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AI-driven multiomics in health: our 2025 roundtable insights

We are in the big data era. Clinically relevant biomedical information is being generated at an unprecedented rate—approximately 40 exabytes (or one billion gigabytes) of genomic data are produced annually.

Generating comprehensive biological insights from these data often requires integration of different data modalities, as is the case with multiomics, to gain a more holistic understanding of disease mechanisms and identify novel biomarkers. Consequently, enthusiasm for the potential of AI-driven multiomics to improve health is growing. But what multiomics encompasses, how it intersects with artificial intelligence (AI), and what its real-world impact might be is subject to uncertainty within the health sector.

The PHG Foundation recently convened a multidisciplinary group of clinicians, researchers, academics, policy makers and industry professionals to address these questions. Here is what we found.

What is multiomics?

Multiomics does not have one clear definition. For the purposes of this roundtable we considered multiomics as the integration of different biological data layers, such as genomics, proteomics, transcriptomics or metabolomics, and newer ‘omics such as radiomics (based on imaging data) or pathomics (analysis of cell and tissue structures).

How could AI-driven multiomics add value to healthcare?

Extracting clinically relevant insights from multiomic data which often involves numerous interconnected data points across different biological layers, such as DNA, RNA, and protein, is a significant challenge. Traditional statistical methods often struggle with this complexity, but artificial intelligence (AI), particularly machine learning and deep learning approaches, could play a transformative role. AI can help identify new associations and discover patterns in the data invisible to the naked eye.

By leveraging advanced computational models, AI can also effectively reduce the 'noise' or random variation in the data and pinpoint clinically actionable signals. In healthcare, AI-driven multiomics could add value via:

Research to inform clinical care

Combining genetic data with other 'omic data, and using AI to understand the functional links between a genetic variant and biological activity, such as transcript or protein expression, could help better classify genetic variants and ascertain disease pathogenicity. This is particularly true for complex genetic conditions with multifactorial origins.

In addition, using AI to analyse multiomic data might help identify new patterns and associations in diseases, which could aid in developing new biomarkers.

Clinical pathway

AI-driven multiomics could have applications across multiple parts of the patient pathway:

Risk prediction

- Polygenic scores, when integrated with multiomics data, could provide more reliable risk predictions for developing diseases such as heart attack or stroke over the next 20 years.
- Enhancing existing screening tools, such as the Hba1c test, with proteomic biomarkers could help identify at-risk individuals and detect prediabetes subtypes that may otherwise be overlooked.

Disease diagnosis

- AI-driven multiomics offers promising applications for early detection, diagnosis, and clinical decision-making for conditions such as cancer, rare diseases and dementia. For example, AI solutions that analyse genomic data in consort with data from facial recognition software may enable earlier and more accurate diagnoses of rare diseases. Diagnostic yield could also be improved by 15-20% for some conditions through the addition of further layers of information to genomic data.
- In cancer care, AI-driven multiomics could offer deeper insights into tumour biology and accelerate clinical decision-making. For example, AI can help differentiate between cancerous and benign tumours in clinical results, and thereby help prioritise clinical cases.
- Multiomic approaches may also serve as confirmatory tests. In cases where high levels of the protein Prostate-specific antigen (PSA) are identified, which can be indicative of prostate cancer, additional confirmatory testing using genomic or transcriptomic approaches could lead to more accurate diagnosis.

Prognosis and treatment

- AI-driven multiomics could help stratify patients by disease severity and help develop a more tailored treatment strategy. For example, in sepsis, AI-driven multiomics could predict the severity of the condition, better identifying high-risk individuals, and helping clinicians decide on early interventions.
- In ovarian cancer, AI-driven multiomics could predict whether chemotherapy will be effective in a patient six weeks before surgery, avoiding unnecessary treatments and improving patient outcomes.
- AI-driven multiomics could also help to determine whether chemotherapy is appropriate for patients with stage 2 and 3 colorectal cancer, reducing the risk of unnecessary side effects.

Where are we now?

Currently, the application of AI-driven multiomics is in its infancy and is largely limited to research. Cancer research is where AI-driven multiomics is most mature, with pockets of innovation and investment in rare diseases, infectious diseases, and autoimmune conditions. However, efforts remain fragmented, lacking a coherent, joined-up strategy.

A clear, overarching vision for what AI-driven multiomics can achieve in healthcare has yet to emerge. However, some examples of promising developments are:

- The Southampton Inflammatory Bowel Disease (IBD) project, which uses a swallowed camera to collect real-time data from patients with IBD, demonstrated how AI-driven image processing combined with genomic data could become a standard part of everyday clinical practice.
- Nightingale Health has developed a metabolomics platform that is being explored for clinical applications, particularly in predicting health risks and monitoring disease progression.
- Multiomics is being used to phenotype research participants with infectious diseases, with AI analysis providing insights into the patient's condition, disease severity and to identify biomarkers. Efforts are underway to bring this approach to clinical settings.
- Researchers in collaboration with the MHRA are developing guidance for the regulation of mRNA genomic cancer vaccines, where AI analyses genomic and transcriptomic data to personalise vaccine design.

While there is some way to go in terms of research studies and evidence gathering, some existing opportunities and enablers to help make this a reality are that:

- **Compute is not a limitation:** Several NHS and academic sectors have access to cloud servers and high-performance computing facilities to analyse multiomic data.
- **Good quality data are available:** existing genomics and imaging datasets in the NHS are valuable resources that may be used to train relevant AI-models. Additionally, retrospective NHS datasets (currently underutilised) offer potential value.
- **Collaborations:** there is appetite that could be harnessed among scientists, clinicians, and industry for collaboration and scale-up.

What needs to happen?

Multiomics has the potential to improve clinical pathways and health outcomes, but it is still an emerging technology that needs support to achieve this potential. The potential for multiomics to shift the focus from disease diagnosis to prevention also remains unexplored. In addition, the following challenges need to be addressed:

Technical and data challenges

- **Inconsistency across 'omic platforms:** Different 'omic platforms may generate data that are not directly comparable, creating a significant barrier for integrating multiomics data across different research settings or hospitals.
- **Data quality:** Clinical data are often noisy and imperfect, and training AI models on poor-quality or incomplete data could lead to inaccurate results. Therefore, data selection needs to be careful and may limit what can be achieved.
- **Data fragmentation:** The lack of standardised data collection and sharing methods makes it difficult to compile large, comprehensive datasets for analysis.

Implementation challenges

Confidence in multiomics: Clinicians are not used to interpreting multiomics data, which can create a gap between data generation and real-world clinical application. There needs to be

greater awareness within the clinical community about the tangible advantages AI-driven multiomics provides, such as improved diagnostic accuracy, better treatment stratification, and personalised medicine.

Data infrastructure: In the UK, the healthcare systems are not adequately prepared to handle the volume and complexity of data generated by multiomics technologies. This includes the need for better IT infrastructure to store, process, and analyse large datasets effectively.

Interoperability: Healthcare IT infrastructure is often fragmented. A technology or model that works well in one hospital may not easily transfer to another, which limits its scalability. There is also concern about how AI models trained on one platform (e.g. one provider of proteomic service) might perform when transferred to another.

Ethical and legal considerations

- **Diversity and equity:** Certain demographic groups (e.g. ethnic minorities, low-income populations) are underrepresented in 'omics research. This disparity could lead to biased results and inequitable healthcare outcomes. Additionally, disparities in uptake across hospitals, where some institutions adopt AI-driven multiomics faster than others, could further widen healthcare gaps.
- **Informed consent:** Questions arise about whether patients are prepared to receive unexpected or potentially distressing information, such as being informed about a vascular problem from an ophthalmologist with the use of AI-driven multiomics.
- **Data privacy:** Genetic data are difficult to anonymise as they are highly specific to individuals, and even small variations can serve as a fingerprint that can be linked back to a person. This leads to concerns that the integration of multiomic data with wider patient data, particularly where this is driven by AI, could lead to patient re-identification and undermine patient privacy.

Regulatory considerations

- **AI as a medical device:** Regulatory bodies, such as the MHRA, face challenges in evaluating AI models as medical devices. These evaluations might be further complicated by the addition of multiomic data.
- **Uncertainty in validating multiomic models:** Unlike genomics, where there is an established "ground truth", many other 'omics are relative and do not have absolute units, making them difficult to benchmark and validate. Regulatory agencies need to develop the expertise to deal with multiomics models.
- **Regulatory standards:** There is a lack of clear, universally accepted standards for the development and validation of AI-driven multiomics models. It is also unclear who would validate external software. If that is to be hospitals, then upskilling of existing staff may be needed. There is also a need for "sandboxes" to test and refine new AI-driven multiomic technologies before they are fully integrated into clinical settings.

Questions people are asking about AI-driven multiomics for health

While the UK has significant strengths in genomics, data infrastructure, and science, the health system is not yet ready to support the potential use of AI-driven multiomic tools at

scale to transform prevention and care. Efforts remain fragmented and largely research-focused, with limited application in real-world health settings. Unanswered questions remain, including:

- Are we addressing genuine clinical problems with multiomics and is AI required to realise the potential?
- Should multiomics be genomics-led or will it require an entirely new operational and conceptual framework?
- How can we optimise lessons learned from the implementation of genomics into clinical practice?
- There is a tension between multiomic tests being developed and validated as generic population-level tests versus their personalisation for individuals. How do we deal with this discrepancy?

Considerations that could help answer these questions

- Industry-wide standards for data formats and integration methods of multiomic data.
- More real-world clinical trials and studies to validate the effectiveness and safety of AI-driven multiomics approaches.
- Stronger collaborations between AI developers, clinicians, and regulatory bodies to create a unified strategy for implementation.
- Development of a clinical utility framework to define and measure impact of AI-driven multiomic solutions.
- Is there a need for a system-level vision for the role of AI-driven multiomics in healthcare and what might this look like?

Practical requirements that could help make AI-driven multiomics a reality include:

- Development of a comprehensive policy direction to navigate the complex AI-multiomics landscape.
- Determination of the different stakeholders that need to be involved in this AI-driven multiomic discussion.
- Assessment of the need for and who might offer clear leadership for the multiomics agenda, not just in terms of research and innovation but also in eventual clinical adoption.
- Ascertainment of when and how AI-driven multiomics knowledge could be embedded into clinical education pathways.

If this topic interests you, please do get in touch, we are always keen to discuss opportunities for collaborations. Email: intelligence@phgfoundation.org

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We would like to thank the participants in the roundtable for contributing their valuable time and insights.