Department of Health & Social Care

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Chair, Science and Technology Committee
House of Commons
Westminster
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Dear Chair

I am responding to your predecessor's letter of 1 November regarding the interim findings of the Committee's previous inquiry into commercial genomics, which I read with interest. I understand that, in addition to the over 80 written submissions, the inquiry managed to hold two oral evidence sessions but was not able to conclude or publish a final report.

Before responding to the Committee's letter, which raises a number of important issues, it may be helpful to set out the policy context and the Government's ambitions for genomics.

The UK is a world leader in genomics research. In 2013 we set up Genomics England and, in partnership with the NHS and other partners, launched the 100,000 Genomes Project to sequence 100,000 whole genomes from rare disease and cancer patients. We reached our goal of sequencing 100,000 genomes in December 2018, providing the largest and most complete dataset of its kind in the world. Researchers are using this dataset to carry out research and develop new treatments and diagnostics which will benefit this and future generations of NHS patients. We are now going further and have committed to sequencing 1 million whole genomes by 2024, 500,000 in the NHS and 500,000 from samples held by UK Biobank.

In addition to diagnosing and treating disease when patients start to show symptoms, genomics also has the potential to transform healthcare and help us detect and prevent disease earlier.

As you know, the Government's manifesto set out our ambition to make the UK the leading global hub for life sciences, accompanied by investment in world-class computing, health data systems and genomic sequencing. Genomics is therefore of strategic importance to extending the UK's lead in the life sciences and transforming the way that care is delivered in the NHS. The Government believes that the NHS, industry, academia and of course patients all have an important role to play in achieving our national ambition. We will soon

publish a National Genomics Healthcare Strategy, which will set out how we can become the world leader in genomic healthcare over the course of the next ten years, focusing on the themes of diagnosis and personalised medicine, prevention, and research.

Commercially, genomics is still an emerging industry; nevertheless, companies developing and offering genomics technologies are an important constituent of the life sciences sector and contributed £1.9bn in turnover in 2018. Commercial genomics, made available either by direct to consumer genetic testing companies or companies offering sequencing and genome analysis services, plays an important role in our life sciences economy and could offer important opportunities and innovations in genomic healthcare.

Commercial genomics is an emerging and evolving field and I welcome your letter highlighting a number of important issues. Our priority is to ensure that new technologies which can improve health are introduced in a safe and equitable way. However, some of the issues you raise are complex and it would be inappropriate for me to respond fully to the Committee's interim conclusions at this time without more detailed consideration, and in some cases consultation, and an understanding of whether the Committee intends to continue with this inquiry.

Below, I have set out the Government's current position on the Committee's interim findings. The Committee may also wish to refer back to the Government's written submission to the inquiry which set out current and developing legislation, in particular for medical devices and medicinal products, which ensures that the area of commercial genomics is regulated consistently and meets applicable standards of safety, quality and efficacy to protect the public and patients from potential safety issues in a rapidly developing area.

Inquiry theme 1: Validity of genomic test results

Interim recommendation: *The Government should ensure that medically-relevant genomic tests available in the UK have demonstrated clinical utility for the conditions and consumers that the tests are being sold for, with any information that would be needed to enable external assessment of such validity also required. It should review the extent to which future regulations will achieve this, and whether or not such measures could be implemented before 2022.*

Government view: Clinical utility can be defined as the benefit/risk balance of a test as determined through clinical use. It is determined in part by the actions taken based on the result of the test and the subsequent health outcomes.

Requirements for clinical utility can significantly increase the amount of evidence required before a novel innovative genomic test can be first made available for clinical use.

The forthcoming regulations on *in vitro* diagnostic medical devices (IVDs) will require that genomic tests must be suitable for their intended medical purpose. For example, a genomic test to predict disease risk must have the analytical and clinical performance that is appropriate for that medical purpose. The test must also have sufficient evidence that correlates the genomic markers being tested with the ability to predict the specified disease.

For rare diseases the threshold may be low, but for common diseases, where more evidence can be generated, the threshold will be higher.

It will not be sufficient to say that a test can simply detect or measure a genomic marker. To qualify as an IVD, the genomic test must have a medical purpose and must have sufficient clinical evidence that the genomic marker is correlated with the medical purpose of the test.

Inquiry theme 2: Consent and reproductive choice

Interim recommendation: *The Government should review different models of consent to determine which might be unsuitable for obtaining informed consent. It should also consider prohibiting the genomic testing of asymptomatic children, other than in certain cases.*

Government view: The 100,000 Genomes Project used a broad consent model, which was agreed after careful consideration and external advice from ethicists and legal experts, and which maximised the size of the dataset available to researchers. By contrast, a dynamic consent model requiring each participant to opt into every research project would have resulted in much smaller, idiosyncratic datasets for each project, severely limiting the benefits of the overall programme. This view is strongly supported by the Genomics England Ethics Advisory Committee.

Regarding the genomic testing of asymptomatic children in a healthcare context, such testing has proven clinical benefit in both the prevention of childhood conditions and in helping to diagnose an affected sibling. The current approach to genetic testing in asymptomatic children in the NHS is nuanced and evidence-based and considers concerns around feedback of findings for late onset conditions, particularly where there is no immediate intervention.

Regarding the genomic testing of asymptomatic children in direct to consumer context, it is an offence under section 45(1) of the Human Tissue Act 2004 to have any bodily material (that is, any material which has come from a human body and which consists of or contains human cells) intending to analyse the DNA in it without qualifying consent, subject to certain exceptions. This offence applies to the whole of the UK. In relation to analysis of DNA manufactured by the body of a person who is alive, "qualifying consent" means consent given by the person from whose body the material came or someone with parental responsibility of the person is a child.

Further information on the role of the Human Tissue Authority and consent and DNA can be found at https://www.hta.gov.uk/policies/about-dna-and-role-hta

Generally, if children have the capacity to give consent for themselves, consent should be sought directly from them. Once young people reach the age of 16, they are presumed in law to be competent to give consent for themselves for their own surgical, medical or dental treatment, and any associated procedures, such as investigations, anaesthesia or nursing care.

Those under 16 are not automatically presumed to be legally competent to make decisions about their healthcare. However, the courts have stated that under 16s will be competent to give valid consent to a particular intervention if they have "sufficient understanding and intelligence to enable him or her to understand fully what is proposed" (i.e. Gillick competence).

Interim recommendation: *The Government should review the regulation of antenatal and prenatal genomic tests, to ensure that all providers of such tests provide adequate and non-directive support to prospective parents taking these tests.*

Government view: Antenatal and prenatal genomic tests and other tests offered direct to consumers are likely to be regulated as 'self-tests' in the new IVD regulations. This will include tests offered directly to consumers from outside the UK. In categorising these as self-tests, the regulations would require that results must avoid misleading information and be easy for the intended user to understand and interpret. The information provided should include a statement clearly directing the user not to take any decision of medical relevance without first consulting the appropriate healthcare professional. The IVD regulation will not extend to clinical practice in the provision of genetic testing services.

Inquiry theme 3: Support available for consumers taking genomic tests

Interim recommendation: *The Government should review the case for requiring commercial providers of genomic testing to provide counselling for highly predictive genomic tests even where the condition in question may be treatable, and for tests related to family history. This should include consideration of the qualifications or registration such counsellors should achieve. The Government should also consider the responsibility of such providers to support the training of genomic counsellors, and if appropriate introduce measures securing this support. Several commercial providers of genomic tests indicated to us that they would be willing to contribute to the training of qualified genetic counsellors in the UK—the Government should follow up on this and engage with providers to ensure sufficient numbers of counsellors are trained for future demand.*

Government view: Currently the main route for training genetic counsellors in the NHS is through the Healthcare Scientist Training Programs (STP) commissioned by Health Education England (HEE). On completing the three-year STP with a Master's degree in Clinical Science, students are eligible to register as Clinical Scientists in Genetic Counselling with the Health and Care Professions Council. The Modernising Scientific Careers Framework is being reviewed to increase the access routes into the framework and develop flexible training opportunities. HEE would be willing to work with all stakeholders to consider the genetic counselling training and registration requirements for those working in the commercial sector and to explore how they may support an increase in training commissions for genetic counsellors.

The IVD regulation will have a limited and clearly circumscribed role in the provision of counselling. The regulation will likely require that the Member State ensure individuals are provided with relevant information on the nature, significance and implications of the genetic test and that there is appropriate access to counselling for genetic tests that provide

information on untreatable diseases. The regulation will recognise that the UK may wish to be more protective or more specific around counselling and informed consent. The IVD regulation will not extend to clinical practice in the provision of counselling services.

Inquiry theme 4: Regulating testing conducted abroad

Interim recommendation: *The Government should work with relevant regulators to review this issue [of consumers buying tests that are outside the UK regulatory framework] and identify strategies to minimise the availability of genomic testing to UK users that does not comply with UK regulations.*

Government view: Where a manufacturer offers a clinical genetic test to anyone in the UK (regardless of how it is advertised, e.g. internet, third party seller, or otherwise), or they use a clinical genetic test in a service that is offered to anyone in the UK as part of a commercial activity, then the device will need to meet all of the relevant requirements of the IVD regulation.

Clinical genetic tests used in the UK or offered direct to UK consumers will need to meet all of the same requirements as devices for self-testing (for example, classification and labelling).

The MHRA will have a range of compliance measures in place to minimise the availability of genomic tests that do not comply with UK regulation. Where the offending business fails to bring their activities into compliance with the regulations, website 'takedowns' will be considered and where necessary, MHRA will work with other national regulators to enforce the legislation.

Inquiry theme 5: The potential impact of direct-to-consumer testing in the NHS

Interim recommendation: *The Government must monitor the impact of direct-to-consumer genomic testing on NHS resources as the use of such tests grows, and ensure that the NHS workforce has sufficient guidance and training to deal with any changes as a result of an increase in demand.*

Government view: The Government will work with the NHS and others to explore how to build the evidence base regarding any potential impact and costs to the NHS arising from direct-to-consumer genetic testing.

Interim recommendation: *The Government should be ready to amend the regulation of commercial genomic testing to ensure that such testing does not lead to inequality of access to NHS diagnosis or treatment.*

Government view: The IVD regulation will set out requirements regarding the manufacture and provision of all IVDs including genomic tests. So long as these are met, there are no regulatory restrictions as to how these devices should be supplied or used within the national healthcare system. However, the regulation will recognise that the UK may wish to adopt or maintain measures which are more protective of patients, more specific or which

deal with informed consent. Any further measures relating to access of NHS diagnosis or treatment are unlikely to be covered by this provision.

Interim recommendation: *The Government should also consider introducing a levy on providers of direct-to-consumer testing to contribute to any costs incurred by the NHS as a result of consumers seeking support following a genomic test.*

Government view: The Government will work with the NHS and others to explore how to build the evidence base regarding any potential impact and costs to the NHS arising from direct-to-consumer genetic testing.

Inquiry theme 6: The opportunity for increased genomic data collection to support research

Interim recommendation: *The Government should engage with commercial providers of genomic tests to explore the potential for results obtained directly from consumers to be incorporated into publicly-obtained data and used for research (subject to appropriate safeguards). It should consider supporting the development and use of interoperability standards as well as digital infrastructure to support such data-sharing. It is important also that the Government ensures that the NHS receives a fair share of any commercial benefits accruing from such data collection and usage.*

Government view: The Government will invest in world-class computing and health data systems that can aid research, such as the ground-breaking genetic sequencing carried out at the UK Biobank, Genomics England and the new Accelerating the Detection of Disease project, which has the potential to transform diagnosis and treatment.

To enable this, the Government is committed to protecting the confidentiality of patient data. There are several key safeguards in place – encompassing legislation, security standards and toolkits, independent advisory bodies and a national data opt-out – which ensure that data is used across the system in a safe, secure and lawful way.

The Government agrees that the NHS should receive a fair share of benefit when it enters into partnerships involving data. The Government has published five guiding principles to ensure partnerships between the NHS and commercial partners are fair to the NHS and protect patient privacy. These principles can be found here:

https://www.gov.uk/government/publications/creating-the-right-framework-to-realise-the-benefits-of-health-data/creating-the-right-framework-to-realise-the-benefits-for-patients-and-the-nhs-where-data-underpins-innovation

These principles are being developed into a more detailed policy framework which the Government intends to publish later this year. Alongside the policy framework, the government is establishing a National Centre of Expertise which will support NHS organisations to agree beneficial terms with commercial partners. The Centre will host legal and commercial experts that all NHS organisations can access for support and advice.

I hope this response is helpful. Should the Committee decide to continue with its inquiry, the Government will look forward to receiving the final inquiry report.

Best wishes

Nicola

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