The Academy of Medical Sciences

Response to the Science and Technology Committee's inquiry into commercial genomics

April 2019

Summary

- The UK has unique strengths in health data and data sharing which could be supported through the development of data analytics capabilities and investment in genomic healthcare initiatives, and their infrastructure.
- Specifically trained genetics nurses and counsellors should be available to support those receiving, or considering asking for, genomic tests. This should be alongside the development of decision-support tools, and a raised awareness of the benefits and limitations of genomic information across all healthcare professionals.
- There are potential benefits to patients from commercial testing, provided that they are supplied with adequate support to be able to act on the information appropriately, and could be particularly transformative for early diagnosis.
- It is important that genomic data, regardless of its origin, can be linked with other data types that are also valuable for research and healthcare; the sharing and linkage of genomic data arising from both commercialised and non-commercialised sources should be considered.

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. The Academy is monitoring the rapid development and application of emerging technologies, including those based on genomics, and welcomes the opportunity to contribute to this inquiry.

3. Our response is based on our previous policy work in this area. As such, it places a predominant focus on the benefits of patient data for research and clinical care, and emphasises a need to ensure proper regulation and governance systems are in place to adequately support data sharing and safeguarding.

Health, or other, benefits that consumers can derive from using commercially available genomic testing

4. Advances in genomic technologies present a range of far-reaching opportunities: from facilitating fundamental research into the genetic determinants of diseases, to supporting the development of more targeted and effective diagnostics and treatment therapies. These advancements can ultimately be translated into patient benefit and realise the ambition of personalised and preventative medicine.[1,2]

[1] Academy of Medical Sciences (2017). *Response to the House of Commons Science and Technology Committee inquiry into genomics and genome-editing*.
[2] Academy of Medical Sciences (2017). *Supplementary response to the House of Commons' Science and Technology Committee's inquiry into genomics in the NHS*. https://acmedsci.ac.uk/file-download/40713555

5. As an example, understanding the genomic basis of disease can provide a powerful and unbiased approach to identify relevant and potentially novel drug targets. This information may be able to improve the success of pre-clinical drug development efforts and pave the way forward for personalised medicine.

6. In our recent horizon scanning survey, genomics was one of most frequently cited areas considered to have transformative potential (together with artificial intelligence (AI)).[3] Respondents to that work felt that advances in genomics could be expected to accelerate the shift towards population stratification and personalised treatments.

7. Notably, this survey also revealed that increasing understanding, and clinical use, of genomic data could support the earlier diagnosis of disease – and in certain areas such as cancer medicine, potentially allow a more accurate diagnosis. The identification of genetic diagnostic markers for neurological and psychiatric conditions could be particularly transformational, given the increasing prevalence of these conditions and current challenges in diagnosis.

8. An increased understanding and application of genomic technologies could also inform preventative strategies, allowing pharmaceutical and non-pharmaceutical interventions to target people based on their genetic make-up in an effort to prevent ill-health before disease has even occurred. As speculated in our 2015 meeting on a stratified approach to diabetes, it may be that, in the future, an individual could undergo genome sequencing at birth to produce a stratified profile that could help to direct lifestyle choices and prevent disease.[4]

9. However, we believe the value from genomic data will only be realised if it is linked with other data, such as phenotypic information. This can create a richer dataset of information, which is key for a stratified approach. Oversight of the collection, access and linking of genomic data, and how this information is applied in research and healthcare, is essential. We believe that learnings can be drawn from the success of the 100,000 Genomes Project in the handling of genomic data and managing its use in individual care, as well as its successful establishment of committees who oversee aspects such as ethics and data access.[5,6]

10. There are potential benefits to patients from commercial testing, however, this is contingent on individuals being supplied with adequate support so that they can act on the information appropriately. To realise the wider benefits, genomic data will need to be linked with other data and those linked resources will need to be accessible for clinical and research use and governed by appropriate oversight.

The industrial strategy opportunity for genomics within the UK biotechnology sector, and how the Government could support UK growth (including for exports)

11. The value of genomics for the UK has previously been stated in our response to the 2017 House of Lords' Science and Technology Committee inquiry into Life Sciences and the

[3] Academy of Medical Sciences (2018). *Horizon Scanning*. https://acmedsci.ac.uk/policy/policy-projects/horizon-scanning
[4] Academy of Medical Sciences (2015). *Exemplar clinical pathways for a stratified approach to diabetes*. https://acmedsci.ac.uk/viewFile/57cfd3c90098c.pdf
[5] Academy of Medical Sciences (2017). *Supplementary response to the House of Commons' Science and Technology Committee's inquiry into genomics in the NHS*. https://acmedsci.ac.uk/file-download/40713555
[6] Academy of Medical Sciences (2018). *Response to the Wellcome Trust consultation on oversight: emerging science and technology*. file://ams-fs-01/CloudUsers$/Rachel2/CloudFolders/Downloads/20151017.pdf

Industrial Strategy, where we highlighted the importance of national genomic data resources such as the datasets developed through the 100,000 genomes project.[7]

12. More specifically, we believe that access to these data sets—provided appropriate mechanisms exist to safeguard inappropriate use— is a unique strength to the UK's health research, and provides an incentive to the wider life sciences sector to be based in the UK.

13. We also recognise that the application of genomics and related technologies is likely to see significant growth in the near-term future. As a result, new healthcare products and therapeutic strategies can be predicted to build on these approaches, facilitating a step change in personalised medicine and an opportunity for the UK to lead in this area.

14. In addition, the UK's unique strengths in health data and data sharing, and the emergence of national genomics data resources, create a unique opportunity for economic growth in data analytics. Investment in genomic healthcare initiatives and their infrastructure, could help ensure that the UK becomes a global leader in the development, manufacture, and commercialisation of innovative products and services based on genomic data.

The extent to which currently available genomic sequencing and interpretation can provide accurate and unambiguous health results, for healthy and ill sections of the population

15. While the Academy believes that genomic data has far-reaching potential, we have previously cautioned that the interpretation of genetic testing can be subjective, or even ambiguous, in some cases.[8] To address this, we have suggested that knowledge sharing processes and the establishment of a centralised database of genomic information could help to support consistency in these decisions.

16. In previous work exploring a stratified approach to cardiovascular disease, we have also noted that a potential obstacle to the utilisation of genomic data more widely could include a reluctance from clinicians to impose a genetically-based diagnosis on a currently 'well' person.[9] However, this could be overcome with improved training on patient counselling and by communicating the benefits of diagnosis before symptoms emerge.

17. Similarly, we reiterate that there must be a carefully considered, standardised approach to incidental findings during genetic testing, and suggest that the model developed by Genomics England can be used as an exemplar where feedback is only provided on high probability incidental findings that may impact health.

The counselling or other support offered for those receiving, or considering asking for, commercial genomic test results, and whether this is to the standard required

[7] Academy of Medical Sciences (2017). *Response to the House of Lords' Science and Technology Committee call for evidence in Life Sciences and the Industrial Strategy*. https://acmedsci.ac.uk/file-download/25500651
[8] Academy of Medical Sciences (2017). *Supplementary response to the House of Commons' Science and Technology Committee's inquiry into genomics in the NHS*. https://acmedsci.ac.uk/file-download/40713555
[9] Academy of Medical Sciences (2016). *Exemplar clinical pathways for a stratified approach to cardiovascular disease*. https://acmedsci.ac.uk/viewFile/57cfd5170e1de.pdf

18. In our 2017 response to the House of Commons' Science and Technology Committee's inquiry into genomics in the NHS, we raised a number of important considerations with regards to the necessary skills, training, and capacity needed to support the accurate interpretation and application of genomic data.[10] In particular, we cited a need for specifically trained genetics nurses and counsellors, and highlighted that the development of decision-support tools could help such staff understand and interpret genomic testing alongside other experts such as geneticists.

19. However, we also wish to emphasise that there is a need to raise awareness of the potential benefits and limitations of genomic information across all healthcare professionals, and a need to embed genomics into clinical education for both specialist and non-specialist training.

20. Realising the true value of genomic data to support health will require a multidisciplinary approach that should include all staff involved in the patient pathway - from geneticists and bioinformaticians to nurses and clinical specialists. Genomic data will also increasingly necessitate transdisciplinary and collaborative working across traditional clinical boundaries to involve multiple disease areas and specialities.

21. It will be vital to ensure that all those involved in the interpretation and application of genomic data - researchers, clinicians, patients, and the wider public - have a robust understanding of what genomic data can inform, and what it cannot, will be vital to better facilitate its adoption to improve healthcare.

22. Similarly, there is an increasingly urgent need to improve data and informatics skills across all areas of healthcare provision in order to harness the rapid advances in genomics technologies. Our *'Health of the Public in 2040'* report recommends that training in quantitative skills should be incorporated at an early stage for current and future researchers. It adds that training pathways for 'informatics for health' should be enhanced for healthcare professionals, in order to build a critical mass of expertise that can take full advantage of the genomics revolution.[11]

The potential benefits and risks for the NHS that arise from the increasing availability of commercial genomic testing

23. We wish to emphasise that genomic data, in the context of the NHS, is beneficial not only for clinical care, but for research. In our response to the House of Commons Science and Technology Committee's inquiry into genomics in the NHS, we noted that the data held within the NHS are a unique strength for the UK.[12] We added that genomic data should not be considered in isolation, but that integration of genomics with other data such as phenotypic information will create a richer dataset that is key for a stratified approach. Therefore, it is important to ensure that genomic data, regardless of its origin, can be linked with other data types that are also valuable for research and healthcare.

24. We therefore believe that the sharing and linkage of genomic data arising from both commercialised and non-commercialised sources should be considered, and draw attention to

[10] Academy of Medical Sciences (2017). *Supplementary response to the House of Commons' Science and Technology Committee's inquiry into genomics in the NHS*. https://acmedsci.ac.uk/file-download/40713555
[11] Academy of Medical Sciences (2016). *Health of the public in 2040*. https://acmedsci.ac.uk/policy/policy-projects/health-of-the-public-in-2040
[12] Academy of Medical Sciences (2017). *Supplementary response to the House of Commons' Science and Technology Committee's inquiry into genomics in the NHS*. https://acmedsci.ac.uk/file-download/40713555

our recent policy report on '*Our data-driven future in healthcare*.'[13] This report outlines some essential principles for the use of patient data to keep public trust. Importantly, the principles aim to enable organisations, including the NHS and industry, to respect and protect the privacy, rights and choices of patients and the public, by helping to provide safeguards to support the use of patient data in ways that are fair and equitable. It also highlights a need to include patients and the public as active and meaningful partners when deciding on the use of patient data.

What data obtained from genomic testing could be used for and if sufficient protection is in place for consumers using commercial genomic tests

25. Participants at our recent horizon scanning events identified significant ethical and privacy issues relating to the storing and sharing of genetic data, and the protection and privacy of patient data is a key consideration raised by the clinical application of genomics.[14] We therefore wish to emphasise that active engagement with patients and the public to discuss data privacy and usage policies is required to build confidence and support this rapidly advancing field, and to ensure that it is at the core of the UK's increasingly stratified approach to medicine.

The regulations or standards that commercial genomic tests are currently subject to, and if any new or strengthened regulations or standards should be introduced to mitigate any perceived risks associated with commercial genomic testing

26. We have previously noted that a national approach to the commissioning of genomic services will help to ensure equity of access across the country, and add clarity around where responsibilities lie for funding for tests and, more widely, as to where this fits within specialised and local commissioning.[15] We also support the need for 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.

This response was prepared by Dr Rachel Brown, Senior Policy Officer, and was informed through the Academy's previous work in this area. For further information, please contact Elizabeth Bohm (Elizabeth.Bohm@acmedsci.ac.uk; +44(0)20 3141 3217).

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[13] Academy of Medical Sciences (2018). *Our data-driven future in healthcare.* https://acmedsci.ac.uk/policy/policy-projects/use-of-patient-data-in-healthcare-and-research
[14] Academy of Medical Sciences (2018). *Horizon Scanning*. https://acmedsci.ac.uk/policy/policy-projects/horizon-scanning
[15] Academy of Medical Sciences (2017). *Supplementary response to the House of Commons' Science and Technology Committee's inquiry into genomics in the NHS*. https://acmedsci.ac.uk/file-download/40713555