







CPM Annual Report 2023-24

October 2024





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https://cpm.ox.ac.uk/



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何鴻燊博士醫療拓展基金會 Dr. Stanley Ho Medical Development Foundation

From the Director

Welcome to our annual report for 2023-2024, which looks back at another successful year of CPM activity. Over the past year we have strategically shifted our focus towards delivering a number of high-impact one-day events. These carefully curated gatherings have assembled diverse perspectives and fostered meaningful dialogue about a range of subjects relevant to personalised medicine. These events have emerged as some of the most influential in the CPM's history, generating high-quality outputs and establishing key networks that foster ongoing collaborations including funding applications.

The summer of 2023 marked a milestone for the CPM as we celebrated our tenth anniversary. We took this opportunity to review our strategy, helped by an external facilitator John Hatwell of Andect Consulting. A summary of this review - taking us to 2030 - is available here: https://cpm.ox.ac.uk/strategy/

This strategic review allowed us to sharpen our focus on delivering scholarly activity and engagement with the broad, and evolving, field of personalised medicine. We have identified six broad areas to which our activities can be mapped; three 'pillar themes' - Diagnosis and Treatment, Evolving Health System Boundaries, and Risk and Prevention - and three cross-cutting themes - Equity, Experiences of Personalised Medicine, and Sustainability. Together they provide the foundation of our forward-looking agenda, and we will use this framework to guide our events, activities, and research throughout the coming year: https://cpm.ox.ac.uk/themes/

As a small but highly driven team, I am very proud of the significant strides we have already made and I am confident this new roadmap will allow us to continue to punch well above our weight in achieving tangible impact across the sector.



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Professor Anneke Lucassen, Director, Centre for Personalised Medicine

Overview

About the Centre for Personalised Medicine

The Centre for Personalised Medicine (CPM) is a partnership between St Anne's College and the Centre for Human Genetics (CHG), part of the Nuffield Department of Medicine (NDM) at the University of Oxford. Our aim is to drive multi- and inter-disciplinary collaboration, share knowledge, and facilitate impactful research that resonates with public, professional, academic and policy-influencing audiences. Through a diverse portfolio of activities including conferences, academic meetings, public lectures, podcasts, blog posts, and position papers - the CPM fosters meaningful engagement, and advances the field of personalised medicine. At the CPM:

We define personalised medicine as the application of insights from what we can measure about individuals to inform their healthcare.

Whilst the practice of medicine has always involved tailoring treatments based on available data, the term 'personalised medicine' has gained prominence over the past two decades, reflecting advances in science and technology that allow us to incorporate new and diverse data types into healthcare decisions. These data types - ranging from genomic and epigenomic information to proteomic, socio-demographic, and early life exposure data - help to refine predictions, treatments and interventions within healthcare. Our work emphasises the integration of all these data types, ensuring a holistic approach to the personalisation of healthcare for all.

Funding

The CPM was established in 2013 through a generous donation from the Dr Stanley Ho Medical Development Foundation to St Anne's College and financial support from the University of Oxford through the (then) Wellcome Trust Centre for Human Genetics. The Foundation has renewed, and expanded, its funding to take us to 2030 and the Nuffield Department of Medicine continues to support the CPM now that Wellcome Trust Centres have ended.



People

Many people help the CPM team deliver its activities. Our core team, comprising the Director, research fellows and administrative staff, are supported by a Steering Group and External Advisory Board. Learn more about the individuals who contribute to our work here: https://cpm.ox.ac.uk/meet-the-team/

This year, we said goodbye to Katherine Wood and Padraig Dixon, our outgoing Junior Research Fellows (JRFs), and welcomed new JRFs Ali Kay, Sally Sansom, and Emilie Wigdor, who joined us in Michaelmas term. We were also delighted to welcome back Rachel Horton following her maternity leave in July.

Our Steering Group has seen important additions with the appointments of former CPM Director Simon Leedham as co-chair, as well as Holm Uhlig, Sarah Wordsworth, Angeliki Kerasidou and former JRF Padraig Dixon. Their experience and insights will be an important steer for our activities over the next year. We offer huge gratitude to former co-chair of the Steering Group, John Todd, who stepped down from his role as Director of the CHG. His support of the CPM over the years has been tremendous. We extend congratulations to Dennis Lo, a member of the CPM's External Advisory Board since 2013, on his appointment as the new Vice-Chancellor and President of The Chinese University of Hong Kong.

Beyond the core team, Steering Group and External Advisory Board, the CPM is fortunate to collaborate with a wide network of experts. We wish to extend thanks to Kate Lyle, a senior researcher at CHG, who has been instrumental in several CPM research and engagement projects, and Nicholas Pitt, who has helped to develop our forthcoming event, *Songs of Genomics*.

CPM core team, November 2023: L-R Dr Susie Weller, Dr Padraig Dixon, Dr Ali Kay, Catherine Lidbetter, Professor Anneke Lucassen, Thea Perry, Sally Sansom, Dr Emilie Wigdor, Dr Sarah Briggs



Summary of activity 2023-2024

Flagship events

This year we have focused on holding several impactful one-day events, often in collaboration with partners. Our aim here is to bring together different viewpoints and encourage conversations that go beyond the event to develop, for example, research, policy or guideline development.

1. Familial genomic information: Scope, context and implications for practice

We partnered with the PHG Foundation and the British Society for Genetic Medicine to hold a meeting which discussed the ethical, legal, and social challenges that arise when genetic testing reveals information that could be important for family members.

Background

The management of genetic and genomic findings that come from the testing of one person, but which may be relevant to members of their family, has been debated in healthcare for decades. Clinicians often face uncertainty when trying to balance the personal and familial nature of such information. While professional guidelines such as those from the General Medical Council advise weighing patient confidentiality against preventing harm to others, the legal framework has provided limited clarity until recently.

The 2020 High Court ruling in *ABC v St George's Healthcare NHS Trust* marked a pivotal shift, establishing a legal duty to consider disclosing confidential information without consent when another person is at serious risk. However, the implications of this judgment remain unclear to many healthcare professionals and hospital legal teams, and communicating these risks to family members may not happen because of these uncertainties. Although familial risks may only be discovered through the testing of one person, some argue that such test results are not bound by confidentiality in the same way, since the information they provide is not uniquely identifying one individual.

The workshop

The workshop took place at St Anne's College in November 2023. Anneke Lucassen and Peter Mills (Director of the PHG Foundation) opened the discussions by exploring the tensions created by one person's test result being of potential interest to their relatives. Helena Carley provided an overview of how this situation is currently managed in the clinical landscape, Michael Parker examined the ethical context of familial information sharing, and Jonathan Montgomery provided perspective on the law and expectations of public ethics. Jonathan Zimmern's analysis of the implications of the *ABC* judgment rounded off the talks.

The invited audience, representing diverse sectors including patient organisations, clinicians from various specialties, lawyers, and academics, contributed to the rich

discussion that was held around these introductory talks. The talks and discussion were transcribed and analysed and a report from the meeting will be published soon. A briefing paper can be found here: <u>https://cpm.ox.ac.uk/familial-disclosure-event/</u>

2. (Re)conceptualising personalised medicine research

Our second flagship event explored the diverse range of personalised medicine-related research projects and approaches that are happening in Oxford. This showcase, devised by CPM Fellows Susie Weller and Ali Kay together with Kate Lyle, a researcher from the CHG, was held at St Anne's College in November 2023.

Background

The event brought together an audience of around 50 early-career academics and clinicians to explore diverse perspectives on personalised medicine, what it is perceived to be, and how it might need to be re-conceptualised. It encouraged discussions and teamwork, giving newer researchers a chance to share their work alongside more experienced academics.

The showcase

The event began with an introduction by the Principal of St Anne's College (and CPM Steering Group co-chair) Helen King. It was followed by a session highlighting the diverse expertise of the CPM Fellows:

- Sarah Briggs: Personalised medicine and sustainability
- Padraig Dixon: Genomics and health insurance
- Ali Kay: Acceptability and patient perspectives
- Susie Weller: Public views on personalised care

The second session, chaired by Ali Kay, featured *PechaKucha* presentations from eight early-career researchers, including new CPM JRF Sally Sansom. William Cooke was voted to have delivered the best presentation with his talk, *Reflections on prenatal testing as a dad, obstetrician, and researcher*.

The day concluded with a panel discussion chaired by Michael Parker, with panellists Anneke Lucassen, Julia Frost and Julian Knight.

We plan to introduce a research showcase as an annual fixture, with a different aspect of personalised medicine explored each year.

For more information, you can watch the talks here: <u>https://cpm.ox.ac.uk/watch-our-lectures-interviews/research-showcase/</u>

A Pecha Kucha presentation consists of 20 slides, each displayed for 20 seconds, moving automatically. This format ensures concise, engaging presentations. The term originates from the Japanese word for "chit-chat".

3. Genetics and insurance: Complexities in the genomic era

For our third flagship event, in May 2024, the CPM collaborated with the British Society of Genetic Medicine to host an academic meeting at the Wellcome Collection in London.

Background

This event brought together invited attendees to explore the evolving relationship between genetics and insurance. Health professionals who encounter the question of how the insurance industry can, and cannot, use genetic information also attended. The day's discussions centred on if, and how, the insurance code of practice might be updated, with recent advances in technology meaning that different types of genetic and genomic data are now more routinely available.

Some of the background issues are summarised here: <u>https://cpm.ox.ac.uk/genetics-and-insurance-complexities-in-the-genomic-era/</u> and in this CPM publication: Genomics and insurance in the United Kingdom: increasing complexity and emerging challenges. Dixon P, Horton RH, Newman WG, McDermott JH, Lucassen A. Health Econ Policy Law. 2024 May 16:1-13.

The workshop

The day began with presentations from key stakeholders across patient groups, industry, government, and academia, focusing on emerging issues in genetics and insurance:

- Ana Hallgarten la Casta, Department of Health and Social Care: *The Code and the 2023 consultation*
- Sophie Peet, Genetic Alliance: *A summary of patients' and families' experiences and concerns*
- Padraig Dixon, University of Oxford: *Genetics and insurance in the UK: Increasing complexity and emerging challenges*
- William Meredew and Rebecca Ward, Association of British Insurers: *Insurance industry perspectives*

Following these presentations, the discussions shifted toward examining whether the current *Code on Genetic Testing and Insurance* ("The Code") needs revision to ensure clarity and fairness in 2024 and beyond.

A summary paper of the event is being finalised and follow up discussions with DHSC (the Department of Health and Social Care, responsible for the Code) are ongoing throughout the remainder of this year.



Our annual lectures

Each year the CPM holds two high-profile standalone lectures, an Annual Lecture and a Dr Stanley Ho Memorial Lecture.

Genomic medicine: Up close and personal

The CPM Annual Lecture 2024 was given by John Burn at the Sheldonian Theatre, Oxford, a fittingly prestigious building for such an event. Professor Burn, a leading figure in clinical genetics at Newcastle University, shared insights from his distinguished career in a lecture entitled *A 50-year odyssey in genetics*.

Professor Burn acknowledged that the terms "genetics" and "genomics" are often used interchangeably; he considered that genetics focuses on familial traits, while genomics encompasses the broader study of any biological information that can be sequenced and analysed. He highlighted the pivotal role of entrepreneurship in driving innovation, offering examples from spin-out companies he has been involved with, and noting that technological advances provide the necessary tools to frame and address key research questions.

The lecture featured several impactful examples of genomic medicine's influence on patient care. These included point-of-care DNA testing for HPV, the study of familial traits tracing back to the 18th century, and his pioneering work on Lynch syndrome, the most common form of inherited cancer. It was particularly noteworthy that many Oxford-based researchers in these fields were present in the audience, as Professor Burn acknowledged their contributions, emphasising his view of himself as just one part of a larger collaborative system of discovery.

Following a lively question-and-answer session, around 60 attendees, from students to senior professors and clinicians, moved to the adjacent Divinity School for a dinner. This provided an excellent opportunity for further discussion and networking among professionals with a shared interest in genomic medicine.



https://cpm.ox.ac.uk/watch-our-lectures-interviews/cpm-annual-lecture-series/



Revisiting genetic determinism: Evidence from large population cohorts

This year's Dr Stanley Ho Memorial Lecture was delivered by Caroline Wright at the Oxford Martin School. Professor Wright is Chair of Genomic Medicine in the Department of Clinical and Biomedical Sciences at the University of Exeter UK and Academic Director of the NHS Rare and Inherited Disease Genomic Network of Excellence. Her research focuses on the clinical applicability of genome-wide assays for rare diseases, with a particular emphasis on analysing exome and genome sequence data in rare paediatric disorders. Professor Wright's work is advancing the understanding of novel genetic causes of disease, improving the interpretation of rare genetic variants, and investigating the penetrance of pathogenic variants. She has also explored the policy and ethical challenges surrounding the integration of genome-wide sequencing into healthcare.

In her lecture, designed for a general audience as part of the Oxford Martin School's widereaching programme, Professor Wright shared valuable insights into these topics, sparking a stimulating question-and-answer session. We were grateful to Ian Huen of the Dr Stanley Ho Medical Development Foundation for introducing this prestigious lecture.

https://cpm.ox.ac.uk/watch-our-lectures-interviews/dr-stanley-ho-memorial-lectures/

Other CPM events, activities & engagement

Watermark engagement award

The CHG gained a prestigious Silver Watermark Award from the National Coordinating Centre for Public Engagement (NCCPE), a nationally recognised quality mark for engagement and the highest that can be awarded at the first time of application. The application was put together by the then Public Engagement Officer for CHG Brian Mackenwells together with Anneke Lucassen as chair of the public engagement working group for CHG. The application was praised for its strategic approach of using the CPM effectively and for emphasising context as a central element to public and community engagement with research. The feedback singled out "your significant commitment to embedding support for public engagement and your preparedness to take robust action to reflect on and enhance your support" as well as a "culture where public engagement is the norm, and all staff have time allocated to it'. Anneke Lucassen remains the chair of the working group, which draws on members of CPM and CHG to move forward the public engagement agenda of CPM, CHG, and more widely of NDM.

Student induction session at St Anne's College

Sarah Briggs spoke to all new St Anne's College medical students at the beginning of the academic year, in October 2023, exploring what personalised medicine is, highlighting how students could become involved, either directly, or via the affiliated student group, the Oxford Personalised Medicine Society.

Centre for Human Genetics Lunchtime Talks

The CPM has two slots per year to present at the CHG fortnightly Lunchtime Lab talks, designed to encourage interdisciplinary discussion. In November 2023, two researchers, Leah Boyle and Emily Parker, gave talks about their work with the CPM. Dr Boyle, a surgeon from New Zealand, presented her qualitative evidence synthesis exploring barriers and facilitators to engagement in breast cancer screening. Dr Parker, a paediatrician, spoke about her work on green inhalers and the difficulties of implementing environmental policies in the NHS.

New CPM JRF Emilie Wigdor presented her research at a second Lunchtime Lab Talk in April, showcasing the variety and breadth of CPM research interests within the team and promoting the CPM and its work to the CHG community. Her talk was entitled *Investigating the role of common cis-regulatory variants in modifying penetrance of putatively damaging inherited variants in severe neurodevelopmental disorders.*

Understanding the causes of dementia: How far have we come?

Emma Anderson from the Division of Psychiatry at University College London delivered an online lecture about the aetiology of Alzheimer's disease and related dementias, including genetic factors and their interaction with other factors such as environmental ones. Although research has come a long way, Professor Anderson highlighted there is still much work to be done to fully understand the causes of Alzheimer's disease and related dementias. The lecture took place in November 2023, and it was chaired by Padraig Dixon.

Spotlight on neuroscience

The CPM hosted a St Anne's College Subject Family Evening on the topic of neuroscience in February 2024. The speakers were Jun Ong: Examining the effect of Galectin-3 on neurons and glia; CPM JRF Emilie Wigdor: Sex differences in autism: National Registry data and common genetic variants; and CPM Steering Group member Francis Szele: Building brains to study madness. This event was offered to the whole College community. It was organised and introduced by Sarah Briggs and followed by a lively discussion facilitated by Ali Kay, which included discourse on the public perception of neuroscience research.

Can it happen again? Informing pregnancy decisions using personalised risk information

Ali Kay gave a talk with Anne Goriely on their current research on personalising recurrence risks for couples whose child's serious condition is attributed to a *de novo* (new) genetic cause. This talk, which took place at St Anne's College in March, covered both the assessment strategy PREGCARE and qualitative research with genetics practitioners on the implications of this personalised assessment for couples, and genetic counselling provision.

Exploring health inequities in personalised medicine

The CPM welcomed back Leah Boyle and Jason Torres (a former CPM JRF), to present their work at our second event exploring health inequities in personalised medicine. Hosted at St Anne's College in June and chaired by Anneke Lucassen, the evening commenced with Dr Boyle's presentation on her qualitative evidence synthesis, focusing on the barriers and facilitators to engagement in breast cancer screening in different international contexts. This was followed by Dr Torres' presentation, which outlined insights from his work with the Mexico City Prospective Study to map disease risk. CPM Fellow Susie Weller provided a commentary, drawing out common themes across the two papers. Together, the talks illustrated what different disciplinary perspectives, approaches and methods offer for our understanding of health inequities in personalised medicine, and what can, and should, happen to address them.



CPM art competition and exhibitions

Designed to encourage creative responses to issues in science, our topic this year was screening newborn babies for disease. We received over 70 entries from young people aged 11-14 from across the UK.

The winning entry, shown above in full and close-up, was by Laranya, aged 13 from Worksop College, Nottinghamshire. Laranya produced this artwork using the letters GATC the types of bases found in a DNA molecule. Laranya wrote, "When viewing the picture up close you only see the letters, but when you look from a distance you can see the face of the baby. This shows that when examining our DNA, you must look with a microscope, and these tiny proteins make up a whole person."

The panel of judges comprised: Rachel Horton and Ali Kay (CPM JRFs), Kate Keohane (St Anne's College / Ruskin School of Art), Brian Mackenwells (Centre for Human Genetics), Melville Nyatondo (Oxford Personalised Medicine Society) and Taisiia Sazonova (Oxford Personalised Medicine Society).

https://cpm.ox.ac.uk/centre-for-personalised-medicine-art-competition-2023-24/

Entries from the 2022/23 competition have been exhibited at the Northern Ireland Science Festival, the John Radcliffe Hospital in Oxford and at the Research Showcase at St Anne's College.

The CPM also exhibited artwork depicting patient journeys from research into patients' experiences of genomics at the CHG lunchtime talks.

Perinatal mental health scoping review

Conducted in collaboration with the Centre for Reviews and Dissemination, University of York, the project was designed to outline the breadth and types of evidence available from research in the field of perinatal mental health, and to help identify potential new research avenues. This research was commissioned and funded by the PAM Foundation. Susie Weller and Kate Lyle led the collaboration.

https://cpm.ox.ac.uk/perinatal-mental-health-evidence-mapping/

Mass Observation collaboration

The CPM has been collaborating with Mass Observation on an initiative designed to gauge wider public perceptions of genetics and health. Mass Observation is an ongoing national social research project that directs a panel of public participants to write about a diverse set of topics, known as Directives. Susie Weller, Rachel Horton and Anneke Lucassen commissioned a Directive on "Genetics and health in our everyday lives"; analysis of this rich data set is in progress.

Future of healthcare: The Oxford prescription

Anneke Lucassen joined researchers from across Oxford's Medical Sciences and Mathematical, Physical and Life Sciences Divisions to contribute to a report compiled by Citi Global Perspectives & Solutions (Citi GPS). You can read the full report here: https://www.citigroup.com/global/insights/future-of-healthcare

The Gift

Anneke Lucassen took part in a BBC Radio 4 series that explored direct to consumer ancestry tests. On the one hand they are often thought of as a fun gift to provide information about which part of the world we stem from - yet on the other they are sold as powerful means of personalising health risk predictions. Episode 5 of focussed on the complexities of such health predictions and how they often provide far from clear answers. You can listen to the series here:



https://www.bbc.co.uk/sounds/brand/p0gd2dgb

Race and Health observatory award

Together with the Genomic Medicine Service Alliance (GMSA), Anneke Lucassen was part of a successful bid to examine health inequalities and ethnicity in the NHS Genomic Medicine Service. The research is exploring how terms such as ethnicity, race and ancestry are used and understood in the practice of genomic medicine, and particularly how these social constructs are then translated to interpret genomic variants and their link with diseases.

Cancer and climate change

In a collaboration with Oncologists United for Climate and Health (OUCH), Sarah Briggs chaired a session in November in which some of the key challenges for the global oncology community were discussed, acknowledging that cancer care itself has a significant carbon footprint and environmental impact.

Linked lives: The impact of genomic testing on families

Anneke Lucassen spoke at the Festival of Genomics and Biodata in London in January. This annual festival is aimed at the entire genomics and biodata community and has several thousand attendees. Professor Lucassen discussed some of the CHG research that explores how families engage with genomic testing, and she showed the CPM artwork and animation resulting from this work.

Economic considerations in personalised medicine

Padraig Dixon took part in Imperial College's Personalised Medicine Student Society symposium *Precision therapeutics: A genetic revolution*, which took place in January. His talk highlighted some of the opportunities and challenges of using genetic data in insurance, the value of innovative but expensive pharmacotherapies, and the economic implications of using polygenic risk scores in medical screening.

Utility of polygenic scores in healthcare

Anneke Lucassen spoke about the use of polygenic scores at a UK National Screening Committee online seminar in February. She discussed research insights from polygenic scores, but highlighted how these often translate to very modest clinical utility for individuals, which is often contrary to expectations.

Niche to Normality! Genomics for nurses, midwives and HCPs

On Rare Disease Day, 29 February, Anneke Lucassen and Padraig Dixon participated in a webinar which focused on the patient and family experience. Professor Lucassen led a session focusing on the challenges of translating detailed molecular genetic information into useful clinical information. Dr Dixon discussed what patients need to know about genomics and insurance, and how this area is evolving in the United Kingdom.

Precision medicine lectures

In March, Sarah Briggs delivered lectures on personalised medicine to the MSc in Experimental and Translational Therapeutics (Department of Oncology and Department for Continuing Education), and to the MSc in Genomic Medicine (CHG).

A Greener NHS: How could / should healthcare change?

Sarah Briggs gave a lunchtime Senior Common Room research talk to members of St Anne's College in April, discussing the actions the NHS is taking to make healthcare more environmentally sustainable, and preliminary results of qualitative research she has recently undertaken looking at public understanding and attitudes towards sustainability in healthcare.

A personalised predictive risk-based approach to clinical medicine

Anneke Lucassen delivered this talk in May to the Medical Pilgrims, a group of senior clinical academics from around the UK, she outlined the work of the CPM and our strategy going forward. The meeting was held in Oxford and CPM Steering Group members Julia Hippisley-Cox and Holm Uhlig also spoke.

Clinical genomics and next generation sequencing

This regular week-long European Society of Human Genetics summer school in Bertinoro, Italy, took place in May. It is aimed at professionals in clinical and medical genetics. Starting from the basic notions of medical genetics and bioinformatics, Anneke Lucassen gave a lecture on some of the common ethical issues encountered in genomic practice, as well as two workshops on population genomic screening.

European Society for Human Genetics conference

Anneke Lucassen and Helena Carley (CHG) led a workshop on population screening using genomics at the annual Conference in Berlin, Germany in June. They highlighted the importance of context in interpreting predictions that genetic variants impart.

Deepening our understanding of risk

Anneke Lucassen gave an invited lecture at the Cancer Prevention Research Conference in Boston, United States, in June, exploring personalised risk prediction. Her talk focussed on the possible futures promised by advances in technology, and highlighted the hurdles that need to be navigated before these can be successfully implemented in society.

Enriching engagement

Ali Kay gave a talk to the science journal club at Magdalen College School in June. The session included newborn screening using genomics and the implications of testing as well as an animated film exploring genetic variation. The students gave thoughtful responses about evaluating the pros and cons of expanding the range of conditions currently tested for in the UK Newborn Screening Programme.

Oxford University Personalised Medicine Society

Report from Maximilian Lin (MBiomedsci), OUPM President 2023-24

The Oxford University Personalised Medicine Society (OUPM) was founded by students at the University of Oxford studying a variety of subjects, but united by a common interest in personalised medicine. We work closely with the CPM to host a variety of education, speaker and career events throughout the academic year for our community of 1000+ lifetime members. In the 2023-24 year, these have included:

- A welcome event to get to gain insights into the society operations and encourage networking amongst members
- A series of introductory R coding workshops

O/U/P/m

- An Oxford Bio hackathon, in collaboration with the Oxford University Biotechnology Society and the Computer Science Society
- A talk by Dr Ron Zimmern (CPM External Advisory Board) on *Personalised medicine: Its public health genomics antecedents*
- A biotech start-up panel session, in collaboration with the Oxford Founders Society
- A guest lecture by Professor Dean Fennell on *Translational thoracic cancer research*
- A talk by Professor Munir Pirmohamed on *Pharmacogenetics*
- An entrepreneurship panel session in collaboration with the Oxford Entrepreneurs Society
- Our flagship Annual Symposium, with seven student speakers and keynotes delivered by John Todd and Francis Szele, both from the CPM Steering Group
- A conference on *Inclusion and interdisciplinarity in science*, in collaboration with the Oxford Women in Science and Technology Society
- A guest lecture by Professor Andrea Cipriani on *Precision psychiatry*, in collaboration with the Oxford Psychology Society
- A visit to the headquarters of Nanopore Technologies
- A talk by the Chief Technology Officer of Immunocore, Dr Annelise Vuidepot, on *T*-*cell receptor biotechnology*

Additionally, we brought out educational newsletters and published two editions of our

popular magazine *The Gene 'Zine*.

We would like to express our deep gratitude for the support of the CPM, without which we would not have been able to organise such successful events!

https://www.oupm.co.uk/





The year ahead

We are busy planning a range of activities around our core themes during the year ahead, and have the following lined up, but with more to come!

- Clinical Genomics workshop: An event exploring how clinical genomics can guide care for individuals with monogenic inflammatory bowel disease. This workshop is aimed at paediatric and adult gastroenterologists, trainees, nurse specialists, and other specialists involved in monogenic IBD. *(Theme: Diagnosis and Treatment)*
- Songs of Genomics: A musical project that collaborates with a contemporary music ensemble to create pieces inspired by patients' lived experiences with genomics in healthcare. *(Theme: Experiences of Personalised Medicine)*
- Accountability in Al workshop: In collaboration with the Oxford Internet Institute, this workshop will focus on trustworthy and explainable Al systems, with applications in personalised medicine. *(Themes: Risk and Prevention, Diagnosis and Treatment)*
- Newborn screening through genomics: An event exploring newborn screening, cohosted with *Cambridge Prisms: Precision Medicine* journal to explore what success would look like in the context of a rare disease screening programme. *(Themes: Risk and Prevention, Diagnosis and Treatment)*
- **Cancer Vaccines symposium**: An event exploring cancer vaccines, organised in collaboration with OxCODE (Oxford Centre for Cancer Early Detection and Prevention). *(Theme: Risk and Prevention)*
- **ReNU syndrome**: An event describing the recent discovery of ReNU syndrome, with lead author (and former CPM JRF) Nicky Whiffin among the speakers
- Young People's Art Competition: A competition inviting young artists to explore how personalised medicine affects our planet, reinforcing our commitment to sustainability. *(Theme: Sustainability)*
- **Podcast Series**: We're embarking on our third series, exploring all six of the CPM's core themes.

Further details on all our forthcoming events can be found: <u>https://cpm.ox.ac.uk/events/</u>

Appendix: Publications arising from CPM work

- Challenges of using whole genome sequencing in population newborn screening. Horton R, Wright CF, Firth HV, Turnbull C, Lachmann R, Houlston RS, Lucassen A. BMJ. 2024 Mar 5;384:e077060.
- Genomics and insurance in the United Kingdom: increasing complexity and emerging challenges. Dixon P, Horton RH, Newman WG, McDermott JH, Lucassen A. Health Econ Policy Law. 2024 May 16:1-13.
- Glowing gels and pipettes aplenty: how do commercial stock image banks portray genetic tests? Horton R, Boyle L, Weller S, Lucassen A. Eur J Hum Genet. 2024 Apr;32(4):456-460.
- Ancestry, race and ethnicity: the role and relevance of language in clinical genetics practice. Redman MG, Horton RH, Carley H, Lucassen A. J Med Genet. 2024 Mar 21;61(4):313-318.
- Genomic data: building blocks for life or abstract art? Horton R, Lyle K, Weller S, Ballard L, Lucassen A. Front. Young Minds. 2024. 12:1249534.
- Discussion of off-target and tentative genomic findings may sometimes be necessary to allow evaluation of their clinical significance. Horton RH, Macken WL, Pitceathly RDS, Lucassen AM. J Med Ethics. 2024 May 9;50(5):295-298.
- Realistic expectations are key to realising the benefits of polygenic scores. Sud A, Horton RH, Hingorani AD, et al. BMJ. 2023 Feb 28;380:e073149. doi: 10.1136/bmj-2022-073149.PMID: 36854461
- Why digital innovation may not reduce healthcare's environmental footprint. Samuel G, Anderson GM, Lucivero F, Lucassen A. BMJ. 2024 Jun 3;385:e078303. doi: 10.1136/bmj-2023-078303. PMID: 38830688; PMCID: PMC7616622.
- Ethical preparedness in genomic medicine: how NHS clinical scientists navigate ethical issues. Sahan K, Lyle K, Carley H, Hallowell N, Parker MJ, Lucassen AM. J Med Ethics. 2024 Jul 23;50(8):517-522. doi: 10.1136/jme-2023-109692. PMID: 38320848; PMCID: PMC11287646.
- Population screening requires robust evidence-genomics is no exception. Turnbull C, Firth HV, Wilkie AOM, Newman W, Raymond FL, Tomlinson I, Lachmann R, Wright CF, Wordsworth S, George A, McCartney M, Lucassen A. Lancet. 2024 Feb 10;403(10426):583-586. doi: 10.1016/S0140-6736(23)02295-X.
- The ethical challenges of diversifying genomic data: A qualitative evidence synthesis. Hardcastle F, Lyle K, Horton R, et al. Cambridge Prisms: Precision Medicine. 2024;2:e1. doi:10.1017/pcm.2023.20
- Polygenic risk scores: Colloquium held at the Centre for Personalised Medicine, Oxford. Dixon P, Briggs S and Lucassen A. Cambridge Prisms: Precision Medicine, 2023, 1, e32, 1-3
- Immortal data: a qualitative exploration of patients' understandings of genomic data. Lyle, K., Weller, S., Horton, R. and Lucassen, A. 2023. European Journal of Human Genetics, 31, 681-686.
- Beyond regulatory approaches to ethics: Making space for ethical preparedness in healthcare research, Lyle, K., Weller, S., Samuel, G. and Lucassen. A. Journal of Medical Ethics. 2024 49: 352-356
- Fostering habits of care: Reframing qualitative data sharing policies and practices. Weller, S. Qualitative Research, 2023 23(4): 1022-1041.
- Professionals' views on providing personalized recurrence risks for de novo mutations: Implications for genetic counseling. Kay, A. C., Wells, J., Goriely, A., & Hallowell, N. Journal of Genetic Counseling, 2024 00, 1–13.
- Providing recurrence risk counselling for parents after diagnosis of a serious genetic condition caused by an apparently de novo mutation in their child. Kay AC, Wells J, Hallowell N, et al Journal of Medical Genetics 2023;60:925-931.
- Focussing attention on physicians' climate related duties risks missing the bigger picture: towards a systems approach to health and climate. Samuel G, Briggs S, Hardcastle F, Lyle K, Parker E, Lucassen A. Journal of Medical Ethics 2024;50:380-381
- Big qual: A guide to breadth-and-depth analysis, Weller, S., Davidson, E., Edwards, R. and Jamieson, 2024. London: Palgrave Macmillan.
- Balancing the rights of the pre-symptomatic child to be found with the risk of harm to others from the screening process. Lucassen A, Horton R. Eur J Hum Genet. 2024 <u>https://doi.org/10.1038/s41431-024-01689-6</u>



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