**Genetic Diagnosis and impact on mental health**

**The G Word transcript**

**Vivienne:** Hello and welcome to the G Word. My name's Vivienne Parry, and I am the Head of Public Engagement at Genomics England.

People tend to think of the impact of a genetic diagnosis purely in terms of treatment or the impact on future physical health. We do not often think about the impact on mental health, both on the person directly affected and on the wider family. For some, there's crippling anxiety about the future. For others, there's guilt about what this diagnosis might mean for wider family members, and there's grief too, as the future imagined for a child is suddenly taken away. All of this, and I have only scratched the surface, is played out in families already vulnerable and deeply stressed by illness in the family.

So, in this Mental Health Awareness Week, we thought we would look at the impact of a genetic diagnosis on mental health and what we can do to reduce this burden for families. With me, three people are each seeing this from a different viewpoint. From the perspective of the parents and the family, Kym Winter is the Clinical Director and founder of Rare Minds, a non-profit organisation providing specialist online psychotherapy and counselling services to rare disease patients and family members. Kym has been a psychotherapist for over 25 years. Then, from a researcher's point of view, Helen Dolling. Helen is a researcher at the Centre for Family Research at the University of Cambridge; her research investigates the psychosocial impact of the diagnostic process on parents through her involvement in the Next Generation Study of 521 families. She is now involved in the Peregrine Study, which follows on from this. Finally, from the Genetic Counsellor, Melanie Watson is the Lead Genetic Counsellor for the Wessex Clinical Genetic Services and an Honorary Senior Lecturer at Southampton University. She provides genetic counselling to individuals and families affected by a risk of genetically related disorders, with a special interest in psychiatric disorders.

So, first of all, what are the main issues that parents face or even individuals face when given a genetic diagnosis? Kym, let us start with you.

**Kym**: Well, I think the impactsare profound and complex, Vivienne, and it depends very much on the condition that is being tested for, certainly with families and parents that we see. There may be the initial relief of getting a diagnosis, but then you are plummeted into a world of continued and ongoing uncertainty. Very often treatments may or may not be available, you may be dealing with the impact on your wider family and the opinions and views that everyone has about how it should be dealt with or managed. It is not unusual for us, and certainly the families or individuals that come to us, to hear of it being a little bit like a small bomb going off in your family. The impact on relationships has a ripple out effect over time. So, I think receiving a diagnosis is always a life-changing event.

As you said in your introduction, you are coming to terms with a different imagined future for yourself or your child. You are dealing with grief and loss around what you had hoped things might be, perhaps, and the guilt and the anxiety of what it is going to mean, and those fears for the future. So, it is always very profoundly impactful, and that impact, I think, unravels over time, too. Different families, different individuals processing news at different times, as I am sure you know, Melanie and Helen will confirm.

**Vivienne:** Melanie, how are diagnoses received?

**Melanie:** Diagnoses can be received either in an environment that's quite intense, in an acute environment when a child's still under acute care, such as intensive care; or they can be sort of received in a more organised way, where you had the opportunity to prepare the family; or there may be a family history so they're already aware of that condition in the family. So, it can be received in very unexpected circumstances, in an acute situation, or in a more managed situation where people are aware of this family history.

**Vivienne:** And there is also the impact when it comes completely out of the blue and the baby or child appears perfectly healthy.

**Melanie:** Absolutely, and that can be a complete shock, and I think at that time of having that shock, you're given an awful lot of information, or sometimes even a lack of information, but you're given quite complex information, a genetic test result, which maybe doesn't make sense, like you say, in the way the child appears or in your own experience. So, as Genetic Counsellors, we are very much attuned to sort of assessing someone's family history, interpreting what has happened in the family before, and trying to hang that on the experience within the family and the experience of that individual to help them make sense. When I am training new genetic counsellors, I will say it is about finding that shared meaning with the genetic information, but also that individual and family experience.

**Vivienne:** Helen, what does research tell us about the impact on mental health?

**Helen:** I suppose one thing to maybe mention, first of all, is that when we talk about rare diseases or various genetic conditions, it's such a broad base, that saying how it affects one is not necessarily going to be generalisable to a different patient group or different condition, but also that even within individuals within the same family, you can see quite different responses to being diagnosed with or receiving a diagnosis in one's child. So, the responses can be very different, and research shows that, in general, getting a diagnosis does not seem to have long-term effects necessarily in terms of having depression or anxiety related to it. However, there are significant individual differences in different conditions, and depending on what that diagnosis entails, it can sometimes also have very specific impacts on whether it possibly demonstrates the likelihood of a life-limiting condition. The parents, obviously, or children with that condition, will have very different experiences from somebody who has a stable disease with maybe quite mild symptoms. So, it's difficult to give a broad answer to that question because research obviously looks at either specific conditions, say X-linked.

**Vivienne:** Now, Kym, I guess what we're talking about here is diagnosis and we all think, oh, that's a certain thing, but actually uncertainty is a great part of genetic conditions because you don't know how they're going to play out necessarily, and even the diagnosis itself may be less certain than people imagine.

**Kym:** And thathow to live with uncertainty over time is a really strong theme that emerges with the couples and families and individuals we work with. Of course, we know there is a strong association between uncertainty and anxiety, we saw that through the pandemic in the ordinary population, and it is well understood as a link now.

Just going back to one of the things that Helen was saying, the idea that a diagnosis is processed sort of immediately is not something we see in the families with which we are working. We run ‘dealing with diagnosis’ workshops, and when we first started running them, we assumed that they would be for individuals or couples in the first year or so post-diagnosis. What we found was that people were wanting to sign up for them, perhaps 1, 2, 3, 4, or maybe 5 years, post-diagnosis, because they had not really had the opportunity to process that information and the meaning for their lives and the impact on their relationships. Coming together with others to have that sort of conversation was enormously helpful, and that theme of uncertainty figured very highly in those workshops too. How do you live with the uncertainty over time of when a condition's symptoms may start to emerge or the prognosis for your own life, or your child's life over time? So, it is a really interesting one about how diagnosis is processed over time, not just in the moment of delivery, which we often focus on, too.

**Vivienne:** And as you said, Kym, it is a bit like a bomb going off, and one of the effects of that bomb is often marital or relationship breakdown. Sadly, I mean, I used to be an agony aunt on a national newspaper, and so often I would have letters from parents or a parent of a child that had had a genetic diagnosis, as I mentioned in the introduction, about what that impact is for the rest of the family. I mean, Melanie, families are difficult enough as they are. I mean, heaven knows when we all get together at Christmas, you are wanting to, you know, lock the door immediately and not let them in again, but these things can have a terrible impact on the wider family, can't they?

**Melanie**: Absolutely, and when we first see someone, we definitely spend a lot of time around that sort of psychosocial assessment around the family, and, you know, whether there is any isolation, what support is available, who are the key people in your family that you would seek support from, and very much what is the family story so far, because that will give you a sense of what's there to support that person going forward. And, also, we spend quite a lot of time going back and looking at how they have coped in situations of uncertainty or unexpected news before and identifying people's coping strategies, whether they sit with uncertainty well, and whether they are real information seekers. I think that makes a big difference within families; you will have people who have different approaches to dealing with such emotional news.

**Kym**: We see that a lot in the work that we do, Melanie, and I just think it is so interesting hearing Melanie talk about it because often with couples that we work with, we are focusing very much on those different coping strategies. Are you someone who tends to go, "Actually, let's just not talk about it too much; we will deal with problems as they arise”, or are you the person who has to become the expert in that particular condition? And then the other partner is going, "Oh, do we never get time off from this?”, and the tensions that arise because of that. Then there are the tensions in the wider family, too, about who thinks which approach is better. So, you never quite know. I think there are often ways in which couple relationships are fragile, and a genetic diagnosis will worm its way into those fragilities. We could say it is an opportunity to look at a couple's relationship and help it be more robust. But, you know, there are lots of ifs and buts in that process, but certainly couples and families can need a lot of support with that, you know, adjusting.

**Vivienne:** So, let us concentrate now on that support and the best practice. So, what kind of support do parents need? Helen, what does your research tell you about the best kind of support that parents should be given?

**Helen:** This is a really fascinating topic that is emerging in my research. I have to say that, obviously, the analysis is ongoing, so I have to frame it in light of that. What I'm seeing is that those individual needs between fathers and mothers, and often it does go down, are quite different. The gender lines are quite different, parents’ ability to take on that information also varies, and the way that they respond to the diagnosis is quite different in at least the couples that I have interviewed, and I've interviewed mothers and fathers separately. One of the things they have really appreciated and stated is having that interview on their own to talk individually about their concerns and their experiences, which are very unique and often different from the person that they are in a relationship with. That, of course, is sometimes related to their own background as a child growing up in their own family, the experiences they had, and their relationship with the parents, all of which play into how they then experience parenting transitioning to parenting as a new parent with a child with a disability or a genetic condition, and the way that they reflect on the usage of social support groups is also quite different.

Often, it tends to be mothers, but not always, who are the ones that may seek out social support through Facebook groups or condition-specific online groups, and the fathers either benefit, sometimes vicariously through the spouse, by having snippets of that information delivered to them, but at a time and pace that they can cope with. Often the fathers who have looked for these support networks say that there are actually very few fathers on those social online groups, or they cannot cope with the level of information, especially if it is quite difficult to take on board the information that is shared on those platforms.

So, there are definitely very different needs. Then there is a communication aspect as well that was already referenced before me: parents are negotiating these new boundaries and new ways of living together and making a future in a situation that is sometimes uncertain for extended periods of time, but also unknown in terms of what to expect, or how to parent a child for whom the condition might be such a rare one that there is very limited information that they can rely on to help them navigate that. So, yes, parents' differences are quite different.

**Vivienne:** So, Melanie, you’re both teaching baby Genetic Counsellors how to navigate these very tricky waters, but what is your advice for Genetic Counsellors on how to lessen the impact on mental health of a diagnosis?

**Melanie:** I think it is very much about getting the sense of the individual in front of you. It's always talked of as Genetic Counsellors being a comprehensive educational process for genetic counsellors, and I quite often say to them, "Yes, there are lots of information that we can give somebody, but it's whether it's fertile ground to receive that information at that point in time”, so you can give them the information, but quite often it's around recognising that the grief they're going through is a process, the education is a process, and the support is a process. So, we very much try to take a sort of drip-by-drip approach to talking about it in the family, when it feels like the right time to actually give them that information.

Quite often, it is the individual or family member in front of you that knows when the right time is, and they will come back to you. So, we always have our families under review because that is how they stay with us, and they might come back to us another time during the lifespan when someone's planning to have a child or a grandchild and they want more information at that time. So, it is supporting where they are at that moment, giving them as much information as you can, but then maybe reinforcing that information during the process of the emotions that they are processing from having that information in the first place.

**Vivienne:** So, Kym, what are you hearing from parents and families about what works for them and what does not?

**Kym:** Well, if we're talking about diagnosis, just to follow on what Melanie is saying, I really think that people take time to process diagnoses at different times in different ways, so that feeling that they can return, preferably to the same person again, perhaps to ask the same questions again, and have time to process it in their own particular timeframe, and you can't predict that. You know, we have people who've come to us, perhaps given a diagnosis maybe five or six years ago, and may not have had any particular symptoms; it may not be particularly impacting their lives, but then they come to us and say, "So I had this diagnosis a number of years ago. I cannot even remember particularly who I saw or what happened, but it was this, and now I am having to really deal with it”.

So, I think this notion of being able to process different aspects of a diagnosis at different points is really important. You have already mentioned how it impacts couple relationships, and I think that is something that is going to be increasingly important to attend to as genomics and genetics really take forward supporting couple relationships, communication and coping strategies. So, it is not one thing, I think we are saying that, you know, people need help with.

**Vivienne:** We shouldn't forget the intense financial strain this can place on families, because often one member of the family at least will have to give up work in order to care for an affected child, and there are all sorts of things, you know, whether it's travelling to hospitals and appointments or specialist equipment, there are all sorts of financial burdens that can just tip over that already difficult situation that so many people find themselves in now, and that's an added burden. So, is there enough support for people with children with a genetic diagnosis from existing mental health charities, for instance, or do you think they do not quite understand the problems that are being faced? Let us go to you, Kym, on that one first.

**Kym:** Well, what we hear from the individuals and families we work with is that it is quite difficult, first of all, to access mental health support if it is in relation to the diagnosis or living with a genetic condition. I think that goes down often to the general lack of awareness around rare conditions generally, so some mental health care practitioners may be quite apprehensive about dipping a toe into that area. A lot of services are stretched, and they are closely benchmarking their services accordingly. So, if someone turns up with an unusual condition they have not heard of, they may see it as not under their remit. We also see clients who come to us who have tried to access things like IAPT (Improving Access to Psychological Therapies) or other psychological services and have said they are not really fit for purpose for their needs. You know you need time to process the meaning of a diagnosis for you as an individual in your life. For anxiety, you know, CBT (cognitive behavioural therapy) can be helpful, but it is not really the whole picture, or the whole answer.

I mean, I do think, to go back to what Melanie is saying, that a lot of people we see are not given diagnoses by genetic counsellors in a timely, calm, or considered manner. They may happen sort of ad hoc, by letter, or over the phone, with very little follow-up information or signposting. I think people often feel quite stranded and abandoned. You know, that is something we need to get better at moving forward: training healthcare professionals themselves to deliver diagnoses and growing the genetic counsellor workforce, too, which can then work with people like myself.

**Vivienne:** We're always keen to grow the Genetic Counsellor network. Melanie, can I come to you because I know that you have a particular interest in psychiatric disorders. Does the diagnosis of a psychiatric condition have a greater impact on mental health in the family than non-psychiatric conditions?

**Melanie:** I would not say so specifically. It is all those emotions that you get with any other genetic diagnosis—that sense of grief and loss of the imagined future or someone's own identity—but also those usual emotions around anything that has inherited elements of anger, guilt and blame. There is a little bit of stigma and taboo around mental illness and psychiatric illness in particular, and we have actually spent quite a lot of time getting genetic counsellors to be comfortable discussing psychiatric illness. It almost feels like we are talking about bombs going off in a sense, the bomb going off a psychiatric illness. It is our own fears of our own mental health. We all know there is a fine line between us and them. We all go through times in our own lives where we have acute mental health periods, when we have periods of stress, and I think there's a lot of fear around that, and quite often with a genetic diagnosis that might have a psychiatric element, maybe that isn't always discussed to the same extent as if there's a physical element to the condition, and we are very much training the new workforce in genetic counselling to be comfortable with that element.

**Vivienne:** And there is always, of course, with psychiatric conditions, but also with other genetic conditions, that you are healthy, but you are looking for a symptom all the time, so you are anticipating. Helen, I wonder whether anticipating the future when you have not actually had symptoms when you are diagnosed is something that you have encountered in your research.

**Helen:** Parents talk about hypervigilance and being mindful of the possibility of their child deteriorating if that is their anticipation. It has not been a major theme that I have noted in the 91 interviews that I have conducted. In literature, there are definitely references to that quite frequently within specific conditions. So, parents whose children have seizures are constantly, for example, experiencing hypervigilance, anticipation, and uncertainty about when the next one might happen. So, they live with that day to day, especially if seizure frequencies are quite present in their day-to-day lives.

I did want to come in on the support of mental health and parents accessing that, if I may. In response to what Kym and Melanie were saying earlier, acknowledging that they need support, especially if it is to do with anxiety, depression, or mental health-related issues, is quite a barrier for some parents. First, they are thrown into a situation where perhaps previously they have been entirely independent, strong individuals who manage their lives in amazing ways, and suddenly they feel that they don't know how to go on from the moment of diagnosis, and that may not even be genetic to start with – perhaps it starts with symptoms or being hospitalised, and so on. The process may take quite a few years, and then we talked about the adaptation to the diagnosis or the new life over time and how that can be variable. For some parents, that might be a personality trait, or it could be personal circumstances and life experiences and the way that they have always dealt with complexities or challenges in their lives before.

Crossing that barrier of asking for help is a major milestone, and the complexity and, in my interview, certainly some of the fathers in particular, but often also mothers, aren’t related to necessarily one socioeconomic background, but it can be in any background that this is an obstacle to admitting that one needs support to manage their mental wellbeing and knowing how to access that or how to best go about it. So what parents are in effect saying is that if it's framed in a way that this is a mental health issue or support, even the word support in itself can be experienced as something quite negative and something that is not necessarily available to them in the concept of their own personality and the individual identity as a strong person.

**Vivienne:** Kym, do you want to come in there?

**Kym:** Yes, I was actually agreeing with what Helen was saying about the stigma around mental health being very real. I think what we are very keen, within Rare Minds and probably all of us on this call, to say is that it is perfectly normal and understandable to need to have some time to talk to someone to process the news of a diagnosis and the meaning of it for your life. That does not mean you have a mental illness or a mental health problem; it is a perfectly ordinary part of adjustment, and I think the more that we can integrate those sorts of mental and emotional wellbeing aspects into ordinary rare disease care and diagnosis, the better for everyone. We will all get more comfortable with those sorts of conversations.

**Melanie:** Very much, so just to echo what you are saying, we very much try to normalise those emotions and that you'd expect to have those emotions associated with this type of diagnosis and that those emotions will, you know, go up and down and you will face new things throughout different stages of your life where they might come to the floor again and you go through a period where you feel able to cope. You recognise the burden of the disease, the physical demands of that, and the emotional demands of that on the day-to-day lives of that whole family and that individual. Unfortunately, as genomic counsellors or genetic counsellors, we were given appointments with patients with the initial diagnosis, but we do not have the capacity to follow up all the time because there are so many people having a new diagnosis that the capacity to follow up is increasing all the time. I quite often get people who keep asking, and we will make or find the capacity to see them as a follow-up, but if they are not asking, we do not really know what is happening to those people.

I think that is the worry. The people that have that diagnosis and then disappear, you know, we give them information about support groups and charities, and sometimes there is quite a lot of investment in specific diseases, but for the rarer diseases, there is very little out there. Then they are left with the usual mental health services that a GP can offer. As someone alluded to earlier, quite often a counsellor, a general counsellor, will be able to offer support but may not realise the impact of that particular genetic condition. Then they feel slightly lost when they are having that counselling because this person does not really recognise what they are going through or the burden of their specific disease.

**Vivienne:** So, let us now look at the very practical, and I am going to give you my Vivienne Parry magic wand in a moment. What would be the ideal situation if you were a parent with a recent diagnosis listening to this, Kym? How do they access help and support? What is the best route for them?

**Kym:** Well, the first port of call is usually through patient organisations that do a fabulous job of supporting emotional wellbeing and providing information around specific conditions and also generally, and you are probably all aware here of great organisations like Genetic Alliance UK or unique ones like Gene People. A number of those are increasingly using services like mine that have counsellors who are trained in the impact of a rare condition both generally and specifically, so I think it is a case of persevering. It is a case of finding your way to the best support for you at the right time. That might be couples therapy, or it might be individual therapy. Patient organisations are a really good place to start. I do think we need to get better at encouraging healthcare professionals to sign up for them. We still have people who find their way to us, and they say, "Oh, we just happened to Google you. We just happened to come across you”. You know if we are going to be delivering a diagnosis.

**Vivienne:** We tend to be good at giving out literature and information about the physical manifestations of a particular condition, but we are not so good at dealing with the support. Melanie, what is the best way for parents to get the best information and support?

**Melanie:** As Kym was saying, it is very much around online posting, and we definitely make sure we do that, and we do that in a written form. We either give them information within the appointment about specific charities or organisations that may offer future support or as many weblinks as we possibly can. I think the other aspect of it is actually thinking about them in the wider sort of network of their ongoing healthcare in an ongoing sense and making sure that we educate the wider healthcare professionals, such as nurses, about some of these rare conditions and the impact of those on families so that they don't feel isolated when they go further along the process of the diagnosis and there are other healthcare professionals that are aware of that impact.

**Vivienne:** We should say that there is nothing like having other people who have been through the same thing to lean on; that is so, so important. Helen, where are the big gaps in terms of the research in this area? What don't we know that we really need to know to help support people with their mental health during diagnosis?

**Helen:** There is obviously a wide range of research. What we know is that mental health cannot necessarily be separated from physical health and other needs as well. So, families have emotional, cognitive, behavioural, and informational needs, as well as social support needs. There is a whole range of research that is kind of converging on these aspects, showing that parents go through similar processes in some ways, but the timing of those can be different and very individual, as we spoke about before.

For some people, they get on with life as if nothing had happened and try to normalise things, and then it may hit years down the line. For others, the early process of getting the diagnosis is a real shock, especially if it has come unexpectedly out of the blue, or very early on. However, the genetic diagnosis can be helpful in adjusting to the diagnosis and moving forward with their lives by having relevant information and support put around the family and the individuals within that, according to their own personal needs as well.

So what research is showing is that different groups in Canada, Australia, America, Europe, and the UK, all researching genetic conditions and rare conditions in particular, are gradually finding very, very similar things. It is not necessarily the diagnosis per se that makes a difference; it's the processing of that diagnosis and the support that is there. And of course, there are risk factors that individuals have; they may already have had prior mental health difficulties in the past and may be more vulnerable. They may have had a difficult childhood as a child growing up in the family, and that can also contribute to how they are processing their parenting journey and managing the situation. Then, as Vivienne, you pointed out earlier, there are also financial difficulties that may emerge as a result of having to parent a child or having a genetic condition oneself, where one has to give up work in order to manage that situation, and of course, many conditions impact sleep, which is also a factor and causes further vulnerabilities.

**Vivienne:** So, what I am hearing from all of you is that we do not give mental health support the focus that it needs and that it is really something that we need to concentrate on in the future. Particularly, as I said, because Genomics England is giving more and more of these kinds of diagnoses out to parents and we need to make sure that they're fully supported, not just with the kind of information that they need but with the support that they need through organisations such as yours, Kym, and of course through genetic counsellors like you, Melanie, and there is still some research to be done. So, thank you, all three of you, for being with us today. We really appreciate the time that you have given us.

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