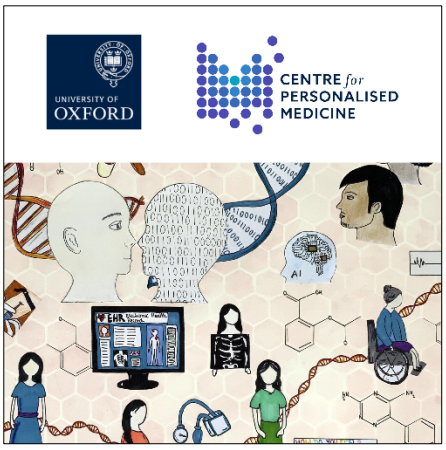
Centre for Personalised Medicine podcast

2023-2024: the year in review



(Our podcast logo features a section of the artwork 'A Lifetime of Measures' by Aneesa, aged 12, from Oxford High School, the stunning winning entry to our 2022-23 Youth Art Competition).

SPEAKERS

Rachel Horton, Anneke Lucassen, Susie Weller, Padraig Dixon

Rachel Horton
Hi, I'm Rachel Horton, a Junior Research Fellow at the Centre for Personalised Medicine, or CPM. In this mini episode between our podcast series, we're taking stock of the year that we've just had, and looking forward to another exciting year for the CPM. I'm joined by Anneke Lucassen, the CPM director.

Anneke, this year's been a big one for the CPM, and there's been a real focus on strategy. Could you talk us through what's been happening?

Anneke Lucassen
Yes, thank you, Rachel. It's been great to focus on our strategy- with our renewed funding up until 2030, that seemed a really appropriate time. And one of the things we've been able to do by really thinking carefully about what we want to do going forward, is identify the themes that underpin the work of the CPM.

So we've thought about themes that are important for our work. And there's three broad, what we call 'pillar' themes of: diagnosis and treatment; risk and prevention; and then how health system boundaries are changing, perhaps for people getting their own testing before they get to health care. And then we've got three cross cutting themes: issues of equity,

which are really important to all those three pillar themes; issues of sustainability, including environmental sustainability; and then how people *experience* personalised medicine and how we need to think about these experiences.

Rachel Horton
They're such fascinating themes with so much to talk about in them. And we're also moving from a sort of small seminar, sort of model to really exploring in depth some of those themes over events, and you tried that in the past year.

Anneke Lucassen
That's absolutely right, it's partly coming out of the pandemic, where we were forced to hold a lot of online lectures, and we felt that our most impactful meetings have been the ones where we got different experts together and thought about a theme, rather than a one-off lecture. So we thought it was really important to focus on those a bit more. And going forward, we aim to have a combination of the two. So have in-depth days, or most of a day, to concentrate on a topic, and then have, in between that, lectures, as we've always had, or seminar series.

Rachel Horton
Sounds really great. And over the past year, the CPM has held several events where we've had those sort of, more in-depth conversations that you mention, and we're going to hear a little more about those. In November, we had a meeting to explore the challenges raised when genetic testing finds something in one person that might be relevant for others in their family. And Anneke, could you talk us through what happened that day?

Anneke Lucassen
Yeah, that was a really interesting day from my perspective. Because, well, as you know, Rachel, this has been an issue that in the specialty of clinical genetics or genetic medicine, we've thought a lot about over the last thirty years, which is when you do a test on one person and that diagnoses a genetic disease or condition, that might also give you information about family members, and then the question arises of how those family members should be alerted, is that the patient in front of you has that responsibility? Is it you that has a responsibility because you know of those relatives? Does it depend on what you found and what sort of treatments are available?

There is obviously no one answer to that, but the day looked at that particular issue on a conceptual level, and it also focused that on a court case that had happened in back in 2019 called the *ABC versus St George's* court case. That's a very seminal decision in our field, where a judge ruled that the health professional has a responsibility to weigh the interests of relatives in the balance with their duty of confidentiality to the patient in front of them. So essentially saying that you do need to *think* about relatives when you're diagnosing in one person.

For me, that was a particularly interesting day because we had about sixty people in the room. We had experts around the court case, and lawyers and clinicians present. But the most important bit was that we had patient representatives from a diverse group of people who might avail themselves of genetic medicine also taking part in the discussions. So we

had really rich discussions that we hope will feed into an update of professional guidelines on how to handle consent and confidentiality in genomic medicine.

Rachel Horton
Thank you. And then in November we also held our first research showcase, which focused on ‘reconceptualising personalised medicine’ and we’re going to hear more about it now from Susie Weller, who's a research fellow at the CPM. Thank you very much for joining us, Susie.

Susie Weller
Oh, thank you Rachel. So in our research showcase, we brought together lots of different researchers and stakeholders to explore the latest thinking on personalised medicine. And one of the things that we were especially keen to do was to facilitate interdisciplinary discussion, but we also wanted to support early career researchers to help them share their work alongside established scholars. So, the focus of the showcase was on ‘reconceptualising personalised medicine’, and we wanted to explore the ways in which personalisation conjures up different things in different contexts, so from genomics and targeted therapies on the one hand, to patient choice and autonomy on the other.

So we started with a series of presentations from CPM Fellows. We heard about Sarah Briggs’ work on personalised medicine and sustainability; we heard about Padraig Dixon’s research on genomics and health insurance. Ali Kay spoke about her work on acceptability and patient perspectives, and I really enjoyed talking about my own work with colleagues on public views on personalised care.

After that, we had a PechaKucha session for postgraduates and early career researchers. So a PechaKucha is a short lightning presentation where the presenter shows 20 slides or images and has just 20 seconds to talk about each slide, so it's a really exciting and engaging format, and we heard some great presentations about the work people are doing in this area. And William Cook won the prize for the best PechaKucha, for which he focused on his own reflections on prenatal testing as a dad, as an obstetrician and as a researcher.

And at the end, we had a panel session chaired by Mike Parker. Where Anneke Lucassen, Julia Frost and Julian Knight spoke about ‘what I talk about when I talk about personalised medicine’. So it was a great day, and we really looking forward to next year's showcase, which will focus on personalised prevention.

Rachel Horton
Thanks, Susie. And then in May, we had a meeting exploring the evolving relationship between genetics and insurance. And Padraig Dixon, who's a recent Junior Research Fellow, who's now on our steering group, is kindly joining us to tell us more.

Padraig Dixon
Yeah, thanks, Rachel. So this event was held, as you said, in May, and it was held in collaboration with the British Society for Genetic Medicine, and the main idea of the event was to explore some of the increasing challenges that we have observed coming from existing and emerging issues that might challenge current arrangements in the UK about

how genetic information can and should be used for insurance purposes. And we started from the view that, to date, despite some individual difficulties, genetic information probably hasn't had wide ranging impacts on insurance decisions in the UK, but there's a strong case for thinking about how current arrangements might change, and if they're fit for purpose.

So we invited patient representatives, academics, clinicians, representatives from the insurance industry, as well as colleagues from the Department of Health and Social Care to discuss some of those challenges. And the discussion focused around the Code on genetic testing and insurance, which has existed in various forms for about 25 years. That covers two types of genetic tests: diagnostic tests, which confirm or rule out a diagnosis, and predictive, which are about predicting future disease risk.

A big focus of the discussion was whether that distinction between predictive and diagnostic testing is fit for purpose. So one set of comments from our participants was that healthcare professionals may not be aware of when each definition applies, particularly as some tests blur the distinction between diagnostic and predictive testing. And an example there was perhaps someone is being tested for *BRCA* status to support a diagnosis of breast cancer, but of course, establishing *BRCA* status would be predictive of future risk for ovarian cancer.

And there was also a really useful discussion about how the original motivation to the Code might have originally applied primarily to strong genotype phenotype links? But as genetic testing has developed and has been established perhaps, or has been undertaken in people who don't necessarily express the phenotype, or have a family history of the phenotype, labelling individuals and perhaps creating the potential for unnecessary disclosures for insurance isn't helpful.

Perhaps a final big topic that's worth mentioning is the issue of variants of uncertain significance? So as research has continued to evolve, some of those variants will be classified, perhaps as benign, perhaps as pathogenic, and again, all of those evolving bits of information will complicate that distinction between diagnostic and predictive testing.

I guess some of the wider issues- we were thinking a little bit about the use of direct to consumer genetic testing, whether that might affect insurance eligibility, whether restrictions on how much individuals have to disclose might increase adverse selection, so you might be labelled high or low risk by insurance companies. And of course, ultimately, all of this comes back to the need to have an ethical, fair set of arrangements for dealing with genetic data in relation to insurance. So we need insurance companies to be commercially viable, but we also need privacy and fair treatment of individuals with different genetic profiles.

Rachel Horton
Thanks, Padraig. So they were some great events where we really explored some very relevant issues to personalised medicine in a lot of depth. And along with these events, we also held our two annual lectures, which are always such a great opportunity to hear from amazing people working in the field of personalised medicine. Anneke, what were your highlights from the annual lectures?

Anneke Lucassen
We had two annual lectures, one from Professor Sir John Burn, and that was a real showstopper lecture in terms of his career and how he'd influenced genetic medicine. What was interesting for me was that so many people, people he cited, were in the audience.

And then Professor Caroline Wright did a fantastic lecture looking at genetic variants and how they might mean something different depending on how you find them. So if you find them in the context of a family history, they might predict or diagnose a disease much more strongly than if you find them in a population setting, and she outlined beautifully, with data to support it, why that was the case.

Rachel Horton
And I should just say that both lectures are available via our recently redesigned website, along with loads more content from the past year. So please take a look, because there's loads to see.

We've got seminars, research, talks, schools outreach, some incredible entries to our youth art competition on newborn screening. And that's all on our website, and you can also see more in our annual report.

So those are the headlines from 2023 to 2024. Anneke, what do we have to look forward to in the coming year?

Anneke Lucassen
So we've got lots of exciting things planned, and there will be more to come still. We've identified three main themes that we're focusing on this year. It's not an exhaustive list, but we're starting with these. So one is newborn screening through whole genome sequencing, linking that to the large research study that's happening in the UK, called the Generation Study. The second is on precision prevention initiatives, cancer vaccines, and whether we can delineate high risk groups and vaccinate them against a cancer rather than treat them for a cancer. And then the third is direct to consumer genetic testing, where people buy kits online, or from a chemist to predict things about their future health.

Rachel Horton
And do we know yet who the annual lecturers are going to be?

Anneke Lucassen
Yes, we have one lined up in February, and that's going to be Philip Ball, who is a writer and scientist, and I've really enjoyed his books, *The Music Instinct* and *How Life Works*. I think that will be a really interesting take on attempts to personalise medicine. And watch this space for the other annual lecture.

Rachel Horton
Brilliant. Thank you. So there's so much exciting coming up over the next year, and we're also working on a new podcast series where we'll be exploring the themes we've identified in the CPM's new strategy: diagnosis and treatment; risk and prevention; evolving health system boundaries; equity; sustainability; and experiences of personalised medicine, and

we're kicking off with an episode on what personalised medicine means for us. So please do look out for that.

Thank you so much for joining us to look back at 2023 to 2024. It's been a great year for the CPM, and we're excited for the next one. Please follow us on social media or sign up to our mailing list to keep in touch and find out more.