Hi, I'm Amanda Pichini, clinical lead for genetic counselling at genomics England. You're listening to the G word. Through the conversations we have on this podcast we hope to bring the benefits of genomic medicine to everyone. Today on Genetic Counselling Awareness Day, I'll be joined by Dave McCormack, member of the participant panel to Genomics England, Sarah Levine, consultant, genetic counsellor at the Centre for reproductive and genetic health and chair of the Association of genetic nurses and counsellors. Janice Bailey, a cardiology clinical nurse specialist and preregistration genetic counsellor, and Heather Pierce, genetic counsellor with a neural net Project at the University of Cambridge. Genetic counsellors are a highly specialised group of healthcare professionals, who have the expertise to help patients and families understand complex information relating to genomics, as well as provide guidance and emotional support. Whether that's to understand their family history, make informed choices about genetic testing, or come to terms with a result or a new diagnosis. They also deal with some challenging issues, like whether someone wants to know if they're going to develop a condition, how they're going to break the news to their family members, or if they're dealing with stigma within their family or community. Genetic counsellors increasingly work in innovative roles to in my role at genomics England, I provide clinical expertise and leadership to how we design and deliver our services and programmes like the newborn genomes programme, supporting our engagement and how we interact with our participants and ensure they can make informed decisions about genomic research. As genomics becomes more commonly used in health care, more and more people are likely to require genetic counselling and more healthcare professionals needing to rely on their expertise. Welcome to the G word.

So welcome, Sarah, Heather, Janice. And Dave, thank you so much for joining us today to share some of your experiences with the G word listeners. I'd like to start just generally by asking if you could each tell me a little bit about your journey to becoming a genetic counsellor, and what you do in your day to day role. Sarah, perhaps I'll start with you first.

Hi, I'm Sarah Levine. And I'm a consultant genetic counsellor. And I started out because I was actually just studying genetics as an undergraduate degree at Birmingham University. And I loved the Science, but I got to say, I never really enjoyed being in a lab. And I didn't know quite what that meant, and where that would lead me. And then one day in a lecture in my third year, they happen to make a fleeting mention to genetic counselling without any explanation. And I dashed off to the lecture afterwards. And I said, well, what was that you said? And he says, I think that's something that doctors do. And I thought, Oh, that's a bit that's a bit disappointed. But I went straight to the library, and I looked it up. And then I discovered actually, it was a profession. And then I stumbled across the fact that there was at that time, it was sort of fairly new MSC master's programme in genetic counselling in Manchester, and I was really lucky to be accepted onto that programme the following year. And after I completed it, I joined the team at guys Hospital in London, initially on research projects for cancer families. And I spent most of my early years in genetics, doing cancer genetics. And then in I think it's 2005 or so I got the opportunity to move into a different areas sort of specialism within the genetics team at guys, which was PGD, as it was called their pre Implantation Genetic Diagnosis, which is when we do IVF treatment for patients or couples who have genetic conditions are at risk of having a child with a genetic condition so that not because they need fertility treatment, but so that we can test the embryos and choose an embryo that is unaffected by the genetic condition to give them the opportunity to start a healthy pregnancy. And I was lucky enough to be able to join the team that offers that service that guys and I worked in that area for many years. And then I left guys to move to a specific IVF clinic called the Centre for Reproductive and Genetic Health, which is a big Centre for PGD in London, and I've been there since 2017. And now that is basically the kind of the genetics of the fertility world and PGD treatment is my day to day role. And you also I know wear a different hat where you are Chair of the Association of genetic nurses and counsellors. Do you want to tell us a little bit about what that organisation is? And I'm sure we'll hear more about that as we go along.

So the association of genetic nursing counsellors the agency is basically the representative body For the profession, and so people join it in order to get the various things that we can provide for the membership, which are, we do conferences and professional development courses and things like that. But we also are there to be a representative body to the outside world. And obviously, that a lot of that is talking to higher up hearts of the NHS, about the future of the development of the profession and how we fit in to NHS structures. It's talking with our colleagues from other specialities who we work closely with and their organisations.

I mean, I know there's only a little over 300 or so genetic counsellors in the UK at the moment compared to the over a million people working in the NHS. So having that central body that brings us all together, I think is pretty crucial. Yeah, thank you. Heather, do you want to tell us a little bit about your role as you've got a slightly different one?

Sure. Currently, what I'm doing is working as a genetic counsellor with a research project called neural net, which is being coordinated by the Department of Paediatrics at the University of Cambridge in cooperation with other hospitals in the region. And what it's designed to do is look specifically at rare diseases having to do with neuro development and mental health issues in the paediatric population. And specifically, the project that I'll be working on for the next couple of years, is doing whole genome sequencing, which is looking at the entire genome of children with a clinical diagnosis of cerebral palsy. And so one of the reasons for doing this is to sort of developed the pathway for doing whole genome sequencing in a clinical setting, because mainly, it's been done in a research setting, for the most part. So getting that pipeline developed to be done at Addenbrooke's, where I work, but also to look at the genetics of cerebral palsy, which traditionally hasn't been regarded as a genetic condition. But with recent studies, which have mainly occurred in Australia, about a third of children with cerebral palsy have been noted to have a genetic cause for their symptoms. And it's quite different from what I was doing before, which was full time. But I'm really looking forward to the opportunity to doing full time research for the first time in my career.

That sounds really fascinating. And so how are you finding that you're bringing your skills as a genetic counsellor into what that research project is doing?

So the the research project itself was primarily focused on just looking at the genetics of things. So actually looking at the the gene changes that we'd find in these families where the kids have cerebral palsy. And so what I have been able to bring to the table is actually looking on the impact on the families of doing this type of testing. So in addition to just looking at the the genetics, we're also looking at attitudes towards genomic testing for these families, again, because it's not really a population that's been approached about genetic testing before, but also the psychosocial impact of doing these types of tests and families, to see what their attitudes are, and whether it's actually something that they're they want to be offered in the future. It's not currently something that's offered on our current genomic testing directory for cerebral palsy. So we hope in part from this study, to give evidence that it's both reasonable from a standpoint of there being enough children with genetic causes to justify the testing, but also something that the families would actually want us to do. So that's what I hope to bring to the table here.

That's really important. Thank you. Janice, I'll turn to you. Could you tell us a little bit about your journey, and you're not just a genetic counsellor, but you have another role as well?

Yeah, thanks, Amanda. Yeah, I came to on genetic counselling quite a bit later than everyone else in my career, probably say, I've been a specialist cardiac nurse for many years, I've always worked in cardiology. And within the role that I was doing, I was sort of developing services for patients with INHERITED HEART conditions, and also families who were affected by, you know, family members that had got inherited heart condition. And so So to understand a little bit more about what these families needed, I went on some local genetics courses at my local university. And to be honest, I was just hooked quite quickly. I think the impact of a genetic condition on the patients I'm meeting and the possible effects on their family led me to really try and understand more and more about the genetics that were causing their condition, but also learn more about how I could support the families and support the patients. I was lucky then, after doing the courses to get to So to get a job in Bristol, on the 100,000 Genomes Project, which was a national project to the transformation project, to map the genome of sort of about 80,000 people with rare diseases and cancer, and I was very much involved with meeting the patients and the families, and doing the consenting alongside doing some more academic study. And and after the 100,000 Genomes Project completed, I think it's maybe three years ago, I was lucky enough to offer the offer to post as a trainee genetic counsellor to develop my skills further. So at the moment, I'm training and day to day I'm spending time in different specialities within genetics really, to try and gain experience, you get a wide experience of different inherited conditions, and sort of develop my understanding and my confidence, really, and I'm still working as a specialist cardiac nurse.

I'd say sounds really busy. I mean, I think there's quite a good amount of overlap between the skills that nurses have and that genetic counsellors have How do you find that in terms of the the differences or the similarities in the two jobs that you're balancing now?

Yeah, I mean, absolutely, I think we all have a core skill set that it sort of overlaps. Really, I think, spending more time in genetics has helped me understand more, I think conditions are very much with a patient in front of them. That's what their concern is. And I think with genetics, understanding more than its families as well, and how that might impact other family members, not just the patient in front of us. So helpful, you know, hopefully, I've been able to take that back into the nursing field and with my colleagues there, and, you know, look at the whole family, and future generations, as well as the generations we've got in front of us now.

That's great. Thank you. Now Dave, would love for you to tell us a little bit more about your and your family's experience with genetic counselling why you needed it? What was it like for you?

Thank you. Yes, we've been involved in working with and being supported by Georgina Hall, principal genetic counsellor, and her team working with Professor Green block, and his team. Within the rare genetic disease clinic. I was born with a rare genetic condition called micro and up Samia, which means the rest of the growth of the eye in utero. So the structure of both my left and right eye are weakened to having to have grown properly, there is bits of missing long story short microbes. Dalmia is small eyes, the eyes haven't grow properly, they're smaller in size and less complete in their structure than most of the people's eyes. As a result of that, I was born and raised as a child with very limited vision.

I've had some working vision in my left eye, not so with my right eye. And over time, it has deteriorated and this is a reality. For most people with my set of say conditions and up to Dalmia no eyes or Microsoft Belavia small eyes, that by adulthood or there abouts. Not all but a number of people will lose most of the working vision Now mine's been going down very gradually, since 2015, when I was diagnosed with to further conditions called My macular atrophy, which is where the central part of the retina is dying off. So I'm going blind. It's not a question of if but when. And I also have a delightful condition called diagnosed called Charles Bonnet syndrome, which is quite interesting because I have visual hallucinations. And this is not me having a psychotic episode, although I can assure you to begin with I thought I was until it was advised by the ophthalmologists. No, it is in this respect. It's quite standard. So I was losing my vision. As I say I was working as a an Sen, Education Manager to local authority riding stables of Sen. Ironically, your educational health and care plans for disabled children, people and families. I've always been involved in that line of work, or for many years in education, training and employment. And my son was going downhill very quickly. So I had to leave that role to get some rehabilitation use of a long cane, a text and logic software, those kinds of things. As part of that, I was sent in and out of Manchester, quite a lot, having various tests and assessments. And in March 2017, I went for a consultation. And whilst they were the ophthalmologist, I was asking all kinds of awkward questions as I do, because I'm very nosy and curious. And it just so happened that Georgina was present. And she said, you're asking some really valid questions about the why and the how and the when and the what she said I don't have any answers for you currently, I really don't. But what we could do with your consent is put you or refer you on to a new research project new then called the 100,000 Genomes Project, which is already been mentioned. And so at that point, I join 100,000 Genomes Project Ever sample by blood signed a lengthy consent form. And he also agreed to additional findings. So not only am I hopefully gonna find out at some point why I was born with this condition from a genetic diagnosis, but also why it's deteriorating, or has been from 2015. And also why all these other things have been occurring as well as any additional finding. So is there anything else that they might find, in addition to the reasons behind my sight loss? For example, I recently was diagnosed with osteoarthritis. Every is locations throughout my body are the two linked question we don't know. But if there is a genetic answer, that links into the sight loss, and the osteoarthritis, I would like to know, and I'm sure you guys would like to know, too, in terms of supporting other patients. So that's how I got involved with that. And Georgina, and the team were brilliant in explaining the different steps and stages. The key question for me is, was, if I start a family, my wife started probably, should I say, Well, this condition be passed on to my children? And the answer was, it literally is 5050. Flip a coin, because the nature of the initial tests and assessments of my condition is, my children may inherit this condition, or then again, they may not, it's occurred that both of my children, both my boys have not inherited this condition, which has led to some speculation as to whether it's held on the female line by condition rather than the male line. My wife doesn't have any sight loss any inherited conditions. And therefore, as the male with this condition, I have not passed it on to my children. They haven't come up with anything conclusive yet. But obviously, as technology improves, long reads of the whole genome develop, and techniques to understand what the data is telling us improve and develop, I'm hoping that at some point, so some clear answers will be given. And just to conclude, I am involved with the participant panel, working with genomics England limited to support the voice of the patient, not just those who have gone to the 100,000 Genomes Project. But now as we mainstream genome sequencing and genome testing, into the whole genome service across the NHS, and having my patient voice there. And those directions. And the guidance that I mean, says that we as patients might be able to give is essential, I'm very honoured to be able to support and develop all that work.

Thank you so much, Dave, for sharing your story. I think you've you've really demonstrated how much people with rare conditions become experts in their condition naturally, so and that's why it's so important that healthcare teams seek to understand your experience in order to be able to help support you in the best way possible, even if there aren't always answers available. So thank you very much for that.

And any playmate just to add to that excellent point you just made. It's one thing reading an abstract or getting involved in a piece of research, but you can't be lived experience. And as you say, that's why the patient voice is absolutely essential for that.

Absolutely. So I think we hear genetic counselling or genetic counsellors, and that automatically sort of sparks very different reactions or understandings in people. Because there aren't too many genetic counsellors, not everyone, at least not yet encounters a genetic counsellor across their lifespan. So I'd be interested to know whether you think there's any misconceptions about genetic counselling? Maybe you've encountered them in your practice, or in your experience, and what that might be? Perhaps, Heather, I'll come to you first on this one.

Yeah. So I think there's a lot of misconceptions. I think one that's particularly topical right now that we're mainstreaming things is what is the role of genetic counsellor if the genetic tests for diagnoses are being done through the mainstream pathway if a genetic counsellor isn't involved in the actual genetic testing itself up front, and what I'm trying to do is sort of close the loop because what I've noticed is that some of the consultants that are ordering these genetic tests aren't necessarily referring the patients back to genetics, for advice about how to deal with the condition, and how to talk to their families about it. And I think that's something that is a misconception amongst Consultants is the role of genetics after a genetic diagnosis has been made. And so what I'm trying to do right now is educate from both sides, both the patients and the clinicians to make them aware that really, we're still of great value to people even after they have a genetic diagnosis, probably especially after they have the genetic diagnosis. So I think if there was anything I was going to try to fix at this point, in terms of misconceptions, it would be that

Sarah Do you want to go next?

Yeah, sure. So, I mean, I was thinking about this beforehand, and that there's actually a few common misconceptions that occurred to me. One of them is I think that the word counselling sometimes throws people off. And I think different people react in different ways. Because for, you know, the general public, the word counselling, sometimes would be imagined to be more of a long term process of regular appointments and being psychoanalyse. So you know, that that's quite different. Often, genetic counsellors will see their patients perhaps, or particular stages in life. It's not an ongoing sort of psychotherapeutic relationship in that sense. And so I think that, for some people who are offered genetic counselling, they might worry that they're going to be psychoanalysed, when they don't want to be or something like that. What we're really here for, is to be able to explain something that's really quite complicated science, but how that fits into your life and your family and your values and that your decision making. And having that view is the counselling bit, rather than, you know, people having to come along to steal their whole life story or their innermost feelings about everything else, to us. And I think that that's an important misconception. I also think there's another kind of flip side to that from, sometimes from all colleagues who are doctors, which is that I think they sometimes hear that there's a genetic counsellor around. And, you know, I hope that doctors listening won't be offended when I say that sometimes they think, Oh, my patients crying, let's go find the genetic counsellor. That's, that's the person we need right now.

But hopefully, for those listening this that'll provide some clarification and reassurance. Thank you. Janice, what are your thoughts?

Yes, I mean, I would echo exactly what Sarah and Heather said. And so anyway, I've taken it a slightly different thinking of a different stance really, in that patients, I think, often don't understand what the genetic testing is. So not only do they not know how to dress a counsellor is, they don't know, what is this appointment with genetics. And in my experience, I've met quite a few people who've sort of watched a lot of TV, and maybe that's been in the DNA programme, or they've watched something about DNA in crime, and they're sort of coming to this appointment, they're going to meet this this person with this strange title, and what's going to happen to them, you know, they know they might have something in the family, but they also, I think, sometimes wonder what will be found out in the genetic tests. So will something else be found out when they've been part possibly of in a crime or, you know, something else about their family or even about themselves that they weren't expecting to know or not wanting to know. So I'm sort of thinking I think it's really important for the genetic counsellor to sort of explain what what they are their role is, but also what the genetic test is for?

Absolutely. A Dave, how about you

my experience of working with the, with the team from Manchester, first and foremost, they made it very clear that they could not give any absolutes. That is not what this is about, to try and make sense of the data. And when it comes to for me, and I haven't had a confirmed genetic diagnosis yet. I'm still waiting. The testing continues. It's ongoing, but as and when it comes through, I'm very clear that Georgina and the team's role will be to interpret the probability, because we are dealing not in absolutes, the technology is still in its infancy, some parts we've made up to massive leaps and bounds, which we can see in the treatment of certain forms of cancer. Absolutely fantastic. But in other areas, we're still very much right at the start of the journey, trying to get that messaging across. And I do need to make an important point here. When we look at how the 100,000 Genomes Project was communicated back in its inception in 2012. That morphed from, we will unlock the secrets of the human genome to we will sample 100,000 genomes, two very different messages. They both meant the same thing. But the way that messaging came across, and the way it was received, initially, there was euphoria, amazement, excitement, which then ended up into a bit of a trough into a bit of annoyance and frustration and irritation, which you will see on the 100,000 genomes Facebook page, if you'd care to look at some of the responses from patients like me who've been on the project, and in my case, waiting six years, haven't had an answer yet. Now, I understand the reasons why. And that's why I'm not going some from some time cross. A number of patients haven't had the clarity of information. And so if it's one message or one lesson that we can learn from this, and I'm determined, given my work with the Northwest genomics Medicine Service, as their interim chair of the patient, and public voice panel is that whatever information you are communicating? You do exactly what it says on the tin, clarity of messaging, what can you provide? How can it be provided? What does this mean for you? The patient's I know that it's so important. And I think partly, one of the key roles of the genetic counsellor is to support with that clear messaging,

as just that I was thinking, listening to what you were saying the other day, which is such a great point, managing expectations. And what maybe in genetic counselling speak, we call contracting upfront is so so crucial. I just

want to jump in there. Because when David was explained that it really reminded me of a common misconception that I experienced in PGD in the IVF clinic, which is that, you know that I think that like you're saying, Dave, that sort of promise of genetic technology is sometimes it's, it's been hyped up in people's minds, whether it's from the media or whatever, I'm not sure. But they come to us thinking that once we've put through the IVF, and we've got made their embryos in the lab that what we're going to be able to do is fix the genetic problem in the embryos and give them a kind of, you know, almost like a genetically engineered baby. Now, whether or not people would want that design the babies and all of that gets, you know, flashed around the media regularly, even though it doesn't exist. But that actually, it's not what we're doing. You know, it's much, much earlier, much earlier stage in technology than that. And what we're doing is quite basic, we're just testing the embryos and choosing, but I often find myself having to manage people's expectations of what actually is possible right now, because I think people get carried away with the idea of the science.

Absolutely. Excellent point. Moving on a little bit to some of the different things that you've all been doing in your in your careers. Janice, I wonder if you could tell us a bit more about your experiences with participants, you mentioned that you've taken families through the process of consent and have been supporting them with return of results of the 100,000 Genomes Project. Were there any particular sort of pivotal or memorable moments in that experience that you'd like to share?

Yeah, I think it's been really, really nice to think back to when I was working on the 100,000 Genomes Project, and meeting the participants I think, was such a privilege. And I think one of the standout sort of moments if you like, was meeting a whole families affected by inherited conditions, because again, generally, we just meet one, one patient and their family member or a partner. But actually, in quite a lot of appointments. In fact, many appointments, I have multigenerational families in the clinic room in front of me, but not enough chairs, you know, we have to get more space, which was just amazing. I worked mostly on the rare disease side, and mostly with families with heart conditions, renal conditions, and also eye conditions. So we had a lot of families that came together. And I think it was really important for them to come as a family, and not just on their own. And one family in particular, I remember we had an older lady come and she had already lost her sight. She came with her son, who was losing his sight. And, and his daughter who was in her early 20s, who are though her sight was fine, they could see changes in her eye test to know that she she had the condition, and she had her young baby with her. So we had Ford and aerations in the room with us. And I think the effects of that condition on that family was really apparent and quite devastating for them really, but the way they supported each other, and helped to clean, okay cared for each other through what was happening to them.

This really highlights that that family impact if you keep coming back to that when you deal with genetics, you're not dealing with just one person. Now Heather, you've you've also worked and trained as a genetic counsellor in a different country. Like myself, I'd like to know some of your reflections on what you think are some of the differences between areas of practice or ways of working in the UK versus elsewhere?

There are a lot of things that are fairly constant in the way that we deal with families. So I think the amount that we support families, and the way we support them is very similar. But I think there's a lot of differences in the infrastructure of how we provide those services. And I also think there's probably a lot well, I know there's a lot more variability in the scenarios in which we work in the US versus the UK. In the US, there is probably more of focus, or at least there was at the time I was there was now quite a while ago, about 810 years ago, was much more of a focus on sort of doing genetic education rather than so much of the actual support of the genetic counselling process. So the psychotherapeutic counselling, and part of that was because we also had to spend a lot of time when talking to patients about the practicalities of whether their insurance, their medical insurance was actually even going to pay for the genetic testing. So a lot of time was taken up with these very crass discussions about well, you might actually not be able to access this test you really need, because your insurance won't cover it. So I think that we are very fortunate here to not have to worry about that type of thing. Now we are quite restricted in what we can offer genetic testing for and who we can offer it to. But it's accessible by the entire population, which is much different than it is in the US. Another difference in practice, which is probably changing now more than it had been in the past is that I think genetic counsellors work in a much broader spectrum of positions in the US than they do here in the UK. And, you know, I think part of that is regulation. So, in the US, I was actually working for a couple of clinical laboratories that just did genetic testing. And I played a very large role in actually writing the genetic test reports and, and doing interpretation of variants, which was fascinating. And I think part of the reason I got to do that is because I'm trained as a PhD in genetics. So I do have a little bit of, you know, background, but I think that's something that traditionally hasn't really been so much of a role of genetic counsellors here in the UK. But now I think that we're changing our training methods to be much more crossover with other aspects of science, we do a lot of cross training in the scientist training programme, with bioinformatics, and with the other specialties,

such as say, Get, we're really, I think, a product of the the country or the culture that we live in. And being such a small workforce, I think it's really helpful that we can do a little bit of our own thing, because each country is different. But also seeing what's happening in other parts of the world really helps us kind of reflect on maybe areas that we haven't yet tapped into that we could or ways of doing things better seeing what other people are doing. And you mentioned about training programmes. So Sarah, I know there's been a lot of growing interest in the profession over the years, there's also a huge demand. Can you speak a little bit to how someone could train to become a genetic counsellor, and what that's been like in the UK.

In the UK, there's a few different ways to become a genetic counsellor. And Heather already mentioned, the scientist training programme, we call that the STP. And that's an NHS training programme. And it's the same programme that's used as headless ng to train other scientists in the NHS. So liberatory scientists and various other professional groups. And so we've been very lucky to be allowed to develop a genetic counselling sort of branch of that programme. But if you're into that programme, then it's basically a three year training programme. It's a sort of a paid NHS post while you're in it. And the requirements are fulfilled to apply for professional registration at the end of it. So genetic counsellors, once they've done their initial training, have to go well, it's a voluntary process, but go through a process of registration with our genetic counsellor Registration Board to show that you meet the kind of competencies of the profession before we have the STP but also still now there's the way of doing it where you do an MSc, a master's programme in genetic counselling, which prior to the SDP was how a lot of people did, it's how I came into it. There's two current programmes, one in Cardiff and one in Glasgow, and those are two year courses. And when you come out of that, you would need to then apply for a trailing post. So it's sort of a junior genetic counsellor job in order to get enough clinical experience to fulfil the requirements to do your GCP registration a bit further down the line. And then the final route to be a genetic counsellor is that, like Janice, actually, you know, there are many people who come into our profession who are already qualified healthcare professionals, nurses, midwives, clinical psychologists and others, who are already practising in a clinical area, but then get drawn into genetic counselling in the way that Janice did. So there are a few ways to become a genetic counsellor. But we are a tiny workforce in a very large, major organisation at the NHS. And that means that yes, there is huge demand for us and huge interest is still small numbers of people that are being able to get trained as genetic counsellors each year. Hopefully we'll be able to keep growing as time goes on

a clear need and whether we can meet that demand in the coming years. Absolutely. That's

the challenge. Yep.

So Dave, I'd be really interested in your views. I mean, as genetic counsellors were often described as very reflective and reflexive healthcare practitioners, we always want to do better, as I'm sure many people want to do for their patients. So what are some ways that you think things could be improved, particularly thinking about the role of the genetic counsellor and what we can do to help patients and families

Thank you. Excellent question. The answer for me alongside what the ground we've already covered on the kind of expectation management, I think there's two or three other components. So they, first and foremost, it's wherever possible being patient centred, what do I mean, trying to get a lot of sides and to understand to unpick where that patient and or their family are coming from, what did they want out of this process, they may not understand initially. So having the curiosity and the ability to pose those key questions in an accessible format, which is my second point about the absolute importance, vital, vitally important to use the language, they terminology, it's keeping the sense, but stepping the language down to an accessible format, which patients and their families can understand and can follow. We use language, it surrounds us all the time we use it in our day jobs, as you know, socially. And knowledge is power, language is power. And so in order to be patient centred to get alongside the patient, and their family to understand what they want out of this genetic counsellor interaction, understanding what that interaction will involve, what it can offer them that expectation management, within all of that is the use of accessible language, which everybody can access, everybody can, can understand. So that ultimately, you've equalised that power relationship, the patient and their family have an equal stake in that power relationship, to help the whole genetic testing process be a bit less scary, and a bit more understandable, and a bit more approachable. And something that the patient or family will then ultimately want to engage with, and be able to provide more information, because they're comfortable, they're safe, they understand what's going on what's being asked of them, and what may come out at the end of this process of, of genetic testing. So what can be improved? I would say those are for starters.

Thank you, Dave, I think those are really helpful things for us to keep very much at the heart of what we do. And as we think about what we can do better in ways forward. Final question. So I'd love to hear your thoughts on what you think might change for the profession over the next say, five to 10 years. Sarah, who I'll start with with you.

I think that what's really clear right now is that the NHS as a whole has a major priority. And you've heard the word lots of times during this discussion of mainstreaming, which basically means that we're, the NHS is hoping for genomic testing to be rolled out much more widely to be much more accessible to not be held just in the small number of hands of the genetic professionals that have clinics and and to be available, really on the ground. You know, in speciality clinics for different types of health conditions. I'm sure Janice will talk soon about the kind of way that plays out in cardiology, but there's many, many settings in fact, every setting where you could do genomic testing, and that undoubtedly is going to bring changes and possibly challenges to the work of genetic counsellors. And actually, a lot of genetic counsellors, or some genetic counsellors are already moving into mainstream roles. I myself don't work in a genetics clinic anymore. I work in an IVF clinic and Janice has obviously in a cardiology clinic and actually have a you're in a paediatric setting now, so there's lots of genetic counsellors who are already moving to the mainstream to deliver genetics, you know, at the coalface, you know, in and with working with our colleagues from those specialities rather than just internally with our own colleagues it who are geneticists, and I odd seeing some roles, whereby you've got some genetic counsellors only a few at the moment, but who, for example, work across a whole hospital trust, and sort of float from one speciality to another being the kind of genetic expert in that hospital, who can help with education for their colleagues who can help pick up the more complicated cases and deliver the kinds of processes for genetic testing, almost acting like a kind of a consultant who's kind of available to everybody. And I think again, that's a very exciting role. Luckily, I think that as a profession, when we think about our future, we are really well equipped with a very wide range of skills. And that makes us I think, able to adapt and, you know, be used in lots of ways by the NHS by different settings, to to fill in some of these gaps and as I say things like education and policy and delivery, are things that I think we can also get involved in. So I feel very positive. I think that, you know, adapting to change is something that we, as our kind of clinical practice, we help our patients with that, you know, most of our families who we talked to, are very much having to cope with adapting to a massive change in their lives have a genetic diagnosis, well, sometimes I think, when as a professional group, we can see the change coming to us, and we sort of have to be genetic counsellors to ourselves to be able to, you know, kind of apply that and, and adapt and cope with having to adapt.

So it's got some really, really salient points, I think, you know, it's a huge change. It's a huge change for the whole NHS, you know, genetic counselling and and other members of the multidisciplinary team, but I think it can work really well. And I think, you know, Dave said about putting the patient at the centre of what we do, is really important, and mainstreaming and in many ways does this. Within cardiology, we've worked really hard over the last few years to make mainstreaming work. And what we found is the collaboration and between the clinical genetics, so with the genetics counsellors, and the clinicians, the cardiologists and the specialist nurses is the most important thing. And it's a really good time to share our knowledge and their knowledge and educate each other. And there's things we can learn from them. But also, we can help really with, yes, I mentioned about the more complex patients who may have sort of more family challenges or psychological problems that navigating that genetic test, and the result can be more, more difficult for them. So that's a place where we come in both to help support our clinical colleagues. But, you know, take that work on and work with those patients as well, you know, I think is really important, and bringing the care for the patient their pathway under one roof, rather than sending them to one separate department.

Just to pick up my last couple of points there. From from Janice and Sarah, I can I think I can say this, perhaps where you may not be able to but as a as a patient, and as someone involved in advising various committees nationally and regionally across the Northwest, I think I'm able to make these two points. And that is, you are an honourable but small community. And this is going to be mainstreamed across all seven Genomic Medicine Service regions. In England, certainly, I believe we need two things, we need resources, you need resources in real terms, you need money, you need input to develop more larger training courses to reach a larger group of trainees coming through as the interest and the requirements increases, the demand increases. And you also need the how do I put this the promotion of the Korea groups of genetic counsellor to to raise the profile and raise the awareness to raise the the benefits of what you as genetic counsellors can actually do so that other clinicians, or the medical professionals understand your central integral role in supporting the patients?

Hey, I think I think your points are really, really important. We mentioned earlier about my role as as a nurse and as a trainee genetic counsellor, and that in the overlap, and I think we need, you know, it's good to remember that a lot of other health care professionals have some really, really good skills. And what a lot of the need is just a little bit more confidence, or is it more understanding or just a little bit more time to think about some of the issues that a patient with an inherited condition that's having genomic testing might have as opposed to someone coming in for just a clinical test? So I think for many patients, and if if some condition is talking to a patient's about an inherited condition, it fits to talk about the genomics of it and the genetics of it. I think the genetic counsellors we have have got a real opportunity to give their experience and knowledge to other members of the team to help them upskill themselves, so that they're not becoming you know, half genetic counsellors, but they have some of those skills and understanding of what these conversations are with with patients and their families and what support they might might be needing. salutely

there. Yeah. So I think, you know, bringing all this together, I think, you know, what we could really do to push this forward is to provide whatever evidence we can to those fun Doing bodies to those people in positions of power and public policy to demonstrate what our value is. And so I think research is a very large part of that. So research demonstrating what our value is, and being involved in research and how to best provide these services to patients and their families. So I think we've, we are so underpowered from a workforce standpoint that we spend a lot of our time seeing patients, but we also have to carve out that time to do research and to what the best way to do that for our patients is, and be involved with those research initiatives so that we can prove our value. And also looking into the best way to leverage the experience of other professionals into providing those services to patients and also how to best educate those other professionals and how to mainstream those services. So that's one of the reasons that I want to be involved at the front end of this neural net project is so that we can foresee some of those potential problems with providing results to patients in a timely fashion, and dealing with those expectations. So I think research is is very key and something we can't overlook in all of this, in actually arguing our case for why we need more support, and how we can be more integrally involved in the mainstreaming effort.

So in general, I think we just need to watch this space as a huge amount of opportunities. It sounds like out there for genetic counsellors. Thank you. It's it's been a really interesting discussion. I really want to thank all four of you for joining me today to share your experiences, your incredible expertise, and to celebrate the role of genetic counselling on genetic counsellor Awareness Day. Thank you very much. That's all for this episode. Thank you for listening to this discussion about the G word and for joining us on this journey to highlight and debate the implications of genomics as it comes to the mainstream of health care and society. You can find out more about the association of genetic nurses and counsellors@www.agnc.org.uk or on Twitter at the AGNC if you have any views on these topics, or have a person in mind you would like us to interview do write to us at podcast at genomics england.co.uk. Remember to subscribe to the G word on Apple podcasts, Spotify or wherever you listen. And then if you've enjoyed listening giving us a five star review really helps other people find out about the series. See you on the next episode of the G word