# **How can Genetic Counsellors improve care through research?**

# **The G-Word Transcript**

**Amanda:** Hi, and welcome to The G Word. My name is Amanda Pichini, and I'm the Clinical Lead for Genetic Counselling at Genomics England. Today is Genetic Counselling Awareness Day, and I'm delighted to be joined by Emma Walters, member of the participant panel to Genomics England, and Jonathan Roberts, NHS Genetic Counsellor and Clinical Content Developer at Genomics England.

We'll also hear voices of genetic counsellors throughout this episode from the recent World Congress on Genetic Counselling. Genetic counsellors can help people understand and adapt to the impact of genetics and genomics on their health, and they can take on many roles in the clinic, in education, in policy, or in research.

Genomics is becoming part of clinical care and research at an unstoppable rate, and this means more and more people will need the support that genetic counsellors can provide. Today, we'll talk especially about the roles genetic counsellors have in research to help improve care for patients and families.

So welcome Emma and Jon. Thanks for joining me today. Let's start with some introductions. Emma, over to you.

**Emma:** My name's Emma Walters. I'm part of Genomics England’s participant panel. But actually, my most important role is as a mum to my son Joshua, who is a young man that's involved with the 100, 000 Genomes Project, which is how we first got involved with genetic counselling.

**Amanda:** Jon, do you want to introduce yourself as well?

**Jon:** Hi, my name is Jon Roberts. I'm a genetic counsellor. I currently work in the NHS, at Addenbrooke’s Hospital and also at Genomics England on the generation study. I work clinically and I also have an academic background with a PhD in science communication and I'm particularly interested in how science is represented in broader culture and popular culture.

**Amanda:** Thank you very much both. So Emma, I'd love to start with you. You mentioned you're a mum to Josh. Could you describe your and your family's journey with genomics and how is genetic counselling part of this journey?

**Emma:** So Joshua was born in 2013. He was extremely premature. So he was born at 26 weeks gestation and we spent quite a considerable amount of time obviously negotiating that NICU journey, which is never straightforward.

Joshua has had problems since birth in that he doesn't breathe when he's asleep. So when he's awake, he breathes perfectly normally, and when he's asleep, he doesn't. So essentially, he's a typical man and can't do two things at once. Kind of about two years into our story, we kind of looked into whether we thought there might be a genetic component to this.

So we'd heard about this condition, which was known as Central Hypoventilation Syndrome, or the old term that they preferred not to use is Ondine's Curse. And so we initially were genetically tested for the gene change associated with that, which is the FOX2P gene. So that's where we kind of initially came into contact with genetic counselling, over at Queen's Medical Centre.

Obviously I'm very conscious that I'm doing this with a recall bias. So, this is all kind of, eight years back for us. But our genetic counselling consisted of a telephone conversation, so we never actually met anyone face to face at all. It was done over the telephone, and I did check this with my husband the other day.

It was actually only done with me, so actually my husband was never involved in the genetic counselling at all and it was done whilst I was at work, so I remember being sat in my office at work, with everything that goes on in the NHS in the background, attempting to have these conversations regarding family history and going through all the medical family history that's really important when you're looking at genetics.

But we were also entered at that time onto the 100,000 Genomes Project, which they explained to us was very new, it was very cutting edge. But we found that Joshua didn't have the gene change on the FOX2B gene, which is what we originally tested for, and they thought, potentially, that it was a variation that hadn't been seen before. They still assumed that was where the error was. I remember us talking a lot about having no findings. There was a lot of discussion about the fact we may never get, you know, we may not get an answer. That there might be quite a lot around, actually, us not being able to get any genetic findings at all.

But I actually don't recall anything being discussed about unclear findings. So actually, things that are likely pathogenic, that are variants of uncertain significance. And we didn't really ever touch on the uncertainty that findings might bring at all. So that was never really discussed with us.

A really long period, obviously, between being initiated into the project and getting a finding, which was really unexpected. So we were actually found to have, what at the time was classed as a likely pathogenic change in a gene, called ATP1A3, which is a neuromuscular sodium potassium pump subunit, I believe.

And it's, it's been linked to some conditions known as Alternating Hemiplegia of Childhood, CAPOS syndrome, and Rapid-Onset Dystonia Parkinsonism, none of which Joshua has. So, he has a very, very rare phenotype associated with this gene change. And we were given that information face to face by our geneticist, a 20 minute/half hour appointment and she was very honest at the time. She said, honestly, this stuff's really cutting edge and we don't know what it means at this point. We honestly don't know; we've not seen this before and it's going to take time because as we gather more and more information, that's potentially where we might be able to tell you a little bit more down the line.

But I'd put some comments here that we've never been offered any form of counselling post this at all. So actually, from that initial meeting where we were given the gene change diagnosis and this was found, we've never actually had any counselling. Unfortunately, there's been no signposting and no opportunity for those follow up questions.

Because I think obviously, as a parent and I'm sure as a patient as well, you kind of take in this stuff cold. And then you go home, and you Google and you start looking into things and then you have these questions and unfortunately, we've never really had that ability to go back and ask those questions.

**Amanda:** Thank you so much for sharing your story with us. It sounds like it's been a very long and stepwise journey for you and your family and that there's some things that perhaps you thought could have been done differently or improved if you were to walk back through the process and think ‘if it could happen again, there's some things I might've liked to have done differently’, like your husband having been involved, for example.

**Emma:** Absolutely. And it's quite interesting really, that we're actually just re-entering back into the genetic world again. We had an endocrine appointment yesterday and we're being re-referred back into genetics as part of the GMS now, so as a slightly different part and obviously we'll be meeting the team down at Addenbrookes Hospital, which we've not met previously.

In some ways I think it's quite interesting for me to see have things changed over the last few years at all. Certainly, I think my preference would be that it should be a family conversation. You know, we're both parents to Joshua, and it's difficult for me to talk about these things with him.

Number one, he doesn't have the medical background, but number two, he never received this counselling, so I'm almost trying to do this and educate at the same time, which is quite difficult. And I think certainly the ability to ask those follow up questions would have been so, so beneficial. And unfortunately for us, it wasn't there.

**Amanda:** Thank you so much.

Jon, I'd love to hear your perspective as a genetic counsellor working in the NHS. And I'm sure over time, you've also encountered things that you've maybe felt in the profession can be improved to do better to address the different and changing needs of your patients. What are your thoughts?

**Jon:** So, I think your experience is really important because if we're going to improve clinical care, we have to understand what went well and what didn't go well, which involves research. It's really interesting that you've talked about discussions about uncertainty, and I think that’s something we could reflect on. Both in terms of how clinical genetics works, but also how genetics is talked about in mainstream care, because we know that with more genetic testing may come more uncertainty. We have to figure out how to have those conversations with patients, especially when genomics is being done at scale. In terms of the research that is happening, there are some genetic counsellors who are working on studies like the 100,000 Genomes Project involved in the consenting process.

My first genetic counselling job was on a study called the deciphering developmental disorders project, which in some ways was a precursor to the 100,000 Genomes project. And those conversations did try and allow patients to have discussions around uncertainty, what the results mean, the timeframes. The challenge is finding space and the right people to give people the chance to have those conversations.

I think some of what you said, I was reflecting on the difference between genetic testing and genetic counselling, and sometimes they get talked about in the same breadth because many people having genetic testing will have genetic counselling, but they're two separate things. And there are genetic counsellors and other researchers who are doing important work, looking at how we evidence the value of genetic counselling separated from genetic testing.

And I think that links back to what you were saying in terms of the way in which once the test had been done, you haven't been given that genetic counselling afterwards to go through. What does this mean? How do I make sense of this? So, for example, there's genetic counsellors like Jehannine Austin in Canada, who works in the field of psychiatric genetics.

We've done a lot of work looking at the role of genetic counsellors and helping people make sense of their family history and any tests that may or may not have happened and evidencing that separate from any testing. It works in psychiatric genetics because psychiatric genetics lends itself to that because historically there hasn't been as much testing you can do, but we know there's still value in understanding the heritable aspect of psychiatric disorders based on the family history and increasingly testing that can come in.

So, I think one way in which research could improve, the experiences of people like you, Emma, is a clear understanding and evidence base of the value of genetic counselling. So even when the testing has been completed in the sense that there's no further testing that can be done either clinically or in research, people still think of genetic counselling as a useful thing that people could have to make sense of what's going on.

I think research can kind of contribute to improve it through that respect. I think we could also look at research that is trying to understand how we can do genetic counselling better. In particular, research that's looking at measurements of genetic counselling. So ,it's quite hard sometimes to clearly identify what the benefit of genetic counselling is because it's centred around these psychological metrics of things like empowerment, but there's also work going on looking at things like ‘how we can measure empowerment’, ‘how we can measure the control that people feel’, ‘how we can measure the impact of genetic counselling’. And I think that feeds into improving the experience for people like you Emma who have gone through that journey and thought this could be done different because it sounds like if you have been offered, for example, genetic counselling that had been done in a way that felt more inclusive to you and your family, it is really important to hear those experiences and reflect on what the impact is, but also measure the impact of what that would be so we know how to do our work better.

**Emma:** I was just going to come back on something that you said, Jon, that was actually really interesting for me to reflect on. And as you know, I work in the healthcare profession, but I'd never really thought about the fact that genetic testing and genetic counselling had these almost two distinct purposes. To me, everything kind of came under one umbrella in one area. And it was actually quite interesting for me to reflect on the fact that actually, Genetic Counsellors, to me, it sounds like almost have their own identity within this journey. And that's actually something really interesting for me to reflect on. And I do wonder if perhaps, that that's something that you need to almost get hold of and really push forward. You know, what it is that you do that is so powerful to patients and to participants, what do you offer that's so unique and so powerful for us?

**Jon:** I completely agree. And I think that's where research and clinical practice can really feed into each other, because we will only make that compelling argument through clear evidence.

That can be evidenced in lots of different ways. It can be evidenced through things like qualitative studies that allow patients to explain and show that experience, to tell those stories of how beneficial it is to have genetic counselling when there's no testing available or the testing's been done and it feels like, from a clinical point of view, there's no further testing.

That's not the end of the story, that genetic counselling can still be really valuable, for we can evidence that as well through showing the impact of these things, like there's a genetic counselling outcome step scale that measures things like empowerment. To show that value, aside from just the testing, requires research to be done in lots of different ways to ensure that counselling identity and that patients can access that counselling.

I think another way in which research can really push that agenda forward is understanding who isn't accessing that counselling. So when people get through the door and they experience genetic counselling, often that can be really valuable and they can suddenly start to make sense of all this testing, this family history. They can have a chance to talk about how they feel about it. That's really valuable and we can work to evidence the value of that. We also know that there are some people who never even hear about that and never get through the door. So, another really important piece of research that needs to continue, and it's happening in various ways, but needs to be pushed forward I think is understanding who doesn't get to those services, who don't we see and why.

**Amanda:** Thank you both for those really important reflections. I think what strikes me as well, hearing what you've both been saying is there's a real need to bring both the genetic counsellors and what they're doing in their healthcare roles and the patients themselves and their experiences together, whether that's to help define what are the right research priorities? What should we be looking at? Who are we not accessing? It's only by bringing those two perspectives together that patients will then feel that their needs are best met and that genetic counsellors can focus on the right types of research questions that can help improve that care, which is really important.

Jon spoke to some other genetic counsellors at the World Congress about what research genetic counsellors should be doing. Here's what they had to say.

**Jon:** Great. Hi, we're here at the World Congress. Could you introduce yourself, please?

**Manisha:** My name is Manisha Chauhan. I'm a genetic counsellor at Sydney Children's Hospital in Australia.

**Jon:** Fantastic. And could you tell me some research that you think we should be doing as genetic counsellors?

**Manisha:** Yes. So, we all know the value that we provide in a hospital setting, and I work a lot with subspecialty medicine in lots of different areas. They can see the value of what we do, but we need to have outcomes measures that really demonstrate our value and from a health economics perspective and from an efficiency perspective and taking patient views into account of how we work and what we provide in that setting is anecdotally is very useful.

**Amanda:** Let's head back to the Congress where Jon asked, what are the key skills that genetic counsellors have that make them good researchers?

**Jon:** Thank you so much. And could you tell me what are the unique skills you think genetic counsellors have as researchers?

**VO:** Yeah, I kind of always intuitively known that, but just being here, at this conference just made me realise it even more and be almost proud to be a genetic counsellor and part of the research community as well. I think what struck me about all of the presentations I've been hearing in the last few days is the importance of co-participation, right? And collaboration with our participants in research. I think we do that every day in clinic. We are people who are empathetic, or people who centre the client or the patient first. We are trained in communication, and we are trained in informed consent and I feel all of these components make us, or set us up to being, great collaborators with our research participants.

**Jon:** So we're here at the World Congress, could you introduce yourself please?

**Alison:** Tēnā koutou katoa. I'm, I'm Alison McEwen. I'm a genetic counsellor from New Zealand and I work for a university in Australia.

**Jon:** Thank you. And, um, could you tell me about some of the skills that genetic counsellors have that you think make us good researchers?

**Alison: Y**ep, absolutely. So, I think genetic counsellors are excellent communicators and listeners. We usually have really strong organisational and planning skills. We have a really deep curiosity about our clients and our work, and I think all of those things translate beautifully into research.

**Amanda:** Now, let’s get back to the interview. So, Emma, building on what Jon was just saying there about thinking about who's not having access to genetic counselling, what's your perspective? What do you think is the impact of not having access to genetic counselling?

**Emma:** It's been really interesting listening to Jon talk because it's made me reflect really on whether we actually ever did have genetic counselling. I think on reflection, and again, I do come back to the fact this is for us, coming up seven years ago now, but it felt to us very centred around our participation in the 100,000 Genomes Project and really making sure the consent was there with regards to that. Which is interesting particularly with what Jon was saying about separating genetic testing from genetic counselling, and I think our experience was that those were very grouped together.

As you say, it's finding those people who don't know what they don't know. So, if you don't know when you're having genetic testing, that you have access to genetic counsellors, that there are these people that you can talk through this uncertainty.

What do you do if you have a result? What do you do if you don't have a result? What do you do if we have something that we're not sure is the cause for your symptoms or your condition? I can't, I think, put too high a point on how important access to that is. But it's something I personally within our family felt was hugely missing within our experience. And it makes you quite sad in a way that we never really had access to that. It would have been so helpful and so powerful. And as Jon was saying, it's how do you evidence things that aren't numbers and aren't results driven? And I think, qualitative research is absolutely the way to go with that, or your mixed methodologies. But actually, there's a huge, huge amount to be said for those things that you can't test with numbers, that you can't evidence. But actually, it doesn't mean it's any less important. If anything, to us as a participant, to us as a parent, it's more important. Because these are the things that impact us on a on a day-to-day basis.

**Jon:** That's a really valuable reflection. And I think that experience. allows us to show how research can really have a meaningful impact for patients because it touches on academic issues which centre around the definition of genetic counselling and the definition of genetic counsellors.

Genetic counsellors have a professional identity and trying to pinpoint what that identity is, is a theoretical issue that can occupy us and feel a bit navel-gazing at times. Lots of genetic counsellors work in the NHS but do other bits to their roles that aren't necessarily genetic counselling. At the same time, genetic counselling is sometimes provided as an activity by people other than genetic counsellors. And drawing up all these fine distinctions and identities and working out exactly what activities is can feel a bit like an academic exercise that is only interesting to the people who are only genetic counsellors and clinical geneticists and those sorts of people. But actually it has a real impact because when you start to identify and clearly define those activities and professionals, professional identities and professional activities, you can then start to implement them in a healthcare system in a way that is really meaningful for patients and make sure they don't miss out on those key aspects that we're talking about, particularly around, I think, uncertainty. And I think that's something that's going to be a key part of genetic counselling and genetic counsellors work is helping people make sense of uncertainty, because you can only do that by understanding your feelings and the context in which that uncertain result has come about.

Everyone makes sense of uncertainty in an idiosyncratic way, and we can't get rid of uncertainty. Genetic testing might give us more information, but with that information may come more uncertainty. And you can only take that so far with an educational lens of helping people understand it. You have to help make people make sense of it and that is a genetic counselling practice and the discussion you're having about what you've missed out on can really link those academic discussions into genuine patient benefit to make sure people are accessing those services that will really be a benefit.

**Emma:** I think it's interesting, Jon, for what you were absolutely just saying there in that I am from a medical background myself, I work within the NHS. If I'm struggling to differentiate between genetic counselling and genetic testing, then that tells you that I'm probably not isolated in that.

**Jon:** I completely agree. And it's something where we know in academic research, this distinction is important.

What we can also then do as another bunch of researchers, is look at how can we make that knowledge accessible and compelling to people who are deciding about what patient pathways are about, on where to spend resources. The NHS. along with many other healthcare systems post COVID is thinking carefully about where to spend their resources.

So if you're going to advocate for genetic counselling to get resources and genetic counselling in this specific set, not just to accompany testing, genetic counselling as an activity unto itself to get resources, which are precious. You have to make a compelling argument. So understanding not just what the research says, not just what the patient benefits is, but for how to get yourself heard is another area in itself.

Because as you said, you are not going to be an isolated case. We talk about this distinction a lot within the profession. That's one thing, all well and good. The next level is how can we make this distinction really meaningful when it's taken out to a wider healthcare setting.

**Amanda:** So we're really trying to bring it, as you're both saying, to the heart of what's going to be most important to improve those patient and family experiences, which is often a driver of research.

So carrying on that research thread, Emma, and I know you mentioned that for you and Josh and your family, you were involved in the 100,000 Genomes Project, which was research. Have you been involved in any other research relating to yours or Josh's genetic condition? What was your experience and how has being a participant of the 100, 00 Genomes Project or any other research impacted your perspective on it?

**Emma:** One is that we're involved in something known as the GenROC study, which is quite a new study I believe coming out of Bristol, and it's looking at intellectual disability and particular genes, and whether there's a link between those. Now, I wanted to kind of pick up on the fact that that's actually not our initial reason for referral into the 100,000 Genomes Project. Intellectual disability, actually felt very much to be related to Joshua's severe prematurity. So, we've kind of accidentally fallen into that piece of research as things are changing and we're finding out more and more and more information. So actually, our initial reason for referral into genetics, i.e. his central hyperventilation and need for ventilation, isn't actually the research study we're involved in. It's just been found through, as you say, massive amounts of data generation that potentially Joshua's gene change actually also impacts another part of his life.

So we're involved in the GenROC study with, I think it was Dr. Karen Rowe who's currently running that one. And we did have quite a lengthy discussion because Joshua doesn't only have the gene change. He obviously has lots of other issues going on as well as the extreme prematurity. We were quite conscious that we didn't want to impact their research by adding in a slightly complicated child.

But having spoken to Karen, she was actually really on-board with us coming on and felt it was really important that Joshua's story was heard. So that was one thing we wanted to comment on. And then the other thing we wanted to comment on was that actually we're still very isolated from a research perspective.

So, as I'd commented previously, the gene change that Joshua has is linked to three known conditions, and those conditions are quite well known. There's support groups, etc., for those conditions. But that we're very much in, and it's a term I've picked up from another participant panel member, that we are N plus 1.

So that we, as far as we're aware, are the only phenotype for Joshua with this particular gene change. We're involved with the charity Unique, so we know of three other people through that with the gene change. Again, we don't share the same phenotype at all, and that's a really isolating place for us to be.

Even as somebody who works within healthcare, I can't access the researchers. I would love to be able to get somebody with an interest in this gene change and say, here we are, hello, we'd love to be involved, we'd love to drive your research forward. But there doesn't seem to be that mechanism yet for that to happen.

And again, I understand that takes numbers. But for us, it's still been quite an isolating experience in that we don't really have anything from a research perspective. The, the GenROC study actually, I found out about on Twitter. I was reading Twitter and that's how I found out about it.

And you think, I'm probably fairly social media savvy, I'm fairly technical, but then are we missing people that aren't looking in those places? You know, is there a better way of accessing these patients and these parents and these families?

**Amanda:** Yeah, that's a really important reflection. Thank you for sharing that, that feeling of isolation from a research perspective, especially when a lot of these conditions, the more genetic testing we're doing and the broader that testing is, the more likely we are to find changes or genes or conditions that we have quite little knowledge about.

It sort of links back to what we were saying earlier about uncertainty. And it's extremely important and helpful that there's some umbrella charities like you mentioned Unique or Contact a Family that can really help support families collectively that are in these individually very rare scenarios but can be really difficult to sort of identify or find research opportunities that are relevant for them and that can help improve and learn more about those conditions in the future.

Are there ways that either of you think we can enhance that? How can we make sure, so for example, Jon, research that genetic counsellors might do, how do we make sure that they can find those participants that are individually very rare so that they can access those research opportunities and feed that back for improvement of their care?

**Jon:** It's so important, I think, for genetic counsellors to understand these issues and ensure that they are exploring psychological issues like the isolation. So there are charities like SWAN (syndromes without a name), which exist because of the challenges of having a diagnosis, but not having a clear name or a clear diagnosis that you can place on that, leaving you in quite an isolated place because you want to be able to connect with other people who've had similar experiences and try and make sense of what's happening. And if you don't have that, it can be very, as you said, an isolating experience to feel as if you're the only person this is happening to, and no one really understands what's going on.

And one of the ways that research is driving improvements in patient care is to be able to link up patients who have ultra rare diagnoses, so it can benefit the scientific knowledge, because we get a clearer picture of what exactly is associated with these gene changes, and can also benefit families who can connect with people who've got similar experiences.

I think from a research point of view, it's about ensuring a consistency in genetic counselling practice of raising these issues so exploring if patients have these feelings of isolation, if they're feeling as if they're the only person and what's available and looking at what is current practice? How routinely are these issues raised? How are clinicians aware of what's available? And how often are they actually linking out to these? So some of this comes from research that looks into how well are we doing with genetic counselling practice? And for me, one of the key values of research is to ensure that we continue to improve our practice.

And that involves critical reflection of what we're doing and an honest reflection of what the state of play is, both in our country and in other countries, so that you can then improve that to understand what patients are really looking for when it comes to depth of counselling and are we providing that?

Because if we don't critically reflect on that, then we won't improve our practice, won't give patients what they need. So this is something that has come up in the research network I talked about earlier, which is how do you put patients at the centre of this genomic revolution? And one of the ways to do that is to understand what they really want from genetic testing and genetic counselling, what really matters to them and what really matters to them when it doesn't happen.

Then you can work to improve clinical practice by ensuring that they have access to those services that they need. So research I think could really drive that agenda of ensuring that if there are key factors that would improve people's experience, for example, links out to other charities or links out to other researchers or ways in which you can proactively get involved in research, if you have an optimal diagnosis, understanding what those needs are, and then ensuring that those inform part of our clinical practice.

**Emma:** It was something I was thinking of, and please excuse me if I don't use the right terminology, but I think we also have to be really cautious about kind of assuming causality as well. As I say, because Joshua's primary reason for coming into the study actually wasn't his intellectual disability. And we're involved in this research study that looks at genetic changes and intellectual disability.

But I think, for me, I'm incredibly conscious that actually his whole journey so far may have impacted his, you know, his intellectual disability. You very rarely come out from being 26 weeks premature without some level of disability. For me, I find it very interesting as to sometimes are we looking for causality that isn't there? You know, from a genetic counsellor point of view, to have somebody to talk that through with is actually quite powerful as well.

**Jon:** I couldn't agree more. There's two expectations that I think can lead to assuming causality. One is patient expectations. If you have a child who doesn't have a diagnosis and you find a genetic change, parents are searching for an answer. they desperately want an answer, and you find a genetic change and it may offer some explanation or may offer a certain explanation, but it's risk that you over interpret that because you're so desperate for an answer for all the reasons you've been talking about so you don't feel like finally we feel at least like we've got an answer we've got a community to which we belong and we can move forward with this and that can shape those feelings can really shape how you interpret the result of a genetic test.

On top of that many of the ways in which we are used to talking about genetics, a lot of the framing and the language and the metaphors that we draw on from broader culture do resort to this deterministic view of genetics. By that I mean that genes are assumed to be a very powerful form of information that are fundamental to who you are.

So if you look at the way genes are used in film, or the way genes/DNA is used in advertising, often the assumption is that genes are incredibly powerful, they're part of who you are, they're the essence of your identity, and that language doesn't always do us any favours if we're talking about an incredibly uncertain and nuanced result in the context of a gene change that may or not explain some or someone's condition.

So there are two challenges that can lead us to assume that causality. One is an internal view based on the feelings of people desperately searching for answers. And the second is the language that we're often drawing on from broader culture. And it does require a nuanced conversation with someone to really get to the roots of how they're feeling, what those expectations are, and what this result really means to the context for them, because you can assume causality for all those reasons.

So I think you’re absolutely on the money there.

**Amanda:** So it strikes me just hearing what you're both saying that a key enabler to making research and clinical care really work best between patients and healthcare professionals is trust. Trust is a key element we often talk about in genetic counselling in order to work through maybe a particular issue or a new diagnosis or genetic risk to be able to talk about that as openly and honestly as possible and discuss the full picture of what's impacting that family.

Trust is so crucial. So I'd love to hear from you both. How can we ensure effective communication and trust building between genetic counsellors and patients and families. Emma, do you want to go first?

**Emma:** I think what you just said there, Amanda, is so fundamental. It's around communication and really honest communication.

I think there's something incredibly powerful about somebody saying, ‘I don't know’. And I think sometimes within healthcare, we feel wrong if we can't give somebody a definitive yes/no answer. Then we feel like we've done something wrong or we've missed something. And that's not the case, particularly in such a fast moving world as genetics is.

One of the most powerful things I remember from our journey is our geneticist, our genetic consultant, actually saying to us, honestly, I don't know what this means. That the data just isn't there, and Joshua is so unique at this point, I can't tell you what's going to happen. And I'll just share a little bit about my journey, if I may.

Obviously, we've been through a huge amount over the last ten years, and this all culminated in last year that I utterly broke for six months. I'd still been working in the NHS, and I absolutely hit a wall, completely stopped work for six months and really had to go back and reflect on everything we'd been through over the previous 10 years.

And part of that reflection involved me ringing our geneticist and actually saying, I don't know where to go with this. I don't know what to do next. I don't know whether Joshua's prognosis is life limiting. I don't know whether it's life threatened. And it was very, very powerful for me. And, I'm very thankful that she took the time to let me have that conversation with her and that communication with her, but she was brutally honest and she said ‘honestly, I don't know’ and but we made a plan going forward and I think that's a huge thing to take forward that if you can find a common ground about how you move forward that's a really powerful thing.

So, we just said we were going to touch base in a couple of years, and she would keep her eye on papers because as you know, these research papers are constantly being generated and looking very internationally as well. Although we have a superb system in this country, we also know numbers generates data.

So we need to be looking outwards as well. So for me, it's very much about the honest communication and also about looking into what a family or a participant or a patient needs. What do they need to move forward? And being really honest about what can be offered. You know, we may never get an answer to this. But what would help you feel like you've not been forgotten?

What would help you feel like we're continuing to look for you? And that's very much touching what Amanda said about trust. I guess, because I work within the service, I understand about confidentiality and safeguarding of data. And that's something for some families that is going to be a huge thing, looking at future proofing our genetic service. Are we looking at, insurance issues going forward, are you going to have to tell people that you've had genetic counselling? Are you going to have to tell people you've had genetic testing? Further down the line, is this going to be an issue? I think it's just being really, really honest about where we are and actually what the red lines are for us, I think, with genetic testing and counselling is really, really important about. Actually, what I'm struck by something, somebody said is it's about just because we can do something doesn't automatically mean we should.

**Amanda:** Thank you so much for sharing that. Jon, any thoughts you have to add to that?

**Jon:** Thanks Emma, It's a really powerful sharing of your experience.

I would completely agree with you. One key factor to ensure trust is being upfront about what you do know and what you don't know. That's a fundamental basic of ensuring that there is trust when you are in any healthcare role, particularly in genetics, where there's a lot of uncertainties, being transparent about what you don't know and what those uncertainties are.

And I think something we can do as genetic counsellors is have an understanding of different types of uncertainty and exploring not just what we don't know, but what exactly is it that we don't know? Because uncertainty can come in different forms. There's different types of uncertainty. So I think a deeper understanding of what we mean by uncertainty, what uncertainty is in play, being clear about what we don't know, which again, sounds linguistic, but actually is really useful when you're having a conversation with honesty. This is what we do know, and this is the bits that we just don't know yet. I also think, reflecting what you were talking about in terms of your experience and the challenges that you faced. I think allowing those spaces in a clinical consultation are really important because I think if you are going to have genetic counselling, that's going to be useful.

You need the other person to have understood the world as best they can from your point of view. So, it goes back to a lot of early genetic counselling, was rooted in the philosophy of Carl Rogers and this idea of empathy and unconditional positive regard in congruence. And I think that is still relevant to our practice today, especially in terms of ideas of trust.

You need to know that you've got space to say what you're going to say. And that someone will listen to you and not judge you for it. As a counsellor, you can ensure that patients have space to articulate what matters to them, what their experience has been, and that you can listen with an engaged curiosity without trying to think, what am I going to say next? What's my agenda? Because it's so important for trust to know that the person, and it's something we all experience, I think, when we see any healthcare professional, which is that when they are pushing their own agenda without listening to you, you back off. You close down. If this person isn't going to listen to me, I'm just going to close myself down here and I'll find what I need somewhere else.

So, if you allow for that into the consultation. If someone knows that they're going to be listened to, they're going to take the time to understand the world from your point of view. I think it really does engender a trusting relationship that, yeah, the value of that can't be underestimated.

**Emma:** I think it was interesting there what you said about that utterly non-judgmental part of the counselling role.

It's something I've touched upon with fellow parents and we have a group of parents that I meet with, who all have children with very complex health needs, potentially very differing genetic diagnoses. But we've said that the one thing it enables us to do is to have a space where it's safe for us to say how awful something is, how crap it is. To say, and it sounds awful, but it's true that if we could do this again with hindsight, would we have had our children?

And that's such a powerful thing for us to admit. And I think, as you said, that that ability to have a very safe and a very non-judgmental space, as you say, you can empathise with patients, you can empathise with participants. You can never walk in their shoes, unless you've been on that journey yourself.

But I think it's so important to have that non-judgmental space where people feel it is safe for them to release that emotion, which for me is very much about where genetic counselling has such an important role to play.

**Jon:** You've touched on something that I think is, again, an academic issue that actually feeds into clinical patient experience in a really interesting way which is the nature of empathy and how we empathise.

Because we can't, as counsellors, walk in your shoes. And actually, that's not necessarily appropriate to empathise to say we're going to really understand exactly how you feel, because we can't. What we can do is provide a space where you can speak freely. Because if you can speak freely, you can think freely, and you can make that decision.

Now, we can be attuned to pay attention to what those emotions are and respond to those. So with empathy in that respect. But actually, what we're doing is we're not trying to put ourselves in your shoes because it's not possible. We can just provide a space where someone is paying attention to what you're feeling and gives you a space to talk freely, to make a decision that is right.

So if people, for example, are talking about whether or not to have more children, if they have a child diagnosed, you’ve got a range of decisions, all of which are fine, you can make one. But the key is which one is right for you, and you're only going to get to that point if you say you can speak your mind and know that somebody isn't going to be judging you or jumping in.

So, it's so important to have that space where you can speak freely because then you can think freely, then you can start making decisions that are right for you. So, I think it's a really important role that genetic counsellors can play in giving people that space when they're trying to make these really important decisions about what to do with genetic information.

**Amanda:** Now we're going to hear from some genetic counsellors from the World Congress on Genetic Counselling on some aspects of genetic counselling research that they're excited about.

**Jon:** Hi, we're at the World Congress and could you introduce yourself?

**Jared:** Yes, my name is Jared. I'm a genetic counsellor working currently in Montreal, and I did my training at the University of British Columbia.

**Jon:** Amazing. And could you tell me about a piece of genetic counselling research you're really excited about?

**Jared:** Yeah, for sure. I am actually planning on doing research looking into the ways in which genetic counselling programs are considering diversity when selecting their future cohorts, so that we can improve the diversity in the genetic counselling field in the future.

**Jon:** So important. Thank you so much, Jared. Thank you.

Hello, could you tell me your name and where you work?

**Jehannine:** Yes, I'm Jehannine Austin and I work at the University of British Columbia in Vancouver, Canada.

**Jon:** Fantastic, And could you tell me about a piece of genetic counselling research you're really excited about at the minute?

**Jehannine:** Yes. So, at the moment I'm working together with Kennedy Borle on some work that's all about trying to examine the potential benefits of embedding genetic counsellors within primary care practice settings, rather than where we have historically been, which **a**s you know is usually tertiary care, large academic referral centres kind of thing. I'm really excited about the potential of what genetic counsellors can do in a collaborative multidisciplinary team-based setting with a family practitioner.

**Jon:** Hi, so could you introduce yourself please?

**Kennedy:** Hi there, my name is Kennedy Borle, I'm a genetic counsellor and a PhD student at the University of British Columbia in Vancouver, Canada.

**Jon:** Great, and could you tell me about some genetic counselling research that you're really excited about?

**Kennedy:** Yeah, so I think one of the things that's plagued the profession for a long time is how we actually demonstrate value, especially when we're talking to policy and decision makers. So, there's a lot of really good research coming out of the US, and some that's coming out of my own PhD thesis, that's more about how to communicate value in order to get health systems to fund more genetic counsellors and more genetic counselling.

**Amanda:** Now let's get back to the interview. So as a final question, and given we've put this podcast episode out in particular for Genetic Counselling Awareness Day, I'd love to hear what's the one thing that you'd like to see change, maybe over the next five years or ten years, so that genetic counsellors or the genetic counselling based research can really improve care for patients and families.

So really looking to the future, what's something you'd really like to see change? Jon, we'll start with you.

**Jon:** There's lots of ways to answer that. And one thing I would like to see a continuing improvement of, I think the trend has started, is active patient involvement in research. So, I was going to give a slightly perhaps glib answer because of my interest in genetics and broader culture.

One thing I'd like to see is representations of genetic counselling in films, TV, soap operas and general representation, because I think one thing that unites genetic counsellors, be they in research or clinical practice, is that they do have, I think, based on this kind of Rogerian philosophy, quite an optimistic and inclusive view of people.

And that's something that's always driven me in the profession is this idea that if you give people information and you bring them in, that they can be empowered to solve their own problems, and you do this in partnership. That philosophy, I think, underpins so much of genetic counselling practice. And if you could see that reflected in broader culture, that'd be fantastic, but something I would really like to see change, there was one thing, is researchers changing what they do based on what patients and participants tell them. So real active engagement. Being brought in at an earlier stage, so they are really shaping the agenda. I think something that will be absolutely amazing is if researchers talk to patients, take their views and go, I haven't thought about it like that, let's do things differently.

I think if we can start doing that, and there is trends to do that, and I think if we can continue that trend, I think genetic counsellors, because of that kind of optimistic view of people and that kind of view of partnerships, if counsellors can drive that change, I think that would be fantastic.

**Emma:** So, I was just having a think about, for us as a family, what would be more useful. And I think one thing we've touched upon in previous questions is we've looked about putting patients at the centre of everything that we do. And for me, what would be hugely life changing for us was to be able to do patient initiated follow up.

So people would be able to access counsellors as and when they needed it. And I know that's really difficult, and I know resources are incredibly limited, but it still is very much around what the NHS will provide. This is what we provide, this is when you can access it, and when you've accessed that, that's when it stops.

I think the ability to actually reach out to somebody and say, I'm struggling at this moment and I could really do with that space would be very, very powerful. And then I think the other thing for us is just to remember perhaps that there's two viewpoints to genetics. So I know for our scientists and our researchers that genetics is incredibly fast moving. It's moving in a phenomenal pace and it is, it truly is. But I guess I would just ask people to remember that for patients, that perception can be very, very different. It took us three years, initially, to get from the decision to get involved to even receiving a diagnosis.

We're actually re-entering the genetics world now. And we've been told, realistically, we're looking at least eight months for that initial appointment. So just to remember, perhaps, that there's two sides of looking at it, that while things are fast moving, it actually can still feel very, very slow, and very, very painfully slow to families who are living with this at this time.

**Amanda:** Thank you so much. It's a really important point you've both made, I think, about bringing patients and families and genetic counsellors together. And it's been extremely valuable having this conversation with both of you today to bring your perspectives to our listeners with a clear and powerful message about making sure that we're all thinking about how we can continue this in the future.

Thank you to our guests today, Emma Walters and Jonathan Roberts. And to welcome Connecting Science and the genetic counsellors who participated from the World Congress on Genetic Counselling. If you enjoyed today's episode, we'd love your support. Please subscribe to The G Word on your favourite podcast app and like, share, and rate us wherever you listen.

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