**Centre for Personalised Medicine podcast**

**Season 3 Episode 1**

***What is personalised medicine?***

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(Our podcast logo features a section of the artwork [‘A Lifetime of Measures’ by Aneesa, aged 12, from Oxford High School](https://cpm.ox.ac.uk/centre-for-personalised-medicine-art-competition-2022-23/), the stunning winning entry to our 2022-23 Youth Art Competition).

**SPEAKERS**

Rachel Horton, Anneke Lucassen, Susie Weller, Sarah Briggs, Sally Sansom, Ali Kay

**Rachel Horton**

Hi, I'm Rachel Horton, a Junior Research Fellow at the Centre for Personalised Medicine, or CPM. Welcome to the third series of the CPM podcast where we're going to be exploring aspects of our new ten year strategy. We're kicking off with an episode discussing what we mean by personalised medicine. Joining me to discuss it, I've got several of us from the CPM team.

**Anneke Lucassen**

My name is Anneke Lucassen. I'm Professor of Genomic Medicine at the University of Oxford. I'm also an NHS consultant in clinical genetics.

**Susie Weller**

I'm Susie Weller. I'm a senior research fellow in the Clinical Ethics, Law and Society Research Group, and I'm interested in understanding families’ and patients’ experiences of genomic medicine.

**Sarah Briggs**

My name is Sarah Briggs. I'm a Clinical Lecturer in medical oncology, and an oncology doctor working in the NHS. My research interest is in environmental sustainability in healthcare.

**Sally Sansom**

I'm Sally Sansom, I'm a health economics researcher, and my research interests lie in working out how best to measure the outcomes from genome sequencing for rare disease diagnosis.

**Ali Kay**

And I'm Ali Kay. I'm a researcher based in the Weatherall Institute of Molecular Medicine at the University of Oxford, and I'm interested in the implications of new genomic technologies, both for practitioners and also for couples who are considering their future pregnancies.

**Rachel Horton**

Thank you so much for joining me today to discuss this topic. This sort of comes off the back of a year, I think, where the CPM really focused on its strategy, and I guess, underpinning that, there's questions about, what do we actually mean by personalised medicine? And today we just wanted to reflect on that and what personalised medicine means to us.

**Anneke Lucassen**

Can I start by reflecting on what I thought when I was asked to take on the job of Director of the Centre for Personalised Medicine? Which was very much… what is personalised medicine? If, if medicine is not personalised, then what is it?

And so on the one hand, I can see that ever since medicine’s been around, there have been attempts to personalise it to the patient in front of you, and in that sense, it's nothing new.

But on the other hand, I think there's lots of developments over the last decade, two decades, perhaps, that allow us, through data, new technologies, to tailor our interventions much more to the person in front of us.

So we’re called the Centre for Personalised Medicine, but we could also be called the Centre for Predictive Medicine, or indeed, the Centre for P4 Medicine, which I think is a North American term that stands for predictive, preventive, personalised and participatory medicine. Essentially, they're all saying the same thing, all with a but, implied in there, I think, that that term doesn't cover everything.

So from our perspective, we are just, as a centre, thinking of ways both to improve personalisation of medicine and care, recognising that they might sometimes be in competition, and that there are limitations to how we do that by what we can measure about people. So we're very interested in thinking about the things that personalised medicine *doesn't* capture as well as what it *does* capture.

**Sarah Briggs**

And I think the definition that we have come to in our strategy as a group, is that personalised medicine is using information about a patient to try and tailor their treatment, or prevention, or intervention more to them. So it's sort of what we can *measure* about somebody. And sort of often people think of that in terms of being genetic information, but actually it can encompass a very broad range of different types of information.

**Ali Kay**

Thinking about the area that I work in, I consider personalised medicine in terms of not necessarily people currently being ill, or having a condition, but how they can make informed decisions about the future. So particularly in the case of pregnancy, we can use personalised medicine assessments to inform them on their risk, rather than relying on population-based approaches.

**Sally Sansom**

I absolutely agree that medicine has always been personalised, or at least has always sought to be personalised. But I think the term personalised medicine has been coined in recognition of the developments in big data, and particularly, or at least in the focus of my research, genomics and genomic medicine.

**Anneke Lucassen**

I think that's really interesting to reflect on, isn't it, because to some people, personalised medicine means ‘related to developments in genetics’, but to others, it will be something much broader. As a geneticist, to me, it's much broader, but I'd love to know what other people think.

**Rachel Horton**

It's interesting though. Like I wonder, if you asked, like, people who don't work in the field what personalised medicine means to them, would that connection between personalised medicine and lots of kind of technical data be the connection that most people make?

**Anneke Lucassen**

I think often it isn't, is it? Because Susie, you've done some really interesting research that highlights exactly how people did the opposite.

**Susie Weller**

Yeah. So we've been working with the Mass Observation British national qualitative survey, asking a panel of public participants their views on genetics and health in their everyday lives, and through reading their kind of written accounts of their own experiences, it's really interesting that the focus is much more on personalised care. And some really rich examples, they gave us some really rich examples of ways in which care felt personalised to them, and that often revolved around relationships and interactions and encounters, rather than more medical aspects

**Anneke Lucassen**

Or maybe they… what I remember from those discussions is that people often said the technology sort of passed them by, the technological developments, but it was the contact from the nurse or the doctor that they really valued, and that's how *they* interpreted personalised medicine. Almost the opposite of what we've just been talking about.

**Susie Weller**

Absolutely, very little mention in the accounts of kind of technologies or medicines, or maybe, maybe a few examples of interventions that were particularly tailored. But also thinking about the positive and negative aspects of that as well, who that's *not* including when we kind of tailor things to particular populations… individuals.

**Anneke Lucassen**

Yeah, I think that's a whole bit of the conversation that I'd love to spend a bit more time on, if we can, of who do we leave behind when we're personalising medicine for some people well- who misses out on that? Who gets less personalised medicine as a result? Because I think if we're advocating personalised medicine, we need to have an account of how we deal with those groups as well.

**Rachel Horton**

And the rhetoric around personalised medicine, I guess it's… I remember seeing an interesting talk, I think it was by Eva Winkler, and she had this sort of slide showing the idea of personalised medicine. And it was like all these people who are all like different colours, and they all got each an individual treatment. And then she had this slide where, actually it was the kind of 80% of the people stayed sort of grey. And then, you know, a couple of people got their personalised treatment. But I guess often with personalised medicine, at the moment, that sort of will be the state. There's this promise of these individualised, personalised medicines for people, but then actually a lot of people kind of are in the big block that gets nothing out of it currently.

**Anneke Lucassen**

And I think we've all been very enthusiastic about those promises, and I think that's right, there are lots of advances that we can expect, but now is also a time to think about the people who in that picture stay grey, and how we can serve them better.

**Sarah Briggs**

Yeah, I think one of the key areas that we've spoken about in previous CPM events, one of the areas we want to look at going forwards of sort of equity and thinking about who's included in personalised medicine research and who benefits from it. And we know that a lot of the data that we're using to sort of form the foundations of personalised medicine is quite biased data and doesn't include all members of the population, and that leads to sort of interventions that perhaps won't benefit those members of the population either.

And that's a real challenge going forwards for future developments in personalised medicine, and it applies to AI, which is an increasingly talked about field in, or area of personalised medicine, with the same really big issues around sort of bias in the data that goes into it.

**Sally Sansom**

I think beyond equity, it also really has some really interesting health economics implications. So if personalised medicine is only benefiting some groups, and we think personalised medicine is better and leads to better outcomes, that means there's still a proportion of the population who are, by comparison, potentially having worse health outcomes. And then thinking about the health system burden and other costs associated with that, really it's in ours and society's best interest to be making these treatments available for as many people as possible.

**Susie Weller**

That's really interesting, because that fits the work we've done on public perspectives, and the kind of really interesting points that participants in that made revolved around, the NHS is already overstretched, and that, you know, resources are limited, so we can't necessarily expect that from the NHS in its current state.

**Anneke Lucassen**

And that they might go privately to get that. When actually, it's not necessarily any better in the private setting.

**Susie Weller**

But the perception is that if you want that, then you might have to go down that route.

**Anneke Lucassen**

And it's interesting, also from a clinical point of view, I don't know if you'd echo this, Sarah, with your oncology hat on, that a lot of patients come thinking that with advances, a genome readout, for example, will personalise medicine in a way for them that they're very optimistic about, but that in 90% of the time we can't actually deliver? So I’m thinking of genome sequencing for cancers in terms of targeting treatments, it's only a tiny minority that that helps. And the expectations that people come with are often much higher than that.

**Sarah Briggs**

Yeah, it's interesting. I think certainly in oncology, we always counsel patients when we're talking about whole genome sequencing that the return rate is very low. And, you know, there are challenges associated with trying to interpret the data and find useful ways to apply it.

But I think, I think generally, patients tend to be quite enthusiastic about doing it, because often the sort of situations where we use it at the moment are where really you're coming towards the end of treatment options, and you're, you know, even a small, small chance of producing alternative treatment seems worth going ahead with. But I think that conversation, will probably change over time, as sequencing becomes more widespread.

And some people are worried about it, because, you know, alongside the cancer sequencing, for looking for sort of changes that we might be able to target with drugs, we always sequence the germline genetics, so looking for potentially heritable changes. And actually, that's a very different discussion. And for some patients, that's quite challenging, and not everybody wants that information about their family background, so it's quite a nuanced discussion that we're having to have increasingly in clinics now. And yeah, it's interesting.

**Sally Sansom**

I think that's really interesting to contrast perhaps the oncology and rare disease perspective, where oncology, of course, there's a large treatments focus, as there is in rare disease, but with such a large proportion of rare disease patients being undiagnosed, the personalisation aspect there could just be simply receiving that diagnosis. And we know that genome sequencing style technologies can significantly change the likelihood of receiving an answer in those cases.

So perhaps, while the treatment side of things may be further along, perhaps we're kind of moving towards that in a stepwise change through these technologies.

**Ali Kay**

This is one of the reasons why we've approached looking at personalised medicine through a number of different themes. So our themes relate to diagnosis and treatment, risk and prevention and health system boundaries, and we also have a number of cross cutting themes. We have practitioner and patient experience of personalised medicine, sustainability, equity.

We need to understand the implications. For practitioners in practice who are already very busy, and will it actually make things more efficient, or does it make things a bit more complicated, and then also for the people who will potentially receive that personalised information, and what does that process look like? What does the consent process look like, and what does the decision-making process look like?

**Rachel Horton**

What we're talking about with sort of... the conversations happening around personalised medicine developments, it did make me think again about the work you were talking about Susie with personalised care? Because I get that many definitions of personalised medicine, and our strategy definition is about sort of the sort of way data informs patient care.

But it feels like, I don't know, to me, it feels like there's… you need to have that sort of personalising, as in talking to a person aspect in it. And I can't quite work out in my mind whether that's just because that's sort of good medicine, and if you are doing data-informed medicine, you need to communicate it appropriately to make it good personalised medicine, or whether that is kind of a personalising thing in and of itself.

**Anneke Lucassen**

One of the, one of the difficulties I had with our definition of what we can measure about a person, is that that moves away from that personalising element, doesn't it? And our trust in numbers within medicine is already at an all time high, possibly at the cost of that very personalising element that you talk about. And what we can measure about someone, won't always seamlessly lead to that personalisation that we hope for.

**Susie Weller**

Yes, I think from the work we've done with patients, families and gathering different public perspectives, from their point of view, it's not really about kind of data and measurements and things like that. It's much more about relationships. So thinking through kind of ethical decision making around who they might tell in their family, who a result might be relevant to within their broader family, and are those kind of more complex decision making processes that families go through.

**Anneke Lucassen**

I also like what you've done in your work following... the longitudinal work, where you follow people up, where they start off, correct me, if I've oversimplified this, but they start off thinking about having their genome sequenced in a very technical way, and then over time, they weave it much more into their everyday lives, and the technical aspects fall away, and the personalised care comes to the fore much more.

**Susie Weller**

Yes, yeah. And I think one of the interesting things, sort of following up with some of the things that Sally said earlier, was around, like kind of developments that might lead to diagnosing rare disease. You know, some of our participants have received a diagnosis. Many haven't, but it's what happens after that.

And I think that's where the kind of personalised care also comes back in because they're sort of left with a variant name, and then no kind of other means of getting any support as to what that might mean for the future, or kind of accessing different services and resources. Sometimes it might mean that somebody falls between different support organisations or kind of access to different welfare help.

**Anneke Lucassen**

And what happens increasingly now with electronic patient records and things is that people get given their technical result, their so-called ‘personalised’ result, but there's no personalisation that comes with it, or they have to wait for six months for their appointment that puts that into a context, and that's another level of personalisation.

**Susie Weller**

It actually has the opposite effect, doesn't it? Because if you have to wait six months to understand what looks like it could be quite a terrifying description full of words that don't mean much without an explanation. It's almost the opposite of personalisation.

**Ali Kay**

And indeed, sometimes the charitable sector is then picking up that responsibility, charities like Unique, who are then explaining the test results to families that come forward.

**Anneke Lucassen**

Yeah, I think Unique would say, I hope Sarah Wynn doesn't mind me repeating what she's told us several times, that actually their role has changed quite significantly, that they're now often providing something that the NHS would have provided in the past, in terms of personalising that care.

**Ali Kay**

Another thought that occurred to me when I was listening to you, Susie, is that people may not always *want* personalised information because it brings psychosocial implications. So for example, in a pregnancy context, or thinking about future pregnancies, people may be uncomfortable thinking that the risk is from them or their partner. It brings in feelings of blame or guilt or responsibility.

**Sarah Briggs**

I was just reflecting that on a step, sort of above that, actually there are some circumstances where we talk about personalised medicine, where the patient may not be involved at all. So we talk about population level implications of personalised medicine, which is things like using data about patients, for example, to decide what screening they might be invited to, potentially what medicines that they ought to be prescribed, and that might be… those decisions might be made at a level above a level where patients would have direct input, but it is still using the data about them to direct their own healthcare.

**Anneke Lucassen**

Yeah, and some of the work that Gabby Samuel has done, and I think is allied to your work on environmental issues, is that if you personalise medicines too much to the individual measurements, that you hugely increase the cost of production and thereby have less available monies for others. So you *depersonalise*, you widen those inequities, increase the environmental costs, and depersonalise for many.

**Sarah Briggs**

Yeah, and I feel that's quite a challenge. So in oncology, for example, we're using cancer vaccines in treatment. Some of those are what we call off the shelf vaccines, so they're relevant to anybody with that particular type of cancer. And then other cancer vaccines are really bespoke vaccines that are manufactured for a specific patient. And the difference in financial cost between those two is *huge*, and also environmental cost in terms of, you know, who benefits from the development of one treatment, and those are things we need to probably think about quite carefully, when we consider how we roll out these new treatments.

**Anneke Lucassen**

Yeah, so I guess what we're saying is that we're really interested in personalising medicine, but at the same time we're interested in the downsides, or the costs to doing that for some parts of the population, and therefore inevitably not being able to do it for all.

**Sarah Briggs**

Yeah, and sort of maximising the benefits for people without discriminating against anyone if possible.

**Ali Kay**

I was thinking when I was listening to you about how a non-medical person might interpret personalised to mean individualised. People might feel that personalising medicine is about them, making individual choices to take the test that they want to take, about the areas of their health that interest them, without having to go through a doctor. I wonder if there are sustainability implications from that as well, though, from all the testing materials and all the plastic?

**Sarah Briggs**

Yeah, definitely. I mean, well, all aspects of healthcare have, you know, an environmental burden. I think you know, the NHS has committed to reducing its environmental impact over short, quite a short time frame, as have many other health systems, and we increasingly need to think about, you know, beyond the financial costs, what the environmental implications are for new technologies and tests that were that were bringing out. Places like NICE have started to think about how they might consider environmental burden alongside financial costs with new interventions, and I think that will increasingly be part of the conversation over the next few years.

**Anneke Lucassen**

I guess, following up on what you're saying Ali, that direct to consumer tests, and thinking about personalising medicine *outside* of the traditional boundaries may well have an environmental cost, because there's more people then coming to the NHS, following them up.

**Sarah Briggs**

Accessing. Yeah, absolutely.

**Sally Sansom**

So just going back to some of Anneke and Sarah's discussion, particularly about the costs and benefits of personalised medicine, these are trade-offs that health economists are very familiar with making, although certainly there's many new or more complex challenges when it comes to evaluating personalised medicine interventions.

I think what's particularly interesting is this trend towards considering impacts outside of the healthcare system, particularly in areas such as environmental impacts and then also equity-related impacts, which, of course, intersect with the health system as well. These are certainly much more challenging to include in traditional health economic approaches, although there's lots of work underway in these areas currently, but much more to do.

**Anneke Lucassen**

And I think the equity point, we've raised this in another podcast as well, haven't we? But if you're talking about genomics as a means to personalise, then of course, our global understanding of genomics is *very* skewed towards people of a broadly speaking Northern European ancestry, so that equity issue is *very* difficult to tackle, because that sort of self-perpetuates. The more we gather those genomes, the more we know about them, and the less we know about people from other ancestries.

**Sally Sansom**

Absolutely. I think there's a number of really interesting initiatives happening around the world to address that. But of course, they're starting from a huge back foot or deficit.

**Sarah Briggs**

And I think also there's a responsibility to try and improve access to the benefits of personalised medicine in that, you know, a lot of the investment in the technologies and the companies driving this are based in the Global North, and there's a lot of work going on at the moment to sort of build capacity, both in terms of research and clinical use of a lot of data and applications of personalised medicine, but again, starting from a back foot really. So lots more work to do in that space.

**Rachel Horton**

Yeah, I suppose I wanted to think a little more about the point you were making, Sarah, about how sometimes there's sort of personalisation in... at a sort of a higher… like on a population level, almost because I suppose conceptually, I've always thought of public health and personalised medicine as being quite separate things? Because personalised medicine sounds like it's like, you know, for a person, and public health sounds like it's for, you know, a population. But I guess that's... there's quite a lot of things that sort of fuse the two now, and

where things like the Newborn Genomes Programme, for example, would fit on that spectrum. Can you be like in the Venn diagram of both public health and personalised medicine, or what does it mean to be kind of one or the other?

**Sarah Briggs**

I guess part of that is what your definition of medicine is, and are we talking about treatment, or are we talking about maintaining health? And they're two very different things, but you can use personalised medical technologies, I guess, for want of a better catch-all term to facilitate both of those. So there's definitely overlap.

**Ali Kay**

Also we’ve a tendency to think of personalised medicine as relating mainly to omic data. But actually that sits with traditional family history taking and other tools.

**Anneke Lucassen**

I guess it's what we started off by saying, isn't it? In many ways, medicine or healthcare has always been personalised to some extent, way before genomics was available. So it's got to be broader than genomics. But I agree that recently it's… it's been very… some people see it very much as genetic, genomic related personalisation.

**Sarah Briggs**

I think the array of data that is potentially feeding into personalisation is vast, though, isn't it? You know, it includes things like sociodemographic factors like, you know, where you've been brought up and what you're exposed to in terms of sort of environmental exposures, like pollution, and whether you smoke. And then it can include, increasingly, the sort of data that people might get from smart watches, so you know, your activity levels, and that's another interesting area to think about as well, is how all those new methods of data collection are likely to feed into personalised healthcare.

**Anneke Lucassen**

But often still, the rhetoric is that it's very promising and it's *going* to transform and what we forget is that more data doesn't necessarily make more personalisation, because how you weave all those different strands together is far from straightforward for most people.

**Sarah Briggs**

Yeah, absolutely and often, actually, the most predictive data is the relatively simple stuff, like your post code.

**Ali Kay**

It also ties in with personalised *care*, because if you have, you know, a fantastically quick diagnosis and great treatment, but the person then still needs to go back on with their life, living in their home, living in the town where they live, with their other environmental factors around them, and if the personalised *care* systems don't follow, then they may not experience the benefits of that personalised medicine.

**Rachel Horton**

It's been a really interesting discussion. And I guess just to ask the question again, what does personalised medicine mean to us here at the CPM?

**Anneke Lucassen**

For me, personalised medicine is almost a red herring, you know, the adjective’s not so important, the personalised medicine? It's about how we improve medicine, or, more broadly, healthcare for the good of *everyone*, thinking about the disadvantages of more data, more apparent personalisation, as well as the advantages.

**Ali Kay**

For me, it's about exploring how personalised medicine can enable practitioners and patients to make more informed choices, to improve their health outcomes, but to explore how we can do that in a more equitable and sustainable way.

**Sally Sansom**

I think personalised medicine is a really useful phrase that works as a vehicle for bringing together people who are broadly interested in a similar area, being improving human health through recent advances, particularly in the omics and related data science fields. Who may otherwise find it… not difficult, but perhaps not as easy, to connect. And I'm thinking particularly of myself, as a health economics researcher, sitting here with all of you, you know, we all have such different but overlapping areas of interest and background, and so I think it's really a helpful term from that perspective.

**Anneke Lucassen**

Nice. I think if we think of it as a sort of catalyst to further discussions, that works a lot better, doesn't it, than a term in itself.

**Rachel Horton**

Thank you for joining us for this episode of the Centre for Personalised Medicine podcast. We've got loads of events and activities exploring different facets of personalised medicine over the coming months, so please take a look at our website, cpm.ox.ac.uk, to find out more.