Supplementary response to the House of Commons' Science and Technology Committee's inquiry into genomics in the NHS

October 2017

Summary

- Advances in genomic technologies and an improved understanding of the molecular basis of disease offer significant opportunities for research and healthcare, ultimately translating into patient benefit.
- Access to high-quality patient data, including genomic data, is essential for research. It is important that genomic data is not considered in isolation and is linked with other data such as phenotypic information to create a comprehensive patient dataset. Therefore a robust data infrastructure is needed that facilitates access for researchers alongside appropriate safeguards to protect patient privacy.
- Engagement with the public, patients, healthcare professionals and other key stakeholders on sharing genomic and wider health data is important to foster an understanding of the value of these data, and support informed choices about sharing data.
- Capacity and capability needs to be built amongst the NHS workforce. This includes raising awareness across healthcare professionals and training for all those involved along the patient pathway. Genomics is a multidisciplinary area and consideration needs to be given to capacity in all relevant fields – from geneticists and bioinformaticians to data scientists, genetic nurses and clinicians.
- Implementing a national approach to the commissioning of genomic services will ensure equity of access, and standardisation and consistency of testing approaches across the country.
- Evaluation of genomic technologies will help to drive improvements and build an understanding of 'value' including impact on patient outcomes and economic measures such as cost-effectiveness.

Introduction

1. The Academy of Medical Sciences promotes advances in medical science, and supports efforts to see these advances translated into healthcare benefits for society. Our elected Fellowship includes some of the UK's foremost experts in medical science, drawn from a broad range of research areas.
2. We welcome this opportunity to submit further input towards the House of Commons' Science and Technology Committee's inquiry into genomics and gene-editing in the NHS, following the Chief Medical Officer's (CMO) latest annual report on 'Generation Genome'.[1] Our initial submission to the Committee's inquiry focussed on gene-editing.[2] Therefore this supplementary response covers genomics, which is also the main focus of the CMO's report.
3. The Academy has been engaged in work exploring the advances in genomic research including the integral role of genomics in delivering stratified medicine in the NHS, as well as broader

[1] Annual Report of the Chief Medical Officer 2016 (2016). *Generation Genome.* www.gov.uk/government/uploads/system/uploads/attachment_data/file/631043/CMO_annual_report_generation_genome.pdf

[2] Academy of Medical Sciences (2017). *Submission to the House of Commons' Science and Technology Committee's inquiry into genomics and genome editing*. https://acmedsci.ac.uk/file-download/83063056

topics that form the basis of this discussion such as data sharing and governance and the Life Sciences Industrial Strategy.[3,4,5,6]

4. The dramatic increase in our ability to generate, analyse and interpret genomic data has greatly improved our understanding of human disease and provides new opportunities for patients to benefit from more targeted and effective treatments, with the potential for wider efficiency savings for the healthcare system. This is supported by a growing infrastructure and expanding national genomics data resources such as the 100,000 Genomes Project, which can be built upon to establish a UK-wide routine genomics service.
5. There are areas addressed within the CMO's report that we wish to comment on: the role of NHS data – including genomics data – in research and healthcare; supporting capability and capacity for genomics services including genetic testing; commissioning of services including standardisation of testing facilities; and evaluation research.

NHS data and research

6. The data held within the NHS are a unique strength for the UK. Ensuring nationwide collection of high-quality, standardised patient data, including genomic information, is key for research – both in terms of use in research and for patient recruitment to studies and trials. This research can in turn deliver better patient outcomes and wider system benefits such as improvements in system delivery and enabling identification of new drug targets and development of novel therapies. Understanding the role of genomics in disease is an important component of stratified medicine, which increasingly enables better targeting of medicines to the right patient. In addition, it can support prevention and early diagnosis by understanding susceptibility to disease and prediction of risk to facilitate prevention strategies.
7. Genomic data should not be considered in isolation, and integration of genomics with other data such as phenotypic information creates a richer dataset which is key for a stratified approach. Therefore it is important to ensure that it can be linked with other data types which are also valuable for research and healthcare.[7] Integrating different data types helps to establish a more comprehensive dataset, overcoming some of the challenges of using genomics in isolation. Although there are significant opportunities afforded by recent advances in genomics, there are limitations of using genomics in the treatment of some diseases, such as those where genetics is one of many complex interacting factors. For example, there are multiple factors that may cause or exacerbate psychiatric conditions including environment and social aspects, heritable and epigenetic factors, and biological pathways.
8. Access to NHS data by researchers is essential and the NHS should act as a partner for research. Robust data collection, storage and sharing infrastructure must be established within the NHS (including for genomic data), with appropriate safeguards to protect patient data whilst facilitating access for research and interoperability across different platforms.
9. Engagement of patients, the public and healthcare professionals in discussions around data sharing is needed to build an understanding of, and support for, the value of these data for

[3] Academy of Medical Sciences (2015). *Stratified, personalised or P4 medicine: a new direction for placing the patient at the centre of healthcare and health education*. http://www.acmedsci.ac.uk/file-download/38266-56e6d483e1d21.pdf
[4] Academy of Medical Sciences (2016). *Submission to the British Academy and Royal Society's call for evidence on data governance.* http://www.acmedsci.ac.uk/file-download/41614-583586af15320.pdf
[5] Academy of Medical Sciences (2016). *Exemplar clinical pathways for a stratified approach to cardiovascular disease.* https://acmedsci.ac.uk/viewFile/57cfd5170e1de.pdf
[6] Academy of Medical Sciences (2015). *Exemplar clinical pathways for a stratified approach to diabetes.* https://acmedsci.ac.uk/viewFile/57cfd3c90098c.pdf
[7] Academy of Medical Sciences (2013). *Realising the potential of stratified medicine.* https://acmedsci.ac.uk/file-download/34525-51e915f9f09fb.pdf

research, and enabling informed choices to be made around data sharing. This engagement should draw upon experience from initiatives such as Understanding Patient Data and projects including the 100K Genomes Project, UK Biobank and the Million Women Study. The Academy plans to undertake public dialogue about new sources of health data, which will help to inform these discussions.

Capacity and capability for genomics in the NHS

10. The NHS will need people with the necessary skills to enable delivery of routine genomic services across the UK. This requires raising awareness across all healthcare professionals and building clinical capabilities through embedding genomics in clinical education for both specialist and non-specialist training – described by the CMO as a *'genomics literate'* workforce. Integration of genomics into the NHS will require a multidisciplinary approach across all staff involved in the patient pathway from geneticists and bioinformaticians to nurses and clinical specialists. It will also increasingly span traditional clinical boundaries to involve multiple disease areas and specialities.
11. Alongside training, additional new capacity and resources will be needed for implementation of genomics services such as trained genetic nurses or counsellors.[8] As a multidisciplinary area this will need to consider all stages of service provision from data scientists, geneticists and diagnosticians to those in clinical practice.
12. Decision-support tools can help healthcare professionals to direct patients onto the right care pathway, and understand and interpret the results of genomic testing alongside experts such as geneticists. For example, to aid clinicians in the diagnosis of a specific type of monogenic diabetes (maturity onset diabetes of the young), a probability calculator has been developed which determines overall risk based on a variety of factors.[9] In addition, the genetic test for this condition is accompanied by a lay report, which explains the results for a patient and clinician. However, genomic results may not be clear-cut and so a framework for shared decision-making between patients and clinicians is needed, particularly when considering areas such as predicting risk of developing a condition.[10]

Commissioning and provision of genomic services

13. A national approach to the commissioning of genomic services and associated patient pathways will help to ensure equity of access across the country and clarity around where responsibilities lie for funding for tests, and more widely as to where this fits within specialised and local commissioning.[11] This was emphasised at the recent series of roundtables held by the Academy and NHS England on adoption of stratified medicine in the NHS.[12,13]
14. We strongly support the CMO's emphasis on establishing national standards for laboratory services, to ensure that provision of genomic services is consistent and of a high standard across the country, overcoming variability across different laboratories. Standardisation of tests, including thresholds and cut-offs, will also facilitate a more systematic approach to

[8] Academy of Medical Sciences (2016). *Exemplar clinical pathways for a stratified approach to cardiovascular disease.* https://acmedsci.ac.uk/viewFile/57cfd5170e1de.pdf

[9] MODY probability calculator: www.diabetesgenes.org/content/mody-probability-calculator

[10] The Academy's recent report explores the potential of joint decision-making: Academy of Medical Sciences (2017). *Enhancing the use of scientific evidence to judge the potential benefits and harms of medicines.* https://acmedsci.ac.uk/file-download/44970096

[11] Academy of Medical Sciences (2016). *Exemplar clinical pathways for a stratified approach to cardiovascular disease.* https://acmedsci.ac.uk/viewFile/57cfd5170e1de.pdf

[12] *Ibid.*

[13] Academy of Medical Sciences (2015). *Exemplar clinical pathways for a stratified approach to diabetes.* https://acmedsci.ac.uk/viewFile/57cfd3c90098c.pdf

patient care.[14] It is important that scalability of tests is considered in terms of uptake across the country, as well as whether they are fit-for-purpose for the setting in which they will be delivered. In addition, interpretation of genetic results can be subjective and opinions may differ across individuals, and so knowledge sharing processes and the establishment of a centralised database should help to support consistency in these decisions.

System evaluation and implementation research

15. Incorporating systematic evaluation processes into the provision of genomics services across the NHS will drive improvements and an understanding of the value of such tests and services including efficacy, cost-effectiveness, patient outcomes and other benefits. Therefore it is important to establish a continuous, robust evaluation process for new genomic technologies once they are introduced in the NHS.

This response was prepared by Liberty Dixon, FORUM Policy Manager, and was informed through the Academy's previous work in this area. For further information, please contact Liberty Dixon (liberty.dixon@acmedsci.ac.uk; +44(0)20 3141 3222).

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[14] Academy of Medical Sciences (2015). *Reproducibility and reliability of biomedical research: improving research practice.* https://acmedsci.ac.uk/file-download/38189-56531416e2949.pdf