**How can we support complex patient journeys?**

**The G Word transcript**

Naimah: Welcome to the G Word. This year it’s our 10-year anniversary and as part of this we wanted to discuss the patient journey for people living with rare conditions. In this week’s episode of the G Word, a member of the Participant Panel at Genomics England will be speaking to a genetic counsellor and the CEO of Genetic Alliance UK. Now let’s go over to the interview…

Shelley: Hello and welcome to the podcast today. My name is Shelly Simmonds, and I am a member of the participant panel at Genomics England. I am joined today by Amanda Pichini, who is the clinical lead for genetic counselling at Genomics England, and with us also is Louise Fish, CEO of organisation, Genetic Alliance UK.

So, I'm going to start by asking Amanda and Louise to tell us a little bit about themselves. So over to you, Amanda.

Amanda: Thanks so much, Shelley. So, I'm the clinical lead for genetic counselling at Genomics England. That means that I provide clinical expertise and leadership across a number of our teams and services, but particularly the Newborn Genomes program, which is a large-scale research study that we've been working on with the NHS and we'll be starting later this year. I'm a genetic counsellor by background. I've previously worked in the NHS and before that in Canada, providing genetic counselling to patients and families with a wide range of conditions. Um, and I was involved in delivering the hundred thousand genomes projects in the NHS and helping to mainstream.

Bringing other genomic tests into a variety of, of specialties. I have a particular interest in the education training needs that other healthcare professionals might have, particularly as genomics and genetics is quite new to them, so that we can make sure that when they're facilitating consent or returning results for patients and families, that that can be done in a sensitive and effective way.

Shelley: Thanks, Amanda, Louise.

Louise: I'm Louise Fish. I'm the Chief Executive at Genetic Alliance UK and we are an alliance of over 230 organizations ranging from household name charities like Cystic Fibrosis Trust, through to tiny support groups set up by a mum and dad whose child has a super rare condition, and they want to support other parents in the same situation. So, a really broad group of patient organisations that support people with a really broad range of rare and genetic conditions. We run two long standing projects - one is Rare Disease UK. Um, so we've got a particular focus on kind of genetics in, in rare conditions, as well. And we also run the Swan UK project, which supports people who have a syndrome without a name such people who clearly have a genetic condition. It's not clear what it is and to date, All the medical advances mean, you know, we, we know we've still got a doc lot to learn. We are supporting individuals who still don't have a diagnosis that would help them better understand what their condition is and how it'll impact on them and the rest of their lives.

Shelley: Thank you. I think we're going to have quite an interesting chat today. We're going to share patient journeys and we're going to talk about how organisations like yours, Genetic Alliance UK, can support people living with these rare conditions and their families, whether they have a diagnosis or not. We want to explore how charities kind of integrate themselves into the lives of patients. The role that, you know, you guys play and how you continue to support patients throughout their journey onwards. I think it's also, important to note that patient journey doesn't stop at the day. You get a diagnosis, it's forever changing. It's ongoing process. And it's really important to acknowledge that, even those who don't have a diagnosis are still living a journey and it's going to be, helpful today to explore how they can find support as well, as things go along.

So, I suppose my first question to you both - is how have you seen things change for those living with genetic conditions over the last 10 years. Do you think there's been more awareness. Are more people coming to you for support? Louise? And have your resources increased to obviously keep up with the demands?

Louise: I think there've been massive change over the past 10 years, and they're primarily on the scientific side. You know, the scientific progress that's been made has been incredible, and, you know, the ability to do whole genome sequencing in particular and to do it relatively quickly and relatively cheaply is absolutely amazing. And that will over time make a substantial difference to people's lives and give people some of the answers that they're looking for. But ultimately, you're right, it doesn't change the fact that people are on a journey. So having a genetic condition is a lifelong journey. You know, start when they're born, that the genetic condition's already there in many cases.

And your relationship with that condition, how it impacts you, changes through your life. What we tend to find with our charities is that people get in touch as soon as they have a diagnosis, and they will stay in touch with that charity throughout their lives. You know, they may get in touch when their child goes to nursery for advice on how to talk to the, the nursery school about what support their child will need, and then they'll get back in touch when their child starts at primary school.

For some advice on how to navigate primary school and what adjustments their child might need to, to make the most of their school life. Um, they may be in touch in different years. If they get a new class teacher, they'll certainly be in touch at the transition between, um, primary school and senior school.

To think through what support a child traveling further to a school that's further away you might need. And then we tend to get people back in touch with us if their child's progressing higher education or onto seeking employment opportunities or just going to a different kind of care setting.

At that stage to access social care and support in the community. And similarly, if people are changing between residential care or home, people will be in touch. So, it's kind of the relationship our member charities have with individuals living with a condition lasts throughout a lifetime and change progressively as that life goes through.

Right through to supporting people if they are elderly carers in their seventies, eighties, and nineties, supporting an adult child with a genetic condition. Again, we get people reaching out at that stage as well. So, these are kind of lifelong conditions, and therefore they require lifetime support and a lifetime relationship with charities.

And we'd like to see that kind of thinking, I suppose, extended to the way that NHS thinks about providing services for people with genetic conditions throughout their lifetime and what that journey will look like from a patient perspective through the NHS as well.

Shelley: Definitely I'd agree with that. My 10-year-old son has Duchenne muscular dystrophy, and it is changing, you know, weekly, monthly, the services that you need to access change. You know, the people that you need to see change. So, to navigate that, you know, sometimes you find from the NHS, the support isn't necessarily there, so it's really important to have organisations like yours to help us through to, you know, navigate what stages come next.

I think over the last 10 years, certainly in my case, there's definitely more awareness out there, you know, with the use of social media and. Facebook groups and communities, people are connecting a lot more, which I find really comforting because, you know, I can't imagine receiving a diagnosis like this back in the kind of seventies or the eighties, where you would've just dealt with it alone.

You would've had no one to talk to, no one to connect with. You would've just, you know, I actually... If, if this doesn't sound in the wrong context, I feel quite grateful we've had the diagnosis now. Um, and not all of those years ago certainly would've been very different for us. What support is there available for people who don't have a diagnosis? So how do you help support those journeys? Because in those cases, sometimes the journey to a diagnosis is very long and drawn out, and regardless of the many tests that somebody might have, there are still no answers at the end. So how do you support those people?

Louise: So, I suppose we support twofold. So, because Genetic Alliance UK is a member of innovation, in the first instance, if someone does have a diagnosis, we try and signpost them as quickly as possible to the specialist member charity that can support them. And that might be for a single condition like, um, tuberous sclerosis complex – we signpost people to the TSA (The Tuberous Sclerosis Association). It might be that someone has a metabolic condition, and we can signpost a charity that supports people with a really wide range of metabolic genetic conditions. So, we try and signpost people to specialist support as quickly as possible.

Where it's challenging is where, as you say, someone doesn't have a diagnosis, or they have a diagnosis that’s so rare that it isn't really clear what that means for them anyway. So maybe that someone just can't get a diagnosis because the part of the gene that's affected for them isn't a well-known bit of the gene. So, there isn't a test at the moment that enables them to get that support. And in that case, we try and support them directly at the moment through the Swan UK project. We bring them together with other parents who have children with undiagnosed genetic conditions. And we try to make really clear for those, families and also for the services that support them.

If it's not clear what the condition is, you should still be supporting people on the basis of need. So, you know, you don't need to have a name for your condition to know that your child needs speech and language therapy to help them or physiotherapy to help them. That's what we try to make clear. It is really challenging having that conversation with the NHS.

And one recent development we've been really pleased to see is there is now a syndrome without a name clinic in Cardiff that's been going through a pilot project. Look at how the clinical genetics service can support people who don't have a name for their condition. And we'd like to see that kind of rolled out across England, Scotland and Northern Ireland as well, if possible, to do pilots in all four nations to see how that can help to provide better joined up care.

I suppose the other thing that would be useful to flag up is that sometimes people do know which part of the gene is affected and they get a diagnosis, but it's so rare that they don’t how that will impact on them for the rest of their lives. So, you know, you can get a condition where it's got a name for the part of the gene that is affected, but what that doesn't tell you if you are the only person in the world with that part of the gene affected that's been found so far, or one of maybe five families around the world - what it doesn't tell you is what that will mean. You know, is, is your condition already affecting you as it's going to affect you throughout your life? Is your condition degenerative? Is it going to get worse or progress in certain ways throughout your life? I'm sure Amanda can add more, but you know, sometimes even when you do get a diagnosis of the bit of the gene that's effective, scientific knowledge and clinical knowledge just isn't strong enough to know what that will mean for you and your family, um, over, over your lifetime.

Louise: Absolutely.

Amanda: Yeah, I completely agree with that. I think it's really changed just the kinds of uncertainties and questions that people seeking that diagnosis have. Or when we talk about the diagnostic odyssey, you could argue that we haven't necessarily been taking that away, we're just changing what that odyssey looks like. So where in the past, we were less able to give people answers to the underlying cause, the genetic cause of their condition. We're now able to do that more. We're sort of swapping that in with new questions around, we can tell you what the gene is or what the changes, but we don't know about enough people or that have lived with this condition that we're aware of to be able to give you some kind of anticipatory guidance for what your child's future might look like at some point. I think that provides a lot of other challenges, but also a lot of hope for families because it means that they can connect with other families in that similar situation and think about what those next steps can look like in that community of support.

It's really important that healthcare professionals across the board - and this links to what you were saying, Louise, about having a syndrome without a name clinic - it's really important that we understand what our role as healthcare professionals are to help families understand where they can be signed, posted to for those sources of support. And also, that every family's journey will be different. So they might come across stories in those support groups that really resonate with them and say, well, this is exactly what I and my child have experienced, so it's so helpful hearing that someone else has been through the same thing, but they might also hear stories that completely don't resonate with them, and that might cause more fear or anxiety because that's not the particular journey that their family's had. So, it's important that I think the NHS is able to both signpost but also provide some helpful context to families to say things like, not every story that you read will be the same. There might be things that are similar and different that you and your family experience. And that's really important to have that context when they're linking with those support groups.

Shelley: I think that's a really important point that you've just made, a really interesting point because actually even among people who do have a diagnosis and maybe have a diagnosis of, you know, more common rare conditions. The condition can present so differently, even with siblings in the same family. So, to be very clear with people about what to expect, but also what not to expect, as well, you know, we have to manage that really carefully. And I suppose this is why the support groups, you know, whether that is an organisation like Genetic Alliance UK, whether that's a Facebook community or you know, whatever, it might be, so important to connect people together because even if you know you have a different diagnosis to somebody else, all of the fears, all of the anxieties, all of the worries, all of the services, they're all the same. So, we are going through something really similar, even if it's not quite the same name or the same, you know, gene or, you know, everything is very similar.

And other people going through something like this have that natural understanding of somebody else's story and can help them emotionally. Because that's another thing that there isn't enough of out there is your kind of psychological and emotional support. And often you do find that from other parents that's how things are. And I've found that particularly special for me to have people there that just get it. That you can phone up at 10 o'clock at night and say, “Oh, I'm worried about this” Or “oh, do you know how do I apply for this?” So, you know, having that lived experience is really important. I suppose that brings us on to the next question, which is for Amanda, what can organisations like Genomics England do to work better with charities like Genetic Alliance UK moving forward? How can we improve relationships and partnerships?

Amanda: I think it starts with, I guess, recognising where Genomics England sits in that kind of wider ecosystem in the UK. Crucially that we work very closely with the NHS. We're not an organisation that’s providing that healthcare directly. But we have a role in working with the NHS to either help with some of that sign posting, to provide some of that support and training in some of the systems that the healthcare professionals use to facilitate genomic testing, particularly whole genome sequencing.

We also work very closely with a number of researchers, and often those researchers are also not directly interacting with those patients and families. So, where we can provide more signposting and awareness to help with that understanding about what a patient or family's journey is like with a rare disease.

And who are some of those patient organisations out there who often are very helpful in advocating for, in championing the use of research in their patient organisations. I think we have a role in potentially helping to make some more of those connections happen. I think one example certainly that we've been exploring with Genetic Alliance and other patient charities is in our work with the Newborn Genomes program where, like Louise said earlier, people interact with charities at different points in their lives. Some points that are going to be much more crucial for certain people than others, with the Newborn Genomes program we would be providing. A small number of families with an early diagnosis for a genetic condition that should be treatable in the NHS, but regardless of where or when or how that happens, that's going to be hugely impactful for that family. When they hear that news, and we know it's important that those families should be able to quickly access timely information and support when they need it. And that I think also provides an interesting challenge for Genetic Alliance and other charities who are maybe more used to providing families with information and support when they've had symptoms and there's already a suspicion of a diagnosis. So, when they go to get that information, they're coming at it at a different time point and from a different perspective than parents in the Newborns program who may be with a child who has no symptoms yet.

So how you sort of interact with that information and find support from other families in that community could be very different. So that I think also presents an opportunity for us to work more closely with charities like Genetic Alliance, so we can think about how can we make sure that the information presented to parents at the time of that first call of a new potential diagnosis from the program means that they are receiving the right kind of information support at the right time.

And that's something that we'll be exploring closely in the coming years. So, I think there's quite a few ways in which we can work together, both in the clinical and research sides to make sure that we're helping to amplify and signposts to the kinds of information support that charities like Genetic Alliance and others can provide.

Louise: Yeah, I think that's right. And I think one of the challenges that actually it's quiet, it's almost easier to have the conversation on the research side. So, we're working really closely with Genomics England to kind of look at how that program might work and, and that, as Amanda says, the emotional journey is very different. If your child is unwell and you want to find out why and it's almost a relief to the diagnosis in terms of your emotional journey. If you've got a newborn baby and you haven't noticed anything different at all with that newborn baby - that's a very different journey when you get the news that there may be something about your baby that's going to require additional support and that you need to go for further testing. So we are, we're kind of working really hard with Genomics England to learn and to think that through. I think where the challenge comes for us is that that diagnosis, whether it's through a research program or through routine clinical practice, like prenatal screening or the newborn heel prick test. That moment of diagnosis and finding out about a condition is just the start of a lifelong journey for the individual with the condition and their family because that genetic condition is going to be lifelong, as we've said, and it's going to impact on someone's lives, is it going to change at different kind of key points throughout their lives, as well as kind of their own health journey when they personally are affected by the condition, when it might make them unwell, when it might be easier to manage, when it might be harder to manage. Kind of all of that journey is, is a lifelong one.

And, and I suppose one of the challenges we are looking at moment is how do we also talk to the NHS about how routine clinical services. We'll support people on that journey and how can we join that up and offer much better coordinated care for people both within the NHS, you know, so when someone has a rare, complex condition, they can be into touch, you know, they can be in touch with kind of 15, 20, 30 clinicians focused on different aspects of their care and joining up that care and making sure that someone is taking a leadership role in it and pulling together a multidisciplinary team to coordinate the care.

That in itself is really hard at the moment and is not happening routinely for most people. And we know from the work that the amazing Concord study is done that only four out of 10 children and one out of 10 adults have a care plan in place. And that's even just having a bit of paper that says you've got a care plan, let alone whether that care plan's being delivered.

So, there's a lot more we need to do to work with the NHS to make sure that the care from the health service is joined up and coordinated for people. And then beyond that, how does that coordination reach out to education, to housing, to benefits to social care? The bit that almost should be simplest is if the NHS has someone who understands your child's condition, that it should be possible for their school to be in touch and to find out how that condition's going to affect them on what support the school might need to put in place through an education, health, and care plan. But those links out to the other services aren't there either. So, for us, there's a lot of work to do that's not just around the diagnosis, but it's about ensuring that lifelong care and support is delivered in a coordinated way. And as more people are getting genetic diagnosis, through this amazing kind of clinical advances, how do we make sure there's also investment into the clinical services that are going to support people throughout their lives?

Shelley: Absolutely. I think you've touched on a, a really, really key point there is, it is brilliant all the scientific advances for giving a diagnosis, I find it incredible what we can do. But you know, we can't just give people a diagnosis and send them on their merry way. We have to, you know, we have responsibility for these people if we are. You know, rolling out this sort of work and programs. And I touch upon a point you made earlier. My son received his diagnosis when he was 11 months old and he wasn't meeting his milestones, which obviously that triggered us to see a doctor and a paediatrician and have lots of tests done and the absolute heartbreak and trauma and shock of a diagnosis for him. Even though we already kind of knew there was something, it was just, and I can't describe that it's unexplainable. So, my heart goes out to these families who may receive some news as part of, you know, newborn screening who have mapped out their life already with their new baby and you know how their life is going to be and will receive a bombshell of news because it is unexpected because their baby is really tiny. And yeah, we need to make sure that they get the best possible experience.

It's just so important you know, how we give a diagnosis, when you hear that news, speaking from my own experience when I heard those words that your son has Duchenne muscular dystrophy. You hear that, you know, life expectancy and all of these terms you remember that day for the rest of your life, that stays with you forever. And there are some people who have had an experience of a diagnosis that has been far from up to, you know, in my opinion, a clinical standard or a, you know, in an ethical way. And I really hope with the newborn program that we can change that for people who are coming after us in many aspects of life. But you are absolutely right, Louise, when you say that things aren't joined up. It's still like that now. And my son's 10 and we've been on this road a very long time. It’s still clinics on these days, but another clinic you need to see is only on, on another day. And it's all that, that extra burden is quite huge as well when you are already dealing with so much. So, there is lots of work to do to make things better for families like ours.

Louise: Absolutely. And you know, I hope the newborn screening program provides a real opportunity, cause obviously it is a research program and it's just going be looking at a hundred thousand babies.

I understand in the first instance, obviously Amanda, you're far closer to the program than I am, but that opportunity to learn about what that good moment of diagnosis and communication can look like. Because you're right, when you hear that news, it's totally overwhelming. And actually, you take in very little of the rest of that conversation.

And actually, you know to have that conversation face to face and delivered sensitively, as Amanda's mentioned, but also to have the opportunity. So, you know, we're going to talk you through it. We're going to give you a letter to take away with you with all the information in it, you're going have a number on the letter to call back and ask us about it because we know that all of the questions are not going to come into your mind right now. We're going to have a follow up appointment in a couple of weeks when, when you've had time for the news to sink in, so you can come back and ask some of those questions face to face. All of that really matters, as does making sure alongside that conversation, there is a really clear pathway of this is what will happen next. This is how the NHS will support you over the next few stages of the journey, and this is what the pathway looks like going forward to support you through your lifelong relationship with this condition. And these are the patient organisations that help you. All of that package of support needs to be there and needs to be right from the beginning. And it, it's not at the moment, but this program, this research program offers us an opportunity to show what a difference it makes if we get that right.

Shelley: Definitely. And you've just triggered something in my mind there, and when we went up to Great Ormond Street on that day, back in September 2014, all of those years ago, we were told the diagnosis and we were given some leaflets. And then we were kind of, you know, the appointment finished. We were sent home. And I was having a tidy up at home, a clear out, a couple of years ago, and in one of the kitchen drawers buried right at the bottom were these said leaflets that I had obviously hidden away when I got home.

They'd never come out of the drawer since. And it really took me back to that day and how, you know, some people on the day will want all of the information in the world about this. What does this mean? You know, how can I fix this? What can I do? You know, they want, whereas other people won't want to know a thing, they won't want to hear it.

They won't want to be a part of those conversations on the day. So, we have to, you know, take into consideration as well that everybody's different. So actually, we might come up with what we think is a really good protocol for giving a diagnosis and what should happen at the appointment. But actually, in reality, I suppose, the clinician, the consultant, who is the person delivering that news, needs to really read the room and think about what the best thing to do is in those circumstances. So, like you say, you know, “here's something with all of the information in, we can talk about it now if you want to. If you don't want to, that's absolutely fine, you can take it away with you. Many other people to date have had.” And that's really encouraging, um, for me as well because like I say, it stays with you for the rest of your life.

Amanda: I think what you're both speaking to is that balance between needing structure and flexibility, and that can pose a real challenge, I guess, in terms of funding NHS services for sure. But it can come down to even just those simple communication skills, which in theory, every healthcare professional should want to and be able to do really, really well.

But it's about understanding also the nuances around. Like you've both said, the lifelong and the familial nature of a genetic diagnosis. So, it's about reading the room and understanding how people will take in that information differently and how that affects different members of the family. So that definitely individual with that rare condition who may be a child or an adolescent or an adult, and also their parents, and also their siblings and their carers. So, there's a huge amount of people that need support and where genetic services are often used to kind of taking care of the family, that's not necessarily always been the norm for lots of other specialties who are really coming to learn a lot about how they could provide those services on a more familial level and thinking about the lifelong nature.

So, although you might be a paediatrician, the family in front of you is also still thinking about what the adult life is going to be like for their child or what they as adults or other members of the family are going to need to deal with. So, it's about needing to appreciate that kind of wider lifelong and family nature of a diagnosis and needing to be flexible around that. That's, I think, really important.

Shelley: I'd agree with that. We talked about earlier the internet and social media in particular, which has kind of given patients’ knowledge and power to become experts in their conditions in the lives that they have. How do you see the skills, this you know, knowledge and expertise that people have been exposed to. How does that bring added value?

Louise: I mean, from my perspective, I think increases in technology have been phenomenal for communities with rare conditions because if there are only five people with your condition around the world, now you can talk to them. You know, you can Skype them in Australia and you can speak to someone in America.

And that ability to reach out to other families who are kind of walking in your shoes and on a very similar journey to you is so important. And that just wouldn't have been possible in the same way before. The other thing, it's made it much easier for patient organisations to do it in the rare disease community is reach out to people all over the UK.

So before joining at Genetic Alliance UK, I ran one of our patient organisations as Chief Executive there and you know, we had really struggled to provide regional services because we were such a small charity. It's really hard to provide, you know, coffee morning in Dundee one week and you know, a coffee morning in Kent the next, you know, that's a real challenge when you've only got a couple of members of staff.

But actually, to be able to do that virtually once a month via bringing together everyone from a virtual coffee from all over the country who wants to join, to get to know one another, to share their experiences and, and come together. It's not the same as in person, but when you are supporting kind of 300 people scattered across the country, it can make a massive difference and you can get a group of people together fairly regularly to keep in touch with one another.

So yeah, from my perspective, that kind of technological support in terms of peer-to-peer support has made a massive difference.

Amanda: I just add that also another area or age group that's often overlooked are adolescents and young people. I think the internet and social media provides a really helpful avenue for that group to get support. You’re often dealing with so much in life transition already, and that's just compounded by having a rare condition or dealing with a cancer diagnosis, for example, or already starting to think about. What you're maybe having your first relationships and starting to think about, well, what's my future life looking like if I was thinking about having children and with my condition?

All of those questions are kind of starting around that time, and it's a much more digitally savvy age group who I think is really then able to benefit from these additional ways of giving support.

Shelley: From a mental health perspective as well, for teens who are, you know, starting to learn more about their own condition, sometimes leaving home is difficult, whether that's from a pain perspective or an inaccessibility perspective. So, to be able to connect with your peers online, through, you know, gaming's really popular and having those common interests. To be able to do that, obviously from the comfort of your own home is really, really valuable. Which that shouldn't be kind of underestimated sometimes, you know, teens don't want to talk to their parents.

I think also what's really important is, being able to talk to your peers about, you know, more intimate stuff like, you know, sex and relationships and, you know, if you do have a rare condition and it is difficult to, to go out and about and you know, being able to actually talk to people in the same situation as you about those things that you probably wouldn't talk to your parents about.

All of those things just really, really valuable and actually get people talking about. Real life stuff rather than thinking things are to taboo to talk about. So, you know, in the eighties, if you had this diagnosis, who would you have spoken to about, you know, non-parent stuff? Who would, or where would you have gone? There was probably nowhere to go for that kind of thing. So, from that perspective, social media's just really, really a brilliant concept.

So, as we look forward, how can families who are interested in research get more involved? In that side of their condition. You know, I suppose if we think about a clinical trial, some people they want to get on a clinical trial. They want their child to be on a clinical trial. They want to feel like they're doing something. Other people don't want the commitment of a clinical trial. They don't want the upheaval in their lives for a clinical trial. If you are a family who does want to know more about a clinical trial, what can families do?

Where can they go? And also, I suppose another question that links to that is how can we change the barriers to getting on a clinical trial? So, for example, clinical trial entry criteria are really quite specific. So, it's this sex of person with, you know, this genetic change. They have to be between these ages. They have to be able to walk this far. They have to be able to do - although we do understand why they're in place, they are real barriers for lots of people. And when you have got a small population to start with actually getting people involved and finding people is really difficult. So, what are organisations like Genetic Alliance UK doing to influence in this space and trying to make changes to policy and respective those hurdles that people face?

Louise: So, we do quite a lot of work in this space. First and foremost, kind of doing some work actually with the pharmaceutical companies to try and make sure that wherever possible they have someone with a patient perspective, or a carer perspective involved right from the beginning when they're devising those trials.

To think through some of those issues you've raised Shelly, which are absolutely right about how can they make the trials inclusive as possible and extend the opportunity to try a new treatment to as many people as possible. Um, and we've definitely seen a lot of progress in that in the last few years.

Companies are much better at getting patient organisations and individuals involved at an early stage in developing their clinical trial protocols. The second challenge is undoubtedly kind of making sure that families are aware of the opportunity to get involved with particular trials, and that is really difficult.

So, um, we're really used to doing research with things like clinicaltrials.gov to find out whether there are trials for particular conditions and if there are, whether there are trial sites in the UK. But that's a really roundabout way of people finding out about them. So, we are doing some work at the moment.

In fact, we've got be part of research, which is a platform which encourages people to get involved in clinical research. They're trying to make that platform more patient friendly for people from the rare disease community. And they're actually coming to talk to all of our members in two weeks’ time about how they can get people involved in testing their new site to see if it's going to work for people with rare conditions.

So, if they are able to develop it in a more user-friendly way that makes those opportunities clearer. That'd be really helpful. The third bit of the jigsaw, so it is a real challenge, is trying to ensure the NHS continues to be a really kind of friendly place for clinic clinical trials to take place.

And that in itself is a real challenge at the moment. Brexit didn't help in terms convergence between. Rules in the UK or the potential for convergence between rules in the UK and rules in other European countries and for rare disease conditions. Obviously, the trials are generally multi-country because there aren't enough people in the UK to kind of fill a clinical trial cohort.

So, we need to make sure we're coordinating with Europe and with the rest of the world. So, there are whole load challenges at the moment. But there is also a whole load of opportunities because there are more and more companies developing new treatments for people with rare conditions. So, we are really actively engaged in trying to address some of those challenges and make sure that we can provide as many opportunities as possible for people to take part in trials and know when they're taking place and to get involved.

Shelley: I think also there's the support side of being on a clinical trial as well. So yes, we need resources to deliver trials in hospital settings, and we need qualified nurses and, and you know, trial coordinators. But we also need support for families on clinical trials as well. So, you know what to expect as time goes along.

And also, I guess in the circumstances of where maybe a clinical trial is unsuccessful, how a family's supported through that, because that can be quite a difficult time, quite an upsetting time when people have pinned hopes on, you know, a, a treatment becoming available because they've put their child through this, and they want the outcome to be favourable.

But obviously the nature of clinical trials that isn't in the majority of trials, that isn't the case. And sometimes I feel the support for those families isn't there. So, as well as, you know, lobbying for greater populations and more relaxed, entry criteria, it's, it's really the outcome as well at the end. What do we do there too?

Louise: Absolutely. And I think that's why it's so important to have people from the patient community involved in designing the clinical trial protocols up front so that all those data have been thought through. And you know, we often see our member charities challenging, you know, companies who are saying, right, we want, you know, we want take blood tests every week. Well, actually that's going to be a huge burden for the family. Do you need it every week? Would once a month be enough? Have you thought through what that's going to mean for pulling a child out of school if you're working with a paediatric population?

So just bringing that real common-sense voice right from the beginning can help people think through the, all the different stages of that journey and how it will actually impact the lives of the people who are, who have given up their time to take part in the trial.

You know, it's a huge burden for families when they do that and. As you say, Shelly, people hope they're doing that because it will have a positive result. So, managing that expectations and being realistic with people about what likelihood of that is. It's really important. And it's also listening to families at the trial design stage.

Shelley: So, you know what's actually important to people, uh, what do they want to get from this treatment? Because what pharmaceutical companies think people want is not actually, you know, what people want. You know, in the case of my son who's been a wheelchair user his whole life, for me and probably him, it's not about him getting up from his wheelchair and being able to, you know, run a hundred meters. That's not really what I want out of a clinical trial. I want him, you know, looking forward, when he is 18, to be able to go to the pub and lift a pint up to his mouth. Or I want him to be able to get his own hair or, uh, you know, really small things like that. And that is actually really meaningful to people's lives.

And I think sometimes that's missed. And I know lots of, lots of charities and organisations are, are banging this drum. All of the time, and it just doesn't feel like we're being listened to.

Louise: Helping their child and the whole family to sleep better. That to them was an outcome that really mattered. You know, not having seizures in the night so the whole family wasn't being woken up. Those are the kind of things that companies aren't thinking about, but for families it can make a massive difference. Some of the, the outcomes that really make a change to people's lives for the better.

Shelley: When we look forward to the next 10 years, what do you see Louise as being the important role for charities to play for those with genetic conditions and their families? Where do we go from here?

Louise: So, from my perspective, I think we've had incredible success around the scientific developments and that's fantastic and we shouldn't take that for granted. You know, I don't want to be kind taking that for granted because it's been such huge progress made so swiftly. So that's absolutely incredible. And I think the challenge for the next 10 years for organisations like Genetic Alliance UK and the challenge for supporting, the challenge is to kind of trying to spread that now through, you know, making sure that the science is working great, kind of, how do we make sure that moves through to make sure people are getting a diagnosis both quickly and delivered well and sensitively. And Amanda, I think you've talked about that really eloquently. And then how do we make sure there's a clear pathway for people with all the different conditions that are diagnosed and also where a diagnosis is not possible. What is the pathway going to be like to support people throughout their lives?

Because we're going to be getting more and more people through genetic diagnoses, which is fantastic due to the science. There are more people with genetic conditions living for longer because of the advance in medicine we're seeing. And that's brilliant as well. But it means a responsibility for all of us to work with the NHS to think through what that means and how that support can be better structured and delivered and may develop both to, to help people live their lives to the full. So that for me is the challenge to kind of see that journey onwards.

[00:42:25] Amanda: Yeah, and I think there's a very important role, again, that genomic signalling can help to facilitate here in terms of identifying what some of those research priorities can and should be in bringing our participants together with those research groups and charities who are often championing the research that should go on and what the outcomes of that are.

Not just doing the research, I think, but bringing it back to communication skills again, which I think are so important. How we communicate those research aims and priorities clearly and transparently is so crucial because that's what helps sets expectations around things like this is why doing this piece of research is so important. This is how we came to this conclusion. This is why there might need to be some exclusion criteria sometimes, or this is what we hope to gain and how we anticipate it will help members of this particular community and what we might be able to do in the event that this may not work because this is research. So, it's again, having those, those avenues for communication and the flexibility to recognise that people's life journey with those rare conditions or with cancers will change the more we're able to do more research and bring in more therapies and treatments and support. So, with those advances and opportunities will have to continually shift our thinking about what the role of rare disease charities are to continue to support those families who are going to be going on different journeys.

Louise: Yeah, I think that that, just to build on that, cause I think what I'm trying to say not very eloquently is, is our challenges territories is both to help shape the research and to help make sure that the research is addressing the things that matter most to people with living with rare genetic conditions in their families, but also to make sure that the benefits of that research actually reach patients and their families and really improve people's lives in the longer term. So, it's a kind of dual challenge from a charity perspective.

Shelley: Lots of work to do. Lots of work to do together as well. You know, we are, as individual groups, very small with our rare conditions, but together we are an army. So, let's look forward and see what we can do together. I just want to thank you both for talking to me today.

Shelley: I've really enjoyed the conversation, listening to your different perspectives, and I hope the listeners enjoy our discussion as well. So, thank you both very much. Thank you.

Louise: Thanks for the opportunity.

Amanda: Thank you for having us.

Naimah: Thank you for joining us for today’s episode. That was Shelley Simmonds speaking to Amanda Pichini and Louise Fish. If you enjoyed this podcast and would like to hear more like this, please subscribe to the G Word on your favourite podcast app. Thank you for listening.