



Policy workshop on genomics and artificial intelligence

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Acadamh Ríoga na hÉireann
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Policy workshop on genomics and artificial intelligence

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Executive summary

Genome sequencing and large-scale genotyping are rapidly entering healthcare practices in various settings. Rapid and large-scale genomic analyses are already improving diagnosis of rare diseases, and it may soon be possible to tailor treatments based on genomic variants. Work is underway to explore the potential for genomic information to predict the risk of future diseases, but more research is needed to establish its clinical utility.

At the same time, artificial intelligence (AI) has emerged as a powerful tool with many possible applications in healthcare. AI tools may help to analyse genomic information more effectively and open up new approaches to use genome sequence information to advance health.

Alongside these opportunities there are also challenges that need further debate, as genomics and AI enter healthcare practices. In November 2024, the Royal Irish Academy and the UK Academy of Medical Sciences held a joint meeting in Dublin, Ireland to take stock of progress in the development and use of AI-driven tools for genomic medicine in the UK and Ireland, as well as to explore opportunities and challenges.

Discussions at the workshop identified a set of priority issues, across three interrelated domains, that need to be addressed to realise the promise of genomics and AI.



1. Technical opportunities and challenges

Participants identified many potential applications of AI in genomics. Those of greatest potential opportunity include approaches to better understand the role of **genetic variants** in disease by integrating multiple types of data, for example identifying their role in developmental disorders, and providing prognostic information and guiding choice of treatment in **cancers**.



Technical priority areas

- 1.1. Explainability:** Overcoming explainability challenges and ensuring the development of transparent tools.
- 1.2. Verification:** Establishing systems for rigorous independent evaluation of new tools, drawing on standardised datasets to assess performance in a diversity of populations.
- 1.3. Prioritisation:** Identifying areas of medicine where AI and genomics could have the greatest positive impact, and where it stands to have significant negative impacts.

In addition, the ability of AI to integrate multiple types of **'omics' data** (large-scale data on gene activity, cellular proteins and metabolites) opens up new opportunities to dissect the underlying mechanisms of diseases. Furthermore, use of AI systems to analyse and classify **biological images**, and to extract information from the **scientific literature**, creates powerful new ways to connect genotype and phenotype.

Participants highlighted several technical challenges. These include the lack of **explainability** (and transparency) inherent in many AI applications – how AI systems arrive at their outputs is often not understood. Current genome datasets

are dominated by data from individuals of European descent. This **lack of representativeness** in genome sequence databases, which are used as training sets for AI, is important since some tools perform poorly when used on data from individuals of a different geographic ancestry. The importance of **independent validation** of tools, including their performance across multiple populations, was stressed.

It was also felt that innovation should be **targeted** where it could achieve greatest public health impact, for example by identifying and incentivising investment in priority areas.



2. Practical and implementation opportunities and challenges

There is much enthusiasm to integrate genomics and AI research and innovations into healthcare but there are also practical barriers, including variation in **data formats**, which makes it difficult for AI systems to identify patterns and make accurate predictions, and lack of **interoperability** of IT systems.



Implementation priority areas

2.1. Data standards and interoperability:

Development of common global data standards and interoperable IT systems. The Global Alliance for Genomics & Health (GA4GH) has been leading progress towards this goal.

2.2. Practical barriers and enablers: Exploring factors likely to impede or accelerate the introduction and use of AI and genomic tools, and related research in health systems.

2.3. Education and training: Assessing potential implications for health workers' professional development, training and education.

2.4. Consumer protection: Strengthening regulation of direct-to-consumer testing.

Genome sequencing generates large quantities of data. Navigating confidentiality and privacy, and the various regulatory frameworks (such as General Data Protection Regulation, GDPR) is difficult, particularly as genomics often straddles both research and healthcare environments, each with their own governance arrangements. A **federated approach** to storage of such data is increasingly being adopted, in which original/source data remain securely held in one environment in which AI or other software tools are trained or tested.

Participants also highlighted implications for health **workforce training**, as well as the need to consider **acceptance** of AI tools by health workers, often at an individual level.

Many commercial companies are developing AI-based tools for use in healthcare. While involvement of the **private sector** can bring in innovative approaches, commercial pressures were felt to raise risks, such as high cost, technology lock-ins and promotion of products of limited clinical utility.

Direct-to-consumer marketing of genetic and genomic tests provides an additional source of genomic information, which patients may bring to consultations with healthcare professionals. However, there are many concerns about the quality of this information and the limited support given to consumers on interpretation of test results.



3. Ethical, social and legal opportunities and challenges

Multiple ethical, social and legal challenges relating to genomics and AI were discussed – some long-standing concerns relating to genetics and others more specific to AI and genomics. These included **data privacy**, **discrimination** based on genetic data, and the impact of **biases** in training data.



Ethical, social and legal priority areas

3.1. Public and patient engagement and involvement: Raising awareness of potential AI and genomic medical applications, and ensuring patients and the public are involved at all stages of prioritisation, product development, service design and creation of regulatory/governance frameworks.

3.2. Trustworthiness: Identifying the key principles that underpin public trust in AI and genomic applications. Building legislative and regulatory frameworks and governance mechanisms based on these principles as one approach to help facilitate public trust.

3.3. Addressing inequities by design: Promoting product development that considers a diversity of target populations from the outset.

3.4. Sustainability: Encouraging R&D that minimises the environmental costs of AI development and exploring possible mechanisms, such as procurement policy, to shape product development and production practices.

Participants also highlighted the **environmental and social costs** of AI tool development. Data processing and AI applications are energy-intensive and contribute to climate change. Importantly from a societal perspective, personalisation of health risks using AI tools has the potential to undermine the principles of social solidarity and risk-sharing that are central to insurance and publicly funded health systems.

Building and maintaining **public trust** in genomic AI tools was felt to be essential. Public and patient involvement and engagement were seen to be critical to the development of **trustworthy applications**, with **deliberative approaches** seen as particularly valuable to explore the complex and nuanced range of issues associated with AI and genomics.

The workshop concluded that the synergy of AI and genomics has great potential to improve healthcare but should not be rolled out without careful attention to multiple technical, practical and societal challenges.

Strong engagement with patients and the public is necessary to guide the direction of innovation and to ensure that any new applications have public trust and support. There was unanimous agreement that there were major opportunities for the UK nations and Ireland to work together, and with other international partners, to ensure that the potential of AI and genomics to health is realised in a responsible and equitable way.

Introduction

The past decade has seen major technological advances in both artificial intelligence (AI) and genome sequencing and large-scale genotyping (assessing genetic variation at hundreds of thousands of sites across the human genome). Many are enthusiastic that the combination of these two fields could deliver major benefits in multiple areas of health.

Both the UK and Ireland have recognised the importance of the two domains to the future of healthcare. In the UK, the NHS genomics strategy, Accelerating Genomic Medicine in the NHS,¹ sets out a vision for genomics in the NHS in England, combining implementation of genomic tools at all levels of the health system with a focus on integrated research and innovation. The strategy is aligned with other health and life science strategy documents, including 'Genome UK: The future of healthcare',² the NHS Long Term Plan, the UK Life Sciences Vision, and the UK Rare Diseases Framework. Scotland published a five-year genomic medicine strategy in 2024,³ while Wales and Northern Ireland are signed up to the Genome UK strategy. Likewise, Ireland has developed a National Strategy for Accelerating Genetic and Genomic Medicine in Ireland.⁴

In November 2024, the Royal Irish Academy and the UK Academy of Medical Sciences held a joint workshop in Dublin, Ireland to explore the state of the art in AI and genomics, and their interaction. It examined the opportunities offered by technological advances, as well as the challenges that need to be considered to make progress.

¹ Accelerating Genomic Medicine in the NHS (2022). <https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/>

² <https://www.gov.uk/government/publications/genome-uk-the-future-of-healthcare>

³ <https://www.gov.scot/publications/scotlands-genomic-medicine-strategy-2024-2029/>

⁴ <https://www.hse.ie/eng/about/who/strategic-programmes-office-overview/national-strategy-for-accelerating-genetic-and-genomic-medicine-in-ireland/>

The growth of genomics

Since the first human genome was sequenced, the cost of genome sequencing has fallen dramatically and the speed by which sequence information can be generated has increased exponentially. This means that whole-genome approaches are becoming more affordable in healthcare, where previously only targeted testing was possible. Data obtained through large-scale population studies such as UK Biobank, Our Future Health and the **Generation Study** are being used to assess how genomic variants contribute to disease over the course of a lifetime.^{5,6}

These advances in genomics have required the development of **bioinformatics**, the analytical tools and pipelines required to filter and sift the millions of variants within any genome, to find those that cause particular health conditions or predict the risk of disease.

Genomics is therefore transitioning from a research tool to one that might help diagnosis in the clinic, or contribute to risk prediction within public health. Genetic factors play a part in almost all health conditions – from being a major component in single-gene disorders such as cystic fibrosis to contributing to the risk of more common conditions. Many medical applications are possible, including:

Providing diagnoses in rare diseases

Many families have undertaken long diagnostic odysseys to find an explanation for their child's rare disease. In these selective cohorts (e.g. 100kGP) genomic approaches can increase the diagnostic rate, but even in these groups around 75% remain undiagnosed, leading to a continuation of the distressing uncertainty. Genomic sequencing in non-selective groups, such as programmes to sequence the genomes of all newborns, may lead to predictive uncertainty for many families, where genetic changes are detected but the consequences, if any, are not known.

Targeting oncology treatments

Identifying the specific genetic changes driving the development of a patient's cancer can indicate which treatments are most likely to work best.

Pharmacogenomics

Certain genetic factors affect how well a particular drug or treatment might work, or whether (and what) side effects are likely. Genetic testing could therefore be used to guide treatment choice and dosing.

Disease prediction

For most common health conditions, a combination of many genetic variants influence, to varying degrees, the risk of developing a disease or manifestations of that disease. Genotyping individuals can be used to generate a polygenic risk score, which provides an indication of their likelihood of developing a condition, enabling populations to be stratified into high-, moderate- or low-risk groups. Those at highest risk can receive tailored behavioural advice, be monitored more carefully, or be given preventive treatment. However, for the most common diseases, polygenic risk scores do not change the absolute risk by more than a few percentage points. Thus, polygenic risk scores are best used alongside other risk assessments. They may also have value in diagnostic assessments and in predicting the likelihood of particular disease outcomes.

⁵ <https://www.genomicsengland.co.uk/initiatives/newborns>

⁶ <https://ourfuturehealth.org.uk>

Advances in AI

In recent years, AI has become particularly associated with so-called **large language models** and **generative AI**, such as ChatGPT. These applications are trained to identify patterns in large text datasets, which they can use to generate responses to user queries.

Generative AI applications have rapidly become everyday tools and embedded within many other digital systems. Consumer-focused applications have been trained on datasets composed of text, but the same learning principles can be applied to other types of data, including genomic data which, like language, consists of a string of characters (A, C, G and T). Just as strings of characters in text make words and sentences, strings of DNA characters make up features of genomes, such as genes or gene control elements.

There are multiple ways in which AI might be integrated into healthcare, for example:

Task automation

Generative AI could generate outputs, such as summaries of consultations or interpretations of test results. Efficiency gains may be possible through automation of routine administrative tasks.

Data integration

Genomic information can provide insights into disease risks. Additional information can come from other kinds of genome-related data, such as gene expression data (transcriptomics), protein synthesis data (proteomics) or other 'omics' data. More specialised AI tools can integrate these different kinds of data to provide more refined insights into the risk of disease, diagnosis or prognosis (prediction of disease course). AI could also integrate genomic data with functional and behavioural data ('phenomics') and with data on environmental exposures affecting health ('exposomics').

Extracting and organising knowledge

The rapid growth of scientific knowledge, captured within academic publications, needs to be assimilated. AI tools have the potential to identify and summarise information held within millions of scientific papers.

Interpreting and classifying images

As well as words, AI systems can be trained on visual images, learning what visual patterns are associated with particularly important aspects of an image. They can therefore learn to extract key diagnostic and prognostic information from images, such as those generated by medical imaging technologies (e.g. X-rays, CT, MRI).

At the workshop, participants heard about some of the latest developments in the application of machine learning/AI in genomics and how efforts are being made to translate these advances into routine medical practice. Participants also discussed some of the obstacles and enablers that affect the speed of translation, as well as the social, ethical and legal complexities associated with AI and genomics in healthcare. Several key themes emerged from these discussions across three broad and interrelated domains.



1. Technical opportunities and challenges

Opportunities

In presentations and discussions, participants identified multiple areas where research is identifying new opportunities for the application of AI in genomic medicine.

One key area is the understanding of **genomic variation**. A central challenge in genomics is to understand the biological consequences of genetic variants. When a genome is sequenced, millions of variants will be identified, and the biological relevance of many of these is often certain. They may be making a critical contribution to a disease process or be of little or no health significance.

Furthermore, an understanding of the genome requires an **assignment of function to its component parts**. This includes the location of genes, but these make up a tiny fraction (1%–2%) of the genome. Regulatory regions that control the expression of these genes also need to be identified and mapped.

As discussed by Professor Ian Simpson (University of Edinburgh), '**foundation models**' have been developed for genomes, analogous to large language models for text but trained on DNA sequences. They can be used to predict particular functional structures within genome sequences, such as genes or regulatory regions. HyenaDNA, for example, analyses DNA sequences in a way that can deal with the discontinuities of structural elements in the genome, with component parts of some control elements being separated by many thousands of nucleotides.⁷

The understanding of genetic contributions to common health conditions such as diabetes or cancer has advanced significantly through **genome-wide association studies (GWAS)**, which assess the association of hundreds or thousands of genetic variants with a particular health condition. Usually, the vast majority of variants make only a tiny contribution to risk. However, in combination, they can account for a significant proportion of the genetic component of disease risk. Many genetic variants identified in GWAS of complex traits are non-coding and it is challenging to characterise their function. AI systems are beginning to offer a way to collate and interpret data on these variants and to make predictions regarding their characteristics.⁸

Oncology is an area where AI systems may be of particular value for interpreting complex genomic information. Cancers are, in effect, genetic diseases, caused by mutations that drive uncontrolled multiplication of cells. Cancers are typically highly heterogeneous and complex – cancer cells accumulate mutations as they divide (including both single nucleotide changes and large structural rearrangements, duplications or deletions). AI is helping to collate and integrate genomic data from cancer cells with data on gene activity, protein levels and other cellular characteristics.

As noted by Professor Simpson, AI systems offer a way to integrate multiple types of data and to identify features of an individual cancer that are most biologically relevant, for example to its

⁷ Nguyen E *et al.* (2023). *HyenaDNA: long-range genomic sequence modeling at single nucleotide resolution*. <https://doi.org/10.48550/arXiv.2306.15794>

⁸ Kathail P, Ayesha Bajwa, Ioannidis NM (2024). *Leveraging genomic deep learning models for non-coding variant effect prediction*. <https://doi.org/10.48550/arXiv.2411.11158>

prognosis or responsiveness to treatment.⁹ These insights can be used to guide care and choice of treatments. For example, in prostate cancer an AI system was able to identify mutations associated with poor prognosis.¹⁰

Another area of interest is the use of '**digital twins**' – combining datasets to generate dynamic computer models of cancers and patients. As discussed by Professor Walter Kolch (University College Dublin), these 'virtual patients' can be studied to shed light on disease processes, to support targeted therapy development, and to underpin tailored diagnostics and treatments.¹¹

Professor Kolch described other applications of AI, including tools to unpick the **evolutionary history of cancers**, so that the likely sequential acquisition of mutations can be predicted. '**Deconfounder**' tools are being developed that can more accurately identify gene expression signatures associated with particular types of cancer, by removing the effects of mutations that perturb gene expression but do not contribute to cancer development.

Genomes are often described as 'blueprints' or instruction manuals for cells, tissues and bodies, but such descriptions shroud the fact that the **pathway from genotype to phenotype** is complex. For example, the impact of genes may depend on the presence or absence of many other factors, including other genes, and epigenetic and environmental factors. The phenotypic consequences of genetic variation may therefore vary between individuals. As a result, it may be difficult to link a diffuse set of phenotypes to underlying genetic causes. There is potential to leverage AI in **phenotypic analysis**, to link phenotypic variation to its underlying genetic roots.

Phenotypic analysis remains important in cancer, with **pathology investigations** being an important part of cancer diagnosis. Professor Aedín Culhane (University of Limerick) described how AI can contribute to such **image analyses**. One example is detection of **perineural invasion**, when cancer cells become attached to nerve fibres, which is useful to detect as it is associated with poor prognosis. The human eye struggles to detect this feature consistently, and AI image classifier systems offer the prospect of more consistent assessments.¹²

Much phenotypic information can be found in the **scientific literature**. However, this information can be difficult to extract and to contextualise at scale. These issues could be overcome with AI tools, although the heterogeneity of information within free-form text presents a major challenge. As part of the Primary Annotated Resources to Advance Discovery in Genomic Medicine (**PARADIGM**) Wellcome Discovery-funded award, described by Dr Michael Yates (University of Edinburgh), methods have been developed to map descriptions of phenotypic features to a standardised taxonomy.^{13,14} This approach will facilitate automation efforts to link genotypic and phenotypic information, which is currently a labour-intensive manual curation process.¹⁵

⁹ Ryan B, Marioni RE, Simpson TI (2024). *Multi-Omic Graph Diagnosis (MOGDx): a data integration tool to perform classification tasks for heterogeneous diseases*. *Bioinformatics* **40**, btae523.

¹⁰ Elmarakeby HA et al. (2021). *Biologically informed deep neural network for prostate cancer discovery*. *Nature* **598**, 348–352.

¹¹ Zhang K et al. (2024) *Concepts and applications of digital twins in healthcare and medicine*. *Patterns* **5**, 101028.

¹² Pantanowitz L et al. (2020). *An artificial intelligence algorithm for prostate cancer diagnosis in whole slide images of core needle biopsies: a blinded clinical validation and deployment study*. *Lancet Digital Health* **2**, e407–e416.

¹³ Yates TM et al. (2022). *Creation and evaluation of full-text literature-derived, feature-weighted disease models of genetically determined developmental disorders*. *Database* **2022**, baac038.

¹⁴ <https://paradigmgenomics.org>

¹⁵ Yates TM et al. (2024). *Curating genomic disease-gene relationships with Gene2Phenotype (G2P)*. *Genome Medicine* **16**, 127.

Challenges

Bias: A common challenge across both AI and genomics is the risk of **bias due to unrepresentative datasets**. AI tools need to be trained, and most genomic training datasets lack genetic diversity, being based on data mainly from individuals with European ancestry.¹⁶ Despite calls to address this over the last decade or so, the understanding of genomic variation remains very skewed. Much of human genetic diversity, which is greatest in sub-Saharan African populations, is poorly represented in public data sources.

As discussed by Dr Daniel Murphy (University College Dublin), the **human reference genome** is a global standard for research and genomic medicine. However, the widely used GRCh38 assembly (released in 2013) was derived from just 20 individuals of mainly European ancestry, with 70% of the sequence coming from a single donor. This lack of diversity introduces reference bias, reducing variant detection accuracy in underrepresented populations and potentially leading to missed or incorrect diagnoses.

A more inclusive approach is to use a **pangenome** model,¹⁷ which integrates multiple individual genomes to better capture human genetic diversity. It moves away from the simple linear sequence of the standard human reference genome, incorporating structural variation (deletions or insertions) that can be important contributors to genetic disease. However, it is more computationally challenging to compare newly derived sequences with this more complex genomic representation and its adoption will require new tools, changes to data storage and representation, and broad community collaboration to update current practices.

Another approach discussed at the workshop is '**synthetic DNA**' – artificially generated DNA sequence. DNA of any desired sequence can be generated and used to test and validate AI tools. Furthermore, the growing capabilities of DNA synthesis are opening up new opportunities to engineer and study genomic diversity.¹⁸ However, use of synthetic DNA may also raise a variety of ethical issues.¹⁹

Priority areas

1.1. Explainability: Overcoming explainability challenges and ensuring the development of transparent tools.

1.2. Verification: Developing systems for rigorous independent evaluation of new tools, drawing on standardised datasets to assess performance in a diversity of populations.

1.3. Prioritisation: Identifying areas of medicine where AI and genomics could have greatest impact.

Genomic data complexity: Interpretation of genomic data also presents technical challenges. The properties of a particular stretch of DNA are not solely dependent on its sequence. **Epigenetic effects**, the chemical modification of DNA nucleotides (e.g. methylation) or of histones (e.g. acetylation), can have a profound impact on the biological impact of a stretch of DNA. For this reason, '**omics**' technologies can provide more insights than 'raw' DNA sequences, but add complexity by requiring the generation and integration of multiple types of data. In addition, the genome is **not stable over time**. Somatic mutations occur, changing genome sequences, in response to external

¹⁶ Popejoy AB & Fullerton SM (2016). *Genomics is failing on diversity*. Nature **538**, 161–164.

¹⁷ Liao WW et al. (2023). *A draft human pangenome reference*. Nature **617**, 312–324.

¹⁸ de Boer CG & Taipale J (2024). *Hold out the genome: a roadmap to solving the cis-regulatory code*. Nature **625**, 41–50.

¹⁹ Villalba A et al. (2024). *The ethics of synthetic DNA*. Journal of Medical Ethics; <https://doi.org/10.1136/jme-2024-110124>.

DNA-damaging insults and during cell division. In many cancer cells, DNA repair is disrupted, leading to the rapid accumulation of mutations.

Lack of explainability: A frequently mentioned challenge at the workshop was that of 'explainability'. Many AI systems are conceived as '**black boxes**' – it is unclear, even to developers, how they have generated their outputs. In the medical context, care providers may feel that the lack of explainability may undermine public trust. Such systems are also incompatible with principles of transparency.

The explainability issue is being addressed in technology development, through '**explainable AI**'. AI systems are being developed that 'self-inspect' and report on how they reached their outputs. Alternatively, explainable AI can also be used to probe the internal processes of black-box AI systems.



2. Practical and implementation opportunities and challenges

While research continues, genomics is rapidly being integrated into routine healthcare systems. This hybrid state of genomics can pose practical challenges to governance, since research and clinical practice have different mechanisms of oversight.

Key challenges include **storage** of large amounts of data, **linkage** of genomic data to related patient data, and protecting **data privacy** while ensuring access for research, as well as for clinical purposes.

Appropriate access to data is crucial for research and clinical care. While electronic health records can provide integrated access to data, this makes anonymisation more difficult, and privacy harder to protect. There is a growing trend for genomic data to be maintained in safe environments in which third-party analytical tools can be deployed. As discussed by Professor Culhane, such a '**federated**' **approach** can ensure compatibility with General Data Protection Regulation (GDPR) and safeguard privacy while minimising the risk of data silos. Use of **common data models** can facilitate analyses across sites.

More than 20 countries in Europe have signed up to the **1+MG Declaration**,²⁰ which aims to embed a common framework and data architecture for genomic data to facilitate international studies and advance personalised medicine.

Genomic data are particularly important for cancer, but their interpretation should ideally draw on corresponding patient data to enhance the understanding of how environmental factors contribute to disease risk. The relative contribution of intrinsic causes of somatic mutations (for example, mistakes introduced during cell division) and extrinsic mutagenic insults (such as exposure to mutagenic chemicals or radiation) remains uncertain, but environmental factors are likely to be a significant contributory factor.²¹ Linking cancer genome data with environmental data can shed light on environmental exposures driving cancer.²²

²⁰ <https://framework.onemilliongenomes.eu/about-the-framework>

²¹ Wu S *et al.* (2016). *Substantial contribution of extrinsic risk factors to cancer development*. Nature **529**, 43–47.

²² Senkin S *et al.* (2024). *Geographic variation of mutagenic exposures in kidney cancer genomes*. Nature **629**, 910–918.

From 2021, genomics will be part of the **European Health Data Space**,²³ designed to promote safe and secure research on health data across the EU. On an even wider scale, adoption of global common data standards, such as the **Observational Medical Outcomes Partnership (OMOP)**, can facilitate global data-based collaborations.²⁴ Closer to home, various efforts are also being made to utilise health data for cancer research across the whole of the island of Ireland, with more standardised data systems.

In the UK, a systematic attempt is being made to integrate AI into NHS England's **Genomic Medicine Service**,²⁵ an initiative described by Professor Eamonn Sheridan (University of Leeds). Launched in 2018, the service aims to leverage advances in genomics in a range of areas of medicine, integrating the work of multiple stakeholder organisations around Clinical Innovation Hubs.

As for the AI dimension, a key role is being played by the **Genomic AI Network (GAIN)**,²⁶ a multidisciplinary and multisectoral community of practice that will generate evidence on AI applications and their implementation within the NHS. In addition, **Genomic Networks of Excellence** have been established in multiple therapeutic areas.

The goal is to create a new digital infrastructure and model for genomic data access within the NHS, built around a **Unified Genomic Record**. This will provide a common platform for 'users', such as clinicians requesting genetic tests, and 'providers' delivering genetic services.

The platform is built around a three-level model, including: a mixed centralised/federated data storage foundation; an intermediate 'interoperability' level that will ensure access from multiple different types of users and systems; and a user-facing level providing the interface through which service users interact with the Unified Genomic Record.

Challenges

Data standards/interoperability: Participants highlighted a range of practical implementation challenges. Foremost among them is the diversity of data sources and lack of **data standards**. Limited interoperability of IT systems is a further barrier to research and the development of integrated services.

Training: New genomic and AI technologies will have implications for many kinds of health workers. As well as **workforce training needs**, there will be a need to consider health workers' **attitudes to new technologies**, including the potential for resistance to AI tools, for example in general practice. Potentially, lessons could be learned from the mixed take-up of existing clinician decision-support tools, to identify human factors associated with effective development and adoption of AI tools.

Evaluation of tools: The **evaluation and validation** of AI tools for genomics was another area of concern highlighted by participants. Specific issues included the lack of transparency in black-box systems and potential for in-built biases because of limited diversity in training datasets. The importance of validation was highlighted to ensure that tools provide reliable results for specific populations, such as those from ethnic minority backgrounds, and the need for independent evaluation of products.

²³ https://health.ec.europa.eu/ehealth-digital-health-and-care/european-health-data-space_en

²⁴ <https://www.ohdsi.org/data-standardization/>

²⁵ <https://www.england.nhs.uk/genomics/nhs-genomic-med-service/>

²⁶ <https://genomicainetwork.nhs.uk>

Priority areas

2.1. Data standards and interoperability:

Development of common data standards and interoperable IT systems aligned with global standards.

2.2. Practical barriers and enablers:

Exploring factors likely to impede or accelerate introduction and use of AI and genomic tools and related research in health systems.

2.3. Education and training: Assessing potential implications for health worker professional development, training and education.

2.4. Consumer protection:

Strengthening regulation of DTC testing.

Commercial development: Participants noted potential tensions in the role of **commercial developers** in the development of AI products for use in a public healthcare setting. Potential issues include the opaqueness of proprietary systems and the need to deliver profit for the developer, which can generate pressures to implement products without clear demonstration of clinical utility.

A sepsis-detection tool was used as an example. This has been introduced into US emergency departments to alert clinicians to signs of sepsis, and hospitals were penalised financially if they did not use it. However, an evaluation at sites using the tool found it offered few, if any, benefits. It alerted clinicians in only half of sepsis cases and, on average, did not lead to earlier detection than traditional methods.²⁷

Regulatory authorities in the UK, such as the Medicines and Healthcare Products Regulatory Agency (MHRA) have recognised the challenges posed by AI-enabled medical tools,²⁸ and the need to balance regulation with a policy imperative to promote innovation. The MHRA is working with other bodies, including the National Institute for Health and Care Excellence (NICE), on an integrated digital regulations service.²⁹

Other risks highlighted included the potential for high costs, as well as the risk of **technology 'lock-ins'** if proprietary solutions are adopted. It may be challenging to determine value for money and to compare the cost-effectiveness of different tools.

Another challenge identified was the exclusion of the academic sector from the development of foundational models, given the huge development costs. More streamlined systems, such as DeepSeek, suggest that this challenge might be surmountable.

Direct-to-consumer (DTC) tests: Participants also highlighted concerns regarding DTC marketing of genetic or genomic tests, bypassing health systems. The genetic information provided to consumers is often of more limited value than consumers – and, indeed, health professionals – expect.³⁰

Regulation of DTC companies operating internationally presents challenges, and concern was expressed about what happens to personal data if companies fail. For example, one well-known company, 23andMe, has recently experienced financial difficulties.³¹

²⁷ Ostermayer DG et al. (2024). *External validation of the Epic sepsis predictive model in 2 county emergency departments*. JAMIA Open 7, ooae133.

²⁸ <https://www.gov.uk/government/publications/impact-of-ai-on-the-regulation-of-medical-products/impact-of-ai-on-the-regulation-of-medical-products>

²⁹ <https://www.nice.org.uk/about/what-we-do/digital-health/multi-agency-advisory-service-for-ai-and-data-driven-technologies>

³⁰ Horton R et al. (2019). *Direct-to-consumer genetic testing*. The BMJ 367, l5688.

³¹ <https://www.bbc.co.uk/news/articles/c4gm08nlxr3o>



3. Ethical, social and legal opportunities and challenges

Individually, AI and genomics raise a range of ethical, social and legal issues. The meeting focused on issues raised by the combination of the two.

As discussed by Dr Peter Mills (PHG Foundation), a 2020 report from the **PHG Foundation** highlighted key issues relating to the application of AI in genomics,³² including concerns that need to be addressed before public acceptance and effective implementation of AI and genomics into routine healthcare can be achieved. These concerns included the risk of perpetuating or exacerbating bias, the lack of transparency inherent in black-box solutions, the need for robust regulatory, legal and governance frameworks, and the importance of building trusted and trustworthy systems.

As summarised by Harry Farmer (Ada Lovelace Institute), the **Nuffield Council of Bioethics**, in partnership with the **Ada Lovelace Institute**, has also examined AI-powered genomic health prediction. Its remit was more focused, concentrating on the derivation and use of **polygenic risk scores (PRSs)**, and their promise to advance personalised preventive approaches to health.³³ Although strategy documents such as 'Genome UK: The future of healthcare' express such ambitions, the value of PRSs in personalised prevention may be more limited than sometimes suggested. Combining PRSs with conventional risk scoring systems typically leads to only modest improvements in accuracy of disease risk prediction, when compared to the use of conventional risk scores alone.³⁴ Given this, it is unclear if the cost of their use, both in terms of money and data-sharing requirements, can be justified by resulting improvements to population health. A focus on genetic factors might also shift attention away from social determinants of health, which are often far more influential, and place the responsibility for disease prevention primarily on individuals.

The Nuffield Council report also highlighted the risks of **genetic discrimination** and lack of clarity on issues such as **consent** and **data governance**. Lack of genetic diversity in the analyses used to identify risk genes also raises questions about the applicability of polygenic risk scores to ethnic minority populations.³⁵

Ireland's AI and genomics strategy, first published in 2022, has a strong emphasis on public trust and ethical considerations. More general AI strategies have also embraced the need for strong ethical frameworks,³⁶ although ethical considerations received less attention in more recent iterations.³⁷

An important driver is the need to secure **public trust** in AI applications. As discussed by Dr Heike Felzmann (University of Galway), trustworthiness can be promoted through an '**ethics by design**' approach. Key elements of this approach include ensuring that core ethical principles are incorporated from the beginning of development, involving key stakeholders throughout, ensuring practical design decisions match ethical principles, and by taking into account the contexts in which applications will be used. At the EU level, attempts have been made to establish frameworks and

³² <https://www.phgfoundation.org/publications/reports/artificial-intelligence-for-genomic-medicine/>

³³ <https://www.nuffieldbioethics.org/news-blog/new-report-says-the-nhs-should-not-widely-roll-out-ai-powered-genomic-health-prediction-technology-yet/>

³⁴ Sud A et al. (2023). *Realistic expectations are key to realising the benefits of polygenic scores*. The BMJ **380**, e073149.

³⁵ <https://www.nuffieldbioethics.org/news-blog/new-report-says-the-nhs-should-not-widely-roll-out-ai-powered-genomic-health-prediction-technology-yet/>

³⁶ <https://enterprise.gov.ie/en/publications/national-ai-strategy.html>

³⁷ <https://enterprise.gov.ie/en/publications/national-ai-strategy-refresh-2024.html>

criteria to underpin ethical AI development.³⁸ The Government of Ireland has also published guidelines to ensure the ethical use of AI systems in the public domain.³⁹

Professor Anneke Lucassen (University of Oxford) pointed out that while generative AI has already shown tremendous potential (for example, in medical image analysis) the powers of predictive AI are much more speculative and often overstated.⁴⁰ Furthermore, genetic determinism may give an illusion of certainty to genetic findings that are usually more probabilistic. She reiterated the point that using genomics to confirm a diagnosis has been a major success story in rare diseases, but that inverting this model, and expecting genotypic variants to predict a future phenotype, is much more difficult. This misunderstanding has already led to misleading and potentially harmful applications, such as the use of PRS to select the supposedly healthiest embryos for implantation.⁴¹

PRSs are based on genetic associations across a population as a whole. Using such population-level data to make individual predictions introduces considerable uncertainty. For example, PRS for coronary heart disease, which performed similarly at the population level, generated highly variable individual-level estimates of risk, with one in five patients having at least one score both in the highest 5% risk and the lowest 5% risk. This emphasises the need to find new strategies to communicate this uncertainty to patients and clinicians.⁴²

Priority areas

3.1. Public and patient engagement:

Raising awareness of potential AI and genomic medical applications and ensuring patients and the public are involved at all stages of prioritisation, product development, service design and creation of regulatory/governance frameworks.

3.2. Trustworthiness: Identifying the key elements that underpin public trust in AI and genomic applications and building legislative and regulatory frameworks and governance mechanisms based on these principles.

3.3. Addressing inequities by design:

Promoting product development that considers a diversity of target populations from the outset.

3.4. Environmental and social impacts:

Encouraging R&D that minimises the environmental costs of AI development and exploring possible mechanisms, such as procurement policy, to shape product development and production practices.

Even when genes are known to make a major contribution to disease risk, such as some cancer-causing genetic variants, DTC test results may not be all they appear to be. For example, most DTC tests test for a minority of all known *BRCA* variants, potentially providing false reassurance, and high rates of false positives have been reported in cases of DTC offering secondary analyses, leading to medical mismanagement.²⁸

Advances in AI and genomics also have some other underrecognised downsides. These include **environmental impacts**, due to the high energy costs associated with AI development and storage costs of genomic data, and the **social harms** caused by the need for cheap labour to train models.⁴³

³⁸ <https://digital-strategy.ec.europa.eu/en/library/ethics-guidelines-trustworthy-ai>

³⁹ <https://www.gov.ie/en/publication/2127d-interim-guidelines-for-use-of-ai/>

⁴⁰ Narayanan A & Kapoor S (2024). *AI snake oil: what artificial intelligence can do, what it can't and how to tell the difference*. Princeton University Press, Princeton.

⁴¹ Furrer RA et al. (2024). *Public attitudes, interests, and concerns regarding polygenic embryo screening*. JAMA Network Open 7, e2410832.

⁴² Abramowitz SA et al. (2025). *Evaluating performance and agreement of coronary heart disease polygenic risk scores*. JAMA 333, 60–70.

⁴³ Samuel G et al. (2024). *Why digital innovation may not reduce healthcare's environmental footprint*. The BMJ 385, e078303.

More philosophically, genomic approaches to medicine are typically framed within the context of personalised approaches to medicine. Although this may benefit individuals, it also has the potential to undermine key aspects of **social solidarity** (such as the risk-pooling that underpins insurance schemes and public funding of health systems). A focus on 'me medicine' rather than 'we medicine' runs the risk of worsening health inequities.⁴⁴

Challenges

Participants emphasised that potential harms needed to be rigorously identified and mitigated. These include the potential for discrimination, possible implications for insurance, biases in tools/applications that disadvantage certain groups, and misleading application of genetic information. These issues can be addressed in a range of ways, for example through formal regulation and legislation, and systems of clinical governance, but also by shaping the culture in which AI and genomics are practised.

Potential concerns, for example around data privacy, need to be balanced with ensuring the availability of data for research so that health benefits – and harms – can be adequately assessed.

Participants also noted some important opportunities, particularly around **public and patient engagement and involvement**. For example, Ireland has gained considerable experience in the organisation of **citizens' juries** to develop recommendations around contentious policy areas.

As discussed by Dr Derick Mitchell (IPPOSI), the **Irish Platform for Patient Organisations, Science and Industry (IPPOSI)** has used citizens' juries to explore issues such as access to health information and use of genomics. It recently organised a deliberation focusing on AI in healthcare. The process aims to build consensus around the values seen as important to the jury, and future priorities. Its conclusions echoed many of the issues discussed at the workshop. A juror-led report summarising the conclusions of the AI in healthcare project was published in February 2025.⁴⁵

Workshop participants suggested that deliberative approaches such as citizens' juries, although more labour-intensive, had advantages over approaches such as cross-sectional opinion surveys. Issues such as genomics and AI raise complex and challenging issues and may require difficult trade-offs. Different groups have different interests. Deliberative consultations provide opportunities for these issues to be aired and discussed, leading to more informed and nuanced judgements.

⁴⁴ Dickenson D (2013). *Me medicine vs. we medicine: reclaiming biotechnology for the common good*. Columbia University Press, New York.

⁴⁵ IPPOSI (2025). Citizens' jury: artificial intelligence in healthcare in Ireland. <https://ipposi.ie/wp-content/uploads/2025/02/Citizens-Jury-Verdict-Report-21.02.25.pdf>

Conclusions

Genomics and AI are both fast-moving areas, and each has the potential to significantly advance healthcare. A fusion of the two offers many opportunities but also presents major challenges – some already well known and some new.

The workshop heard about many possible applications of AI and genomics. These applications are potentially transformative, but their potential harms also require evaluation before they are implemented. The NHS is a complex, highly devolved health system, with a diversity of health platforms and multiple stakeholder groups with different interests. Introducing new AI and genomic tools, and ensuring research can be integrated with care, is a formidable challenge.

The explosion of interest in AI is generating an ever-growing set of applications. Ensuring these are effectively evaluated is a growing challenge. Introduction of new systems needs to consider not just the potential benefits and costs but also the potential for harm, such as genetic discrimination or widening of inequities.

Participants emphasised the need to engage with patients and the public, particularly through deliberative approaches that provide time to explore complex and sensitive issues in depth. Such consultations can help to create legislative and regulatory environments that safeguard public interests but do not stifle research or its application to benefit health. It was acknowledged that this would not be easy in a complex, fast-moving and international field.

It was also suggested that efforts should be focused where positive impacts may be greatest. Many claims are being made about the power of both genomics and AI. Participants stressed the need to take a considered and evidence-based approach to determine whether the reality can match the hype, to evaluate clinical utility and to prioritise accordingly.

In the UK, health is a devolved function. Yet the UK nations and Ireland, face similar challenges and can benefit from collaboration in research and in learning how to implement new applications within health systems. Such collaborations will also need to embrace wider international initiatives aiming to harmonise and standardise health data systems to facilitate international collaborative research.

Annex 1: Agenda

Wednesday 27 November

Time	Item
14:30 - 14:45	Welcome and overview of the workshop Co-chairs: Professor Helen Firth FMedSci , Consultant Clinical Geneticist, Hon. Professor of Clinical Genomics, University of Cambridge and Dr Olga Piskareva , Senior Lecturer in Biology, Department of Anatomy and Regenerative Medicine and Foundation Year Cycle Director, RCSI University of Medicine and Health Sciences
14:45 – 16:30	Session 1: Setting the context for the workshop <i>Session chair: Professor Anneke Lucassen</i> This session will provide an overview of current state of AI and genomics in Ireland and the UK. How is it currently being used and what are the ambitions for AI and genomics in each country? 1. Professor Ian Simpson, Professor of Biomedical Informatics & Director UKRI Artificial Intelligence CDT in Biomedical Innovation 2. PHG Foundation 3. Dr Daniel Murphy, Assistant Professor in Bioinformatics at the School of Biology and Environmental Science, University College Dublin Discussion and Q&A
16:30 – 17:00	Break
17:00 – 18:15	Session 2: What is currently possible <i>Session chair: Natalie Frankish, Genomics England</i> This session will provide examples of what is currently possible and how AI is being applied within genomics in the UK, Ireland and elsewhere. What have some of the successes been, what have we learned so far, where do the limitations and challenges lie? 1. Professor Walter Kolch MRIA, Director of Systems Biology Ireland (SBI), University College Dublin – ‘<i>Precision Oncology using Digital Models</i>’ (15 mins) 2. Professor Aedín Culhane, Director of the Limerick Digital Cancer Research Centre lead of the All-Island eHealth-Hub for Cancer – ‘<i>From isolated data silos to federated learning: the potential of collaborative cancer research</i>’ (15 mins) 3. Professor Eamonn Sheridan, Consultant in Clinical Genetics, Leeds Teaching Hospitals and University of Leeds – ‘<i>Current implementation in NHS & Genomics England and future plans</i>’ (15 mins) Discussion and Q&A
18:15 – 18:30	Close Day 1 Co-chairs: <i>Professor Helen Firth FMedSci and Dr Olga Piskareva</i>

19:00 - 20:30	Networking Dinner
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Thursday 28 November

Time	Item
09:00 – 09:10	Welcome to Day 2 <i>Workshop co-chairs: Professor Helen Firth FMedSci and Dr Olga Piskareva</i>
09:10 – 10:30	Session 3: How we can leverage AI's potential <i>Session chairs: Dr Catriona Inverarity and James Dickinson</i> This session will focus on the opportunities for safe and effective use of AI to genomic medicine. How can we use AI in the future, how can we use it as a safe and effective tool? 1. Dr Michael Yates, Senior Clinical Research Fellow, University of Edinburgh – 'AI-driven phenotype mining with application to diagnostics' 2. Professor Damien Woods, Professor at the Hamilton Institute, Maynooth University Harry Farmer, Senior Researcher, Ada Lovelace Institute
10:30 – 11:00	Break
11:00 – 12:15	Session 4: Ethics, regulation and barriers to implementing AI <i>Session chair: Professor Thérèse Murphy</i> With this session, we aim to delve into the questions related to ethics, regulation and barriers to implementing AI. With the presentations, speakers will map out existing challenges from researchers', patients' and systems' perspectives. • Dr Oliver Feeney, Institute of Ethics & History of Medicine, University of Tübingen & Centre of Bioethical Research & Analysis, Discipline of Philosophy, University of Galway – 'AI in genomics: some ethical and regulatory challenges' (15 mins) • Dr Derick Mitchell, Chief Executive, Irish Platform for Patient Organisations, Science and Industry (IPPOSI) – 'Involving patients and the public in decision make around AI and Healthcare' (15 mins) • Professor Dave Archard, Emeritus Professor of Philosophy, Queen's University Belfast (former Chair of Nuffield Council) – 'Ethical Challenges of AI implementation' to address inequalities? (15 mins)
12:15 – 13:15	Lunch
13:15 – 14:15	Session 5: Breakout groups Participants will be divided into three smaller groups focusing on: 1. Ethical and technical principles for the operation of AI in genomic testing 2. Policy - How to harmonise all the key points from all the reports from genomics & testing 3. Clinical implementation – how to take this forward

	Questions to be discussed in each group: 1. Are there research areas that need to be developed? 2. What are the gaps in this area? 3. What are the risks and the opportunities? 4. What are the next steps? What actions need to be taken and by whom, regionally and nationally, both in the short (1–2 years) and long (2–10 years) term? Session outcome By the end of this session each group will be ready to feed back during the plenary session summarising their discussion and highlighting key points raised. The group will have nominated a presenter, with support from the other members.
14:15 – 14:45	Session 5: Feedback Each group to feedback – 10 mins per group
14:45 – 15:15	Break
15:15 - 16:15	Session 6: Plenary discussion <i>Workshop co-chairs: Professor Helen Firth FMedSci and Dr Olga Piskareva</i> During this session, participants should discuss and reach consensus on what actions need to be taken and by whom, regionally and nationally both in the short (1–2 years) and long (2–10 years) term. Participants should highlight where opportunities lie to work together.
16:15 - 16:30	Closing <i>Workshop co-chairs: Professor Helen Firth FMedSci and Dr Olga Piskareva</i>

Annex 2: Attendee list

Steering Committee

- **Dr Helen Firth FMedSci**, Consultant Clinical Geneticist, University of Cambridge
- **Dr Olga Piskareva**, Lecturer Anatomy and Regenerative Medicine, RCSI University of Medicine and Health Sciences
- **Professor Amy Jayne McKnight**, Professor, School of Medicine, Dentistry and Biomedical Sciences, Queen's University Belfast
- **Professor Anneke Lucassen**, Professor of Genomic Medicine, Director of the Centre for Personalised Medicine, University of Oxford
- **Professor Thérèse Murphy**, Professor, School of Law, Queen's University Belfast
- **Professor Desmond Tobin MRIA**, Full Professor of Dermatological Science, University College Dublin, School of Medicine
- **Professor Ian Simpson**, Professor of Biomedical Informatics & Director UKRI Artificial Intelligence CDT in Biomedical Innovation, University of Edinburgh
- **Dr Joe MacDonagh**, Lecturer, Technological University Dublin

Participants

- **Professor Aedín Culhane**, Professor of Biomedical Sciences (Cancer Genomics), University of Limerick
- **Dr Alexander Deng**, Clinical Lead for NHS Genomic AI Network, and Programme Lead for the NHS Fellowship in Clinical AI
- **Dr Alice Vajda**, Scientific Programme Manager, Research Ireland
- **Professor Andrew Green**, Professor of Medical Genetics, School of Medicine, University College Dublin
- **Professor Andrew Wilkie FMedSci**, Nuffield Professor of Pathology, Honorary Consultant in Clinical Genetics, University of Oxford
- **Dr Avril Kennan**, Chief Executive Officer, Health Research Charities Ireland
- **Professor Bert Rima MRIA**, Professor Honorary Professor, School of Medicine, Dentistry and Biomedical Sciences Centre for Infection and Immunity, Queen's University Belfast
- **Catriona Inverarity**, Senior Health Technology Assessor, King's Technology Evaluation Centre
- **Dr Chaitanya Erady**, PHG Foundation
- **Dr Claire Kilty**, PPI Council Member, Irish Association for Cancer Research
- **Professor Damien Woods**, Professor of Computer Science, Maynooth University
- **Dr Daniel Murphy**, Lecturer/Assistant Professor in Bioinformatics, University College Dublin
- **Professor Dave Archard**, Emeritus Professor of Philosophy, Queen's University Belfast
- **Professor David FitzPatrick FMedSci**, Joint Head of Disease Mechanism, MRC Human Genetics Unit
Institute of Genetics and Cancer, University of Edinburgh
- **Professor Denis Shields**, Full Professor of Clinical Bioinformatics, University College Dublin
- **Dr Derick Mitchell**, Chief Executive of the Irish Platform for Patient Organisations, Science and Industry
- **Dr Dervla Kelly**, Lecturer in Medical Education, University of Limerick, and Young Academy Ireland Member
- **Professor Eamonn Sheridan**, Consultant in Clinical Genetics, University of Leeds
- **Dr Elaine Kenny**, Adjunct Associate Professor, Trinity Translational Medicine Institute, Trinity College Dublin
- **Dr Emma Dorris**, PPI Ignite Network Programme Manager, University College Dublin
- **Harry Farmer**, Senior Researcher, Ada Lovelace Institute
- **Dr Heike Felzmann**, Associate Professor in Ethics, School of History and Philosophy, and the Data Science Institute, University of Galway
- **James Dickinson**, Director, Genomics Artificial Intelligence Network
- **Dr Janna Kenny**, Clinical Researcher, University College London

- **Professor Jenny Taylor**, Translational Genomics, University of Oxford
- **Professor Joe Marsh**, Personal Chair of Computational Protein Biology, Institute of Genetics and Cancer, MRC Human Genetics Unit, University of Edinburgh
- **Dr Julia Foreman**, DECIPHER Project Leader, European Molecular Biology Laboratory, European Bioinformatics Institute
- **Julie Taylor**, Science Technology, Innovation Officer, Foreign, Commonwealth & Development Office, British Embassy in Dublin
- **Dr Kevin Fee**, PhD researcher, Queen's University Belfast, and Young Academy Ireland Member
- **Dr Laura Brady**, Digital Health Innovation Lead, FutureNeuro Research Centre
- **Professor Mary Ward**, Professor of Nutrition and Dietetics, Ulster University
- **Dr Michael Yates**, Senior Clinical Research Fellow, University of Edinburgh
- **Natalie Frankish**, Head of Scotland and Public Affairs, Genetic Alliance UK
- **Professor Noel Lowndes MRIA**, Royal Irish Academy Science Secretary, and Chair, Biochemistry & Director, Genome Stability Laboratory, University of Galway
- **Dr Oliver Feeney**, Ethics of Genome Editing Research Unit, Institute of Ethics and History of Medicine, University of Tübingen
- **Dr Peter Mills**, Director, PHG Foundation
- **Dr Priyank Shukla**, Senior Lecturer in Stratified Medicine (Bioinformatics), Ulster University
- **Professor Richard Houlston FRS FMedSci**, Head of the Division of Genetics and Epidemiology, Institute of Cancer Research
- **Dr Ross Murphy**, Lecturer in Health Data Science, Ulster University
- **Professor Sally Lynch**, Clinical Professor School of Medicine, University College Dublin
- **Dr Sarah Hunt**, Variation Resources Coordinator, European Molecular Biology Laboratory, European Bioinformatics Institute
- **Shane Gordon**, PhD candidate, Ulster University
- **Dr Shane McKee**, Consultant in Genetic & Genomic Medicine, Belfast Health and Social Care Trust
- **Dr Susan Byrne**, Senior Lecturer, Royal College of Surgeons in Ireland
- **Tanya Bridgen**, Senior Policy Analyst (Biomedical Ethics), PHG Foundation
- **Dr Teresa Maguire**, Director of Research Strategy and Funding, Health Research Board
- **Professor Walter Kolch MRIA**, Director Systems Biology Ireland, University College Dublin

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- **Abigail Bloy**, Senior International Policy Manager, Academy of Medical Sciences
- **Fionnuala Parfrey**, Programme manager, Royal Irish Academy
- **Jessamyn Briers**, International Policy Officer, Academy of Medical Sciences
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