**MedTech Chat Podcast**

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**Respondent: Charles Steward**

**Welcome to MedTech Chat where we discover the latest healthcare tools, device technology, as well as research approaches.  We’ll be talking to designers, insight professionals, and other executives to better understand how MedTech is helping patients and those caring for them now and in the future.**

**Today I am very excited to be talking to Dr. Charles Steward. He has spent around 28 years working with the human genome on the Wellcome Genome Campus in Cambridge, United Kingdom. He spent the first 22 years at the Wellcome Sanger Institute, which is where he did his PhD. He is currently the Patient Advocacy and Engagement Lead at Congenica, a digital health company that uses rapid genome analysis to investigate the genomic basis of rare diseases, also based on the Wellcome Genome Campus. His particular interest is in the genetic causes of developmental and epileptic encephalopathies. Charles is the father of two children with severe neurological disorders who have been through numerous UK-based genomic studies. Although, nothing has been found yet to explain their challenges. He is passionate about how patient advocacy and engagement can drive positive change for people, families and caregivers affected by rare genetic disorders. Thanks for joining me today, Charles.**

It is great to be here Tom, thank you so much for inviting me.

**Maybe you can tell us the kind of work you have been up to.**

Yes, sure. I want to say thanks for the introduction. I am essentially a genome scientist by training. As you’ve said I have been working on the human genome for 28 years and I have been doing that on the same campus near Cambridge in United Kingdom. Some people might say it’s a lack of ambition that I have never moved away. But I would say that its absolutely not. It’s the most amazing work that we do and it is on the most beautiful campus. So, if you ever get a chance to visit Cambridge you should pop along to the campus and see the amazing work that we do.

**Yes, I might take you up on that.**

Yes, you would be very welcome. Yes, I started in 1994, really as a lab technician. I studied for my degree, while working there. And gradually sort of worked my way up. Working in different areas in the human genome, putting the human genome together at first, so building the actual sequence of the human genome. And then eventually I moved into bioinformatics - the area of biology the uses computers to try and understand genome sequences. And that’s where I began to look specifically at genes on the human genome. At this time, I was also doing my PhD. I did my PhD at the Sanger Institute. And interesting actually - I should just say that the Wellcome Sanger Institute is most famous for generating around 1/3rd of the human genome. Which is the largest proportion of any institute around the world. We are pretty proud of that, I would say. And it was 2016 that I moved to Congenica. And Congenica is based on the Wellcome Genome Campus. And it’s a software company that actually spun out of a research project at the Sanger Institute called the Deciphering and Developmental Disorders study which used genome sequencing to investigate rare developmental disorders in children and we are essentially a spin out of that. And the software we produce is used in the NHS, our health care provider in England, as part of a big study called the 100,000 Genome Project to help clinicians look at patient’s genomes and to understand why they might - if they have a genomic disorder or not. We work with various other groups as well. But that in essence is what we do. We produce some software which clinicians and researchers use to investigate the genomic bases of rare disease very quickly because of course that’s the key thing. Often with these rare disorders you need to get a diagnosis as quickly as possible because in some circumstances, particularly things like neurological or SMA. If you are intervening quickly, you can stop progression of irreversible disorders. Anyway, the reason I went to Congenica was through some sort of pretty bad family circumstances, as I would say. My daughter, who was born in 2012, was born very premature and after around six to eight months of age she developed really a nasty type of epilepsy that caused massive developmental regression. Something called West Syndrome or infantile spams. She also has severe cerebral palsy. And because of that, she was enrolled into genome studies in the UK including The 100,000 Genome Project. It’s the worlds leading large scale national genome project and was instigated by a guy called David Cameron, who was our ex-prime minister few years ago. And he had a son, who had severe cerebral palsy and a very nasty type of epilepsy as well called Ohtahara Syndrome. And sadly, he died at the age of six and because of that pretty much The 100,000 Genomes is because of his son. It was set up because of his son. Our daughter was put through this study, to see if we can find anything. Nothing could be found and then my son was born, he was even more premature than my daughter. And he too has cerebral palsy. Very severe. He cannot talk, he cannot stand. And he had his genome sequenced as well. We have put them through various different genome studies but not found anything. Yet, I am convinced we will find something soon. Anyway, the reason obviously I am with Congenica was through my experiences of having two severely disabled children and it meant I could use my background in human genome to work more closely with patients, clinicians and researchers to really bring the patient to the center of this new emerging or very exciting technology that is whole genome sequencing the clinic. So that is what I do, I am the patient advocacy engagement lead at Congenica and I put the patient at the center of everything we do and I am the voice of the patient internally to the rest of the members of staff at Congenica. But also, I represent the patients externally and I am an advocate for them. And I also engage with them to either offer advice on scientific things or advocacy work and also speak a lot with clinicians and researchers. It is amazing when you speak to researchers, the number of researchers, who will sit in the basement of their, I mean this is a massive generalization, but please forgive me just for this, imagine your researcher is sitting in the basement in their lab with their favorite gene or favorite disorder and their whole life’s work. But often that would be completely siloed from patient, yes, it is a fascinating disease in the sense a lot of these diseases are fascinating they are terrible but fascinating. But if you put a patient, someone with that disorder in front of them, suddenly everything changes and then they realize. It’s not just an amazing project. It is actually sort of life changing. And I have seen that firsthand myself. You are getting in front of people who have worked, researched epilepsy and told them stories about what we have been through an suddenly everything changes. Comments like “God, I wish you had been there when I was doing my PhD, it would’ve put it into perspective so much better.

**I really appreciate you sharing your background, your experience and your knowledge. This is such important work that you are doing and it’s interesting how your professional experience as well as your personal experience have really come together, and you can basically use the challenges that you had in life to now move the world forward and to help others. I really appreciate that. In matter of fact, as we talked about on the other call at my company we have a series called “the patient point of view” where we have people come in and tell us about their journey. Its only for our internal company to kind of refocus as you kind of said to get away from our day to day task of the work were doing and realize why we are doing this? It is to help our clients help their patients have better lives. This is the part of the work you are doing I really appreciate that.**

**One question I have for you is, why do you think getting a genomic diagnosis can help for people first of all, but also secondly for care givers, even if there is no current therapy avenues available.**

That’s a really important question, I think. Because- and I have heard this myself, you go to a doctor and say “can you look at my genome?” What’s the point you can’t do anything about it. Why do you want to know, just manages symptoms? That is one point of view, but I would suggest that I have a different point of view. It can change everything. Let’s just assume you have a type of disorder that, if you know what the genetic cause is, and you can get – there are treatments, simple treatments and you can get in there really quickly before any disease progresses or any irreversible symptoms happen. And I am talking about something like sort of Menkes syndrome, or SMA or even there is certain types of leukodystrophy which are horrible progressive disorders. If you know the genetic basis for your disorder, it is possible to get to know really quickly, because some of these, disorders the damage will be done before you know what’s going on. And if you get in quickly, you can essentially live a pretty good life with some of these things. That’s the perfect example, isn’t it? You sequenced at birth, you find there is something wrong, you can then intervene with treatments. And even if you do what we call a newborn screen, it may be possible to pick up indicators that if you live your life in a certain way you will be more susceptible to things like heart disease or cancer. These are things good to know you can intervene on these sorts of things. But let’s say that you have a child who has a disorder that comes back. A genetic disorder and there isn’t anything that you can do, currently. First of all you can work out if your child – if your chances of having another child having the same disorder, you can work out what this chances could be, because if your child has a de novo mutation, and what I mean by that is, a mutation that occurs in the child after conception between Mom and Dad, chances are that’s not going to happen again. Whereas, if you look in Mom and Dad and see that you have inherited one copy of the gene from Mom one from Dad. Chances are that could be a one in four chance of that happen again or what have you, so it can have an impact on family planning. Also, it can bring around psychological closure, I think for parents. The idea parents have guilt behind a disabled child, and they think it’s something they might have done during pregnancy and if you realize it is actually a genetic disorder. Then that can bring psychological closer. It can also inform you on how your disease might progress. If there are’nt treatments available, you can still put support in place. You can be best equipped for how things might adapt. I think, what I have seen in the patient community is that it allows you to reach out to other people via Facebook or Instagram or any of these other social media channels to find other people around the world, who have the same disorder and you can join together and gain strength from each other. What were seen is were seeing patient groups really driving research, driving the agenda. Getting together, raising money, generating model organisms to investigate that disorder. Some of them are actually now pretty close to treatments working with pharma. I think, these are the incredible things that happen and patient advocates are the most amazing people you ever meet. Nothing inspires you really as much as you having a severe disease or child having a disorder. There are other reasons, but I would say when you’re dealing with a rare disease by its very name means it is very unlikely to find other people. Now 20 years ago, you would have sat at home leading a miserable life on your own without being able to get in contact with anyone but now via the internet you can do a quick google search and you can find other people who also have the genetic diagnosis with the same disorder. And you can really do great things.

**Those are all great points why you definitely want go through an get that genomic diagnosis even if you might not have a current therapy available. So that’s a great point, thank you. I am wondering from your perspective as a scientist and also as a patient advocate, have you seen engagement with patients and family’s improving the quality and relevance of the research?**

Yes absolutely, this goes back to what I just said really. There are some extraordinary examples of patient advocates. I can mention a couple because I know who they are. There is Luke Rosen, from KIF1A. He has a child with a mutation in the gene KIF1A. and Jeff D’Angelo, who set up an advocacy group called CHMP1. He has got a child with a mutation CHAMP1. And these people they are doing extraordinary things. These people are not scientists, they came from completely different careers and suddenly now they are the world leading – they are the world leaders in their disorder. I say this quite often, if you think about pediatric neurologist and the number of disorders they have to know intimately hundreds and hundreds and hundreds of them, there is no way they can know them in as much depth, that each one, it is just impossible for them to know in that much depth. In some respect they going to have to rely on patient groups, who actually understand their disorders more. And it’s interesting where you also get groups of patients together and they start chatting. Its sometimes the most extraordinary things happen they discover what they thought was just a little mild habit at six o’clock in the evening, the other kids are doing it as well. This might be an indicator that there is some other part of this disorder is happening that nobody is really knowing about and by these advocates chatting together they build a better picture of what it is like to live with their disease than the clinician who sees them for 15 minutes or so, he is just not going to know.

**Thank you, yes that is important to know. It is interesting what we do patient journey worker or other kind of research who we do pick up on some of these nuances that your standard physician wouldn’t notice because they don’t have time. I am curious, I know that recently you moved, I believe, I was also wondering is it difficult to balance all the demands of your career and home wise and remain positive and obviously passionate about rare diseases?**

Yes, as I mentioned, I have been working on the same campus for 28 years. Now, that’s not strictly true. Because I am actually now talking to you from the Black Forest in Germany. And we moved here three months ago, because we have such a demand for the children because they can’t stand. We ran out of money and ran out of land at home in England. That was it. We were done. We had nowhere else to build for my little boy. Everything we did we adapted for my little girl and then my little boy came along, and we just had no means of building something or having a home that was accessible for us all. My wife is from Germany from the Black Forest. We decided to sell up. Come over here to Germany to because houses are a lot cheaper, and you get a lot more land and we got all her family around to help us. And we going to have a - we hope we going to have a much easier time over here, because as the children get older, they get heavier because as the children get older I get older and my wife gets older and of course we get weaker. It means that we are going to have – I hope this means that we are going to have an easier life, because its 24 hours a day this thing, this struggle that we are going through. And I think it’s important to have some sort of down time. And at least have the ability to sort of escape for 15, 20 minutes and have a bit of your own time and because you are reminded of the struggles that the children have when you get them up in the morning, when you feed them when you are taking them to the loo, you dress them whatever, put them in their wheelchairs. And it can be relentless. Obviously I am passionate about rare disease because it’s a family thing. I don’t think there is any way I would not remain positive about them but at the same time it’s important that you can have a bit of live that is away from all of that. And I am hoping that a new home will allow a bit of that. will allow easy access for the children to do whatever is they need.

**That sounds like a wise move to me and it brings up images of, I think, like fairytale castle and beautiful scenery. Hopefully it will turn into a great fairy tale ending.**

Thank you.

**Wondering, what excited you about precision medicine in the next say five to ten years?**

I think we have touched on some of the areas we, where precision medicine is already making a difference. But I think as technology, as we get - as it becomes quicker to sequence the genome, as it become cheaper to sequence the genome. As we learn more about what the genome is, it will become standard practice in healthcare. I envisage a time when you go along to your GP and they take a blood sample and sequence your gene and return it in matter of hours or whatever. Let’s not forget people like Dr. Stephen Kingsmore at Rady Children’s Hospital who holds the world record for blood to report from genome sequencing in 19.5 hours. This is already a thing. Admittedly, I would suggest this is more of this is what we can do rather than this is what we can do all the time. But it proves to people as a doctor you can see a sick child in intensive care, take a blood sample sequence it, gets the results back in time for his next ward round in the next day where in theory he would then be able to administer a specific treatment based upon the underlying genome. That’s the sort of thing I would like to see. I think we will also see more understanding of what the genome is actually involved with, because at the moment, if you are looking at things like genes in the genome. They account for - depends how you sort of classify them as such, but they account for sort of one, two, three, four percent of the genome. There is lots of the genome we are not sure what it does yet. And nature doesn’t tend to waste things. I suspect those other regions are very important for something. Whether that’s for how the DNA maintain its shape or its changes it shape or perhaps how those bits of the genome are involved in turning genes on and off. Because we know that not all genes are expressed in all tissues. And that we know that certain genes are turned on and off during developmental stages. Gene expressed in fetus brain may not be expressed in an elderly person. I suggest some sort of examples. Now, I think these are the sort of things we are going to begin to learn more about and really dig much deeper into the properties of the human genome.

**Excellent, sounds like we have a lot of work yet to do but it also seems like there is some low hanging fruit that we can really make a difference by implementing. Sounds like an exciting time to be alive right now. Now, enough of that work for a minute. What do you like to do in your free time to relax and forget about work? I know you need to have some of those breaks. What do you do?**

I haven’t been able to do this in Germany, sadly yet, but I suppose I was trained as a musician as a young age I played the violin, the piano and did a lot of singing. I did a lot of singing in choirs in Cambridge and music is the thing I love. But singing in choirs is my favorite thing in the world I would say. As a leisure activity it is my favorite thing in the world because not only is making music wonderful but it is also when I am singing I can only think about the music I am making. All of those stressors and worries in the brain they just go for a little while. Because all I can concentrate on is making music and it’s wonderful. Afterwards, I feel kind of relaxed.

**That’s great. Yes, I think music is an excellent way to change our mood and to reset. I know often I be humming to myself when I am stressed, some song typically is a holiday tune that I learned in choir, and I find myself singing it. It makes me feel better and I am like “why am I singing this song?” Its relaxing me and getting me to a better place. I appreciate that. Yes, as we are rapping up here curious what historical figure or fictional character do you relate to or are inspired by?**

I don’t know if you have heard of them but both of them have had a massive impact in my life. One in music one in science. And the guy in music is a guy called Sir Stephen Cleobury [ph], who was my choir master at Kings College in Cambridge. And he gave me the most extraordinary teaching experience when I was at the age ten to 13. And the sort of training has helped me in life outside of just singing, I suspect. Very sadly he died of cancer a couple years ago which it just goes to show that these cancer, rare diseases they target anyone. Your just unlucky. It is really, very sad. The second person is a guy called Sir John Sulston. He was the guy who led the British initiative behind the Human Genome Project. He was based at Sanger. I knew him very well. And John was an awesome guy. He used to – I told you Did my degree while I was at Sanger - he used to sit with me in the evening and go through my chemistry homework with me, used to help me with it. He won the Nobel Prize for his work with the microscopic worm C. elegans. But he was humble enough to sit there and explain to me very basic concepts, I now realize, in chemistry and biology. And again, he very sadly, he died recently as well, from cancer. And it just it makes you realize; however brilliant you are, life still has these difficulties that are put in front of us. Those are the two inspiring people in my life.

**That’s great to hear that you had some great mentors that inspired you and as you say it is kind of sad how sometimes life brings us down and you never know what is around the corner for us. But for those of us that are able to be inspired despite of that and to march on passionately and help others like yourself, I think it is important that we are able to do that. Thanks for sharing here your story and your experience here. I am curious as we are wrapping up what places should people go to look for more information about you, obviously I can post them on my site but anything in particular you want people to know to go find you?**

Yes, of course on LinkedIn, that would be the best place to go. I am very open to people contacting me – as they are not trying to sell me insurance or mortgages which I get a few of that in the past few weeks. And obviously you can read about the work I do on the Congenica patient advocacy web page. But yes sure, contact me on LinkedIn should be easy to find. Charles Steward working at Congenica the Wellcome Genome Campus.

**Excellent. Thank you, I have to make sure I post this on my site. And thank you so much for being a guest today. It was a real pleasure, I appreciate all the work you are doing and you seem like not only you are a great scientist a great advocate as well as a great parent. I mean I just applaud you and thank you for joining us today.**

Thank you so much for having me.

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