The Cancer Professionals Podcast

Episode 7 (August 2024) – Genomics and cancer care: Impactful insights and personal stories

(Intro music)

00:00:09 Lydia

Do you want to know the impact that genetic testing can have on someone's life?

00:00:13 Emma

The results came and even now I still remember sitting on the sofa with my dad and my dad cried because he said gosh, I can't believe you know you've inherited this for me. I'm so sorry. It was hard for him because I think it's much easier to deal with the diagnosis if it's yourself. But it's tricky seeing potentially what impact it might have on your children.

00:00:35 Paul

Hi, I'm Paul and my pronouns are he/ him.

00:00:37 Lydia

and I'm Lydia and I go by she/ her. Welcome to the Cancer Professionals Podcast, a podcast from Macmillan. In this series, we chat to a wide range of guests, including health and social care professionals, to lift the lid on current issues faced by the cancer workforce.

00:00:54 Paul

In this episode, we're joined by Kelly Kohut, lead consultant Genetic Counsellor, and Emma Jenkins, a patient representative working with the South East NHS Genomic Medicine Service. Kelly discusses what genomics is, why it's important, and how we can use genomics to improve cancer care.

00:01:14 Lydia

Emma shares with us her personal experiences of being diagnosed with Lynch syndrome following genetic testing and the impact that this has had on her life.

00:01:23 Paul

This episode contains conversations about lived experience of cancer, which you may find upsetting or triggering. Listener discretion is advised.

00:01:33 Lydia

Hello, Kelly and Emma. Welcome to the Cancer Professionals podcast. It's so lovely having you join us today. Can you start by giving us a little introduction to yourselves, Emma, if I start with you?

00:01:43 Emma

Thank you for having me. My name is Emma. Emma Jenkins and I work in education, and I have Lynch syndrome.

00:01:52 Lydia

And Kelly, are you able to introduce yourself?

00:01:53 Kelly

Hello. I'm the lead genetic counsellor at Saint George's Hospital in London, covering the SW Thames region and I'm also doing some original research co-designing a patient's website and decision aid for Lynch syndrome to help people make sense of their genetic risks for cancer and think about making decisions to manage those.

00:02:16 Lydia

Now we know that genomics is such a vast topic, but really today we want to get into a bit of an introduction to our listeners, perhaps for people that haven't heard of the topic before or might have heard of it, but I just aren't really sure what it means. So Kelly, would you be able to start by telling us a little bit about genomics and what we mean when we talk about genomics?

00:02:38 Kelly

Genomics refers to our understanding of the sequence of DNA and all the bits in between that control the instructions that make our body so our genetic blueprint. And over the past few decades, we've had an increasing understanding of the contribution to the chances of people getting certain conditions or diseases. So genomics is important to virtually all areas of medicine and and testing is becoming more and more widespread.

00:03:11 Lydia

And why is it so important that we know about it in cancer care?

00:03:15 Kelly

It can inform the treatment of cancer when someone has a diagnosis, so it might help the oncologist to select a certain treatment. There's things like immunotherapy, which are newer treatments that could be considered. And also sometimes to guide what type of surgery people are choosing, so more and more people are having genetic or genomic testing right around the time that they have a diagnosis of cancer. So it all happens quite

quickly in order that the results can be available to guide those treatment and surgical decisions.

00:03:51 Lydia

And what about for those people that might have had some genetic testing but don't have cancer?

00:03:58 Kelly

Yes. So that often happens later, after a pathogenic variant or a change in the spelling of a gene is already found in someone in the family who has had a cancer. And then, because it's familial, we can then look at other relatives who might benefit from knowing about the availability of genetic testing, coming for genetic counselling to consider that for themselves. And then they could be tested to see whether they carry the familial variant or not, and that will tell them about their chances of developing cancer and their choices in terms of surveillance programmes and risk reducing options.

00:04:36 Lydia

And Emma, listening to what Kelly has said there, I wondered if you would be happy to tell us a little bit more about your story and your experience of genetic testing, if that's OK.

00:04:45 Emma

Yeah, of course. So in 2007, my dad developed a cough, which to begin with seemed fairly innocuous, didn't really worry about it. But then the cough seemed to go on and on. And I think, like, a lot of relationships, it was my mum who said I think you ought to go and get that cough checked out and and although my dad did work in oncology, he said, oh, no, it's just a cough. It'll be fine. It'll be fine. But then Dad always says you know your own body and you know, if something's not right and he realised, actually, it's not just a cough. I think I do need to go to the doctor. So he went to the GP and the GP understandably said Yep, you've gotta cough. Here's some cough medicine. Off you go that didn't work. He went back again. I can't remember how many times it was 2007. So it's a while ago.

And and then in the end, he said. Look, I definitely know something's not quite right. And that's when a blood test was carried out and they discovered that my dad was anaemic and they wanted to try and understand what was sitting behind that anaemia that was causing the cough. And that was when after some further tests, they discovered that he'd got colorectal cancer.

So he had surgery straight away at Maidstone Hospital and it was during the Histology of the tumour that they discovered that he had got a fault with his PMS2 gene and he was diagnosed with Lynch syndrome. So it's quite a lot for a family to take on board, although my father's obviously worked around oncology, it's very different when you're sitting there

with a diagnosis. And I think selfishly as a family, if you are a scientist, your family are often playing catch up because what the specialists are saying to my mom and my dad were very much science led, which is understandable, which was great for my dad because he he loved that sort of science information that was being given. But for my mum it was really tricky because she's trying to take on board the fact that the man she loves has cancer and she's not understanding a lot of the technical terminology that's being given, and my dad said I'll explain it later, I'll explain later.

So he was then very keen and sat down with my brother and I and talked about the importance of going for genetics counselling. But again we were adults so I was sort of 28. And said it's it's your decision if you choose to go, but you are being invited for counselling. And I did sit with him and I did talk to him for quite a while because again, it's a head and heart issue. Do I really want to have a crystal ball that may or may not give a glimpse to my future for something I may or want may not develop later on in life? And I'm really pleased that I did listen to him. I think he's probably quite pleased too. So I made the appointment and travelled to guys hospital and my family said do you want us to come with you? And I said no. I want to go my own.

So I went and I spoke to a wonderful lady. Who made the process really straightforward. Explained what genetics counselling was all about, looked at family history, and we had a really wonderful conversation that really did encompass the head and heart issue that I was experiencing. So concern for my dad, who's only just recovered from surgery, you know, what's my result going to be? Because at that time you think it's going to be 50/50. And what was great for me was that I was given a two week hiatus once I was tested in 2008 to make that decision. Once my blood was taken, would I actually like to have the test gone through? So they contacted me after 2 weeks. And I decided yes definitely. And so they phoned me and said, would you like the test done? I said yes and then the results came and and even now I still remember sitting on the sofa with my dad, and my mum in an armchair, and my dad cried. Because he said, gosh, I can't believe you know you've inherited this from me. I'm so sorry. And I said, listen, you've got nothing to be sorry about. This is, you know, this is just the luck of life's draw. We're all gonna get something. This is nothing you've done wrong. But it was hard. It was hard for him because I think it's much easier to deal with the diagnosis if it's yourself.

But it's tricky seeing potentially what impact it might have on our children, but the letter was quite scary if I'm honest, because the letter said you know, sadly, you've inherited this, and then there was a breakdown of some pretty scary statistics, although the genetics counsellor had talked about. It's one thing to have a friendly conversation in a supportive environment. Seeing something in stark black and white with percentage increase of certain types of cancer is is quite alarming. And then it explained that I would then be fed into a system with my local trust for regular surveillance in terms of colonoscopies.

The thing that I found most difficult though, with the whole process was that was kind of it at that time in 2008. Great strides have been made since then in terms of surveillance, in terms of endometrial type cancers or anything gynaecological. But being 28 and of an age when I may or may not have been wanting to start my own family, being told you should really consider having a hysterectomy at 40 and that was almost a throwaway comment. That's why I'm so pleased that conversations like this are happening today because the strides that have been made since 2008 around support for patients with Lynch syndrome in terms of head and heart issues. It's it's a different generation, it's, you know, it's it's worlds apart. But for me, don't forget about those people who were diagnosed a long time ago because it's kind of you tend to go to doctor Google, which isn't always the best place to go. So that was that was my experience really.

00:10:34 Lydia

Thank you for sharing and that's really interesting to hear. And as you say, it's so important to remember that you are giving someone possibly quite life-changing news so for it to be sometimes a bit of a throwaway comment.

00:10:49 Emma

For me I think when you're talking to patients every day you know what you're saying and you know how to say it because it's it's medicalised. You're talking in a healthcare and a clinical setting. So it's very much your head and the science that's dictating the conversation. I think as a patient who's already feeling quite vulnerable and you never know whether it's going to be a yes, you've got it or no, you've not. And actually, I imagine if you don't have it, there's emotions associated with not having something that someone a loved one has has got. So I think for me it's that. It's trying to remember that it is an emotional conversation and not just about the statistics around you need to do this, this is what's going to happen then and the surveillance programme kicks in. There's a lot that has to kick in in terms of, even for me, do I share this with work? Am I going to? Am I going to be concerned that works? You know, a future employer won't employ me because they're concerned that, oh, gosh, well, she's going to get cancer, so we won't. We won't take the risk of employing her. So there's there's a lot more than associated with just knowing that you're in a system that is going to surveil you for certain types of cancer.

00:12:00 Lydia

Yeah. As you say, it is for a healthcare professional it's their day-to-day. But for you, it's completely new, possibly completely shocking. Well, I suppose you don't really know how you're gonna react to it when you receive that, that news, so, yeah, I suppose for healthcare professionals missing, it's about remembering that and keeping it at the forefront of your mind.

And Kelly, so something that I picked up on, well, two things that I picked up on really that Emma mentioned. The first thing would you be able to explain to us a little bit about what PMS2 is please?

00:12:33 Kelly

Yes, PMS two is one of the four genes associated with Lynch syndrome and I think that since Emma had her genetic testing, we've learned a lot more about the differences between the genes and that there isn't just one lymph syndrome and people now say there's four different types of Lynch syndrome.

Because depending upon which gene the variant is found in, there would be different chances of developing certain cancers, and it's dependent on the organ and the the gender of the person and the age, as well as the gene. So PMS2 is the one that we've probably got the least information about because it was not one of the first ones that was discovered. So back in the early days of genetic testing, it wouldn't have been included.

So that was probably fairly new when Emma had her testing to find out about PMS2, so we now know that there are lower chances of developing the gynaecological cancers, especially the ovarian, so we no longer recommend removing the ovaries. There may be a consideration of hysterectomy, of removing the womb and then, as Emma mentioned, the bowel screening using colonoscopy is something that PMS2 variant carriers would have, so it's important for people to know which gene and actually it's interesting that, you know, when I've spoken to people who were diagnosed and had genetic testing 10 or 20 years ago, some of them didn't even realise that there's more than one gene for Lynch syndrome.

00:14:08 Lydia

I guess people talk about Lynch syndrome probably like day-to-day in healthcare, but actually there's probably a lot of people that don't understand the different types. So that's that's really interesting to hear and hopefully people will go away and do a bit more research into that. So thank you.

And the other thing that Emma mentioned was talking about her dad's reaction. Is it a common thing that you see with family members, like maybe feeling a bit of guilt of passing on that that gene to their children?

00:14:38 Kelly

It is a common reaction and I think it's very natural for people when they are parent to immediately start thinking about their children and we often hear that they say, well, I'm not worried about myself, but I I'm worried about my son, my daughter. So it's a very common reaction and one that we spend time talking about in the genetic counselling appointment to try and help people understand that, you know, there are genetic variants

in every family we happen to have found out about this one and and actually with Lynch syndrome, we have proven surveillance programmes, risk reducing measures that can be recommended. So there's a lot of reassurance that we can give people that we know quite a lot about these genes and the management guidelines and that finding out can actually be powerful for a family, because if they know about the chances of developing cancers being higher because of this genetic reason, then they can start screening earlier, people might start colonoscopy every two years from age 25 or 35.

So trying to help people deal with these natural reactions and emotions that come up and then it can take a little bit of time. But people usually do quite well with the process of having genetic testing and adapting to the results. And where they can get access to genetic counselling, it can really help with that process and communication within the family.

00:16:04 Lydia

I guess it's that taking back control a little bit, isn't it? I guess people say knowledge is power, right? So yeah, that's great to hear.

And Emma, when you were going through the testing, I know you spoken about the fact that you had a genetic counsellor that you could speak to. I wondered if you could give us any insight into what was helpful and maybe what was not helpful with the support you received. I'm just thinking about the healthcare professionals listening it would be really great to hear from you if few tips about what was good and what was not good?

00:16:34 Emma

Yeah, I think I think understanding actually what you were going there to do and what you were going to talk about. So my dad explained it from what he'd understood, which was useful, although he did preface it with. I'm not a genetics counsellor. I'll leave that to them. And again, it might look very different now. So this is this is 2008 but for me it was I remember as clearly as anything taking the train to London Bridge and then the walk up and then sitting in a very sort of medicalised waiting room and then being taken into a room by a lady and I mean this in the best possible way that didn't come across as a scientist or someone who was going to say things to me that I wouldn't understand, I felt I almost felt a warmth from them when I walked in, which helped alleviate some of the fears I had about potentially a difficult conversation.

So we sat down and she talked about who she was, what her role was, how the appointment would play out, that her role was to support me in terms of any questions that I had. And if I didn't understand anything to to to ask, the great thing was she didn't use any acronyms, so I didn't have to say sorry can you just explain what that means please?

And and then we sat down and she'd done a huge amount of work before I'd actually arrived. So there was my family tree, which seemed weird because I'd seen my family tree in family situations, but to see it out in front of you with to see it look like that was that kind of took my breath away, if I'm honest. Because you suddenly become very aware of your mortality and the fact that might someone and a relative in the past still have been here today if they'd have had the opportunity to be tested, you know. So that was really, really empowering in a lot of ways because it made me realise how important being tested for genetics was because what it was going to enable me to do was to have surveillance in terms of colorectal issues, and that meant that it would be far more treatable should they find something that when it's small, rather than waiting until I'm potentially seriously ill, and then it's much harder to to have that, that that sort of treatment process.

We did have some brief conversations in terms of the gynaecological issues and as Kelly said, great strides have been made in terms of identifying the different genes that are involved and and the impact it has on the different range of cancers that that Lynch can impact. And that for me, I think was the hardest because I was at that age where I could have had a family or I might not have done. And I know it wasn't meant as a throwaway comment, but it was very clear in terms of colorectal this is what we're going to do. You're going to be sent a letter every two years. You're going to go for a colonoscopy, you know, that was really as clear as day. But then there was a lot of sort of wooliness around the gynaecological issues. And I said Ohh, you know, well, do I need to go and speak to someone about that? And she said in all honesty, you're not 30 yet. Don't worry until you're 40. Well, 12 years goes by and you suddenly go, oh, I'm 40 now. What do I do? And and even as a patient knowing, do I just go to a GP and say sorry, I was told 12 years ago that I should consider a hysterectomy and and I think if I'm looking back that possibly I was naive and that maybe I should have asked those questions. But I think you're trying to take in so much about everything about what genetics testing is. Will I have it, will I not? What's a colonoscopy going to feel like? You know all of those sorts of questions that that possibly I pushed the hysterectomy to the back of my mind. And so that's why it's great to hear Kelly talking about the changes and that sort of more nuanced approach to surveillance because I think that means that that sort of pathway that a patient is is moving along is far more tailored to their specific type of lynch or any genetic condition for that matter.

And that's why science is. Yeah, it's fantastic. Yes, it's a, it's a looking glass potentially. But at the same time, if it can save you some really aggressive treatment because you can catch something early or do something to prevent a certain type of cancer, then yeah, we we should do that in order to live well. And as long as possible.

00:21:00 Paul

And I think, you know, talking about the conversations it brings us on to another question. But Kelly, perhaps to start with and and Kelly, you've talked about what we mean by genomics and the testing and things that can happen quite quickly. How can healthcare professionals start to broach the conversations about genomics?

00:21:21 Kelly

I think that Emma's touched on that, this, this, that there there needs to be more education of healthcare professionals and there has been a lot of effort put into that over the past few years. There's been a big national programme to increase awareness, education, making sure that there's someone in every multidisciplinary team meeting who's a champion for Lynch. There's guidelines that recommend that all colorectal tumours, all endometrial tumours, should be screened for Lynch. But actually seeing that happen at every hospital has been challenging and has needed some coordinated work to make sure that the resources are there to increase the testing. And that has happened, it's improved, but there's still a long way to go. And when people with Lynch come up against going to see their GP and the GP doesn't know what Lynch is, or they go for a colonoscopy and the person doesn't know what Lynch is. It puts a personal burden on them. And I've learned this first hand from interviews that I've done in my own research with people with Lynch feeling that, you know, they're having to go out and search on their own. Or maybe they're meeting up with peers in a support group or something like that and finding things out that perhaps we as healthcare professionals, could be more proactive in making sure we sign, post them safely to the right information at the right time so they can make decisions that are right for them and feel supported to do that.

00:22:48 Paul (ad)

Before we hear more from Kelly and Emma, here's a quick message about other resources.

So Chris within the cancer workforce, we know that people are talking about and thinking about genomics within their work and why cancer genomics is relevant to the work they do, which is why it's really important to be having this conversation with Kelly and Emma.

What else can people access to learn more about genomics?

00:23:12 Chris

Well, we offer a series of recorded webinars on various cancer genomics topics, along with links to additional external resources, all available on the Macmillan Learning Hub. Additionally, you can access the Genomics toolkit on Macmillan’s website.

00:23:27 Paul

That sounds great. Lots of extra resources to learn and for more details see the episode description. Let's get back to the episode.

00:23:35 Paul

So, Kelly, you've touched on how we can use genomics to start to improve cancer care. Could you perhaps expand on that a little?

00:23:43 Kelly

Yes, I think that if people do have the testing right at the time of diagnosis, then it can give an opportunity for genomics to inform better treatments, more personalised treatment. So I mentioned things like immunotherapy are becoming more commonly considered and people having a very good response to that type of treatment. So it's no longer a one size fits all treatment of cancers in oncology. It's really if we can understand if there was a genomic mechanism for that cancer developing that it can help us to better target the treatments to affect the cancer cells and not so much the normal cells, which gives a lot of side effects from the traditional types of chemotherapy, so more personalised treatment.

00:24:36 Paul

And Emma, just coming back to you, how has the testing impacted on your life?

00:24:42 Emma

I think it's almost two different answers; how it impacted me at the time and how it's impacted me now. Back in 2008, like I said, not even 30 huge amount to try and take in and at the time I won't lie it it it was tough to to take and there was a lot of head work. I'm I'm very much someone who's led by my heart so it was quite, you know, to try and go into that zone of right, this is this is a sensible thing to do. You're an adult now, Emma. Come on, let's think about this. And like I touched on before there was a huge concern. Work is very important to me and I had this real fear about talking to people beyond my circle, if that makes sense, one because you know there's there's up to 300,000 people living with Lynch in the UK and only 5% know about it. So it's not necessarily something that's understood or discussed in the same way that a lot of conditions are. And the thought of the headline that an employer might take away being, well, this person's going to get cancer, even though that's not necessarily the case in terms of being more likely to develop. That was really difficult because I almost went into or do I need to keep this a secret and that's not me at all.

But then I wanted to be open with my employer because obviously potentially I'd need time off for colonoscopies. And interestingly, now my trust run them all day Saturday and

Sunday so I can have it at a weekend, which means again, if you are someone who is nervous about that you you can choose to to have that procedure done at the weekend, if that's what your trust offers. So back then, there was a lot of fear, but I fundamentally knew it was the right thing to do because I'd seen how far my father's cancer had progressed and and distressingly, when you look back at photos of him at that time, you noticed how ill he was. You saw how pale he was. When you're with some every day, those changes, the weight loss, the changing skin tone, you don't notice.

So there, there was a lot of. There was a lot of soul searching with that.

And something really that changed me in terms of of the whole process and and just lit my lynch world on fire was was two years ago, supporting through work a local cancer alliance in a programme developed around helping families and schools and children to be safe in the sun. And they were talking about a campaign that they were working on around a varying cancer and then Lynch syndrome. And I said Ohh, I have Lynch syndrome. And then I explained about PMS2 and then invited me to be part of a a Kent and Medway stakeholder group around Lynch syndrome. And that for me was a real empowering moment to change something that potentially had negative connotations because of the fear factor into something really empowering in the sense that I'm now happy to talk to anyone and everyone about Lynch syndrome. I know more about how much has changed in terms of surveillance and how much has changed in terms of support for patients. And for me, that's what we need to be doing. Things like this, talking to more people so they even though it exists or not being scared to go to a GP and saying, look, you know my family, my dad was diagnosed with Lynch syndrome after this. You know, what can I do about genetic testing?

Because again something that's come up very clearly in conversations with other people is that some GP's are not referring patients to genetics counselling because they think they have to pay for it. So again one of the messages that's coming across from people who are living with Lynch at the moment is how important it is to to remind GPs or anyone who's in the position to refer that a genetic test will not cost them anything. It's a free it's a free service that's provided by the NHS so so that even little things like that. I still think there's a massive way to go. If only 5% of people know they have Lynch syndrome. We also need to make sure that everyone knows what Lynch syndrome is, what it isn't and also how they can access that counselling and to demystify that why would you choose not to and almost understand the rationale that people have behind that? Because if we think about it from that heart perspective, it might see that more people engage with the science if they understand the psychology behind someone would choose not to get involved, that would

that would be for me. But like I said, now I'm more than happy to talk to people about it and I don't have that sort of stigma that I had 20 years ago.

00:29:21 Lydia

It's so important to have people like you who can advocate for those people that you know are living with Lynch syndrome or any other genetic condition and perhaps don't know about it. I think we're very lucky to have you out there spreading the message.

Kelly, I just wanted to come back to you and now you mentioned a little bit earlier about how knowledge of a genetic condition can affect treatment plans for people who do have cancer. Would you be able to tell us a little bit more about that and perhaps provide an example of how the knowledge of that gene could could help with treatment?

00:29:54 Kelly

If you take someone who's diagnosed with a colorectal cancer, for example. And the tumour goes for routine screening and pathology to look for. They use a technique called immunohistochemistry, or IHC. Sorry to use an acronym, Emma, but I've explained what it means. They're looking for whether there's a mismatch repair deficiency, because the genes that are associated with Lynch syndrome are called the mismatch repair genes and their job in the body is to correct mismatches that naturally occur in the DNA. So when there is a deficiency, it can mean that people with Lynch have a higher chance of developing these cancers. But once the cancer is found, it can also inform the types of treatment that might work best.

So for people who have a mismatch repair deficiency, the scientists have found that they produce these things called frameshift peptides. And they have a good response, often to the immunotherapy treatments. So it could be that finding out that someone has mismatch repair deficient tumour can help the oncologist to select the treatment and that might give a better outcome for people that they could perhaps have a better chance of survival of the cancer.

If people have already known that they had lynch and had genetic testing, they may have had an earlier diagnosis because they were in a surveillance programme from an earlier age where they might be having colonoscopies when people normally are not asked to have colonoscopy because they don't have a high enough risk of colorectal cancer. So it can be sometimes that we find cancers earlier where they're easier to treat. There could be the chance for other cancers. So depending on which gene is involved for that family, for Lynch syndrome, there can be other cancer risks, so it could be that depending on someone's age, for example, if they had colorectal cancer and they've already said that

they've definitely completed their family, if they, if they were going to have children. So let's say they were over 50 they might, you know, talk about the chance to consider having risk reducing surgery as well to have a hysterectomy when they're having colorectal treatment or soon thereafter. So it can, it can tell people about their chances to treat this cancer successfully but also thinking about future risks.

Typically we don't remove the entire colon because of Lynch syndrome. But for some people they might consider having a slightly more extensive part of the bowel removed depending on, you know if there's other polyps. But it's it's not like other genetic conditions where there's many, many polyps growing in the colon and people have the colon removed. That's a different set of genes. So usually it's just you know, surgery to treat the cancer that's there and think about, you know, what are the options for future risk reduction for that person?

00:33:12 Paul

And where can we see genomics in the future Kelly? Are there any kind of any exciting advances that we might see soon?

00:33:23 Kelly

There's more and more research and and fortunately, funding going into preventative medicine and personalised medicine. So there's some vaccine trials now for Lynch syndrome and this is an international effort. So that's quite exciting where we might think about not better treatment of cancers, but actually avoiding the cancers developing in the first place. And there's a real opportunity for Lynch because of the way that we've understood the genomic mechanisms behind the chances of developing cancers and how that happens that you know, we can actually consider having people vaccinated to lower their chances of developing those cancers. So that's a better situation to be in than having to go through treatment even if someone survives, there's often side effects and long term things that they experience from going through cancer treatment. So if we can avoid cancers altogether, then we're in a much better position. And that gives a lot of hope, I think, to families in the next generation.

00:34:27 Paul

That sounds really exciting and and Emma, why do you think it's important, people have knowledge about genomics and and Lynch syndrome?

00:34:36 Emma

The things that unite us genomics unites all of us, you know, in good and not bad ways, but it it really does unite us. And and what I've also found is that regardless of what genetic conditions someone might have, fundamentally, we all want the same things. We want to understand more about what that that diagnosis means, not just for us, but for our loved ones. We also want great communication where we understand what we're being told and and to know what that sort of process is to live long as well as possible.

So for me, Genomics is and genetic testing is really important because it's, if we can demystify it so that people feel that this is them taking control of their own life, not their own destiny, because there's a lot of things that can't impact that. But in the same way as when we get into a car, we put a seat belt on, you know that's that's a safe way for me. A genetic test is almost a similar version of having putting a seat belt on because it means that I'm more aware of things if I have something that I think isn't right, I'm more likely to go to a doctor now, whereas before I might have explained it away as something else, so I really would agree with what Lydia said about knowledge being power, and I really agree with what Kelly was saying in terms of it's so important to understand, to genetics what it is and the impact it can have and also take hope from the scientific changes that are happening. Because that also gives you more hope for going for that sort of preventative, possibly preventative surveillance.

00:36:12 Paul

And Kelly, where can professionals find out more information to educate themselves?

00:36:17 Kelly

The Macmillan Toolkit is a great example of really accessible education for healthcare professionals, and we've talked about demystifying genomics, and I think that's so important because all healthcare professionals are now recommended to have at least a basic knowledge about genomics and the fact that it can be important for patients to have access to genetic or genomic testing to give them more information and more choices for better outcomes and treatment and prevention. So they don't have to have the high level of specialist knowledge that we have in the genetics service. In fact, often when people are going through a diagnosis and having genetic testing or genomic testing, it might be just one more thing that's discussed in, for example the oncology clinics. But it's really important that that this is done routinely and offered to people so that they have the best chance of having all the options open to them, and then we always have the genetic service as a specialist referral. Once something is found like Lynch syndrome in a family and then for the relatives to come in their own time for genetic counselling. We will be there to support them and we have things like a National Register for colonoscopy screening now,

which was a big step forward. So there are ways that we could become more organised to better support families with Lynch syndrome. But the most important thing that we can do is listen to people like Emma who've lived through through this. They have experience and they can tell us what it was like for them having the testing, going out, trying to find the right information coming up against healthcare professionals who didn't know about Lynch just didn't know about genomics and how we can improve, support and care pathways to make it a better experience for everyone.

In my PhD research. I've been co-designing a website with people with Lynch. Other patients in the Community and as part of a patient panel and a big international group of expert stakeholders so that we can create this information hub and signposting to great charities like Macmillan and peer support groups and the NHS website. And help people like Emma who years ago were sort of told that they had lynch and then went off to grapple with it on their own. They were looking up things and meeting people who might have told them things that might have been helpful but maybe not relevant to their situation. So really having a central trusted source of information and support and there's two main sections of this website called Lynch Choices which contain decision aids. So these help people work through kind of what matters to me when I'm making a decision about something like taking daily aspirin to lower my chances of developing cancer or having hysterectomy to prevent gynaecological cancers and working through kind of what does it mean to me at this time? Because we know decision making is so highly personal depending on people's life context, there might be other people in their life who are going to support them or might be influential to them in making that decision, and time is so limited in clinics and and we want to preserve that for the really personalised genetic counselling that can help people to feel supported to make the right decision for them at the right time.

00:39:53 Paul

Emma, anything else you'd like healthcare professionals to be aware of?

00:39:57 Emma

I had a recent experience where I received 2 letters on the same day for my surveillance for my colonoscopy. One from my trust and one from the bowel cancer screening programme. Now I'm fairly intelligent and I thought it was for the same. I thought that this is good, they're really aware of what's going on. So when I was then phoned by my trust and said right, you know, let's make your appointment. We'll have your telephone interview to go through the procedure with you. Made the appointment all fine. I completely forgot about it. And then a week later I get a letter from the bowl cancer screening programme to say

we've booked your appointment for a telephone consultation with your specialist nurse practitioner for this date, which was two days after I was actually having the colonoscopy in my trust. And I then frantically phoned the bowel cancer screening programme, saying I'm so sorry, but I'm already booked in. And they and I said, can I just ask why I'm I'm very confused. I thought this was the same thing.

And it was really useful for me to have this explained in layman's terms. So they were saying, quite rightly, that the bowel cancer screening programme has been there to try and reduce some of the burden on the NHS in terms of surveillance. So taking over that responsibility for people who are having regular routine surveillance in terms of colonoscopies means that the, hypothetically the NHS trusts are in a position to be having procedures for people who are living with cancer or are going through that treatment. I think for me as a patient that was great that I had that opportunity to have that conversation and also through working with the Kent and Medway Cancer Alliance and the SE Genomic Medicine Service Alliance, I was even able to put that question to the head of the bowel cancer Screening programme saying, you know, if everyone's getting 2 letters how do they know? And fundamentally for me, I think it was quite difficult because the bottom line was I understand the rationale of the bowel cancer screening programme. But since COVID, there is a backlog and some trusts are behind in terms of surveillance. So that's again, some people will only receive one letter from the bowel cancer screening programme I was fortunate it's better to have two than than none.

But I think one of the things for me that's quite tricky that I would like patients to have an awareness of is where they fit in that puzzle because fundamentally I was told it comes down to choice. I can choose to have that procedure under the bowel cancer screening programme or I can choose to have it with my trust, even though I'd still have the procedure at the same hospital. It's under that sort of who is responsible for that information. And I think when you're, it's already tricky understanding where you fit and what to do and you're a bit nervous about colonoscopy, but it's choosing is almost a bit alien. I almost want to be told this is what's happening. We've got a bowel cancer screening programme because it's for surveillance you're just going to hear from them. I think if patients are in that position where they're hearing from their trust, who they potentially have a pre-existing relationship with and a separate organisation and they don't realise that they're the same but slightly different and it's down to choice, I think potentially that's muddying the waters. So as a patient, I would almost prefer it and I know everyone's different to be told this is how it is and this is where you're getting your surveillance. As long as I'm surveilled, I don't mind who does it. But it's just knowing where you fit in that that sort of big old jenga puzzle of logistics. I think that for me is really important to just highlight what Kelly was saying about the programme?

00:43:40 Lydia

It's about streamlining that service to make it as simple as possible. Make it really clear. So we're just gonna move on to the three questions.

Before we dive into our regular feature I'm thrilled to announce that our next episode on Genomics will be released in November. From Kelly and Emma's discussion today it's clear that genomics is a vast and fascinating field. We'd love to hear from you. What aspects of genomics intrigue you the most? Share your suggestions by leaving a review or send us a message at professionalspodcast@macmillan.org.uk. Your feedback is invaluable to us.

So the first question, if you could go back in time, what piece of advice would you give yourself?

00:44:25 Kelly

Involve patients and the people who will be using the services in designing those pathways because both parties learn a lot in that process and it's so important to to hear from the lived experience.

00:44:40 Emma

I would encourage myself to advocate more and ask more questions, particularly around the sort of gynaecological issues. I think once the the information had settled, I would have said to myself no, go and ask what they actually mean and what the process is for that, so that again, it's all about empowering yourself.

00:45:02 Lydia

And what change would you like to see to improve the lives of people living with cancer?

00:45:09 Kelly

I'd like to see continued progress in the prevention of cancer and personalised treatment and really supporting people to make decisions that are informed by genomic testing where that's available to them.

00:45:24 Emma

So for me it's around looking at treatments that are tailored to an individual's genes, so that might be around making sure that genomic testing is helping to lead towards really specific treatment pathways or, it might be around opening up the conversation about lifestyle changes that people can make to support that prevention of cancer, because fundamentally what we want to do is live well, not suffer and live as long and happily and

healthily as we can. And so using that information to aid that lifestyle, it would be the advice I'd hope.

00:45:59 Lydia

And finally, what would you like listeners to take away from this episode?

00:46:05 Kelly

Really take a step back and listen to Emma's story and with the benefit of hindsight, hearing about the impact of how it was for her being told about a genetic diagnosis of Lynch and then her experience thereafter because it's it's really important that we listen carefully to people's experiences and think about how we can improve things going forward.

00:46:30 Emma

I think it's about medical professionals and listening to and learning from patients. It's not always negative things. Patients will often tell you it how it feels. So it's separating that head and heart issue and thinking about when you're giving someone a diagnosis, even if it's just for a genetic condition and not a cancer diagnosis. It's around how am I communicating that in a way that the patient isn't left feeling more vulnerable than when they walked in, and if they are, what support are we putting in place so that patient doesn't feel on their own, even if it's terrifying when they leave. And I think it's really important that there is that joined up thinking between practitioners. So that patients aren't sort of caught somewhere in the middle or this nowhere land. So I think you know improving that communication that joined up thinking and having the ability to listen to patients and their ideas, I think would be really empowering.

00:47:27 Paul

Wow. Thank you, Kelly and Emma, for such valuable insights and expertise which you've brought to the discussion. We've loved having you here and we really appreciate you taking the time to join us on the podcast today.

00:47:40 Kelly

Thank you so much for having me. It's been a really interesting and enjoyable discussion and I look forward to hearing from other professionals who listen to this podcast and who might want to learn more about genomics. It's really important for everyone to to know about this and I hope they find it interesting and exciting as I do.

00:47:58 Emma

Thank you so much for having me.

00:48:01 Paul

You've been listening to the Cancer Professionals Podcast, which is brought to you by Macmillan Cancer Support. If you work in health or social care, visit macmillan.org.uk/learning to find out more about our learning hub, where you can access free education and training. For links to the resources mentioned see the episode description.

00:48:23 Lydia

If you enjoyed this episode, follow us so you don't miss our next conversation, where we'll be joined by expert guests to learn more about immunotherapy.

00:48:31 Paul

We'd love you to rate our show and share with your colleagues. Get in touch with us by emailing professionalspodcast@macmillan.org.uk. New episodes are released on the first Wednesday of each month.

00:48:44 Lydia

I'm Lydia.

00:48:46 Paul

And I'm Paul and you've been listening to the Cancer Professionals Podcast by Macmillan Cancer Support.