POLICY BRIEFING

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Putting AI to work in genomic medicine

Artificial intelligence (AI) offers great opportunities to advance genomic medicine, but enthusiasm for these technologies must be matched with collective commitment and investment to tackle serious issues posed by their use in healthcare. Decision makers will want to realise the potential of AI for genomic medicine by speeding up delivery whilst minimising harms. We outline seven practical policy actions to help achieve this goal.

Meeting the genomic challenge

The strength of AI is its ability to make sense of large complex datasets far more quickly and effectively than human intelligence, making it a perfect partner for genomic analysis. The AI techniques machine learning and deep learning allow the streamlining of analytical problems in genomic medicine. Their use is increasing, driven by:

- Advances in high-performance computing
- Resurgence of deep learning
- Growing availability of resources for building machine learning models
- Growth of large genomic and biomedical datasets

AI can support clinical research and practice

- **Directly facilitating clinical genome analysis,** for example in facial analysis to help diagnose birth defects or in the identification of genetic variants, including those that are difficult to accurately detect
- **Improving understanding of genomic variation in health and disease and accelerating discovery in genomic medicine.** Current research includes studies into how cancers evolve, determining which genetic changes could be drivers for tumour growth, improving the efficiency and accuracy of CRISPR, and methods to integrate and analyse genomic data together with other types of data

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Limiting factors

Multiple related issues are hampering the optimal application of AI to improve outcomes in genomic medicine, including:

- **Skills and infrastructure** - no single sector has a monopoly on all the skills, data, resources and infrastructure needed
- **Bias** - this can arise in AI tools due to data limitations, such as under-representation of populations within datasets, availability and preparation of data, as well as how questions are framed; these risk worsening health inequalities
- **Regulation, governance and ethics** - clarity is needed on issues such as when AI tools constitute medical devices, how data protection legislation may apply to their use, where legal liability for harms may lie, and ethical use of data
- **Validation** - in genomic data analysis, where typically a range of dynamic and adaptive algorithms are applied, clarity is needed on thresholds for evidence and validation

Priorities for policy

The immediate priorities to efficiently reap the benefits of AI are to:

- Establish the right conditions for facilitating AI in genomic medicine which includes better digital infrastructure, data acquisition and management, access to specific technical skills, and cross-disciplinary collaborations
- Prioritise the development of AI tools that address well-defined problems that address actual clinical need
- Mitigate AI bias by promoting a diverse workforce and research environment and monitoring and addressing bias within training datasets
- Facilitate research efforts to apply machine learning to well-curated, high-quality genomics and biomedical datasets
- Support community efforts to benchmark, review, and determine the most effective use and integration of emerging new algorithms for clinical genome analysis
- Establish sector-specific strategies to address the complex challenges and limitations of AI in genomic medicine and research
- Establish the clinical governance arrangements for the use of specific AI applications in the practice of clinical genomics

Artificial intelligence for genomic medicine is available at phgfoundation.org

Contact: intelligence@phgfoundation.org

PHG Foundation is a health policy think tank with a special focus on how genomics and other emerging health technologies can provide more effective, personalised healthcare

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