Centre for Personalised Medicine podcast

Series 2, Episode 5

Who's 'the patient' in genomic medicine?

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 Gabrielle Samuel, and in today's episode, we're talking about the meaning of 'the patient' in genomic medicine. To help us think about this, we're joined by Dr Susie Weller, Senior Research Fellow at the Clinical Ethics, Law and Society group at Oxford, and Research Fellow at the Centre for Personalised Medicine, whose research explores the ethical and social challenges that arise for those living and working with genetic and genomic results.

Thank you so much for joining us today, Susie. Please could you start by telling us a bit about how you got interested in this question of who's the patient in genomic medicine?

Susie Weller
Yes, thank you Rachel. Over the past few years, I've worked on a number of projects focusing on understanding patient experiences. And as a social scientist, I'm particularly interested in the wider impacts of long-term health conditions, how individuals and collectives including families navigate and understand the impacts of health interventions, the kinds of resources and support on which they draw, and how caring relationships, identities and practices evolve over time. As you mentioned at the beginning, my current work explores the ethical and social issues that arise for those living and working with genetic and genomic results.

Rachel Horton
So, I'm really interested to hear more about this Susie, because I guess a lot of your work from what I've read, it's kind of challenging this focus on patients as people on their own in a vacuum making decisions. Could you tell us a little bit more about what you've been looking at recently?

Susie Weller
Yeah, so as I think in, in most areas of medicine, it's really common to be focusing on assessing, diagnosing and treating individual patients. Viewing patients as autonomous and independent decision makers stems largely from the ethical principles outlined in the Nuremberg Code, which were intended to protect individuals from a repeat of the unethical medical experiments endured during World War Two. So this idea of personal autonomy forms the cornerstone of contemporary clinical practice and research in many international

contexts.

But for me, genomic medicine really highlights the inadequacies of this individualised way of defining a 'patient'. So we know that patients are likely to have family members who may be directly affected by the outcome of tests in others. And professional guidance is increasingly taking the view that genetic information should at times be regarded as of relevance to families rather than individuals. And also, clinical genetics professionals often see themselves as family practitioners with a wide range of obligations and responsibilities to different relatives, and a long history of talking with and about multiple family members, including across generations.

Rachel Horton
Yeah, I guess in clinical genetics, that's sort of the way I had been taught to think about, thinking about all those kind of genetically-related people who might be impacted.

Susie Weller
Understanding the family history is important for determining eligibility for testing, assessing risk to others, and interpreting the results. So, while a genetic test might provide a diagnosis for some, for others, it will predict future patienthood with varying degrees of certainty or uncertainty. Rather than just being about the individual index patient, some findings might prove more important, at least temporarily, for the health of a relative of a patient who was originally tested.

This really brings into question who we regard as a patient, it's not just about those who are clinically ill, it's also about those who are at risk or healthy carriers. And as genomic medicine starts to become more a part of mainstream care in the UK, it's all the more important to think about the potential implications for those beyond the index patient. And that's where my interest really lies.

Gabrielle Samuel
So if that's where your interest lies, how do you go about researching this area, like, who are the patients that you speak to when you're thinking about these issues in your own research?

Susie Weller
Now, that's been interesting. So this, the work that I'm drawing on forms part of the Ethical Preparedness in Genomic Medicine study, or EPPiGEN for short. And we've been working with index patients and wider family members, and also a wide range of health care professionals. So we're really, as part of this work, we're really keen to understand how future patients and healthcare professionals might prepare for the ethical and social challenges they may face as genomics becomes part of mainstream care.

One of the EPPiGEN projects is concerned with documenting the journeys of those affected by the process. So that might include patients, individual index patients, it might include parents of young patients, also a range of other family members, including partners, and adult children, some of whom may not be biologically related. And we're using a qualitative longitudinal research design, which basically means we speak with participants periodically,

traveling alongside them to help understand their experience. Some of these participants have accessed whole genome sequencing via their involvement in the 100,000 Genomes Project, and some through the NHS Genomic Medicine Service.

Gabrielle Samuel
So I'm quite interested in that, because I can imagine that there are quite a lot of studies that have looked at patients, not just in genomics, but more broadly? And so if you're starting to think about conceptualising patients within genomics, and that kind of individual versus broader conceptualisation of the patient, are there any theoretical models that you can draw on from elsewhere that can inform your research?

Susie Weller
Yeah, to help sort of understand the experiences of our participants, we've drawn on a concept of linked lives, which is from life course theory, often used by sociologists and other social scientists. And this linked lives lens helps us focus on the ways in which the lives of individuals shape and are shaped by those in the networks in which they're embedded. And it helps us think about the ways in which life courses of individuals are intertwined with others. So it's not just about present connections, but it's also about those located in the past. And it's also about future imaginings, so for instance, thinking about potential future grandchildren.

And this approach also links interconnected lives to wider social processes. So events, circumstances and decisions made in one generation shape the lives of future cohorts. So adopting this notion of linked lives as a conceptual tool to explore journeys through genomics has helped us really think about who constitutes the patient, how patients and family members think and talk about what it means to be a patient. And what these understandings might mean for practice, particularly ethical decision-making.

Rachel Horton
That sounds such a fascinating way of looking at things. I guess it makes so much sense really, that we make decisions, thinking about the impact on other people and kind of influenced by them. But it's just not something that I've often seen brought into lots of, you know, when you think about autonomy and what it means, it's so interesting to kind of bring in those that wider circle of people around the index patient- what did you find talking to patients?

Susie Weller
It was very common for our participants to talk about the process as a collective endeavour, this kind of shared journey, albeit experienced from different perspectives. And I guess this is most apparent in the way that parents who've been tested to try and help make sense of a genetic finding in their child discuss their experiences, but we also came across a wide range of other participants talking about it in terms of a more collective endeavour.

Gabrielle Samuel
Could you give us an example of that, like, how they speak in collective endeavours, just so we can get a flavour of what your research is showing?

Susie Weller
Yeah, of course. One example of a more collective understanding was provided by a couple, William and Maggie, we've spoken to William and Maggie on four occasions so far. Together, they have two adult children. And William was diagnosed with a neurological condition about fifteen years ago, and it was suspected that his father also had the same condition. So William's genome was sequenced to explore a genetic cause.

And so far, he's received a letter stating that nothing has been found, so he's living with this probable diagnosis of a rare inherited neurological condition, which could have implications for their adult children and potential future grandchildren. And much of their desire for greater certainty centred around expectations and imaginings regarding their children's futures. And they really hoped that William's, the whole genome sequencing might help provide some certainty.

So in many respects, William was the patient, and much of the discussion focused on him coming to terms with the condition, the onset and progression of symptoms and his pragmatic approach to adapting to physical changes. So for Maggie, her own genetic information played no part in the process. And she would not conventionally be recognized as a patient. But how she described their experiences and how she positions herself within the process, suggested otherwise. She's very much an inherent part of William's journey, not simply in terms of caring for and about him, but also in terms of making sense of their journey through genomic testing as a couple. So she went to appointments, she was very proactive in the management of his condition, and was very concerned about whether it might develop in the children they share.

So encounters with healthcare professionals, moral deliberations about the testing of their children, and anxieties about potential outcomes were similarly felt and shared by them. And across all of the interviews, it was really interesting because Maggie in particular, described the whole process and their experiences, in terms of 'we' and 'us' from being a patient and making challenging decisions and facing outcomes or uncertainties. Their story really resonated with Richard Settersten's work on linked lives and couple formation, where he argues that 'it's the story of *us* that counts'. And that really reflected William and Maggie's experiences.

Gabrielle Samuel
I find that absolutely fascinating, that idea of collective experiences. And thinking like more broadly outside of genetics and genomics, I know, it's not your research area, but I can imagine, like, for example, cancer patients, like when they visit clinicians with their relatives. And that's like, that's quite collective as well? And like with relatives using terms like 'we' rather than 'they', and I just wonder if something like that you've considered this idea of collectiveness and whether it should go broader than just genomics?

Susie Weller
I think so. And I think it will have wider implications, as, you know, genomics, becomes more part of mainstream care and other specialists also have to perhaps think about in these more collective terms, but I do think it certainly would resonate with other areas of research. I do think it's, it's an apt way of thinking for other areas of medicine as well,

actually, that, that genomics perhaps provides a really pertinent example of it but it's relevant for other areas.

Rachel Horton
Such as challenge how to sort of incorporate that, I guess, into clinical practice. In all the computer systems it will be William's name that goes in the box or whatever, yet, it's so evidently happening to Maggie as well in lots of really important senses. Were most examples like that, it was a sort of a couple together, or are there any other kind of ways in which linked lives came to light in your conversations?

Susie Weller
Yeah, absolutely. So there's another really interesting example that focuses more on kind of intergenerational patient identities and one of our participants, is a really good example of this. So she was diagnosed with breast cancer in her 40s and she participated in whole genome sequencing with her sister and her daughter to look for a genetic explanation of their family history of cancer. And like William, she has also received a letter saying that so far nothing has been found.

Past and present experiences of cancer in her family and social networks shaped how Shirley positioned herself and others in terms of patienthood. And in the quest to gain answers, Shirley, like other participants took a much more intergenerational view linking her understandings to those of past generations, whilst also using her own experiences to reinterpret and question the past. So Shirley had lost multiple family members in quick succession. And it was this along with her hopes and expectations for the future health of her relatives that were really motivating factors in her drive to support genomic research, so to, to participate in whole genome sequencing, but also to be part of our study.

And for Shirley, cancer was shrouded in secrecy in the previous generation and by her sister. So for example, her sister had been told she was terminally ill, but had told all the other family members that she was okay, when she wasn't, and in response, Shirley strongly advocated much more openness and this featured in her drive to pursue genomic testing, and in her quest to ensure that her grandchildren, including those in their teens, understood the risks and were vigilant regarding potential symptoms.

As Shirley was all too aware, ensuring timely dissemination of such information could be key to an individual accessing testing, and prompt diagnosis and treatment. But there was also a moral imperative, being open about her own vulnerabilities and the potential heritable risk to others was the right thing to do for the health of future generations. So this was very much presented by Shirley as this like joint familial project, part of this kind of broader fight against cancer. And viewing Shirley's narratives through a linked lives lens really helped us think about the inter and intragenerational connectivity between patients' identities. So her understanding of this clear family history meant she regarded all family members and future generations as potential patients.

Gabrielle Samuel
I think that's really fascinating. If we go along with this, which I think is such an important way of conceiving how people make decisions about genomics testing, like how do you then

think your findings can help genomic medicine that as it becomes more embedded into the health system? And within the structure of, I mean, Rachel was saying earlier, right, a very individualized care?

Susie Weller
Yeah, I think as it becomes further embedded, more people need to be prepared to face a range of ethical and moral deliberations with, and or about the linked lives of those within their networks. The prospect of receiving certain or uncertain findings, and living with a result or results or uncertainties will become more commonplace. So I think it's really important to think about, and prepare for the duties that healthcare professionals may have to those beyond the individual index patient. So this might involve obtaining, recording, storing, sharing, and reusing data from multiple family members, as well as others embedded in the journey, but not necessarily directly involved in testing.

I think it raises important questions about what guidance and support ought to be given regarding how, when, and to whom heritable risks and potential findings are disseminated. In discussions with healthcare professionals, space, I think, needs to be made to incorporate the wider impact on participants' lives. And I think as I mentioned earlier that mainstreaming will mean that such challenges will be encountered beyond genomic medicine. For some healthcare professionals whose specialism lies outside clinical genetics, more nuanced views of both patienthood and family are likely to be necessary.

Gabrielle Samuel
Did you get a sense when you were talking to your patients, that the sharing of information amongst relatives happens already? It's just because I've done quite a lot of research around thinking and talking to patients. And there's sometimes this assumption that it's already happening within the NHS and I was just wondering if you came across that at all?

Susie Weller
I think from the perspective of the participants that we've spoken to so far, it's much more about them being proactive themselves, in terms of sharing it quite widely actually, and being very proactive in tracking down people. So we have several examples of participants who have traced relatives with whom they have very little, very little or no contact, estranged relatives overseas, with this possibility that there, there might be some implications for them, and also others who've traced family through websites such as Ancestry to try and track down as many people as possible. So being incredibly proactive about it.

And I think there's lots of issues there, actually, in terms of what you're what you're actually sharing, how much you know, as the two examples I've given here, actually know very little at the moment, they don't, nothing has been found yet. But they're still sharing the possibility. There's something there might be implications for other family members, quite distant family members as well. But I think the interesting thing that's come from this work so far, is this the involvement of non-biological relatives and the implications for them and how this 'story of *us'* involves more… people in the network. So it was it was more than just about shared decision making, it was about participants' feeling an inherent part of the journey, and experiencing it alongside and with others.

Gabrielle Samuel
I imagine that also raises connotations in terms of support, right? Because if we, if we view non-genetically related individuals coming along the journey with them, then I suppose emotional support issues come along with that, right? And then from there questions of where that support comes from? Have you thought about how that links in as well?

Susie Weller
Absolutely. And we haven't finished all the analysis for this project yet. But I think some of the sort of preliminary findings coming from that suggest a lack of support in that area, and participants feeling quite unsupported emotionally.

Rachel Horton
Thank you, Susie, that has given us so much to think about, and it's been really, really great to hear more about your work. Could I ask a question we ask everyone- if you had to pick one message for people to take away from this podcast, what would it be?

Susie Weller
I think that people are situated in networks of relationships, and one person's decision will have consequences for others. And often these consequences for others are a key influence on the decisions that they might make.

Rachel Horton
And how can we find out more about your work?

Susie Weller
We've published a paper on this aspect of our work in the journal Social Science & Medicine. The paper is called ‘Reimagining the patient’ and it features in volume 297. I also tweet project updates @DrSusieWeller1.

Rachel Horton
Thanks Susie. It's such a brilliant paper and so kind of you to take the time to talk to us about it.

And thank you very much 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.