

[00:03:28] But most often it happens sporadically. So, it is a genetic condition, but not necessarily an inherited condition.

[00:03:34] **Stephen Calabria:** So, if it is not inherited, does that mean that it's just kind of a free for all? Like it's, it could happen to anyone at any time or are there other reasons for that?

[00:03:45] **Dr. Ibrahim Elsharkawi:** Yes. So this, this happens at the very, very early stages of embryogenesis when an embryo is first forming.

[00:03:52] And it can happen to anyone for any reason. It is associated with what we call advanced maternal age. So, the older a pregnant person is, when they become pregnant, the higher the chances of having a fetus with trisomy 21 with Down syndrome.

[00:04:09] But it can certainly happen in, in young people, young mothers, and really anyone at any time.

[00:04:15] **Stephen Calabria:** Nowadays, when do expectant parents first know that their child has Down syndrome?

[00:04:21] **Dr. Ibrahim Elsharkawi:** So these days, with the advanced technologies we now have in the prenatal realm, as early as 10 weeks, we can conduct a test called non invasive prenatal testing, NIPT, and this is a blood test that examines fetal DNA, and it can test for specific kinds of chromosomal conditions.

[00:04:41] It can detect the sex of the fetus and when it comes to detecting Down syndrome, there's about a 99 percent accuracy for NIPT to detect it. You would then need more invasive testing, like what we call a CVS or an amniocentesis to confirm the diagnosis, but with NIPT about 99 percent of the time, you can detect Down syndrome.

[00:05:03] **Stephen Calabria:** How do most expectant parents react after they've been given the diagnosis?

[00:05:10] **Dr. Ibrahim Elsharkawi:** Everyone's a little different, and everyone's different, and everyone's values are different. As of 2016, the data suggested that in the U. S., about 75 percent of people who had access to prenatal testing, and remember, not everyone has access to prenatal testing, chose to terminate the pregnancy.

[00:05:28] So, we do know that most folks in the U. S, will choose to terminate a pregnancy when they find out their fetus has Down syndrome. But certainly a lot of people carry Down syndrome. Carry the term and, and go on to have a baby and a child with Down syndrome.

[00:05:43] There's no right or wrong answer, of course. It's a very hard decision to make for most folks and people make the decision that's best for them and their circumstances and their families.

[00:05:54] **Stephen Calabria:** Could you talk about the stigma attached to Down syndrome and how that's changed over time?

[00:06:00] **Dr. Ibrahim Elsharkawi:** Absolutely. So I think this is a question that would be best answered by someone with Down syndrome or their caregivers.

[00:06:06] But from my perspective, I can, I can certainly talk about how in the last in the last several years, in the last decade, I think we're seeing more and more portrayals of people with Down syndrome in popular culture, in the media.

[00:06:19] I think the discussion around into people with intellectual disabilities in general has shifted and we're talking about it more and people are listening and learning a little more when it comes to talking about people with intellectual disabilities.

[00:06:33] **Stephen Calabria:** In maintaining our resilience, we often have to rely on our social network, be it professional or otherwise. Collaboration with interdisciplinary teams is, I imagine, crucial in managing complex medical conditions like Down Syndrome.

[00:06:50] What does that kind of collaboration typically look like in your work, and what's an example of a particularly successful collaboration you can recall?

[00:06:58] **Dr. Ibrahim Elsharkawi:** Absolutely. A multidisciplinary collaborative approach is essential when providing holistic comprehensive care to people with Down syndrome.

[00:07:06] So people with Down syndrome are at increased risk for many different kinds of medical conditions, and published guidelines regarding surveillance and kind of standard of care for people that are unique to people with Down syndrome involve referrals to many different specialists, neurologists, ophthalmologists, GI doctors, heart doctors.

[00:07:27] And so, we do often refer to multiple subspecialists and we do often have to rely on close collaboration and working together for kind of head to toe holistic care of people with Down syndrome.

[00:07:38] A great example of this, I think, is, I recently evaluated a patient with Down syndrome who had moved here from another state and providing care to her meant I had to get in touch with her other previous physicians who had evaluated her at two other different states from different medical specialties and I had to collaborate with our own neurologists here who, despite being very busy, made time for me and made time for the patient.

[00:08:03] And she's been experiencing some trouble with her mental health and overall functioning over the last couple of years or so. So, in order to best

serve her after I evaluated her, I was able to get in touch with her former providers for guidance and to touch base and discuss the case a little more and discuss her past medical history.

[00:08:23] And I was also able to reach out to folks in our own Mount Sinai Neurology Division and dementia specialists. And as busy as they are, they still took the time to help and collaborate. We all put our heads together and, and we came up with a plan.

[00:08:37] And it was a wonderful example of interstate and multidisciplinary collaboration.

[00:08:42] **Stephen Calabria:** I imagine the treatment for a given patient and their families is completely different nowadays than what it used to look like. What are the biggest differences?

[00:08:53] **Dr. Ibrahim Elsharkawi:** Absolutely. So in the 1970s, the average life expectancy of someone with Down syndrome was around 30 years of age.

[00:09:00] As of 2002, it was about 60. And I think with advances in medical technology, with advances in the kind of procedures we can offer these folks, we're seeing improvements in life expectancy, in quality of life.

[00:09:14] I think we have a, as a society collectively, I think we have improved understanding, it's still not perfect, but I think we've made great improvements over the last few decades in how best to serve people with intellectual disabilities and their families.

[00:09:28] And I think all those things together have led to many, many improvements in the care of someone with Down syndrome.

[00:09:33] **Stephen Calabria:** Let's talk about those families for a moment. What are the greatest challenges they typically face in providing treatment and care?

[00:09:42] **Dr. Ibrahim Elsharkawi:** So I think there are two kind of barriers that I can think of here. So, the first one is, as much as things have improved over the last several years in the last decades, there's still a lot of stigma. There's still a lot of prejudice out there in the world.

[00:09:54] And I think navigating that for someone with Down syndrome and their families must be very challenging. And also with regards to medical

treatment as well, people with Down syndrome have unique health and psychological and neurobiological needs and guidelines, published guidelines that help clinicians, that guide clinicians when taking care of people with Down syndrome.

[00:10:15] And not everyone, not all clinicians are maybe familiar with this or up to date on the latest guidelines or the latest research. Some clinicians may not be able to readily recognize a condition in someone with Down syndrome because it may present in a unique fashion.

[00:10:32] And I think all these things are, are challenges people with Down syndrome face, but I think it would be best to ask someone with Down syndrome and their family as well.

[00:10:41] **Stephen Calabria:** We talk a lot in resilience about realistic optimism. Seeing things as they are, not as we want them to be, while putting as positive a framing on it as possible. Is endowing patients and their families with this kind of realistic optimism something that's a focus of your work?

[00:10:59] **Dr. Ibrahim Elsharkawi:** I think it places too much importance on me for me to claim that this is something that I can do, but it's certainly something I try to do. I think patients and their families, of course, deserve to hear the truth, and the realities and of complex medical conditions, of complex questions regarding life expectancy and quality of life, it's important for them to have the facts, but at the same time, there's so much gloom and doom out there when it comes to genetic conditions broadly.

[00:11:29] And it is, I do see it as my responsibility and my role as a medical geneticist to present the good as well as the bad when I discuss these rare, rare, often rare medical conditions and genetic conditions.

[00:11:42] **Stephen Calabria:** Acts of resilience often require a great deal of cognitive and emotional flexibility. As far as patients' families, is there a certain element of flexibility that they have in order to navigate the caring for their loved one?

[00:11:59] **Dr. Ibrahim Elsharkawi:** I would love to hear from caregivers directly what their experience has been. But in my humble observations and what I've noticed throughout the years, a caregiver of someone with Down syndrome has many challenges that they have to face and deal with because not only do they have to navigate.

[00:12:16] The usual medical and psychosocial conditions that can affect any child or any per any human being. They also have to navigate a world that is often prejudiced and often not set up for people who have different abilities.

[00:12:33] **Stephen Calabria:** Among both family members and just the population at large, what do you think are the greatest misconceptions surrounding Down syndrome?

[00:12:43] **Dr. Ibrahim Elsharkawi:** I think people often, who don't know anyone with Down syndrome or who are not familiar, or who haven't worked with people with intellectual disabilities in general, might be quick to dismiss someone with Down syndrome. They may have different perceptions about what someone with Down syndrome can and can't do.

[00:13:01] People with Down syndrome can be very capable and they can be highly functioning members of, society. They can be someone's friend, someone's colleague. And, if a lot of the time there's a lot of prejudice, I think, among, among folks who don't, who, who are, who, who are ignorant or who may not know someone with Down Syndrome or who may not know better.

[00:13:22] **Stephen Calabria:** We've spoken about the resilience of the patients and their families. Let's spend a moment on the resilience of the researcher. For starters, what moved you personally to pursue this as a career?

[00:13:35] **Dr. Ibrahim Elsharkawi:** So I grew up in the Middle East, and I think things are different now, but at the time where I was, growing up in the 90s and 2000s, there were cultural differences and infrastructure, medical infrastructure differences when compared to the U. S. or other countries in how people with Down syndrome were perceived.

[00:13:54] People with Down syndrome didn't necessarily get, for example, at the time, the developmental or medical treatments that they needed in order to thrive, and then cultural perceptions were pretty negative.

[00:14:05] I think that's also shifted again. Things have improved. But at the time where I was growing up, that was my experience. And then I moved to the US, and I did a two year research fellowship in the Down Syndrome Program at Massachusetts General Hospital, under Dr. Brian Scott Coe, who continues to mentor me and serve as a source of inspiration for me to this day.

[00:14:23] And I got to meet a lot of people with Down Syndrome, and their families and caregivers. And I saw, I Kind of just fell in love with this work,

and I saw how people with Down syndrome can thrive when, when they're set up for success, and, and that's kind of what motivates me and it's something I, I'd like to carry forward even as, in my career as faculty now.

[00:14:43] **Stephen Calabria:** I imagine that researching and treating Down syndrome requires a long term commitment, time, resources, even emotions. What are the greatest sources of difficulty for you in your work?

[00:14:58] **Dr. Ibrahim Elsharkawi:** I think just working with people with genetic conditions and rare disease can be challenging. It can often be emotionally taxing and it's a great privilege to do this work, but it can be sometimes emotionally taxing, I think is the biggest, the biggest part.

[00:15:15] **Stephen Calabria:** How do you then maintain your resilience and emotional well being through what are, I imagine, often difficult cases and setbacks?

[00:15:26] **Dr. Ibrahim Elsharkawi:** I have incredible coworkers. I have a wonderful team here at Mount Sinai. I have, I've been lucky to have wonderful mentors, who continue to be in my life and support me in my career to this day from other institutions.

[00:15:40] I have an incredible network of friends and, and, and that's really it. It's the people in your life, I think, that, that really make all the difference.

[00:15:47] **Stephen Calabria:** As a clinician, someone who's taking care of patients with Down syndrome, are there any folks that you see as role models as far as how you take care of your patients?

[00:15:59] **Dr. Ibrahim Elsharkawi:** I've been lucky to have many different mentors and role models throughout my life. When it comes to the world of, Down syndrome, I really look up to Dr. Brian Scott Coe, who was the first boss I ever had, and who is the director of the Down syndrome program at Massachusetts General Hospital. I've learned a tremendous amount from him, and he, to this day, continues to be a source of inspiration and support for me.

[00:16:23] He's taught me how to collaborate with and partner up with caregivers, people with Down syndrome themselves, the community, as well as how to collaborate with other specialists, in the service of people with Down syndrome in this community.

[00:16:38] He's taught me how to think and how to research the latest evidence based guidelines and how they might apply to people with Down syndrome. He's taught me how to bring a lot of humanity and heart to the work that we do.

[00:16:51] **Stephen Calabria:** Resilience is ultimately about bouncing back from adversity. Can you discuss a situation where you faced criticism or setbacks in your work and how you used it as an opportunity for growth and improvement?

[00:17:06] **Dr. Ibrahim Elsharkawi:** I think the biggest thing for me was decided, was my journey to becoming a physician in the United States. As an immigrant, it can be challenging for many reasons, some of them obvious, to become a physician in the United States.

[00:17:18] And there was a lot of uncertainty back in, in my early 20s when I was first trying to do this. And a lot of criticism and a lot of uncertainty and a lot of setbacks in general. And it's been a long journey, but now on the other side of it, I'm very grateful to be here and I don't forget how much, how hard it once was and how hard it seemed to get here.

[00:17:39] **Stephen Calabria:** You've mentioned in the past a person with whom you previously worked who served as an inspiration for you as far as the great potential that Down Syndrome patients have in leading a happy and productive life. Who was that and what specifically would you say you learned in observing and working with them?

[00:17:56] **Dr. Ibrahim Elsharkawi:** Yes, that's Ben. So he was my coworker. He, he has Down Syndrome and he was my coworker in the Down syndrome program at Mass General Hospital when I worked there. So I first met Ben when he worked in the program when I was a visiting medical student in the division of Medical Genetics at MGH in 2013.

[00:18:12] And then I worked with him as a, as my coworker in, in 2000, between 2015 to 2017. And I got to work with him. I had the privilege of working with him again last year during my fellowship in Boston. And Ben really taught me that before, before I met Ben, it really didn't occur to me that someone with Down syndrome could be my coworker, my colleague.

[00:18:29] But Ben, Ben certainly changed that. Ben does his job excellently. He became my friend. Um, he was someone I could rely on at work. He's been there for, for over a decade at this point and I think that's inspiring.

[00:18:43] **Stephen Calabria:** Now we're launching, or rather you're launching a new Down Syndrome facility here at Mount Sinai that's unique in many ways. Could you tell us about it?

[00:18:53] **Dr. Ibrahim Elsharkawi:** Sure. It's a, it's a Down Syndrome program. So it's a, it's a clinic where I, a medical geneticist and my wonderful team, uh, Rachel Kaplan, a nurse practitioner I work with, and Michelle Torres is our program coordinator. We'll be seeing patients with Down syndrome in clinic, we'll be taking the time to make sure, we'll be taking a full medical history, doing an exam, making sure that the person with Down syndrome is up to date on all the recommended special guidelines that are published and available for people with Down syndrome, studies have shown, previous studies have shown that people with Down syndrome when under the care of usual primary care are often up to date on only 10 to 70 percent of these guidelines.

[00:19:31] So we'll be taking the time to make sure all those guidelines are being met. And also, sometimes medical conditions that are common can look different in the typical population than they can look when they present in someone with Down syndrome. And uh, so

[00:19:44] **Stephen Calabria:** What's an example of that?

[00:19:46] **Dr. Ibrahim Elsharkawi:** An example of that is obstructive sleep apnea, which, in the typical population, presents with daytime fatigue, disrupted sleeping, low energy levels.

[00:19:56] In someone with Down syndrome it may present like that, but it also may present with decreased functioning, decreased mental health, and a clinic like mine would aim to screen for and detect these conditions as early as possible because many of them are treatable and in fact preventable when you catch them or when you know to look for them.

[00:20:14] **Stephen Calabria:** And so, this facility is going to be more observant of these sorts of problems than would a facility that just treats, you know, your average person off the street.

[00:20:25] **Dr. Ibrahim Elsharkawi:** Absolutely. I will say I would encourage folks to use people first language. So I would say patient with Down syndrome rather than Down syndrome patient. And yes, that's exactly one of the goals of this program, this clinic.

[00:20:38] We'd also aim to provide holistic, kind of head to toe, care for someone with Down syndrome while working with their usual primary care physician as well, referring to any specialists that they may need, GI, neurology, and also conducting broad developmental assessments and making sure they're these folks and their families are plugged in with all the resources that are indicated.

[00:21:00] **Stephen Calabria:** How have pop culture references relating to Down syndrome progressed over time and changed over time?

[00:21:08] **Dr. Ibrahim Elsharkawi:** I think we're increasingly seeing more and more portrayals of people with Down syndrome in the media and in pop culture. A show I've really enjoyed watching lately is called Down for Love. It's a dating show that pairs adults with Down syndrome who are looking for love, who are looking for romance. It's really funny. It's really heartwarming. It's really beautiful to see.

[00:21:31] I'd like to share a couple of the ways the people with Down syndrome have described themselves or described having Down syndrome in that show, I think is really powerful. Here are some quotes from that show. Down syndrome allows you to learn differently from other people.

[00:21:48] To me, having Down syndrome is kind of normal. I don't feel like I have a disability. And then some of the things they've said, I think that show of the things they've said when talking about looking for love and looking for romance, I think is, is very relatable to us all. So here are some quotes from that as well.

[00:22:07] Having a relationship with Ellie makes me see a new life with bright colors. Every time I see her, I tremble. Every time that she speaks or smiles, somehow it penetrates every cell of my body and I'm completely in love with her. How poetic is that?

[00:22:21] I am cute and bubbly and smart and sassy. I think any boy would be lucky to date me. This one I think is particularly relatable- if the date goes wrong, I might cry.

[00:22:31] **Stephen Calabria:** I think we can all relate to that one.

[00:22:33] **Dr. Ibrahim Elsharkawi:** Yeah. I want a boyfriend who has a lot of dreams and wants a big life. I hope the boy I meet is kind and honest. Girls, same. I also am looking for a guy who's kind and honest.

[00:22:46] **Stephen Calabria:** What do you wish more people knew about Down syndrome?

[00:22:50] **Dr. Ibrahim Elsharkawi:** I think everyone should have the privilege of knowing and loving someone with Down syndrome. I would love for people to know that people with Down syndrome are people like any of us, they're people with their own hopes and dreams and, and variable human qualities that we all have.

[00:23:04] They can be co workers, they can be friends, they can be social, they can have hobbies. And I think also, of course, it would be important to ask someone with Down syndrome what they would want as well.

[00:23:15] **Stephen Calabria:** Well, that's it for my questions. Was there anything else you wanted to say?

[00:23:18] **Dr. Ibrahim Elsharkawi:** No. Thank you for having me and giving me the opportunity to talk about this.

[00:23:23] **Stephen Calabria:** It's our pleasure.

[00:23:24] Thanks to Dr. Ibrahim Elshakawi for appearing on today's show. That's all for this episode of Road to Resilience. If you enjoyed it, please rate, review, and subscribe to our podcast on your favorite podcast platform.

[00:23:36] Road to Resilience is a production of the Mount Sinai Health System. It's produced by me, Stephen Calabria, and our executive producer, Lucia Lee. From all of us here at Mount Sinai, thanks for listening, and we'll catch you next time.