



Centre for Personalised Medicine

Annual Report 2022 - 23









Funding statement

The Centre for Personalised Medicine (CPM) was established in 2013 through a generous donation from the Dr Stanley Ho Medical Development Foundation to St Anne's College. The CPM has enjoyed a decade of fruitful collaboration with the Foundation and it is delighted to have secured ongoing funding until 2030. This will enable continued activity, both in the UK and further afield, and in particular with the University of Macau.

The CPM is also grateful for the funding it has received from the Wellcome Trust for the period 2013 - 2023, in support of its work.





Executive summary

The Centre for Personalised Medicine is a partnership between the University of Oxford's Wellcome Centre for Human Genetics - part of the Nuffield Department of Medicine - and St Anne's College, Oxford. It is a communication, engagement and research vehicle for a wide range of stakeholders including students, academics, clinicians, the public and policy makers, to explore the benefits and challenges of personalised medicine from a range of perspectives.

The CPM coordinates a wide range of events and activities including seminars, conferences, debates, public lectures, blog posts, vlogs and podcasts.

Personalised medicine is a broad term that aims to tailor healthcare to the particular person using details about them. To some extent, medicine has always been personalised, so for example, details such as gender, ethnicity and Body Mass Index will have been used to personalise medicine to some extent for many decades. However, recent years have seen an explosion in the amount of data that can be generated about individuals, from wearable technologies giving insights into heart rhythms, to analysing entire genomes in routine healthcare; personalised medicine addresses how we can use this vast array of data to more effectively diagnose and treat patients, as well as predict disease in a population.

Some approaches to personalising medicine include exploring why the responses to certain drugs and therapies vary between people. The reasons for this variability might be (a mixture of) underlying biology, socioeconomic factors or environmental influences, and identifying this mix can help identify the best drug to give to the right patient, and/or highlight strategies to prevent disease in vulnerable communities. The overall aim of personalised medicine therefore is to improve prevention, diagnosis and treatment for (groups of) individuals.

Whilst we believe that personalised medicine can be very powerful when done right, it can also raise significant clinical, ethical, legal, economic and societal challenges. It is both these benefits and challenges that the CPM is here to discuss.

I have enjoyed this second year of my Directorship as much as the first and I was very proud to be at the helm as we were awarded seven years of (increased) funding by the Dr Stanley Ho Medical Foundation. I'm delighted that this funding recognises the need to be closely aligned to research into personalised medicine and we have been working on ways to make the CPM embedded in this.

Although our online presence continues to expand - the vlogs and podcasts are going strong - it has also been great to do more face-to-face events. There were many fantastic events throughout the year. Particular highlights for me were Mary Dixon-Woods' Annual Lecture at the Sheldonian Theatre, with dinner afterwards in the Divinity School; and a workshop exploring how personalised medicine can address the needs of under-served populations: the Black Women's Health workshop, led by Plumer Fellow Dr Jenny Douglas.

I am in awe of the abilities of the (Junior) Research Fellows and of Catherine Lidbetter (Programme Coordinator) and Thea Perry (Administrative Officer). They have ably rounded off the first decade of CPM life and I look forward, with them, to the next 10 years!

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Professor Anneke Lucassen Director, Centre for Personalised Medicine



People

The CPM benefits from the talents, insights and wisdom of its core team, comprising the Director, research fellows and administration staff, for day-to-day activities, and its Steering Group and External Advisory Board for strategic direction.

Tim Gardam, former Principal of St Anne's College and co-founder of the CPM, is stepping back from our External Advisory Board. Tim was instrumental in setting up the CPM in 2013, and it simply would not have existed without his insight, dedication and drive in the early years. We are grateful for his ongoing support as a member of the External Advisory Board after leaving St Anne's College, and we wish him well with all his future endeavours.



Tim Gardam

Professor Cecilia Lindgren, Director of the Big Data Institute in Oxford, left the Steering Group in June 2023. We thank her for her enthusiasm and support over 8 years.

We welcome back Dr Ron Zimmern, Chairman of the PHG Foundation, and so helpful to us in the early years of the CPM, to the External Advisory Board.

Over the past year we have welcomed new Junior Research Fellow (JRF) Dr Sarah Briggs, welcomed back JRF Dr Katherine Wood from her maternity leave, and said 'goodbye for now' to JRF Dr Rachel Horton as she embarked on hers. Assistant Professor Nicky Whiffin finishes her term of office as a JRF at the end of September 2023, and Dr Padraig Dixon will remain a JRF for a little while longer when his term of office is up, covering Rachel's maternity leave.



The CPM Team

CPM Team and Steering Group



Professor Anneke Lucassen, Director



Helen King, Co-Chair



Professor John Todd, Co-Chair



Dr Zoi Alexopoulou



Dr Sarah Briggs, Junior Research Fellow



Dr Padraig Dixon, Junior Research Fellow



Professor Julia Hippisley-Cox



Dr Rachel Horton, Junior Research Fellow



Catherine Lidbetter, Programme Co-ordinator



Michael Milad, student representative



Thea Perry, Administrative Officer



Professor Catherine Pope



Associate Professor Francis Szele



Dr Susie Weller, Research Fellow



Associate Professor Nicky Whiffin, Junior Research Fellow



Dr Katherine Wood, Junior Research Fellow

External Advisory Board



Dame Mary Archer, Chair



Professor Jane Anderson



Professor Sir Peter Donnelly



Professor Gary Ford



Tim Gardam



Richard Girling



Ian Huen



Professor Dennis Lo



Dr Frances Rawle



Dr Magdalena Skipper



Professor Joseph Sung



Dr Ron Zlmmern

Year 10 summary: Events, activities and research

Key to demographic groups:

Clinicians and academics

During Students

Schoolchildren

Patient groups and the public

Policy makers

Personalized Medicine in Pregnancy Worldwide workshop: September

DOUG DOUG

Our Director, Professor Anneke Lucassen, gave two talks at a workshop entitled *Personalized Medicine in pregnancy worldwide: Advantages and special considerations*, run by the Global Pregnancy Collaboration and held at Oriel College, Oxford. As well as setting the scene by providing an overview of personalised medicine, she spoke on *Ethical considerations for pregnancy research* at this two-day event.

Oxford Padua summer school: September

DOU DOU

The CPM was delighted to host a group of postgraduate and postdoctoral students and faculty from the Universities of Padua and Oxford to a three-day conference at St Anne's College. This event was all the more welcome because it had been delayed several times due to the pandemic, and it was wonderful to see everyone exploring the conference themes together and having a stimulating time. The conference covered a range of themes: Infection, Liver, Luminal gastroenterology, Regeneration and cancer, Metabolic syndrome and Microbiome and immunology. Expert speakers from the two institutions included Professors Dame Sarah Gilbert, Julian Knight, Patrizia Burra, Simon Leedham, Vincenzo Ciminale and Fiona Powrie.



Each student gave a presentation on their own research on the second afternoon, and many were of an exceptional standard, with the winning talk being given by Jhanna Kryukova of Green Templeton College, Oxford. This conference was supported by the Translational Gastroenterology

Unit (TGU) at Oxford, and it was a 'return match' for a similar TGU and CPM event held in

Padua pre-pandemic.



Royal Society: Living with the eugenic past: October

DOU DOU

Anneke Lucassen spoke at the Adelphi Genetics Forum conference on *Living with the eugenic past*. Her talk was entitled *Genomic medicine, diverse data and the language of race, ancestry and ethnicity*, in a thought-provoking day of talks that explored a broad range of historical and current perspectives, including a keynote speech by former CPM Annual Lecturer, Dr Adam Rutherford.

CPM Freshers' introduction talk: October

DOU

Dr Katherine Wood and Dr Padraig Dixon delivered this introductory talk to medical students at St Anne's College in October. They gave a presentation about what personalised medicine is (from both a medical sciences and social sciences perspective), the role of the CPM, and how students can get involved with the CPM and the Oxford Personalised Medicine Society (OPMS). The talk was well-attended by the new students and there were some excellent questions and an interesting discussion after the presentation.



Royal Society of Edinburgh keynote lecture: October

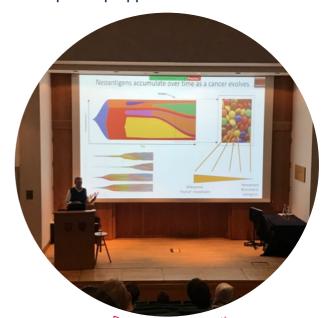
DOU DOU DOU

Anneke Lucassen gave a lecture called *Hopes* and hypes: The role of genetics in cancer prevention and treatment for the Royal Society of Edinburgh in partnership with the Scottish Cancer Foundation. She highlighted how rapid technological advances in our ability to analyse a person's genetic code have resulted in significant, helpful developments in clinical practice and that optimism about further developments remains high and rightly so. Yet this can also sometimes result in a discourse too focussed on these technological achievements, one that assumes that they will seamlessly lead to clear predictions, diagnoses, or treatments and that the interplay of a myriad of other factors (socio-economic, environmental exposures or random) are no longer relevant. Such deterministic discourse appeals to many but leads to promises about delivery that are difficult to honour in practice. Anneke used examples arising from her clinical practice as well as her research group to illustrate where we need a more nuanced debate about the use of genetics in the prediction, prevention, and treatments of cancer. The talk was followed by a lively Q&A session

Personalised cancer vaccines lecture: November

DOUG DOUG

Professor Tim Elliott is Kidani Chair of Immuno-Oncology at the University of Oxford and a world leader in the field of antigen presentation and T cell biology. He has incorporated his discoveries in the areas of antigen processing, T cell regulation and immunodominance into the development of new cancer immunotherapies. Professor Elliott gave a thoroughly engaging introduction to his field in a lecture that used wrapped chocolates as a visual aid to explain how T cells recognise cancer and how this opens up opportunities for treatment.



Professor Tim Elliot

Newborn screening debate: November

DOU DOU

The British Society for Genetic Medicine's annual conference held a plenary debate about Genomic England's plans to pilot whole genome sequencing of newborns, Anneke took part in a 6-member panel debating this. She highlighted how the aim of prompt identification of disease and early intervention in newborns is laudable, but how there can be difficulties in making such diagnoses from a genetic code without a clinical context that directs interpretation. Newborn genomic screening is often represented as having the potential to give both breadth and clarity of diagnosis, but in reality, this screening can generate more uncertainty, and may often require more tests and interventions that are lengthy and use expensive NHS resources. Conversely, other panellists highlighted that without a pilot to investigate the benefits and challenges, we might never know what the value of such screening can be.

RSM Medical Genetics talk - November

DOUG DOUG

Dr Rachel Horton was invited to speak at the Royal Society of Medicine event *Whole genome* sequencing: The future of newborn screening in the UK?, discussing ethical issues relating to newborn genome screening.

Rachel discussed the challenges in predicting what a baby's genetic code might mean for their future, and highlighted issues that newborn genome screening initiatives will need to consider.

Black women's health and wellbeing symposium: November

DOWN DOWN DOWN

The CPM held a multi-speaker day event exploring a variety of perspectives on Black women's health. The event, co-organised by Dr Jenny Douglas of the Open University, with whom we previously collaborated on our Health Disparities conference in 2021, had some exceptionally high-profile speakers, including Baroness Valerie Amos, who introduced the conference, and Professor Dame Elizabeth Anionwu, who fitted us in in the same week she



Other speakers explored recent UK reports into Black Women's Reproductive Health and the Black Maternity Experiences Report, and Dr Faranak Hardcastle presented the findings of a recent CPM-sponsored systematic review on diversity in genomics. The event concluded with a lecture by Dr Douglas: How can an intersectionality-informed approach assist personalised medicine to meet the needs of Black women's health and wellbeing? This important event aimed to explore and discuss the health inequities experienced by Black women in the UK and to consult with Black women about the priorities for health research, and also to learn how these inequities relate to attempts to personalise medicine to meet the needs of Black communities, in relation to a range of health aspects in research, practice and policy. We received extremely positive feedback from a predominantly black and female audience of academics, activists and the public.

School talk: December



The CPM was invited to Bartholomew School in Eynsham, Oxfordshire, to give an after-school lecture to the sixth form about personalised medicine. Rachel Horton enjoyed talking about the CPM and discussing newborn genome screening and direct-to-consumer genetic testing.

The students asked lots of questions and it was a real pleasure to see how engaged they were with personalised medicine and its opportunities and challenges.

College lunch-time talk: December



Anneke Lucassen spoke to a group of St Anne's College Fellows and other academic staff about her research interests and about the CPM. This was an excellent opportunity to promote the CPM to a College audience and to engage in discussion on possible future collaborations.

Guest lecture by Dr Patrick Short: December

DOUDOUDOU

Dr Patrick Short from Sano Genetics delivered an online talk entitled *Accelerating precision medicine research with digital and at-home testing*, discussing how private companies can facilitate the delivery of personalised medicine with surveys, studies and clinical trials. The talk had good attendance with an excellent interactive discussion following the lecture, both with regards to the interaction between the NHS, academics and industrial partners in terms of precision medicine, and also for early career researchers the transition between academic and industrial career paths.

Guest lecture by Dr Shiri Shkedi-Rafid: January

DOUT DOUT DOUT

The CPM was delighted to host Dr Shiri Shkedi-Rafid (senior genetic counsellor at Hadassah Medical Center and Director for the MSc programme in genetic counselling at the Hebrew University of Jerusalem) for a seminar at WHG during her visit to the UK. Shiri gave an extremely engaging talk discussing ethical issues raised by genetic and genomic testing in pregnancies conceived with donor gametes. She involved the audience in considering some of the ethical dilemmas encountered in clinical practice, including issues such as how to respond when genetic screening of prospective gamete donors finds variants of uncertain significance, or what to do when a new genetic diagnosis in a donorconceived child raises the possibility of germline mosaicism in a gamete donor.

Dr Stanley Ho Memorial Lecture 2023: January

DOT DOT

Professor Jeffrey Macklis of Harvard University gave an engaging lecture on the immense complexities of neuroscience and the human brain, and the diversity of neuronal circuitry. He touched on the concept of genetic 'address codes' and specific targeting of RNA to different neuronal compartments. Professor Macklis spoke about these concepts in the context of development, and disease, and how increased understanding can be harnessed therapeutically. The event was chaired by Associate Professor Francis Szele of the CPM's Steering Group and it was a collaboration between the CPM and the Oxford Martin School. The talk was followed by a Question & Answer session with a clearly engaged audience, and a drinks reception.



Guest lecture by Dr Maya Sabatello: February

DOUG DOUG DOUG

Dr Maya Sabatello, Assistant Professor of Clinical Bioethics at Columbia University, gave a thought-provoking lunchtime talk at St Anne's College. Dr Sabatello's focus was on diversity, ableism and trust in precision medicine, with an emphasis on disability ethics. Members of the CPM and the CELS (Clinical Ethics, Law and Society) research team had worked recently with Dr Sabatello on a literature review and synthesis exploring the ethical, legal and social issues in diversifying genomic data. The talk provided an excellent opportunity to continue discussions about the important issues highlighted by her work and the review. Further details of the review, which was co-authored by 4 members of the CPM team among others, can be found on our website. In summary, the need to diversify genomic data, to improve the evidence base for genomic medicine for all ancestries, is well recognised, but is more complex than simply increasing the collection of data from people from a range of ancestries. The team reviewed the literature to understand the challenges of diversifying genomic data to identify key ethical, legal and social issues. Further information on the research follows on page 26.

Guest lecture by Dr Francois Halloy: February

DOU DOU

Dr Francois Halloy, a non-CPM JRF at St Anne's College, delivered an excellent lunchtime inperson guest lecture entitled *Oligonucleotide* therapies for genetic diseases: Current landscape and perspectives. The talk was very interesting and focused on nucleic acid-based therapies and their prospects in personalised medicine. There were some excellent questions and it was a good opportunity to promote the work of a fellow St Anne's Senior Common Room member.

School genomics and ethics workshop: March

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Research Genetic Counsellor Dr Ali Kay and Dr Dale Maxwell, both postdoctoral researchers in the Goriely Lab in Oxford, delivered a genomics and ethics workshop on behalf of the CPM at Bartholomew School in Eynsham, near Oxford. Supported by a grant from the University of Oxford Enriching Engagement scheme (funded by Wellcome), the workshop included sharing an animation entitled *We are all mutants*, designed in conjunction with Scriberia. This introduced the students to de novo mutations and the discovery of 'selfish mutations'.

Ali and Dale also engaged the budding scientists in a highly topical evaluation of newborn genome screening. They presented the group with three different conditions, all with very different ages of onset and varying degrees of risk, severity and treatability. Facilitated discussion groups were used to encourage some lively debate on what matters when evaluating a genetic condition for screening and how to prioritise. Lots of enthusiastic students stayed behind at the end to ask for advice on careers in science. Dr Lindsay McDowell, a teacher at the school, was appreciative of the opportunity to engage their students with current research.

Highlighting the relationship between new technologies, scientific approaches and ethics, this pilot workshop was carefully planned with colleagues in the CPM (Katherine Wood, Padraig Dixon and Rachel Horton) and with ethics researcher Dr Arianna Manzini. It was the first of hopefully many events with local schools,

engaging students with personalised medicine and reproductive genomics.



Ochool genomics and ethics workshop

Opportunities and challenges for Polygenic Risk Scores: April

DOWN DOWN DOWN

This event, in collaboration with Cambridge Prisms and its journal *Precision Medicine*, was very well-attended. Following an introduction to the different narratives around polygenic risk scores from Anneke Lucassen, we heard case studies on the use of polygenic risk scores (PRS) in different diseases. Dr Amit Sud (Institute of Cancer Research) spoke about their use in cancer whilst Professor Aroon Hingorani (University College London) discussed their use in cardiovascular disease, and Professor Cathryn Lewis (King's College London) explored potential applications in neuropsychiatric conditions.



A highly interactive panel discussion followed, chaired by Professor Clare Turnbull (Institute of Cancer Research), in which the speakers were joined by Professor Nick Wald (University College London), Dr Judith Hayward (NHS NE England and Yorkshire), Dr Imran Rafi, (St George's, University of London), Professor Claudia Langenberg (Queen Mary, University of London), and many members of the audience contributed their thoughts. The discussion continued over a College dinner. All of the talks, and excerpts from the panel discussion, were added to the website and YouTube channel, and Anneke and Dr Sarah Briggs are writing an editorial for the journal.

Working with large amounts of qualitative data: The breadth-and-depth method: April



Dr Susie Weller was invited to give a guest lecture on large-scale qualitative analysis as part of the Advanced Qualitative Research Methods module, from the Evidence-Based Health Care (EBHC) programme. It focused on the breadth-and-depth method, developed by Susie and colleagues. The method comprises a four-step process fusing computational techniques for exploring breadth with more conventional forms of qualitative analysis to provide depth. Since the lecture, Susie has been invited to speak to several teams both in the UK and Canada about the method.

Outreach talk to U3A group: April



Taking CPM outreach to a new audience, Anneke spoke to the University of the Third Age (U3A), an international organisation of retired people interested in keeping their brains active, and learning about new discoveries and ideas. Anneke spoke to the National U3A Science & Technology Community about personalised medicine and how the CPM encourages debate about its opportunities and challenges. She introduced the concept of personalised medicine to the group and showcased the CPM and its work.

Annual Lecture: April

DOWN DOWN DOWN

Professor Mary Dixon-Woods, Director of THIS Institute and The Health Foundation Professor of Healthcare Improvement Studies in the Department of Public Health and Primary Care at the University of Cambridge, gave the Centre's 2023 annual lecture at the Sheldonian Theatre. Professor Dixon-Woods' compelling lecture, entitled *Putting the person into improving quality and safety in healthcare*, began by noting that the effectiveness of personalised efforts to target care on those likely to benefit is highly variable in practice.

In particular, organisations with similar resources, environments and populations served may experience very different outcomes. The lecture emphasised the need to appreciate institutional contexts, power structures, and socio-technical contexts to understand how patients and populations might benefit from personalised medicine and other aspects of modern healthcare. Poorly supported top-down directives, and insufficient investment to make and optimise improvements despite onerous data collection initiatives, reflect (in part) failures to appreciate considerations other than effectiveness when designing and implementing interventions. Solutions to these challenges should involve the use of collaborative co-design alongside robust evaluation to develop an evidence-based approach to support decision-making in personalised medicine and healthcare more

broadly. The lecture was followed by a dinner for 60 VIPs in the Divinity School, which was The Radcliffe Camera and the University Church From the University a real highlight for the CPM.





Illustration and animation in patient journeys: May

DOUT DOUT DOUT

Susie Weller and colleague Dr Kate Lyle were invited by the Centre for Research on Families and Relationships at the University of Edinburgh to share their work exploring the role of illustration and animation in communicating patient journeys. Based on their research interviewing patients and families who have taken part in genomic medicine, and exploring the ethical and social challenges associated with this, they reflected on the process of co-producing visual resources with patients, families, an artist and animator to help illustrate the complexities of these pathways.

Subject Family Evening: May

DOUG DOUG

The CPM hosted a College Subject Family Evening, organised by Sarah Briggs, with the theme Personalising Medicine Through Genetics. Anneke spoke about the benefits and challenges of predicting [ill-] health, Nicky Whiffin spoke about harnessing genome sequencing data to find genetic diagnoses for rare disease patients, and Olivia Fisher (a Genomic Medicine MSc student supported by the CPM) discussed her dissertation project, exploring the preparedness of community pharmacists for genetic testing and results.

This was a great opportunity for the CPM to link with College students, with interesting

the subject areas, and research and career opportunities.

discussions of both



WHG lab talk: June

DOUG DOUG

WHG holds regular lab talks where its groups showcase their research twice a year. In June Anneke introduced the work of the CPM and the CELS research group and Rachel Horton, CELS research fellow and CPM JRF, spoke about her research that explores how 'results' are constructed from the soup of variants found in a whole genome sequence. Dr Faranak Hardcastle, CELS Research Fellow and lead for CPM diversity systematic review, presented findings from this review exploring ethical issues in diversifying data (see below on page 26).

Website POUT DOUT DOUT DOUT

The CPM has been working hard over the past few months to develop a new website, which is due to launch in the early autumn and situates us within the Nuffield Department of Medicine. This will feed content to a wide range of audiences including a knowledge exchange platform that our funder is developing, to broaden access to Hong Kong, Macau and mainland China. The new website is a significant update from the previous one and allows our increased activity levels to be more easily searchable, providing more streamlined access to topic areas, research outputs and engagement activities. A new visual identity will go hand-in-hand with the website launch; here is the new CPM logo, which will replace the existing one from the beginning of the new academic year:



Podcasts DOU DOU DOU DOU

The Centre for Personalised Medicine podcast series explores the promises and pitfalls of personalised medicine, and asks questions about the ethical and societal challenges facing the field. It is available via Oxford Podcasts, Apple and Spotify. The second series, focussing on research into ethical issues in personalised medicine, started last year with Dr Gabrielle Samuel discussing the environmental cost of personalised medicine, and Anneke Lucassen talking about why the same genetic finding might mean different things in different people.



This year, we spoke to Dr Lisa Ballard and Anneke about sharing genetic results within families and the challenges this can raise. Rachel Horton recorded an episode discussing newborn genome screening. Susie spoke about reimagining who is 'the patient' in genomic medicine. Dr Faranak Hardcastle shared insights from her research looking at the ethical challenges with diversifying genomic data and discussing why our understanding of genomic variation remains overwhelmingly biassed towards individuals of Northern European ancestry. We also heard from Dr Kate Lyle, who discussed her work looking at why research regulation falls short in genomic medicine. In the final episode of this series we were delighted to hear from Julie Young, from the CanGene CanVar patient reference panel, sharing her experience of navigating a genetic diagnosis for herself and her family.

Vlogs DOU DOU DOU DOU

Katherine Wood has continued to create vlogs for a flash interview series, which have been very popular on social media. A further eight vlogs have been released in the past year. The interviews have covered a wide range of topics related to personalised medicine with various experts in their field, including genetic counsellors, academics, clinicians and the CEO of the charity Unique. For example, Katherine had an interesting conversation with Dr Arzoo Ahmed from Genomics England discussing the Newborn Genomes project, while an interview with Dr Eduoardo Ostinelli focused on precision medicine in psychiatry. A particular highlight was an interview with Professor Ewan Birney, Deputy Director General and Joint Director of EMBL-EBI. Ewan talked about his scientific journey from a young scientist working on the Human Genome Project through the development of The ENCODE Project, to now.

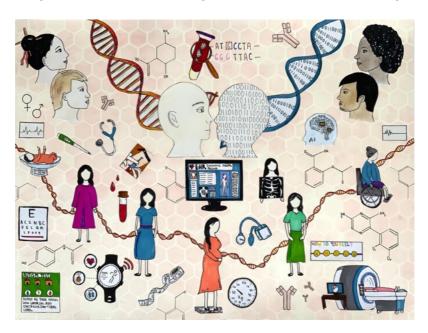
Blog programme p

We released three blog posts this year. The first revisited the 2021 annual lecture by Dr Adam Rutherford. This post pulled out some of the key points made by Adam in his fantastic lecture and put them into context. The second blog reported a collaboration between Genomics England and CPM to write a report around ethical, legal and social issues in diversifying data. The final blog post was written to showcase the winning and finalist entries from a schools art competition.

Art competition and exhibitions DUM DUM

This year the CPM launched its first Art Competition. We were delighted by the response, with pupils in Years 7-9 from schools across the UK sending in entries on a theme of measurements in healthcare.

Our winner was Aneesa, aged 12, from Oxford High School, with this stunning entry:



Aneesa wrote: "My artwork, A Lifetime of Measures, represents everything I learnt about personalised medicine and the fascinating ways changes in health are monitored and conditions identified. I realised traditional measures such as scales or thermometers are not obsolete but complementary to modern measures to develop and provide the right treatment for each person. The "digital twin" is a focus as it is a way to capture all the measurements and use these to assess health risks over time. My art shows how important DNA analysis is and how age, sex and ethnicity though simple are important when deciding the best treatment. It shows measures are important at different stages of life and may mean different things at different stages. I noticed that many chemicals used as medicines have hexagonal rings, so I decided to choose a hexagon pattern for my background to represent known and future personalised medicines."

Our runner-up was this entry from the Art Club at Bartholomew School in Eynsham, Oxfordshire, seen in full and in close-up to reveal some of the exceptional petri-dish art:





The best entries from the competition have been on display at the Wellcome Centre for Human Genetics; St Anne's College Open Day; and will feature in an upcoming exhibition at the Cairns Library in Oxford; and at the John Radcliffe Hospital later in 2023 as part of the Artslink initiative.

Stop Motion and Lego Project Duri

Katherine Wood has been working with Dr Alexandra Martin-Geary from the Big Data Institute (BDI) on a Lego-based genetics outreach project with students on the Creative Media Production course at Abingdon and Witney College this year. Katherine, Alex and two other early career academics wrote short scripts aimed at secondary school pupils as "explainers" about three different genetic conditions (beta-thalassemia, sickle cell anaemia and spinal muscular atrophy) which disproportionately affect under-represented communities, as well as a "primer" focusing on key genetic concepts.

The students were delivered a brief by the team, and in small groups had three days to create Lego stopmotion videos to accompany each script, with checkins with their mentor from the team during that time to ensure everything was going well. Following the project, there was a launch event supported by the CPM to screen the final videos for the first time in front of an audience including the students, CPM members and the wider academic community. The project was a huge success and the students were very engaged and interested in the genetics underlying the scripts.



Maternal mental health review programment

Anneke Lucassen, Susie Weller and colleague Dr Kate Lyle from the CELS team are collaborating with the Centre for Reviews and Dissemination at the University of York to map current evidence around perinatal mental health. Commissioned and financially supported by the PAM Foundation, which supports research into postpartum depression and psychosis, the evidence map will be created through thematic analyses of identified systematic reviews, detailed overviews of systematic reviews, updating of evidence syntheses, and further evidence mapping and/or systematic reviews to include primary research. The first phase of the work focuses on demonstrating the breadth of evidence already available from systematic reviews in the field. After completion of this initial work further discussion and stakeholder interviews will take place with key informants.

Mass Observation study DOM DOM DOM

Susie Weller has been leading a collaboration between CPM, the Clinical Ethics, Law and Society (CELS) research group and Mass Observation. Mass Observation is a well-established archive of daily life in Britain. Three times a year, a panel of Observers are invited to write about set topics, called Directives. The team worked with colleagues at Mass Observation to develop a Directive on 'genetics and health in our everyday lives'. Distributed to the panel in the Summer of 2022, the Directive covered areas such as: experiences of personalised care, genetics in our everyday lives, the issues genetic testing can raise, and future uses of genetics. Responses were received from 139 Observers, the majority of whom provided incredibly rich, detailed accounts. Whilst this was designed as a public engagement exercise, the team has since gained ethical approval to analyse the material and is currently writing a paper juxtaposing observers' understandings and experiences of personalised care and narratives of choice in healthcare policy.

Environmental impacts from the manufacture of personalised medicines DOT DOT

The CPM and CELS group were awarded a small grant to explore the environmental impact of the manufacture of medicines that are made as a result of personalising therapies. Personalised medicines aim to move therapies from medicines designed for use by all people, to ones targeted to certain sub-populations or the specific characteristics of one individual. This move has implications for medicines manufacturing processes, and these implications need to be considered in terms of their environmental costs. This is especially because manufacturing of personalised medicines will

most likely occur in addition to traditional manufacturing rather than instead of it. Our report made a range of recommendations about how the adverse environmental costs of manufacturing personalised medicines should be considered. A summary of the findings was captured here:

Ethical issues in diversifying genomic data

DOU DOU

Genomics England Diverse Data group commissioned CPM and the CELS research group to do a systematic literature review of attempts to understand genomic variation (and thus disease aetiology) on a global level. The sequencing of entire human genomes has now become a routine, relatively inexpensive investigation in healthcare which offers many promises of personalising, stratifying, and targeting healthcare with an understanding of genetic susceptibility. However, research collections (databases, biobanks etc.) that underpin these developments are significantly skewed towards populations of European ancestry meaning that our understanding of genetic susceptibility (or indeed of genetic protection to disease) is less good for many other populations in the world. The need to improve the evidence base for genomic medicine for all ancestries is well recognised, but is more complex than simply increasing the collection of data from people from a range of ancestries. We reviewed the literature to understand the challenges of diversifying genomic data to identify key ethical, legal and social issues. We found that:

- 1. Many research practices are exclusionary and need to change.
- 2. Co-design is key to identifying and avoiding potential problems around data diversification.
- 3. There are wider structural issues that influence researchers' and participants' attempts to generate diverse data. For example, (a) some researchers view data as neutral, but this ignores the social construction of data and technologies, and their tendencies to reflect societal inequalities. (b) Efforts to diversify data should be contextualised (c) Classification and categorisation of populations have political consequences and need to be closely interrogated.

A preprint of the report is available using this QR code and an abridged version is in press in Cambridge Prisms: *Precision Medicine*.

A selection of publications arising from CPM activities

Dixon P, Harrison S, Hollingworth W, Davies N, Davey Smith G. *Estimating the causal effect of genetic liability to prevalent disease on hospital costs using Mendelian Randomization*. Economics and Human Biology, Volume 46, August 2022

Dixon P, Keeney E, Taylor J, Wordsworth S, Martin RM. *Can polygenic risk scores contribute to cost-effective cancer screening?* A systematic review. Genetics in Medicine, Volume 24, Issue 8, August 2022

Briggs SEW, Law P, East JE, Wordsworth S, Dunlop M, Houlston R, Hippisley-Cox J, Tomlinson I. Integrating genome-wide polygenic risk scores and non-genetic risk to predict colorectal cancer diagnosis: A cohort study in UK Biobank. BMJ 2022; 379:e071707

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Scholarships, bursaries and student support

This year the CPM has supported a number of students to attend conferences:

Ricardo Gonzales, a DPhil Candidate in Cardiovascular Image Analysis, attended the 6th International School on Imaging with Medical Applications (SSIMA) in Oradea, Rumania.

Umair Mahmood, a DPhil candidate in Gastrointestinal Oncology, presented his team's work originating from Oxford at the Crick Cancer Symposium in London.

Maria Pikoula, a Graduate-Entry Medicine student:

"I am very grateful to the Centre for Personalised Medicine for awarding me the Dr Stanley Ho Vacation Research Studentship. It has allowed me to perform the foundational work of what will be a large-scale research study in chronic lung disease. This is a great project on which to build on for my future clinical academic career goals."

Susie is currently supervising a research project as part of the MSc Genomic Medicine. Supported financially by the CPM, student Olivia Fisher is conducting a qualitative study exploring the preparedness of Community Pharmacists for genetic testing and results.

Oxford Personalised Medicine Society

The Oxford Personalised Medicine Society (OPMS) was set up by and for students of the University of Oxford who are interested in personalised medicine. It works closely with the CPM. It hosted a number of talks and events through the academic year:



- · A welcome event at the beginning of the academic year
- · A series of Introduction to R Coding workshops
- A collaboration with the Oxford University Science Society with guest speaker Sir Phillip Campbell
- A talk by Dr Sarah Briggs on The use of Polygenic Risk Scores in predicting bowel cancer
- A talk by CPM Steering Group member Professor Francis Szele on The pros and cons of modelling Schizophrenia
- A talk by CPM Steering Group member Professor Julia Hippisley-Cox on work with QResearch, a large consolidated and anonymised database of health records from general practice in the UK.
- The annual Student Symposium at St Anne's College, with Keynote Speaker Professor Bass Hassan
- A talk by CPM External Advisory Board member Dr Magdalena Skipper on *What is the role of a scientific journal: The case of* Nature

The ever-popular magazine *The Gene'Zine* was published each term and distributed amongst the writers, the medical school, Colleges and OPMS members.

"We thank the CPM for all their help, especially in allowing us to book rooms at St Anne's, as well as providing contacts for talks. We are incredibly humbled by all their support."

Michael Milad, OPMS President 2022-23

2023 - 24 Summary of planned activities

We have an exciting range of events and activities in the pipeline for 2023 - 24. A research showcase in November promises to be a real highlight and an opportunity to explore overlapping fields of interest around personalised medicine. We are also planning a further polygenic risk score

event that picks up some of the unfinished debate from this year's successful event. We also plan to host an event to explore how the code on genetic testing and insurance might need to be updated in the wake of new genomic testing possibilities (a background paper was prepared by Padraig Dixon and others here:)



2023 - 24 Strategic plan

Building on achievements and partnerships established by the CPM, and thanks to the generous renewed funding from the Dr Stanley Ho Medical Development Foundation, our strategic direction for the next decade can now be charted.

The first decade of the CPM has seen extraordinary developments in science and technology resulting in ever more detailed understandings of disease processes and in data available to personalise healthcare approaches. Successful translation of these advances into disease understanding, prevention and treatment will require new, improved and global interactions between people, healthcare, research and technology. It will also require research into the obstacles that hinder progress and suggesting means to dismantle them. One example of such research is our collaborative work exploring the barriers to a more diverse understanding of genomics, highlighting how co-production is key to any such attempt.

Our strategic objectives encompass:

Issues around equity: We want to ensure that the benefits of personalised medicine can be made accessible to all through challenging health disparities and devising strategies to democratise the benefits of personalised medicine;

Informed dialogue: We aim to facilitate nuanced debate around how to adapt to the emerging challenges and opportunities presented by new approaches to personalising medicine; for example, how consent processes might need to evolve or be maintained. This is particularly relevant for large scale open ended ventures such as whole genome sequencing of newborns;

Exploring a range of perspectives: we will seek the views of diverse stakeholders on translating technological advances into effective personalised healthcare, acknowledging that this will require a wider lens than the role of genetic and genomic factors;

External funding and collaboration: Seeking additional external funding for research activities whose results and outputs can then be brought into the CPM for global knowledge exchange and dissemination.

Our collaborations extend to the PHG Foundation in Cambridge thanks to the enthusiasm of Chairman Ron Zimmern who re-joins our External Advisory Board this year, fostering a regular exchange of ideas and building on each other's different strengths. We plan to co-host an event in London that looks at how a particular court case (ABC versus St George's NHS Healthcare Trust) has influenced healthcare practices and policy on disclosure of genetic information to at-risk family members. Our workshop on genetic testing and insurance will come a time when the government code of practice is due for renewal.

Our (Junior) Research Fellows bring a wealth of research experience and approaches with them. Exploring the environmental impact of new techniques and data collections, and how this impact may reduce access to PM for some populations, is just one of the exciting areas that we will begin to explore.

We invite engagement and ideas, extending an open invitation for discussions on research, partnerships and growth opportunities. Please get in touch!

Some 2022 - 23 insights

Online reach



Over 1,200 YouTube subscribers 17,000 views in the last year Over 100,000 'lifetime' views



YouTube views from 47 countries

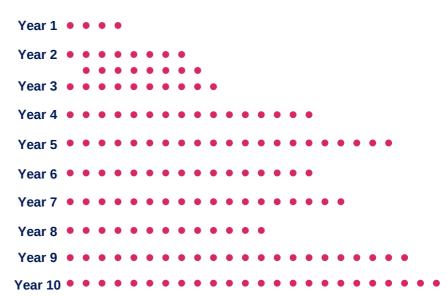


2,255 followers

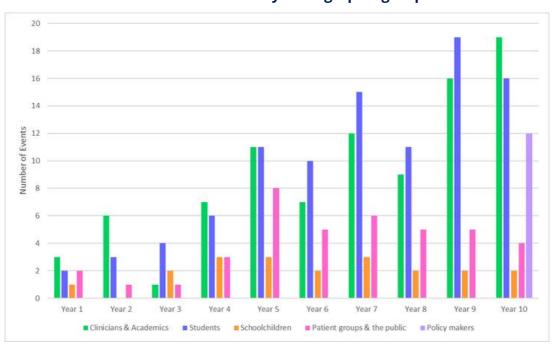


159 followers

Number of events



Number of events by demographic group



Collaborations



























Università DEGLI STUDI DI PADOVA











In February 2023, a signing ceremony took place to mark the renewed funding agreement with the Dr Stanley Ho Medical Development Foundation. Pictured left to right: Professor Anneke Lucassen, Ian Huen, Trustee of the Foundation, Dame Mary Archer, Helen King and Professor John Todd.

Professor Anneke Lucassen said: "I am so delighted that the Centre for Personalised Medicine has received a generous 7 year funding award. This will allow us to research, debate, engage and implement a range of personalised medicine approaches so that developments in technology and data analysis can be translated into improved healthcare".

Acknowledgements

The CPM acknowledges with thanks the Dr Stanley Ho Medical Development Foundation and the Wellcome Trust.

Dame Mary Archer, Chair of the CPM External Advisory Board, and the board's members

Helen King and Professor John Todd, Co-Chairs of the CPM Steering Group, and the group's members

Everyone else who has been involved with the CPM in the past year





St Anne's College



