GENOMICS AND GENOME EDITING IN THE NHS INQUIRY

Science and Technology Committee (Commons)

Response by Genetic Alliance UK, 13 October 2017

Introduction

1. Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 180 patient organisations. Our aim is to ensure that high quality services, information and support are provided to all who need them. We actively support research and innovation across the field of genetic medicine.

2. Almost all single-gene disorders are rare. Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK, working towards the delivery and implementation of the UK Strategy for Rare Diseases, signed by all four health departments in the UK and published by the Department of Health in November 2013.

3. Approximately 6,000 children are born in the UK each year with a syndrome without a name – a genetic condition so rare that it is often impossible to diagnose. Genetic Alliance UK runs the support network SWAN UK (syndromes without a name) to provide peer support and information to families with a child with an undiagnosed condition.

4. There are many thousands of genetic conditions affecting patients and families in the UK. Very few of these have an effective treatment or cure. For most of the patients and families we represent, palliative care, mitigation of symptoms and reproductive choice are the only tools available to address the impact of genetic conditions on their lives. This constitutes a tremendous volume of unmet health need.

5. The topics of this inquiry are important to the patients, families and patient organisations that we represent. Genomic and genome-editing technologies are tools with tremendous potential to further our understanding of genetic conditions, and to develop treatments for genetic conditions.

Generation Genome

6. We welcome the attention paid to genomics by the Chief Medical Officer's (Dame Sally Davies) in making it the topic of her latest annual report, ‘Generation Genome’. The expansive document discusses current progress establishing genomics in the NHS and her views on how we can best

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take advantage of the opportunities provided by advances in genomic technology to improve clinical practice and public health.

7. The report follows almost exactly the structure of the 2003 genetics white paper 'Our Inheritance, Our Future: Realising the potential of genetics in the NHS', which is a missed opportunity to demonstrate the substantial progress toward implementing genetics and genomics in the NHS that has been made in the last 14 years, and the resulting shift in the areas of greatest opportunity. The large number of authors for the report has unfortunately diminished its focus.

8. The report contains some unusual recommendations, with items already happening (for example that NHS England should reprocure laboratory genomics services nationally – recommendation 2), items based on the particular specialist interests of chapter authors (for example that all patients with severe childhood onset of obesity should have access to rapid genetic assessment), and items that are some period away from being proven (for example the section on stratifying preventative efforts for common diseases, a concept which is acknowledged to require more evidence).

9. We welcome the emphasis on 'democratising' genomic medicine and making testing a routine part of clinical care, and urge for this to be expanded beyond the cancer, obesity and infectious disease applications discussed in the report.

10. From the perspective of Genetic Alliance UK, the report's most vital themes are perhaps best summarised in the final paragraph of section 1.1: 'To make this dream a reality across England and secure the vision of NHS transformation needed, as well as build on the 100,000 Genomes Project, we need to: embed national standards; streamline laboratories; and, in a secure environment, agree to use of data for our own benefit and others.'

11. The key piece of ongoing work addressing the first two of these, the NHS Genomic Laboratory Services reprocurement is moving forward and we understand will issue the formal invitation to tender and final service specification in November 2017, with a view to the contract being awarded in March 2018 and implemented by October 2018. This process has been beset by delays, with work beginning prior to 2014. Despite the slow progress, there remain many unknowns about the new draft service specification.

12. We are particularly concerned at the lack of sufficient detail to adequately comment on: the methodology for the development of the National Genomic Test Directory; transition between the current genetic test evaluation system and the proposed new approach; and the proposed quality management and monitoring approach.

13. If implemented well, reprocurement of genomic laboratory provision in a hub and spoke model and the moving of the UK Genetic Testing Network processes into NHS England does have the potential to benefit patients. The streamlining of laboratory structures may lead to shorter wait times and lower costs. A more proactive approach to identifying which genetic tests are needed and how they fit into clinical pathways may avoid some of the duplications and gaps which exist currently due to the reliance on laboratory experts submitting test proposals. National standards and monitoring tied to funding may lead to greater equity of access across geographical regions. However, if the detail of the proposals is not made public with plenty of time for stakeholder involvement it appears unlikely that this best case scenario will be achieved.

14. We understand that the Chief Scientific Officer, as chief responsible officer for genomics, has expressed her wish to reprocure clinical genetic services as a next step. We are gravely concerned about the potential impact on patient care of the uncertainty and chaos of both laboratory and clinical genetic services being reprocured in quick succession. The benefits of reprocuring clinical genetic services are not immediately obvious given the continuing need for expert clinical geneticist and genetic counsellors to be in close geographic proximity to patients and mainstream clinicians.

15. We strongly agree with the view expressed by the Chief Medical Officer, in her report, that we need to move beyond 'genetic exceptionalism', recognising that genetic data is neither uniquely sensitive nor uniquely identifiable. We are aware of several work programmes at NHS England and NHS Digital looking into issues of consent and the sharing of data in genomics, both for routine clinical care and for research. It is important that these are not developed in a genomic silo, but are instead connected to the broader work being done around the National Data Guardian's opt out model programme and the Paperless 2020 framework.

Talking about genome editing

16. At a previous call for evidence for this inquiry the previous Director of Genetic Alliance UK, Alastair Kent OBE, informed the committee about a piece of work that Genetic Alliance UK was in the process of delivering alongside the Progress Educational Trust. This work is now complete.

17. The report is available here: www.geneticalliance.org.uk/our-work/medical-research/communicating-about-genome-editing

18. The key recommendations of this work are in the first instance aimed at scientists who need to talk about genome editing with non-expert audiences, but the majority of them can be broadly applied. The report recommends a focus on genome editing rather than the tools or techniques that carry-out genome editing, and so to deprioritise the use of terms like 'CRISPR' unless necessary. The focus should instead be on the purpose or potential use of genome editing.

19. It is also crucial to be clear about the scope of individual uses of genome editing. We need to distinguish clearly between uses that benefit human health and other purposes such as agriculture. Is this technology available now, soon, or is this many years away? Is genome editing being used as tool for research, or will the genome editing be part of a therapy? Would changes in the law be necessary to use genome editing in this way? The clearer we can be, the better the chance we have of communicating effectively on this subject.

20. We hope the committee finds this report valuable.

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