Summary:

The commercial genomic testing industry is rapidly expanding and is an opportunity for growth, innovation and investment in the sector. However, the genomic data and disease risk factors are statistical and difficult to interpret and have implications for consumer mental health. Therefore, it is imperative that all genomic testing is accompanied by adequate support and genetic counselling to avoid misunderstanding of risk factors and to provide support to patients.

1. Any health or other benefits that consumers can derive from using commercially available testing.
   1.1. There are circumstances where genetic information may lead to specific lifestyle changes, to increased monitoring or taking preventative measures (e.g., mastectomy in the case of BRAC1/2 mutations). However, many conditions that can be diagnosed by genetic testing have poor treatment options. Knowledge of an increased risk for an untreatable disease, where there are no changes of lifestyle that may be beneficial, may cause a consumer only stress and anxiety. In this case, the knowledge of this risk may therefore not be able to be considered a benefit to the consumer’s overall health.

2. The industrial strategy opportunity for genomics within the UK biotechnology sector, and how the Government could support UK growth (including for exports)
   2.1. The Biochemical Society recognises that there is an opportunity for investment and commercial genomic testing could be a growth area. The costs associated with genetic testing (predominantly reagents and kits) are becoming cheaper, opening it up to more people. Furthermore, the technology is becoming more robust. Although costs in some areas are going down, costs of infrastructure (sequencers) may not decrease at the same rate, nor will the cost of training and employing adequately trained councillors, technicians and bioinformaticians. Significant investment may therefore be necessary to address the current shortage of these skilled professionals¹, and prevent any further skill shortages from emerging. This will need to include investment in the STEM education pipeline, training specifically in bioinformatics and continuing professional development for clinicians without a background in genetics, in combination with an immigration system that works to support these vital roles².

   2.2. Despite the opportunity for growth, profits derived from such companies may be many years away due to high set-up costs. For the government to help in this area, it should make sure there is funding to make genomic testing available via the NHS with the appropriate staff, and then seek to invest in commercial development of the technologies.

² Genomics Explained: A guide to genomics and UK excellence in the field, UK Bioindustry Association, 2018, https://www.bioindustry.org/uploads/assets/uploaded/367e0ae6-6fa4-47b1-aa80c77118ca0c83.pdf
2.3. There is an opportunity for the UK to build a reputation for quality and robustness in genomic testing (Brand UK already has an expectation of integrity and quality at a global level). However, this will require government regulation to ensure that quality is maintained.

2.4. There are a number of risk-factors that can be identified by genomic testing for conditions for which there are currently no treatments. Investment in research into treatment options for these diseases, and the fundamental science underpinning genomic testing, must therefore be continued alongside any investment into diagnosis via genetic testing.

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

3.1 Many health conditions are caused by more than one genetic factor, or a combination of genetic and environmental factors, making interpretation of the data difficult. There are relatively few instances when genomic test leads to certainty around future health; cases are often associated with a probability. Interplay between genetic and other factors is not fully understood, nor are the layers of genomic control above the base sequence of DNA.

3.2 Interpreting probabilities, and using this information to inform choice is not trivial. Presence of genetic risk factors does not necessarily predetermine that an individual will develop a condition. As discussed in 2.1, significant investment in the training of scientists, genetic counsellors and clinicians that can interpret this complex data will be required. Without genomic data being explained within the correct context, there is a risk of misinterpretation and misunderstanding of results by the consumer. It is therefore imperative that genomic testing be accompanied by genetic counselling as outlined under 4.

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

4.1 The Biochemical Society believes it is essential to ensure adequate provision of genetic counselling to minimize risk of misinterpretation and misunderstanding of results, and to provide sufficient support and advice to consumers. This includes support in the interpretation of risk factors, which could also have ramifications for individuals’ mental health. Knowledge of the probabilities of acquiring specific conditions must be handled with great care. This level of aftercare must be made available at the outset and be costed appropriately.

4.2 Current genomic sequencing can provide detailed information, largely in the form of probability which is not easily interpreted, and education and support for the consumer is of critical importance. For example, if an individual is informed that their risk of cancer has doubled, without adequate counselling, this is likely to have adverse effects on their mental health. However, whilst “doubled” could mean a risk increased from 25% to 50%, it could equally refer to 0.00001% to 0.00002%. The actual risk, and therefore implications for effect on mental health differ in these two scenarios, exemplifying how both careful interpretation of the data and support to the consumer are critical.

4.3 The level of risk associated with different genetic variations must be explained by a healthcare professional. Indeed, making an informed choice to seek probabilistic information is also not necessarily trivial. However, rapid commercialisation of genomic testing may result in a shortage of qualified clinical geneticists as demand increases. It is therefore imperative that adequate
provision for increasing number of qualified clinical geneticists be made alongside any investment into the technology itself. This may include up-skilling existing professionals and the education, training and employment of experts coming from a breadth of pathways to avoid future skills shortages.

4.4 Consideration should be made as to where responsibility lies for providing genetic counselling. If individuals get genetic testing done that is not carried out by the NHS, but then have to approach their GP for guidance and support, this will place an additional burden onto the NHS. The results from genomic testing are nuanced and involve potentially serious diagnoses which GPs are presently not trained to interpret, making their use as the main source of clinical advice inappropriate. However, any private clinical advice paid for by the testing company risks conflict of interest as there is a commercial pressure to stress the importance, use and predictive power of the test. One potential solution may be requiring that, whilst tests may be requested by individuals, results may only be accessed and released by a registered clinical geneticist (who is bound by General Medical Council professional standards). This would ensure that patients have the opportunity for professional counselling at the same time as receiving their results.

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

5.1 There is vast potential for use of accumulated genomic data, and we have concerns around possible misuse of data for commercial benefit. How and to whom data can be communicated should be clearly controlled through a positive action by the consumer, especially as they may not fully appreciate the value of such data. The availability of a genome sequence could be used by, for example, employers and insurance companies. It is the view of the Society that the selling or passing (i.e. non-commercial transfer) of genetic and any linked medical data to insurance companies must continue to be monitored and controlled\(^3\). As the use of genomic testing grows, this may require legislation.

5.2 Genetic data may also be of use for drug development purposes. If anonymised data is to be made available for these purposes, it will be important for small companies to have similar access to data as larger ones. Without this, there will be a risk of stifling innovation in small and medium drug development companies. There is, therefore, a need for government guidance on the use of commercially acquired genomic data in drug development (and other) research, which must be clearly communicated to the consumer.

5.3 Availability of genomic data will be invaluable to moving forward with medical treatments within the NHS, as is already demonstrated in the 100,000 genome project\(^4\). Embedding the use of genomic data within the operation of the NHS has enormous potential for widespread healthcare benefits, such as following treatment method efficacy, stratifying patients in treatment of cancer or monitoring the benefits of drug use across a spectrum of patients.

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these benefits to be realised, the NHS should be the custodian of genomic data and planning for this should become integral to how the NHS moves forward.

5.4 Security of genomic data on servers must be a priority, especially within the NHS. For commercial companies offering genomic testing and subsequently storing data, consideration must be made and published as to data ownership and processes in the case of company closure.

6 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

6.1 The Biochemical Society believes any form of genetic testing should be subject to strict government/MHRA (or other) regulation and that genetic counselling must be mandatory for all. As described in paragraph 2.3, regulation will be required to maintain high quality of genomic testing. Furthermore, there are a number of ethical considerations that should be made in the regulation of commercial genomic testing. These include: criteria for sample handling; communicating the data; monitoring, reporting and communicating errors or oversight; claims used in the advertising of services and guidelines for liability and patient redress in the case of fraudulent results.

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About the Biochemical Society:
The Biochemical Society promotes the future of molecular biosciences; facilitating the sharing of expertise, supporting the advancement of biochemistry and molecular biology, and raising awareness of their importance in addressing societal grand challenges.

We achieve our mission by:

• Supporting the next generation of biochemists; promoting the opportunities offered by biochemistry and molecular biology through education and training from age 15 upwards

• Bringing together molecular bioscientists; fostering connections and providing a platform for collaboration and networking across our membership and the wider community to ensure a strong future for molecular biosciences in both academia and industry

• Promoting and sharing knowledge; enabling the circulation of scientific information through meetings, publications and public engagement to support innovation, inform decision-making and advance biochemistry and molecular biology

• Promoting the importance of our discipline; highlighting the role of molecular biosciences in
interdisciplinary and translational research, while supporting the fundamental research that underpins applied studies.