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Q1 Following your undergraduate degree, you completed an MSc focusing on microbiology and genetics, which shaped the rest of your career. What inspired this choice?

At the time, in India, there was not a concentrated effort on specialisation available in genetics. My interest in genetics really stems back to sixth grade, when I first saw DNA extraction on an educational video. My family background is very much in medicine, education, and entrepreneurship. I think I got the best of both worlds, and that really stemmed my interest in genetics.

I did my Master's degree at the University Department of Chemical Technology in Mumbai, which is a premier institution in India. There, I had the right mentors who gave me that direction to go into genetics. Genetics has always fascinated me, and I consider myself very fortunate to have been able to finally find my way to this discipline.

Q2 Your research interests include newborn screening. How did your passion for this field develop?

I think it really goes back to when I finished my Master's degree in 1992 and I enrolled for my PhD, which I ended up completing at the University of Auckland, in New Zealand. New Zealand was one of the early countries that started doing newborn screening, and Diane Webster, who is considered one of the leaders in the world of newborn screening, was there at that time. I was able to benefit from this exposure. However, my research interest has been primarily in neuromuscular disorders.

My PhD advisor, Don Love, had just returned from England having completed their postdoc with Kay Davies, and identified the utrophin gene, which at that time was a novel discovery. This piqued my interest in Duchenne muscular dystrophy. My research career has always been

in the area of neuromuscular disorders, but at the same time as working in the hospital, I had exposure to newborn screening, and, specifically, to some of the early disorders that were screened in newborns, such as phenylketonuria. After leaving New Zealand, following my PhD, I went on to do my postdoctoral fellowship at Baylor College of Medicine in Houston, Texas, USA, where I had a much bigger exposure to genetics, genomics, and newborn screening.

Q3 What inspired you to make the switch from academia to industry in 2016?

It is a continuation of my journey after completing postdoctoral training at Baylor College of Medicine. I was recruited by Emory University School of Medicine in Atlanta, Georgia, USA, where I divided my time between running my research lab, of which I was a funded investigator, and running the Emory genetics laboratory. The Emory genetics lab was one of the premier genetic testing labs in the USA, and, at that time, we also received samples from all over the world. It was very much a global exercise, but we did not have labs in the rest of the world.

For a long time, genetics, and especially genetic testing, has stayed in the world of academia. I think that Revvity, Inc. (Waltham, Massachusetts, USA; previously affiliated with PerkinElmer), is one of the very few companies that really took on the initiative in the rare disorders screening space (newborn screening), and in getting the relevant tools to perform the screenings through the U.S. Food and Drug Administration (FDA) approval processes. Obviously, I was very aware of what Revvity was doing at that time. At Emory University, we were also the confirmatory testing lab for any positive results detected by newborn screening. We had our clinics downstairs, where we used to see babies and paediatric patients come through. The time was right to take all the work that we were doing in academia into the industry.

Revvity also approached me at the same time, as they had a lot of interest in going beyond newborn screening and step into the world of next-generation sequencing. If you look back now, after 7 years, it's really interesting to see where we are today; for example, we also have cell and gene therapy in our portfolio. So, my transition from academia to industry was very timely, and I think I have learnt a lot. In academia, where we use products from different companies, we really don't know what goes into developing a solution; the tremendous amount of work, expertise, quality control, and regulatory aspects. I consider myself lucky that I have been able to see how Revvity develops a product, and how it comes to market. That's a huge learning point for me.

Q4 You are currently Senior Vice President and Chief Scientific Officer at Revvity, formerly affiliated with PerkinElmer. You also head Revvity Omics, a network of laboratories offering services across the globe. How important is access to advanced genomic technologies, in your opinion?

In the last two decades, and especially the last decade, it feels like we have switched from old technologies to new technologies overnight. As someone who runs clinical labs, and has run academic research labs, I understand first-hand that the access to the key technologies that enable the labs' work, and how these technologies come to market, is very important. There are two ways to look at it: firstly, from the diagnostics perspective; at Emory, as I mentioned, we used to get samples from all over the world, and now I'm running a global network of laboratories. At Revvity Omics we stress our 'in country for country' commitment, which refers to the importance of understanding regional needs, and making these technologies accessible in each region or country.

> **"We are in a very exciting phase of genomic and omics technologies."**

Inside Revvity, there is an engine where diagnostics and life sciences come together, and that line is always blurred. We are now thinking about the entire pipeline, from the individual, whether it be preventative testing or symptomatic testing, all the way to gene therapies, and the entire lifecycle of the different products that need to come to market, with the omic-based technologies and innovation behind it. We keep that engine very alive and active by looking at new technologies and debate them to understand

what is needed. I am fortunate to have the opportunity to bring that knowledge to the company because I have come from an academic setting.

We are in a very exciting phase of genomic and omics technologies. These technologies are going to not only shorten the time to diagnosis, but also facilitate development and monitoring of new therapeutics, such as biomarkers. As these assays are developed and the technologies evolve, we must also remain alert and adapt accordingly.

Q5 Revvity was the first clinical laboratory to offer whole genome sequencing commercially. How was this new service received initially, and how does it impact patients today?

Around the time I came to Revvity, a revolution of whole genome sequencing had begun, and genome sequencing became relatively affordable. There were also some National Institutes of Health (NIH)-funded projects at that time, such as the BabySeq Project, which is quite well known, and Genomics England had also kicked off its 100,000 Genomes Project. There were also a lot of smaller companies, core laboratories, doing research-based whole genome sequencing.

There was no single clinical laboratory that could offer whole genome sequencing on a clinical basis for healthy newborns, so we launched two programmes: the healthy newborn genome sequencing programme, and a clinical genome sequencing programme in our clinical laboratory in the USA. At Revvity, we are very fortunate that we have our blood cord tissue storage channel, ViaCord, through which we launched whole genome sequencing for healthy newborns. In the second or third trimester of their pregnancies, families using ViaCord services were introduced to the possibility of having their babies' samples tested via whole genome sequencing at birth.

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This was a novel approach, so a lot of processes had to be put in place, such as institutional review board approval, consent forms, proper information on how each gene causes disease, and why we look at certain genes and not others. From a testing standpoint, giving families the opportunity to choose whether they wanted to use our whole genome sequencing services or not, instead of having to enrol in a project, makes this a real-world experience. Hopefully the outcome of these projects is going to show that newborn genome sequencing is useful and necessary. It is an ecosystem, because whole genome sequencing is not just one test; it has so many components. We are also doing clinical whole genome sequencing in certain cases when samples of sick babies or children come to us.

Q6 You have recently launched ultrarapid genomic testing (urWGS) to aid with the treatment of critically ill patients in the neonatal and paediatric intensive care setting. Can you outline how this technology could revolutionise healthcare?

This is truly a turning point for paediatric patients as the earlier you detect a condition, the earlier you can intervene, which is the primary goal. When you have a baby or a child in the intensive care unit, you want a result as fast as you can, so that you can intervene early, and prevent any further damage. This is where ultrarapid genome sequencing comes in. We've been doing this for a while now; the turnaround time is about 5 days, and we can have turnaround results in 48 hours. It is also important to take into consideration that doing ultrarapid genome sequencing in a clinical setting and in a commercial laboratory is different to doing it in an academic setting where the turnaround can be 6 hours, but this would probably cost around 100,000 USD. You have to blend both the viability of running such a test with the accessibility and timely delivery of results, so that physicians can intervene. With the ultrarapid also comes the delivery of a written report within that specified time, and then the follow-up with a genetic counsellor who calls out the result and discusses it with the physician. This is very important, as there are over 7,000 rare disorders and you cannot expect this information to be at a doctor's fingertips. It is our responsibility to give them that information, so that they can act on it.

Q7 As both a medical geneticist and American Board of Medical Genetics and Genomics (ABMGG) certified diplomat in clinical molecular genetics, how do you approach your role at Revvity?

The teams at Revvity are high-functioning, with experts in different areas of technology, such as mass spectrometry, sequencing, and cell and gene therapy. What I am able to bring to the table as a trained medical geneticist is the patient perspective. On a day-to-day basis, in clinical cases, I look at data. We know that a certain gene has a function in the body, and we need to take that further to what is next. I work closely with the team on improving the products we have and the technologies we are looking at, which is an important part of what we do as a company, so we can keep fine-tuning our approaches and be alert to any emerging technologies. We can then talk to different groups and start motivating them to look into developing new products and solutions.

Q8 Which aspects of human genetics warrant further research?

Sequencing, and especially next-generation sequencing, is evolving. Firstly, we are moving from short-read to long-read, which is going to improve diagnosis, given that, even today, we can diagnose only 50% of the cases that come to us, and there is still a lot of discovery left to do. Secondly, this is going to facilitate all the discoveries that will happen in the future. Some advancements have taken place in certain recent methods, such as mass spectrometry, that really stand out. Thirdly, the field is very quickly shifting into looking at therapeutic answers for these diagnoses, with data interpretation, variant interpretation, and amelioration of clinical symptoms Some interesting topics are evolving: how protein–protein interactions happen, whether some therapies are interchangeable, and whether old drugs can be repositioned and repurposed. I think this is going to be an emerging area, which we all have to watch very closely.

Q9 During 2020–2022, you directed three COVID-19 testing laboratories for the National Health Service (NHS) in the UK and the California Department of Public Health in the USA, which together processed approximately 16 million samples. How did you tackle such a huge task during the pandemic, and how do you think this experience has shaped your career since?

The credit goes to the entire team at Revvity, as we really came together, and everyone pitched in. It was a tremendous team effort, with everyone working towards the same goal. Obviously, running two labs in two different countries was not a trivial thing to do, but we had a good cadence set up for our interactions in these labs, and in how we were going to carry out the testing. Revvity also developed a COVID-19 assay that, following an FDA review, earned the recognition of being the most sensitive assay on the market, which played a significant role.

Additionally, the company was able to provide end-to-end automation from sample to reporting. You can only imagine how many different teams were participating: assay development, manufacturing, software team, automation team, quality, regulatory, and clinical team, and so on. We all went through this period where we were reading the news and learning how things kept evolving. Our understanding of COVID-19 is still evolving now, both around the active disease and long COVID. This creates challenges for the labs, especially around the speed at which results are provided. The tremendous strain on the laboratories' throughput was an interesting challenge to tackle, and I think we have all learned that we can only progress if we help each other, through extensive collaboration. All of the people I interacted with, including my colleagues in the field, vendors, providers, and our contracting organisation, were working towards one common goal, which is probably the most important learning point: to keep that focus, and keep going, in spite of adversity.

Q10 You have chaired many international societies and councils, been appointed as Senior Vice President & Chief Scientific Officer of Revvity, and received numerous awards recognising your impressive contributions to the field of human genetics. To date, which of your achievements are you most proud of?

It's very hard to pick one, but if there's a single achievement I had to name it would be the people who trained with me and have gone on to develop their careers. I think it's really amazing to have the opportunity to pass knowledge on and pay it forward. I have been fortunate to train postdoctoral fellows, who have excelled and have now completed training and left to join different organisations. Within the global Revvity Omics force, we have labs in different countries, and to see that passion in each one of the professionals there is just truly amazing. I feel that my contribution is being able to train people who are really carrying the flag forward, and I feel very good about that.

Q11 What is next, both for Revvity and your own career?

The way Revvity was brought together, and the name itself (from 'revolutionise' and 'vita', meaning life in Latin), explain our passion in revolutionising healthcare. Our teams are energised and so well poised to take on these challenges. Now that we are a more streamlined company, it gives us more freedom to be able to look more broadly across the life sciences and diagnostics portfolios and focus on innovation. With my own career, I have really enjoyed my time here at Revvity, and I hope to continue to contribute to Revvity's purpose of expanding human boundaries through science. I'm really looking forward to that.