**Shining a light on rare conditions**

**The G Word transcript**

**Naimah: Welcome to the G Word.**

[Music]

Sarah Crawford: But I would also say it’s okay to grieve the child that you didn’t have that you thought you were going to have. I just think that’s so important. And I think for me, the most difficult thing in the early couple of years was feeling like I couldn’t do that because nobody appreciated that I’d actually lost anything.

[Music]

**Naimah: My name is Naimah Callachand and I’m head of product engagement and growth at Genomics England. On today’s episode, I’m joined by Shaun Pye and Sarah Crawford, who are parents of Joey, who was diagnosed with DYRK1A syndrome at the age of 13, and Sarah Wynn, CEO of Unique, a charity which provides support, information and networking to families affected by rare chromosome and gene disorders. Today, Shaun and Sarah are going to share Joey’s story, and discuss how their role in writing the BBC comedy drama There She Goes has helped to raise awareness of people with rare conditions in mainstream culture. If you enjoy today’s episode, we’d love your support. Please like, share and rate us on wherever you listen to your podcasts.**

**So first of all, Shaun and Sarah, I wonder if you could tell us a bit about Joey and what she’s like.**

Shaun Pye: Yes. So, the medical stuff is that she’s got DYRK1A syndrome, which was diagnosed a few years ago, which means that she’s extremely learning disabled, nonverbal.

Sarah Crawford: Yeah, autistic traits.

Shaun Pye: Eating disorder, very challenging behaviour. She can be quite violent. She can be quite unpredictable. Doubly incontinent, let’s throw that in. She’s 17 but she obviously has a sort of childlike persona, I would say, you know. She sort of likes things that toddlers like, like toys and that sort of thing. But that’s the medical thing. What’s she like, she’s a vast mixture of different things. She can be infuriating, she can be obsessive, but she can be adorable. Occasionally, she can be very loving, especially to her mum.

Sarah Crawford: She’s very strong willed, you know. Once she knows she wants something, it’s impossible to shift her, isn’t it? So, she’s got a lot of self-determination [laughter].

Shaun Pye: So, her obsession at the minute, or it’s fading slightly, which is quite funny, is that she’s become obsessed by – there’s a toy called a Whoozit that she loves, but she became obsessed by the idea of – she was typing buggy baby Whoozit into her iPad, so that’s how she communicates. She’s got quite good literacy skills.

Sarah Crawford: Yeah.

Shaun Pye: And we figured out eventually that what she wanted was she wanted her mum to take her to the park to find a buggy with a baby in it that also had a Whoozit in it that she could steal, and when Sarah explained to her at some length that it was not yours, she would say, “It’s not yours,” that drove her insane with excitement, at the idea that she could steal another child’s toy. So, it’s a good example of her because it’s funny, and, you know, it is funny, and she’s so cheeky about it and she flaps her hands, she’s very hand flappy, and she sort of giggles and she gets really excited, but, you know, the 2,000 time she asked to do that, and we have to walk to Mortlake Green near our house, and to the point where – again, it’s funny when it happens, but you get to the green and she doesn’t even look for the buggies anymore. So, that’s an example.

But she’s a lot of different things, you know, and I suppose the thing that is dawning on us at the minute is that she’s 17, she’s going to be 18 very soon, and, you know, the list of presents that she gets on her birthday is always the same, ‘cos she’s autistic. So, at Christmas, she always gets the same presents. But the idea that, for her 18th birthday, we’re going to have to buy her children’s toys and – you know –

Sarah Crawford: Toddlers’ toys.

Shaun Pye: Toddlers’ toys and everything, it’s sort of hitting home, but that’s something – a bridge we’re going to cross on July 27th [laughter].

**Naimah: Yeah, I can imagine that’s quite a difficult bridge to cross, but it sounds like, you know, Joey’s got lots of personality and you have lots of, you know, lovely times with her as well. I wanted to go back a bit before the diagnosis. So, you mentioned Joey’s been diagnosed with DYRK1A syndrome, but can you tell me what it was like before you both – and a bit about your journey, and when you suspected maybe something might be wrong and what you did first of all.**

Sarah Crawford: I mean, there were hints that things might be wrong before she was born. The measurements were such that they thought there might be intrauterine growth restriction, because basically my belly wasn’t as big as it should be for dates, and that was obviously the working hypothesis. And they actually did a scan, an ultrasound quite late on in the pregnancy, which I picked up when I looked at the report was showing a small head measurement. And I remember querying it with the consultant, who said it was probably measurement artefact and nothing to worry about. But after she was born, she wouldn’t latch on, you know. We had to switch to bottle feeding straight away. She was small, and the head measurement actually was small. You know, you could see on the very early one, they must have taken it kind of three times to try and get it bigger, probably angling the tape measure, and it had been crossed out and rewritten. That was the pattern. So, her head simply did not grow in those early days in the way that you would expect.

So, I was wildly anxious about this right from the get-go, and very adamant very early on that I thought that, you know, she was learning disabled. And to be fair, you know, the GP took that seriously. You know, at the six-week check-up, things weren’t quite how they should be. We got in the system very early on, saw a paediatrician really quite early. So, I was, you know, fairly convinced very early. I mean, I’m a clinical psychologist, I’ve got training in learning disability, a bit more clued into these sorts of things I guess than the general public on the whole, and I think the bigger challenge for us wasn’t so much the attitude of, you know, the healthcare system. It was more trying to debate this with family, who were very much of the, “There’s nothing wrong with her,” kind of mantra.

Shaun Pye: She wasn’t our first child, so we had experience – and all children are different, but because we had that comparison – all children are different. Obviously, there’s not a set thing. But we had a benchmark in our own minds and hearts sort of, to know that she was missing things that he’d hit, and something wasn’t right. And the parental thing – basically, we’re talking about grandparents – it was sort of – there were two approaches that they took, one of which was to tell us nothing was wrong, because they couldn’t bear the idea that me and Sarah were in pain or unhappy. It was just out of pure love. It’s just a natural human reaction to say, “I’m sure everything’s going to be alright.” They were just trying to be supportive because that’s, you know, what they thought they should say.

And then the other approach from other members of the family was again from just a supportive, loving aspect, but it was a sort of, “They just need a bit of tough love, pull your socks up. Lots of children are different and you just need to learn ways of dealing with it.” And the way I describe it nowadays is that they’d mistaken Joey for someone on the far end of a spectrum of abilities or behaviours, whereas she wasn’t really on that spectrum at all. She was on a different spectrum [laughter]. She wasn’t a difficult child. She wasn’t a naughty child, was she?

Sarah Crawford: No, no, she was a child whose brain hadn’t developed.

Shaun Pye: She was a very, very different child. So, all of that has gone on over the years. And genetics wise, we had early genetics testing. Kingston Hospital took quite a lot of interest early on, and then they sort of didn’t take any interest [laughter].

Sarah Crawford: No, it’s more that they ran out of technology, so they couldn’t pinpoint the diagnosis with the technology they had. I mean, the geneticist was excellent, wasn’t he? We really loved him.

Shaun Pye: Yeah, Sarah’s going to like this, ‘cos I’m about to say I love geneticists ‘cos they’re – on the spectrum of doctors, I love them, ‘cos one of the guys we saw just looked at it like a puzzle and he was sort of excited to solve it, and he really wanted to work out – and in a way, you could have walked away from that thinking, “We wanted the bedside manner and we wanted the, “Oh, that’s terrible,”” whereas he really was just a sort of – he was terribly excited about the whole thing, and he wanted to solve a Sudoku, basically, yeah. But me and Sarah walked away from that just thrilled, ‘cos we’re the same [laughter]. There’s not a Sudoku or a crossword that we don’t love finishing. So, we walked away thinking, “These are exactly the people we want involved.”

And so when I say they gave up, that’s not fair. They just ran out of – you know, they can’t spend increasing amounts of NHS money. So, they tried – you know, different genes were mentioned.

Sarah Crawford: Yeah, they tested for a whole load. I mean, his attitude was right from the get-go, you know, based on the history, everything else that had been ruled out, dysmorphic features, those kinds of things – I don’t know if that’s the terminology they would use now – but that this was going to be a chromosomal disorder, and that they would do the tests that were available, but that it was possible that those wouldn’t pinpoint in, but that the technology was changing all the time, and that if they didn’t find it now, they would in the future. And that was how it played out.

Shaun Pye: There was one meeting that I did get a little bit – having said that, I got slightly – but you didn’t – about one of the geneticists, who sat there and said, “We’ll do this test and this test, and if they come back with any interesting things then we can get really excited.” And he used the word excited, and I was sort of a bit, “I’m not that excited by all of this.” But actually I calmed down quite quickly, and in hindsight I really wanted someone to get excited by the idea of working out – but then a long time went past. I wouldn’t say that we lost interest in finding out what her genetic condition was, we didn’t. It’s just it’s something that became less and less – it wasn’t like a holy grail for us. But then the opportunity came along with 100,000 Genomes, and we signed up immediately, and then they did that and it was a few years before that went through the system.

And then out of the blue really, we were asked to go and see a geneticist, and we had no idea that this is what it was. I honestly thought it was just a routine sort of, “We’ve got a few more theories,” or something, and she just said, “We’ve found out what it is.” And that moment is – well, we tried to describe it in the TV programme, but it’s quite hard to describe what goes through your mind when, after 13 and a half years, somebody suddenly says, “Oh, by the way, that thing that happened with your daughter, we’ve worked out what it is.”

[Music]

**Naimah: I wonder if you could talk a bit about what the diagnosis meant for you both.**

Shaun Pye: It was sort of different for both of us, wasn’t it? I was a bit more excited, Sarah was a bit more…

Sarah Crawford: My attitude early on was that, while the label would be nice to have, it wouldn’t make any material difference to anything to do – I mean, it was never going to be precise enough that it would give a map out of what we’d expect for her as an individual, and it wasn’t going to change the fact that there was a severe learning disability. It wasn’t going to change the challenges that we would have over things like schooling, therapies, you know, what the future holds for her. It is useful to have it, but it doesn’t really change the day to day.

Shaun Pye: But what it did change, and this is where Unique is so brilliant and important, is that it puts you in touch with people who have children with a similar condition. That’s the main takeaway from getting the diagnosis. ‘Cos Unique is great, and obviously in a broad sense it’s great, but to actually meet people and be in touch with people whose children have DRYK1A – so, I’ve met quite a lot of them now and I’ve met quite a few of the children. There was a meetup last year, and you just walk in and you just go, “Oh my god, oh my god” [laughter]. Literally girls running around, just the same as Joey, just the same, and the different ages as well. So, there were some in their twenties and there were some just starting out on their – who’d only, you know, very young, been diagnosed. But just to see your life just in front of you [laughter] is very useful. So, that’s the basic takeaway, I would say, from the diagnosis.

**Naimah: Yeah. It must have been really nice to be connected with those other parents and to kind of share experiences as well.**

Shaun Pye: It was, it was. And this applies to most – well, every family from Unique that I’ve ever encountered actually. Nearly all of the DYRK1A – ‘cos it’s spread around the world as well, so, you know, there’s slight cultural differences, but just to see that they are all of a very similar mindset is comforting, ‘cos it sort of makes you think, “Actually, we haven’t been doing this wrong.” It’s a sense of humour thing. It’s an attitude to the world. It’s the way they see their children. It’s the way they see the outside world. I’m not saying we’re all uniform, of course we’re not, but you can see it. When you talk to them, you can just see that they have the same sensibilities as you about the whole thing, and it’s sort of quite reassuring really that, you know, we’re not outliers.

**Naimah: I just wanted to go back to, you know, when you were talking about the bit before the diagnosis, and I wanted to come to you, Sarah, to ask, you know, Shaun and Sarah both described their journey with a lot of uncertainty, but I wondered, could you tell me a bit more about the role Unique plays in this part of the journey for parents?**

Sarah Wynn: Yes. Well, actually I think Shaun’s done such a good job of summing up why Unique exists already, thank you, Shaun. But I think really what we’re aiming to do is to try to alleviate that sense of helplessness and being overwhelmed, and isolation that often families feel when they have a child that’s got additional needs. I think our experience with our Unique community is very similar to that that Shaun and Sarah have described. So, many parents know that there is something – that their child isn’t developing as they would expect. And we hear lots and lots of stories of families going to healthcare professionals and actually not being taken seriously, or like Shaun and Sarah were saying, you know, everybody saying, “No, they’re just a bit delayed, it will all be fine.” And so I think that’s a common experience of many families, that the parents inherently understand and know their child better than everyone else, and it’s very common that families have to wait quite a long time to get to that point where they get to a diagnosis.

And often I think the uncertainty continues after you get that diagnosis, because as Shaun and Sarah said, you get a diagnosis of a rare condition and actually there just isn’t that much information available. So at Unique, we try to help in various ways. One is by connecting families with other families, and that might be other families who’ve got the same condition, but it might also be families who are just going through the same experiences as you are, so you’ve got someone to share your journey with. And the other thing we try to do is to help families understand the kinds of genetic testing they’ve been offered, and a bit about the results of genetic testing. Because of course genetics is something that lots of people haven’t thought about since school, and actually quite often hoped they never had to think about again.

Although we’re a group supporting families and patients, actually a big part of what we’re doing is around translating those complicated genetics terms, and trying to explain them to families, so they can understand the testing they’ve been offered, the results of testing, and really what the benefits and limitations of testing are. Sarah said, often you get a result and a diagnosis from genetic testing, but that doesn’t give you a magic treatment that’s going to cure your child. It’s really important, for all the reasons Shaun and Sarah have already said, just knowing why it’s happened, being able to connect with others, being able to meet others, but actually often it doesn’t necessarily change treatment.

Shaun Pye: I guess one thing I would say, just ‘cos it was important to us, and it’s de novo in our case, but that’s comforting to know. We always suspected it was and we were always told it was, but to have that confirmed means – I mean, we’re not going to have anymore children, but it’s more to do with our son and whether there’s something inherent that could be passed on.

Sarah Wynn: Yeah, it gives you information that you can use for either your own family planning or other family members.

**Naimah: You mentioned that Joey received her diagnosis via the 100,000 Genomes Project. How did that come about?**

Sarah Crawford: I think it was offered, as in the 100,000 Genomes Project was the only way that that was potentially available at the time, that this was effectively a project that was going on to try to answer those unanswerable questions with the technology they had at the time. I mean, it was years between us enrolling in it and getting the answer.

Shaun Pye: It’s so important to me in hindsight the diagnosis, just for all the reasons that we’ve been discussing, but without doing down the role of genetics, there was a period of Joey’s life when we thought we’d run out of road with the testing, and it wasn’t something that really I was obsessed with or occupied my mind massively. It wasn’t like me and Sarah were saying, “We must get back to Kingston Hospital. We must get back to the geneticists. We must write to the NHS. We must insist that they do this.” We’d sort of resigned ourselves to the fact that they’d done all that they could and they hadn’t found it, and that’s what it was going to be. Having said that, when 100,000 came along, we obviously jumped at the chance. We had no misgivings about it whatsoever, ‘cos I think we’d resigned ourselves to the fact that we might never know.

Sarah Crawford: I think I thought that at some point we would, because the technology, the methodology that they’re using obviously was changing all the time, but it didn’t preoccupy me because I didn’t think it would make a massive amount of difference. It probably made a bit more difference than I thought it would, for the reasons that Shaun and Sarah have said, about, you know, particularly the sort of connecting with others, you know, just realising how useful it is to be able to hear about the similarities and differences that other families experience.

Shaun Pye: I think a key point for us, and I’m sure this is true for the vast majority of Unique families, that we never thought that there was a cure. We never ever, ever, ever, ever, ever thought there was. And nobody in our family did. It’s not like anyone was saying, “Oh, with this treatment or that treatment…” Once you know that it’s DYRK1A, there’s obviously things that you can tailor towards her in terms of therapy, you know, there are things that you can do, but we were never under the illusion that if we found out what it was, she could go on and some sort of drug would suddenly make it better.

Sarah Crawford: Yeah, we’re not queuing up for experimental stem cell treatment [laughter] in weird and wonderful parts of the world, you know. What’s happened has happened. Her brain didn’t develop properly in utero and beyond. There is no changing that.

**Naimah: But I guess with diagnosis, and like you said, if you can get some relief from some of those other symptoms that are caused by it, then, you know, that’s some sort of relief for Joey and a bit of help.**

Shaun Pye: Yeah, there are absolutely concrete things that you can learn that will – Joey will never be better, but talking to the other families – eating disorder, that’s one of them. Constipation, that’s another thing. But hearing their experiences, hearing the roads they’ve gone down, finding out that there’s, you know, a unit somewhere in the country that specialises in this, that or the other, these are concrete things. It’s not just about emotional support. It’s absolutely about practical support. But there’s no magic wand, but there are things that, you know, we’ve learnt that can help.

**Naimah: And then Sarah, to come to you then, do you find that families find it difficult to seek out help from Unique once they’ve received a diagnosis, or are they likely to come quite quickly to you? What’s your experience?**

Sarah Wynn: It’s a really good question, and of course we don’t know the ones that never find their way to us. But what we try to do at Unique is to be sort of warm and friendly and welcoming, so that it’s not too daunting. ‘Cos I think all of these things are an extra thing for parents who are already busy and dealing with lots of medical appointments and therapies, so we try to make it as easy as possible to join us. Many, many families do join us at that point of diagnosis, because that’s when they’re looking for more information. Actually, you can get in touch with Unique and if you decided you didn’t want to join us, that’s also fine. So, we have a helpline that you can call. And for some people, joining a support group just isn’t their cup of tea, and that’s really fine. Other people find us a little bit later on, you know, perhaps when their child starts school or, you know, there’s sorts of crunch points where people are looking for extra information or support that they tend to find their way to us.

But one of the things we try really hard to do is to get the word out that organisations like ours exist, so that we can be contacted if people want to. And lots of our families come, like Shaun and Sarah, after the geneticist has told them that we’re there. So, that’s a really important thing for us is that everybody knows we’re there. You can join us and involve yourself as much or as little as you want. So, as we’ve already talked about, one of the things we do is put families in touch with each other, but not all families want that. So, you know, you can join and remain no contact, and stay quietly under the radar if you’d like to. But those people often want their child to be sort of counted in the system, you know. When you say how x number of people have DYRK1A, they want their child to be in that number even if they don’t want to go to the meetups, or they’re not quite ready to do that. And of course people change. So, some people join us and think, “We’re just going to quietly sit here for a bit,” and then change their mind a bit further down the line.

I think, although There She Goes, and what Sarah and Shaun have said about their journey is really similar to many people’s journeys, of course everyone is a bit different, and so people want different things at different times. And what we try to do at Unique is to be those things for whenever families need us.

**Naimah: Yeah, that must be really reassuring for families, knowing that they can come to you whenever they feel ready to more than anything.**

Shaun Pye: Just to jump in quickly as a sort of user of Unique, from the sort of different perspective from Sarah, that is literally how the service presents. That’s not an ideal that they aspire to. That’s what it’s like. So, I can confirm that – I mean, people think different things, and within our DYRK1A group, for example, you know, there’s a broad range of people who think various things, but the one thing about it and Unique is it’s very well self-policed, so people know how to behave. You won’t be subjected to ill informed sort of medical nonsense. It’s very well self-policed, but it’s also very, very occasionally – I’m speaking for the DYRK1A group – the example they gave me was around covid and vaccinations, and, you know, people have very strong views about it, and these forums aren’t the places to be having that sort of discussion.

Sarah Wynn: I think that’s exactly it. One of the ways families can connect with each other is via an online forum, and generally we take quite a light touch in moderating it, because the forum is for the families, and we want them to feel ownership and that it’s their safe space. But yeah, ever so occasionally, it needs just a tiny little bit of input. But yeah, I think Shaun’s right, everybody’s there for the same reason, and that’s to kind of share experiences, sometimes vent about the world, ask questions, and actually celebrate things that other people might not see as such a celebration. You know, lots of our families, their children might be late to walk, and it’s a place where you can celebrate all of those sorts of things as well.

[Music]

**Naimah: So, next I want to move on to talk about There She Goes. So, you mentioned it briefly there, Sarah. So, this is the BBC Two comedy drama, for which Shaun and Sarah were both writers on, and it really draws upon your real-life experiences of caring for Joey. And although the series is posed as a gentle comedy, it also displays really frank and honest emotions experienced by Emily and Simon, who are the parents of Rosie in the programme. Let’s listen to the poignant clip from the series by Jessica Hynes, who plays the mother, Emily.**

Emily: You know, when you’re younger and daydream about what family you might have – so, I was the girl who thought Claire always got away with murder. Or when we found out Ben was going to be a boy, if it would be like you and Soph, you know, dorky older brother, biffy outdoor sister who everyone liked, you know. But in none of my dreams was there a girl who… Yeah, who was like Rosie. Yeah… No one ever dreams of a child like Rosie… You know, and I… I love Rosie, but why do I have to be defined by her? You know, for a long time, I felt cheated by her, because she wasn’t the girl that I dreamt about, you know. She’d taken her place. And then as she got older and I accepted her more, you know, what if it wasn’t that she’d taken her place, what if she just pushed in the queue and then if we started again, then if I had, you know, a normal girl, and then I wouldn’t have to… I wouldn’t have to resent Rosie anymore because I’d have the family that I’d always wanted, and I’d have – I’d have Rosie as well, yeah. [Sobbing] Just after all these years, haven’t I earnt that?

[Music]

**Naimah: Off the back of that, I wondered if you could both tell me a bit more about what it meant for you being able to write for the programme and, you know, what it’s meant in the aftermath as well.**

Shaun Pye: So, it came about - I basically am a TV writer and Sarah’s a psychologist, but it came about primarily because I was trying to think of something to write about and we realised that Joey’s just an incredible character. Those sort of children aren’t featured on mainstream television really at all, I would say. And so we thought it would be an interesting thing to do. But from that sort of slightly selfish motive, I wrote an episode, and Sarah read it and said, “You’re not doing that, it’s not honest enough” [laughter]. So, Sarah came on board as a writer with me and we cowrote it. The whole thing’s cowritten. And it’s the most important piece of work I’ve ever done, I ever will do, and it became far more than just a TV programme.

The first series went out and we had a screening, and Unique came to the screening, along with some of the other charities, and we were so terrified of what the response would be. And the fact that the response was what it was, which was overwhelmingly, “It’s like looking at our own lives on television,” it was recognition. It was nothing to do with whether the stupid jokes were funny or anything [laughter]. It was purely whether – if anyone had turned round and said, “This has got nothing to do with what it’s like bringing up our child,” or our brother or sister or whatever, that would have been quite bad for us, but it wasn’t, and that’s been the overwhelming response since. It’s, “Thank you for putting our life on television, ‘cos it’s not normally on television.”

So, it became that, and so the second series was even more about that, and then the special that we did was almost totally aimed at, we need to tell these stories because there are so many people in this country who this story isn’t being told for them. And it so happened that Joey hit puberty and had some very, very, very problematic behaviours, sort of self-harming behaviours, it happened quite close to her being diagnosed, so we thought this story is just written for us. Joey’s written it for us. So, we just sort of wrote down what happened. That was sort of what it was. And then obviously the response to that was very good. So yeah, and we wanted to feature Unique ‘cos that was such an important part of what we’d been through.

So yeah, it went from me wanting to further my career to that having nothing to do with it, and me wanting to [laughter] tell the story of children with rare chromosomal disorders and learning disability, and that’s what it became.

**Naimah: I’m sure it must have been almost quite cathartic, I imagine, in a way, to share your story that way, and also, you know, give you a real sense of accomplishment to be able to kind of share your story on that platform. Like you said, like it’s never been done before in such a way, and to get that kind of response from other families, it must have really just helped you both in your journey as well, I can imagine.**

Shaun Pye: For me, because it’s what I do for a living, it still retained a certain sense of my job. And, you know, emotionally, obviously, entirely committed to it. All the bits that make you sort of cry, or all the bits that are like, oh my god, Sarah wrote – I wrote all the stupid bits that David Tennant says [laughter]. So, I think it was more cathartic for you. You really had to dig deep into some quite unpleasant memories [laughter].

Sarah Crawford: Yeah, it wasn’t always the most comfortable process, you know. We’d sort of agree – I mean, particularly in the earlier process, we’d sort of have a little think about what we wanted to talk about, and then I’d go off and like kind of delve deep into memory, and just type a stream of consciousness, and I’d be sitting there sobbing [laughter], you know, with tears rolling down my face, you know, just reliving these really awful experiences. But yeah, I think the end process ended up being cathartic, and a lot of that was stuff that I would never have imagined sharing with anybody [laughter], let alone, you know, this huge audience of people, which – yeah, strange how things evolve.

Shaun Pye: Yeah, I think possibly if we hadn’t done this then we might have just tried to not think about these things and not bring them back, and I think we probably wouldn’t have spoken to each other – we may have, I don’t know. I don’t know what would have happened. But I don’t think these things would have come out into the open. And very interestingly, another side aspect of it in the catharsis way is the effect the programme had on the wider family. There were certain members of the family who were really shaken by that programme, really shaken, because they had a set view. Even as Joey got older, they had a set view of the history and what had happened, and they were really shaken by the idea that their – out of love again, there’s nothing bad here, but they were really shaken by the idea that their actions had a detrimental effect on us when Joey was born.

You know, there were people saying, “Well, I didn’t say that there was nothing wrong with her,” and, “I didn’t say this or that,” but actually when you see it presented in the programme then there was a lot of re-evaluation that went on, in a good way, in a positive way and it’s all good.

Sarah Crawford: I think there’s something about seeing it, you know, and especially given, you know, we were so fortunate with the cast because they’re so good at portraying it. And I think there’s a power in seeing things played out rather than just hearing about them in the abstract.

**Naimah: Yeah, definitely. I definitely had moments of crying and laughing, and a range of emotions while I was watching it, so yeah, definitely very powerful. And I guess it’s really great for other families going through similar circumstances, for their families to see what’s happening and, you know, there’s a lot that can be learned from the programme as well. So, you know, it’s, yeah, really a powerful piece that you put together.**

Sarah Wynn: I would really like to echo that. I think Shaun and Sarah have said before that they didn’t do it to represent everybody’s experience, but actually that is exactly what it has provided. I would say that huge numbers of people are really grateful that that portrayal is there, so that they can be seen and heard and understood so brilliantly. But it has provided other families with the opportunity to show it to their friends and family, so that they understand their life as well. And so I think it’s had a hugely positive reaction from our Unique community. And I think it’s not always an easy watch, I think lots of families would say it’s challenging to see it up close in front of you, but I think it’s really cathartic and has been just incredibly powerful at showing these sorts of stories, which, as you said, just don’t get shown very often.

And I think particularly when we think that rare conditions, although they’re individually rare, if you put all of the rare chromosome conditions together, they’re not actually that rare, so these are stories that are going on up and down the country and all over the world.

Shaun Pye: Just to follow up on something Sarah said earlier on about, you can take as much or little as you like from Unique, it’s the same with the show. I’ve had lots of people get in touch with me or talk to me in person and say, “I’m really sorry, I tried to watch ‘There She Goes’ and I can’t watch it,” and I have to say, “Don’t apologise, you have nothing to apologise for. You take what you need from it. If you can’t watch it then don’t watch it. If you can watch it then do. There is literally no right or wrong way of doing this. There really isn’t.” But having said that, the nicest comment – well, one of the nicest comments I’ve seen was on the DYRK1A forum. It was someone who casually referred to it as “our show,” as in the DYRK1A community, it belongs to them, and that – yeah, a little tear, a little tear went down my face [laughter].

**Naimah: Yeah, that must have been a lovely thing for you to read. That’s really nice.**

Sarah Wynn: Also from the Unique and general people who have rare conditions community, it’s been so fantastic for raising awareness about genetic testing and rare conditions in general, and, you know, there just isn’t – because these stories don’t get talked about or shown about very often, it’s been really great from that point of view as well.

**Naimah: And hopefully this will be the catalyst for similar programmes and, you know, more things in the mainstream media as well. And you did touch on it briefly there, Sarah, about, you know, what the programme’s meant for Unique, you know, and the Unique community being very supportive, but have more people reached out to Unique since the programme?**

Sarah Wynn: I think the main takeaway is that being heard, “Our family’s being heard and represented,” which I think is really important. But yes, we’ve got lots and lots of new families that have come to us through watching There She Goes. And it was really fortuitous that when the special aired last spring/summer, it was the evening before our awareness day, which I think was a complete coincidence but actually turned out to be really great timing. So, we got lots and lots of new families get in touch with us, many of whom then went on to join us. But actually what it also did was get lots of members who’d been members for a long time but perhaps had been a bit quiet, or hadn’t been in touch, so it sort of also reinvigorated that engagement from other members who we might not have heard about for ages, and who might have got older children and had been in touch at the point when they were diagnosed, and then hadn’t been.

So, it has just been such a brilliant, brilliant experience to have Unique as part of it. And I think that’s really important. At Unique, we have members from 120 different countries, and the reason is that when you have these rare conditions, you’re really unlikely to find someone in the same town as you, possibly not even the same country with some rare conditions, and so the idea that you can connect with people all over the world I think is really important, particularly in rare conditions.

**Naimah: Yeah, that’s great, and hopefully, you know, it just continues to increase support with Unique and, you know, families know they can still come to you as a resource and as that continues. So, I just wanted to kind of wrap up here and come to the final question. So, you know, your story highlights a lot of challenges, a lot of difficulties, a lot of ups and downs, but I just wondered, Shaun and Sarah, if you had any advice for other parents going through similar circumstances.**

Shaun Pye: Yeah, I think one of the things is what I just said, which is I would tell people there’s no right or wrong way of doing this. I would say, from my experience, don’t be hard on yourself, and you’re going to think that you wish it never happened to you and that’s fine. That is absolutely fine. That’s normal. We’ve all thought that. It doesn’t make you a bad parent. It makes you a normal human being. I would say to get in touch with Unique. I shied away a little bit from help and charities, ‘cos I think it was a sort of pride. I think I had a preconception that it would be glass half full, put on a happy smile, best foot forward, blitz spirit sort of. We have encountered it a little bit over the years, not very much, but we’ve encountered a little bit of, you know, “As long as you love them, that’s the most important thing,” and, you know, which is fine and that is an okay perspective to have, but there are times when it’s just not what you want to hear. I want to be allowed to feel the feelings that I’m having without feeling guilty.

So, I would encourage people to seek support from Unique or from wherever. But, you know, generally, the thing I’ve learnt about people is that the vast, vast majority of people are nice and kind and understanding about this. Not everyone, but most people are good people and, you know, people should remember that, I think.

Sarah Crawford: Yeah. I mean, the first thing I was going to say in terms of advice to other people was something Shaun said already, which is the don’t be harsh on yourself, because, you know, you’re allowed to find it difficult. But I would also say it’s okay to grieve the child that you didn’t have that you thought you were going to have. I just think that’s so important. And I think for me, the most difficult thing in the early couple of years was feeling like I couldn’t do that because nobody appreciated that I’d actually lost anything. The world seems to use the word difference a lot at the minute, you know, “These children are different, they’re differently abled,” but actually it is disability [laughter], and it is more difficult, you know.

There are rewards, there are positives, but, you know, she’s 17 and a half now, our daughter. When our son was 17 and a half, you know, the challenges were different, but they were also nowhere near as big [laughter], and I don’t think that should get lost. Because I think parents need to feel it’s okay to get the help they need and to push for the help they need, and not feel like they’ve just got to kind of put on a brave face and, you know, as Shaun was saying, the attitude sometimes of, “Well, you’ve just got to get on with it.” Because while you do, actually, you know, you do need help to do that. It is difficult.

Shaun Pye: The only other thing I’d say is, just ‘cos Sarah just mentioned it and it gets forgotten, is the siblings thing. The families with Unique will have all manner of different configurations. I can only speak from our own experience, but Joey has an elder brother, Frank, who is, well, in my opinion, the best human being in the world [laughter], and I’m sure in his mother’s opinion as well, but my experience, never forget about the toll it takes on siblings. ‘Cos Frank is a very, very loving brother. Only last night, Joey was typing, “Frank book.” ‘Cos he’s gone to university, she likes looking at pictures of him in the photo albums. She likes looking at pictures of old toys mainly.

Sarah Crawford: Yeah, yeah, she likes looking at her as a baby and the toys they had.

Shaun Pye: Yeah, but it’s not really advice, it’s just, you know, there’s a danger that Joey could have taken over our entire family life, and especially Sarah made sure that didn’t happen and that, you know, we were a unit and he was – but, you know, it is possible that it can swallow up your entire life.

[Music]

**Naimah: Okay, so we’ll wrap the interview up there. Thank you so much to our guests, Shaun Pye, Sarah Crawford and Sarah Wynn for joining us today as we discussed Shaun and Sarah’s journey to Joey’s diagnosis, and how charities like Unique can support families of those living with rare conditions. If you’d like to hear more like this, please subscribe to the G Word on your favourite podcast app. Thank you for listening. I’ve been your host and producer, Naimah Callachand, and this podcast was edited by Bill Griffin at Ventoux Digital.**