Centre for Personalised Medicine podcast

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Sharing genetic results within families

SPEAKERS

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Rachel Horton
Welcome to the Centre for Personalised Medicine podcast, where we explore the promises and pitfalls of personalised medicine and ask questions about the ethical and societal challenges it creates. I'm Rachel Horton and I'm here with Gabby Samuel, and in today's episode we're talking about how one person's genetic results can impact others in their family.

We're joined by Professor Anneke Lucassen, Director of the Centre for Personalised Medicine in Oxford and the Clinical Ethics, Law and Society group, and Dr Lisa Ballard, Senior Research Fellow in the Clinical Ethics, Law and Society group at Southampton, who have done lots of research exploring what confidentiality means when thinking about genetic results, and looking at challenges in helping families share information about genetic risks.

Anneke, would you mind starting off by talking us through why it might be important to share genetic results within a family?

Anneke Lucassen
Some conditions are due to a particular inheritance that may only have been discovered in one person, so other family members may be entirely unaware of the possibility of them having inherited it. So take, for example, a gene variant that causes a very high risk of breast or bowel cancer that may have been discovered in one person in the family because they developed a very young onset cancer, but their relatives may not know about that, and may not know to go and ask whether they too are at risk. And so it may be really important to say to those relatives, actually, you could have a test to see whether or not you've inherited the same variant that causes a high risk of this particular condition.

Rachel Horton
So if we take a genetic result as just being kind of personal to that one person, and don't think about their family, a lot of people could miss out on health care that might be quite important for them?

Anneke Lucassen
Yes, I think that's right. What you're hinting at Rachel, is that we tend to think of medicine as very personal to the individual in front of us and sometimes forget that there are important consequences for others around them. And that might be in the pandemic, where we realised that if you'd been in contact with another person, that they might be at

increased risk of infection. And that therefore, there was a responsibility to try and find those people to alert them to being at increased risk, or to protect them from transmitting it to other people. And similarly, in inherited conditions, you might need to let people know that they're at the same risk of inheriting that condition that you happen to have found in another person. So you know, you may have found it because that person is displaying a beacon, or… I don't quite know what the right word is. But they're displaying some sort of obvious sign of that condition. And that's why they've been tested for it. Other people may not be there yet.

Gabrielle Samuel
Anneke, if there's this need to inform family members, in case they're at risk, how does that work? Because I suppose the processes that are set up at the moment in the healthcare system are very much around individual confidentiality. So how does that process work in terms of informing family members?

Anneke Lucassen
Yeah, it's a really interesting question, because we've just had that sort of experience in the pandemic, of contact tracing. But we've also got a lot of experience, in sexually transmitted diseases, of contact tracing, that we don't tend to think of as being in any way related to that contact tracing in genetics.

And in answer to your question, the practice has arisen rather sort of ad hoc. We usually say to relatives in whom a genetic test has found something, can you let your relatives know? Ask them to get a referral to our service so you too can be tested. And we don't tend to know how often that happens or not. You know, the relatives might live in another part of the country. So we won't know if we don't get a referral, whether that communication has happened successfully or not. Or whether it's happened successfully and the relative has chosen not to be tested.

It's a bit of an ad hoc process that sometimes we're challenged to think about when a relative says “no, I'm not telling anyone else in the family. It's my secret and nobody else can know”. Then I think we sometimes say- or we ought to be saying- “what's my responsibility as a health professional here? Do I need to weigh the interests of these other perhaps unknown people, but known in the abstract to me, in the balance with this person's wish to not communicate that to their relatives?”

I should just say, really, that in genetic practice, it's very common to start with taking a family history from someone. So the very first thing you might do, even before you've done a genetic test is say, “tell me about your children, your brothers, sisters, your aunties, uncles”, and you draw a picture of that, and you often take quite a lot of details of those family members, so that by the time you've got a genetic test result, you as the clinician might know of, say, four or five people in that family by at least first name and date of birth, that you think, “Oh, actually, they have an interest in knowing about this result”. So perhaps there's a particular tension in genetic practice, in that we start off by asking about family details of other people.

Gabrielle Samuel

It makes me think, and I suppose this is to either you, or Lisa, that puts quite a lot of responsibility on to the person who's just been perhaps diagnosed with a particular condition, and I suppose might be in a difficult frame of mind. But also, I suppose, also the relationships that they have with their family members, and how comfortable they are to disclose that information. Are there tools or like, helpful guidance that you provide for them to help them through it?

Lisa Ballard
So I guess before I start talking about the tools, it'd be helpful to talk about, like you said, Gabby, those emotions and things that come up, and what a responsibility that is for the patient. And what Anneke said- in clinic, we truly don't know which relatives get informed and which don't. The only reason we'd know is if they came to clinic. But what the research shows- there's been lots of research studies on patients sharing with relatives, and the research shows that there's a significant proportion of relatives that don't find out this health information, or maybe they do but not in a timely manner, or maybe not all the information. And that research also details the reasons why, and some of the research we've done in our own research group, as well has kind of highlighted that.

And you've already mentioned that really, so that emotional toll, so you've got a patient in front of you that maybe has just been given… say, they're at risk, or they've been given a diagnosis. And then they're not only dealing with that, but then the responsibility of sharing that with their relatives. And there might be aspects of guilt, especially if it's to a child that you've passed it on, or guilt that you don't want to kind of inform your parent that they've passed it on to you. And there's lots of other issues as well, that the literature kind of highlights, things like family dynamics, so people not speaking to family members. There's kind of thinking, "yep, I'm going to tell that person but I don't know what to say, I don't know when's the best time" or they've just had a baby, or they're just taking their exams- it's not a good time. And also, there's that element of misinformation as well. So there's one that crops up a lot in the research of people kind of saying things like, "Oh, well, it skips a generation" or kind of things like that.

So you're right, Gabby, there are tools. So one of the… I think the widely used tool is the To Whom It May Concern letter. So the health professional might write a letter, maybe Anneke can correct me about what exactly is in the letter, but kind of general about, you know, "someone in your family has come to the clinic, and it might be in your best interest to kind of contact us and talk about it".

Anneke Lucassen
Just to agree with you, Lisa, that often, those letters are rather sort of abstract and bland, and they're given to patients to give to their relatives, but they may not find that very easy, or they may wait for the next funeral and then think, "Oh, that's not quite the right time to do it either". We think as health professionals that we've done our job- tick, family communication sorted, but we don't really know where they end up. And I suspect a lot of them end up staying in a crumpled heap somewhere on a pile.

And the other thing I just wanted to add, more adding to my earlier point, but given that NHS resources are so much more stretched than they used to be, we often only see people

once or twice now rather than over several appointments. So we sort of run through this checklist of “got to get everything done, oh, To Whom It May Concern letter, tick, done”. And off it goes without really guiding the patient through what they need to communicate, rather than just pass on the letter, what is the essential information that needs to be transferred, and how they might be helped in doing that? And I think your two apps that you've created, if you can say a bit about those, I think they're really helpful.

Lisa Ballard
Thanks, Anneke. Yeah. So I think, again, some of the research that we've done in our research group shows that patients do want to do this themselves? So like in sexual health and maybe in like infectious disease, to some extent it kind of is done by the health professional, or like by computer programme or whatever. But actually with genetic test results, patients really want to communicate that themselves. So they don't think it's the health professionals responsibility, kind of generally, but they do want a bit of support with that, and that's what we've done in our research group, and with people from other faculties like Web Science at the University of Southampton, we’ve developed some web-based interventions.

So the interventions kind of tackle that stuff that Anneke highlighted that we just don't have time to do in the clinic now. So it kind of it looks at that capability. So what does the patient need to ensure that they can pass that information on? And they've got the skills to have that conversation, so kind of action planning, and maybe developing scripts and stuff. And the opportunity- planning out, when is a good time? Like Anneke said, maybe waiting for the next funeral’s not great. And the time element of that is kind of incorporated into our app. So we've got this option where you can electronically send the To Whom It May Concern letter or your test result, or you can construct your own message, and you can get it out there quite quickly. You can even send it anonymously, if you want to keep it anonymous, or there's like those tricky family dynamics. And then working on that kind of motivation element. So just highlighting, why is it important that that person gets that information? And kind of capitalising on that… those the kinds of elements, those behaviour change elements that we've built into our web app that can do some of that stuff that a busy health professional just cannot get done, and is something the patient can do in their own time.

Gabrielle Samuel
I find it so fascinating, this kind of idea that you're developing an app to really help individuals in this particular situation. And it's really making me think about the relationship between care and healthcare, and also between patient autonomy and paternalism, and they all kind of mix together. And here, it seems that the app is kind of ticking a lot of those boxes to ensuring the needs of the person are met. Are there any instances where that isn't the case? Like is the app out? Is it being used or are there any drawbacks to using app-based systems?

Lisa Ballard
So it isn't in use. We've got a patient group from the 100,000 Genomes Project. So the app has been heavily developed with those patients. So patients that have had a genetic test result. There's certainly drawbacks. I mean, I'm quite passionate, we do a lot of work in our

research group around underserved groups. And that's definitely one thing, I've developed this app with people that aren't from an underserved group- white middle class women really, is the group that I've developed this with. And that's something I would love to do some more work on- how can we take something like this and make sure that it's appropriate for other groups? And I'm sure there's lots of research out there around that. But there isn't an another app that does this, there isn't an app that works in this way, anywhere. So I think that's a great point.

Web applications are incredibly popular, and there's a good reason for it. They're cost effective, they don't take up the health professional's time, but they're timely for the patient. But not everyone has access, and the way it's written and how it's targeted, I think there's so much work that can be done to make sure it's useful for lots of other people, the people that really need it, the people that aren't communicating that for all the reasons that we've identified.

Anneke Lucassen
I can think of a few clinical examples that have come up either in my own practice, or nationally discussed at the Genethics forum, that have taken up an awful lot of time, because people have really worried about whether they're breaching confidence or not. And the usual scenario is- somebody's really angry with their clinician for suggesting they contact their relatives, because they want to be assured that their clinical information is kept confidential. They don't want that shared with anyone. And before you know it, it's become a very sort of adversarial situation where the clinician feels accused of wanting to share information inappropriately, and the relative says, "No, this is my information, you know, I'm not close to my relatives, they haven't been very friendly to me recently, just leave it with me and butt out", only to find that when that information is then followed up, say a year later, or you know, sometime later, that actually that initial communication got way too focused on the clinical information. And that when those people who start off being really angry realise that all that we're trying to do is let other people know there is a test available for them… and it doesn't have to say anything about that first person. It doesn't have to say this person has got cancer, or this person has had a test, and that's relevant to you. All we're trying to do is alert close relatives who we now know are at risk, but they don't know it themselves, that they could have a test… that all the anxiety falls away.

And I've seen people turn right around where they've, you know… one example, where somebody was really angry and didn't want any further contact with a clinician that they had met, when met sometime later, was really anxious to hear that those relatives that she'd initially been angry about, had heard that information, because she realised, after some time has passed, the tension about breaching confidentiality is completely gone because the patient has realised it's not about breaching their clinical confidence, it's about telling family members they're at risk, and then they're completely on board. And I think the app that Lisa talks about can be really helpful in that sort of situation. But not everybody will be using apps. So we've got to also think about other ways of supporting that.

Rachel Horton
It sounds like there's so much potential usefulness in the app, and especially because… I guess, as you said, at the beginning, it's funny that lots of the situations of family

communication that get a lot of attention and worry are the very rare situations where people at least initially feel they don't want to inform their family or might have concerns about what that would involve. Whereas actually, it sounds like there are so many people who would want to tell their family but just don't necessarily have the tools to do that easily.

Anneke Lucassen
Or we've presented it, we as clinicians have presented it the wrong way to them. We've talked about this being an issue that we need their consent for, rather than an issue about warning relatives of something more abstract that they can go and find out more information about.

Gabrielle Samuel
Does having something like an app alongside say, health professionals supporting and helping patients through this, raise questions about… not just where responsibility lies, but how that could be developed, I suppose into guidelines or policies, because… I'm not suggesting guidelines and policies are correct, but… ultimately, there needs to be somebody responsible for this decision?

Anneke Lucassen
I think it has to be a shared decision. But I think you're right, Gabby, that we're not very good as health professionals at thinking that one through logically, and usually, where we get to is "Oh we couldn't possibly have the responsibility to contact relatives, because they haven't been referred to us, they're not our patients. And we're too busy anyway, with doing our day jobs to then go and find a load of other relatives, you know, sort of trying to track down relatives in Australia that we haven't got contact details for".

But that's sort of deflecting from the issue. I think, for me, the issue is, if I know there are specific relatives, who are closely related to this patient in whom I found a result, what is my responsibility to ensure that they get to hear about that? And I think that responsibility that I have, has to be discharged together with the patient in whom I found that result or in whom that result has been found, to work to make a shared decision that these relatives do have an interest in knowing. So how can we best go about letting them know about that thing, they have an interest in knowing?

There's a recent court case that actually was helpful, as well as being a bit of a distraction here, because it was a case in negligence. What was helpful about it is that the judge said that clinicians have a duty to weigh in the balance the interest of the patient in front of them and their confidentiality, with the interest in their relatives in knowing something that puts them at high risk for which they might be able to take measures to somehow reduce that risk. And so we've got to do this balancing exercise, we can't just say, "Well, you know, can't do anything without consent, we'll stop there". That's (a) not good medicine. And (b) it just doesn't make any sense really, in genetic medicine to think that everything can be solved by consent, because genetic medicine is all about how people are related and shared inheritances that may just be found in one person, but don't therefore belong to that one person to veto the use for anyone else.

Gabrielle Samuel
So it's about shared responsibilities from both the healthcare professional and also the patients as well. And then having an app that can be used to support that responsibility as patients go on the journey.

Anneke Lucassen
Yeah, but also I think, recognising that if, you know… let's create a scenario where that absolutely doesn't work, the app doesn't work and the patient doesn't want to tell anyone else. I think these are rare as hen's teeth. There's usually some other explanation but let's just imagine that you have a patient who absolutely does not want their relative to know, because they're worried that that will have some sort of implications for them. I think that clinicians then still have a responsibility to think about those relatives, and to think about communicating the risk to their relatives without breaching the confidence of that patient. So the responsibility doesn't stop with "Oh I've handed something over to the patient". And it doesn't stop with "Oh, I haven't got consent. So therefore I can't do anything". That responsibility continues.

Gabrielle Samuel
It sounds like a minefield, doesn't it, Rachel? So many different stages of thinking through how to address these really complex issues.

Rachel Horton
Absolutely. Anneke, I've heard you talk before about kind of a joint account sort of analogy for thinking about, I guess, these issues, that genetic information might not just be in one person in a family? Would you mind telling us a little bit more about that?

Anneke Lucassen
Yeah, I mean, the joint account analogy, I think has been very useful to think about these issues. But also, people have been quite critical of it. Because we started off by saying, "well, it's like a joint bank account, you've both got rights of access". And then everybody said, "Oh, bank accounts are completely different". And so I actually liked the sort of development of that, that called it a joint account as in a joint family story rather than their joint bank account. But for me, that analogy has been useful, because it's led me to think there is a difference between something that we all inherit- I know that I share more genetic information with my parents than I would with you, Rachel, or you, Gabby, and that I might need to let other people know about their potential inheritance, without that saying anything about how that inheritance has shown itself for me. So if I, if my sister gets to hear of the particular gene that's caused my symptoms, that won't tell her about my symptoms, it just tells her about her own risks.

Gabrielle Samuel
Lisa, from your experience, and speaking to patients, do people have an understanding of genetic risk, and what are their views on sharing genetic information? Does it kind of align with I suppose, how healthcare professionals see this? Or is it a slightly different perspective?

Lisa Ballard

I couldn't comment on their understanding of genetic risk. But what I can comment on is something that kind of relates back to that responsibility. In all my interviews, and that case that Anneke has kind of talked about- that unlikely case where someone refuses, and what is a health professional's responsibility? And I asked every person I interviewed, "do you think it's a health professional's responsibility to contact your relative and share that information, even if you refuse?" And, interestingly, quite a lot, over 50% of patients said, "Well, it's kind of not my responsibility for that person not to hear. So I think the health professional is kind of within their rights to share. It's not fair, either if we've fallen out or I've said "No", why should my relative suffer?" But the caveat to that is that they're just people sat in front of me, without a result to share with their relatives. So it's quite, very theoretical. They don't have all the complexities that we talked about before, but it's… it's quite interesting. Because I think health professionals are often well, quite rightly, very cautious. And, you know, we're all bound by, you know, thinking about confidentiality, Caldicott principles, we have it kind of drilled into us, whereas the patient is a bit freer from that. And to kind of hear that they don't think it's beyond the pale for their health professional to contact their relative, I think it's interesting.

Anneke Lucassen
I think your research, and Sandi Dheensa's research, has shown that patients most definitely construct their inherited genetic information as something different to their clinical information, and that they would say their relatives have a right to know what they too might have inherited, without that saying anything about their clinical information. So I think the patients that you've interviewed over various research projects have a better ability to differentiate genetic information from clinical information, than often the health professionals looking after them do. Because the health professionals often said to us "Oh our patients wouldn't want that". When the patient said, "Oh, well, I'm sure our health professionals would tell our family members anyway". So there was a big misunderstanding, in that research.

Rachel Horton
So curious that there's that really different perspective on what other people might think. If you had to pick one message for people to take away from this podcast, what would it be?

Anneke Lucassen
I think we should think about genetic information much more like infectious disease information, where we wouldn't worry so much about communicating the risk to others, even if those others infer where they might have picked the infection up from. I think we could think about that model much more in genetics than we've been able to do so far.

Lisa Ballard
I guess, like, my main message would be to the research community and health professionals, for us to start thinking about how can we go beyond that, that To Whom It May Concern letter? What can we do to make it as easy as possible? Because the research shows that patients want to do it themselves. And they know the importance, they understand why it's important, but then there are all these other barriers as to why it doesn't get done, or doesn't get done in a timely manner. So, to really start thinking, what can we do to make that job, that difficult job of passing on that information to their relative,

how can we make it as easy as possible, remove as many barriers as possible? And to do to do a bit more around that, I think.

Rachel Horton
Thank you for listening to this episode of the Centre for Personalised Medicine podcast. If you'd like to find out more about personalised medicine and its promises and challenges, please visit the Centre for Personalised Medicine website at cpm.well.ox.ac.uk.