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PEALS aim to produce world class research focused on the social and ethical debates around the contemporary life sciences.

We provide this evidence as a collective of individuals and it does not represent the position of PEALS Research Centre.

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

Commercially available genomic testing is commonly offered as a way of improving consumer access to health. However, in reality it is difficult to see what health benefits a consumer can gain from undertaking a commercial genomic test. It is acknowledged that currently available tests are usually unable to accurately predict susceptibility to disease. The utility of the test depends on the disease and on the personal context: in the UK, those individuals who might benefit clinically from genomic testing, such as members of families with a history of genetic disease, or whose disease is known to have a genetic component, will be able to access tests via the NHS and should have no need for commercial testing. There is no evidence of clinical benefit in providing ‘susceptibility’ tests to healthy people, and furthermore the results of such tests can be highly problematic in terms of interpretation for individuals and their family (we discuss this further, below).

The recent high-profile case of Matt Hancock MP, Secretary of State for Health and Social Care, who undertook a commercial genetic test, illustrates these concerns. Mr Hancock stated that the test results showed he was at high risk for prostate cancer; the test results had caused worry; the test had saved his life and he would be seeking an appointment with his GP to discuss next steps. Clinical geneticists pointed out that Mr Hancock had misinterpreted the results: he was not at increased risk, and there was therefore no reason to be worried. This meant the test was not life-saving, had caused unnecessary concern, and that the subsequent GP appointment was a waste of NHS resources. If Matt Hancock, with above average access to resources and an elite education, can come to such an erroneous conclusion, what does this imply for the average consumer? These concerns are particularly relevant where consumers may purchase genomic testing for what might be considered recreational reasons – out of curiosity, for genealogical research, or to identify supposedly genetically linked ‘talents’ – and be unable to interpret the results appropriately (1, 2).

We should not forget that all services provided by DTC genomic testing have the purpose of making a profit for company shareholders. The profits are realised by the processing and sale, to third parties, of genetic information about individuals collected during the testing, for the purposes of research. DTC genomic testing companies are undertaking medical research on UK individuals, including children, without, we argue, the majority of those individuals realising this.
2. **The industrial strategy opportunity for genomics within the UK biotechnology sector, and how the Government could support UK growth (including for exports)**

We have no comment.

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

We want to draw attention to the fact that the wording used in the question, of tests providing ‘accurate and unambiguous health results’, is unhelpful. Interpreting results of genomic testing is always complex and is constantly evolving. Results can be ambiguous while at the same time yielding clinically useful information.

We must distinguish between genomic testing and interpretation made available direct-to-consumer and that which is not – the latter usually being within a healthcare setting. We will refer to these respectively as ‘DTC’ testing and ‘healthcare’ testing. The UK healthcare setting, particularly the work of Genomics England, has seen some encouraging progress, including: increasing test utility for single-gene, rare disorders; identifying new causal mutations for single-gene disorders and tumors; and using results to guide treatment decisions. However, even in the setting of the NHS and Genomics England (ie of publicly funded healthcare), the complexity of new genomic technologies makes their application to clinical medicine problematic. The ethical implications and societal impacts are poorly understood, and need further investigation (3).

Consumers are likely to assume that genetic testing, whether DTC or healthcare, is always highly predictive and determinative (even when the test is for susceptibility). This assumption about the meaning of results may lead to unnecessary interventions, life-style changes and anxiety. Also, for any given genomic test result it is generally unclear which of the range of possible life-style changes and interventions will be most beneficial. For example, the identification of the haemochromatosis genotype by itself may lead to many more people seeking intervention than are at real clinical risk. Susceptibility testing may also be falsely reassuring: if consumers believe they have been told that they are at low risk of heart disease, for example, then they may continue to indulge in high risk behaviours. In addition, some positive tests, such as apolipoprotein E genotype associated with Alzheimer’s disease, can be imprecise without further careful interpretation. We do not believe there is convincing evidence that companies offering DTC genomic testing are adequately communicating these distinctions to their customers. In addition, we consider that some companies are advertising that susceptibility testing is of higher utility than it really is which is in contravention of UK guidelines (4).

Evidence suggesting relationships between particular genomic markers and diseases or traits has a track record of being modified or even disproven over even short periods of time. The responsibilities of DTC genetic testing companies with respect to this are unclear. We have particular concerns where susceptibility testing relates to serious mental health issues: our knowledge of this area suggests that the available support from DTC genomic testing companies is inadequate.
If consumers rely on the NHS for follow up after receiving the results of a DTC genomic test, there are potentially complex implications regarding changes in time over interpretation of genetic variants. Will the NHS be expected to keep abreast of changes in interpretation of the patient’s genetic information? What would be the standards involved in deciding when to recontact patients about changes in the interpretation of their results? Further research is needed.

4. **Counselling or other support offered for those receiving, or considering asking for, commercial genomic test results, and whether this is to the standard required**

Currently, individuals purchasing a DTC genomic test are not required to undertake pre- or post-test genetic counselling. This is in contravention to the only UK guidelines for commercial genetic testing: The Human Genetics Commission’s ‘Framework of Principles for DTC genetic testing services’ (2010) were compiled by an expert group after extended public consultation, in direct response to the rise of DTC genetic testing. Section 4 of the Principles states that companies offering DTC genetic testing services should provide easily understood information, including:

‘4.1 information about counselling offered in connection with the test including whether counselling is included in the cost of the test and for what costs the consumer will be liable if they withdraw following pre-test counselling’

Section 5 of the Principles states:

‘5.1 Where the test is a genetic test in the context of inherited or heritable disorders, that test should only be provided to consumers **who are given a suitable opportunity to receive pre- and post-test counselling**.

5.2 The counsellor **should have the appropriate skills and competencies** and should be accountable to a relevant professional body.

5.3 After receiving the information provided in part 4 and **receiving any offer of pre-test counselling**, consumers should have the opportunity to cancel purchase of the test without incurring further costs relating to the test’. (Our emphases).

We wholeheartedly endorse these stipulations, given our and others’ concerns about the impact of knowledge about inherited or heritable disorders on individuals, families and family relationships (5, 6). Since the actual impact of genetic test information on the individual will depend on many factors, not all of which are clinically obvious, it seems difficult or impossible to decide ahead of time which aspects will have a severe enough effect to merit professional input. Currently, DTC genomic testing companies leave the decision of whether to undertake counselling, as well as the responsibility and most importantly the cost of arranging counselling, to the individual undertaking the test, with no guidance or prior information that such counselling may be required to make best use of the test findings and to avoid harmful effects. Empirical evidence has shown that being overburdened by information, particularly uninterpretable information, makes people less rather than more able to make informed and reasoned decisions (7).

We consider therefore that information provided by DTC genomic testing companies should be curated according to personal circumstances so that the consumer is not overwhelmed by it. This is
likely to be only feasible if every person purchasing a DTC genomic test kit is **required** to have personal interaction with a qualified professional.

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

We find it difficult to identify significant benefits to the NHS from the increasing availability of commercial genomic testing. While it might appear plausible in principle that DTC testing could reduce the burden on the NHS, in practice we think there is a strong likelihood that the risks, including that of increasing the demand on NHS resources, will outweigh any potential benefit. For instance, there is a strong risk of raising unrealistic expectations in the public mind about what sequence information can currently tell us about health states or prognoses. Most importantly for resource implications, it may encourage unrealistic expectations about the ability of the NHS to respond to genomic information that a person obtains – whether the limitation is simply because of current lack of knowledge, or because of resource limitations. Increasing demand by DTC consumers for the NHS to provide further information, counselling, or interventions effectively means that DTC companies are imposing on the NHS the full cost of following up on initial DTC findings. We saw this in the case of Matt Hancock.

There is a risk that increasing popularity and availability of DTC genomics will reinforce an existing popular trend towards ‘geneticization’, ie the belief that most aspects of human life can fundamentally be described in genetic terms, which neglects other relevant environmental and social factors - factors which may be more amenable to public or clinical health interventions than genomic ones. Conversely, there is also a real risk that if DTC testing companies provide inaccurate or contradictory information, or more dramatically if there are high-profile cases of consumers being harmed by poorly regulated DTC genomic information, there will be damage to public confidence in genomic information in general, including that offered by appropriately regulated and medically supervised health services.

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

We have serious concerns around the Intersection of commercial and state interests in using commercial genomics data (e.g. by police, border agencies, social services) as the implications for when and how such cross-over use could be proportionate or legitimate are not yet properly understood. We note the use in the USA of genealogy databases by law enforcement agencies and would argue that legal and professional guidelines are currently insufficient for the protection of the individual consumer with regards to DTC genomic testing and law enforcement (8, 9).

Another concern is that DTC genomic databases are not representative of minorities and the industry is contributing to racialisation and the misrepresentation of genetic diversity (10). It will be important to put in place measures to appropriately identify, inform and provide protections for, such groups, which may place additional demand on public health and care services.
7. **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**

Commercial genomic tests are not subject to any specific regulation or legislation within the UK. The equipment used for the spit test is regulated by the MHRA as a medical device, but the process of testing, the reporting of test results, and the retention and use of individuals’ data are not covered. Many of these services flout voluntary advisory guidelines, like the European Society of Human Genetics’ ‘direct-to-consumer genetic testing for health-related purposes’, 2010 and the Human Genetics Commission’s ‘Common Framework of Principles’, 2010. Companies conducting the tests are usually located outside UK jurisdiction (and typically outside of the EU), while offering services directly to UK consumers. We believe this raises a number of concerns regarding:

- **Services which, if they were UK based, would contravene UK law**;
- **Services which contravene internationally recognised standards of ethics**;

a) **Services which potentially contravene UK law**

A number of companies offer an Infidelity Test, where individuals are encouraged to submit another individual’s sample for DNA testing, collected from “underwear, bed sheets and or clothes” ([https://www.easydna.co.uk/infidelity-dna-test/](https://www.easydna.co.uk/infidelity-dna-test/)). We would argue that this promotes surreptitious testing. Under the UK Human Tissue Act 2004, genetic analysis of human tissue without the consent of the donor is a criminal offence punishable by a custodial sentence. One company, EasyDNA, who have UK office and web addresses, note on their UK website that submitting another person’s sample for testing without their consent is in contravention of the HTA; however, at the same time they put the onus for obtaining consent on the person submitting the sample. The website also states it is the responsibility of the submitter to inform the donor about how the sample will be used. There is no indication that the company checks whether consent has been obtained or that the company bears any legal responsibility under the Human Tissue Act. If a UK consumer were to order this test from EasyDNA’s USA-based website ([https://www.easy-dna.com/infidelity-dna-testing/](https://www.easy-dna.com/infidelity-dna-testing/)) rather than the UK one, there would be no mention of the HTA or illegality. It remains puzzling to us how a company based in the US, can advertise and supply a service in the UK, which is illegal. UK consumers are left exposed by UK law.

b) **Services which contravene internationally recognised standards of ethics**

**Testing of children**

A number of DTC genomic testing companies specifically encourage the testing of children. For example, MapMyGene provides an ‘Inborn Talent Genetic Test’, which they claim allows parents to discover and nurture their child’s unique talents via genetics ([http://www.mapmygene.com/services/talent-gene-test/](http://www.mapmygene.com/services/talent-gene-test/)). Such claims are not evidence-based and the company misleadingly presents population-based research as pertinent to tests on individuals when it is not. They also claim 99% accurate predictions, which is similarly misleading. The Inborn Talent test includes information about ‘traits’ which might be linked to mental health, including explicit testing for depression. Some companies offer testing for even more severe mental health
disorders including bi-polar and schizophrenia and we consider this testing to be based on dubious
science.

Most medical ethics guidelines (11, 12) state that medical interventions or medical research in
children should only be undertaken when they are in the best interests of, or of benefit to, the child.
To be clear, we consider these tests to be medical, even though they are presented as something
other, and we argue that the processing of children’s genomic data obtained from the tests
constitutes medical research on children. ‘Best interests’ here would normally be understood to be
the benefit of gaining knowledge about a current or future health condition which might be subject
to effective prevention, treatment or care. This must be balanced against the potential harms of
knowing the child’s genetic status, including removing the child’s opportunity to make future
decisions about that status. Given these factors, we can see no benefits in genomic testing of
children outside of a healthcare context.

Informed consent

Notwithstanding the example of surreptitious testing above, we believe that many of the companies
offering DTC genomic testing do not follow international guidelines which say that, for consent to be
informed the person taking the consent must be satisfied that the person giving consent
understands the procedure/test they are about to undertake and the implications of taking it (11,
12). This can not be the case with DTC genomic testing. DTC genomic testing websites frequently use
‘wrap contracts’ where the consumer ticks a box online to indicate that they have read the contract
they are entering in to. The wrap contract may also mean the consumer gives their consent to the
test. The provider has no idea if the individual has even read the contract, never mind understood it.
In addition on-line formats easily conflate informed consent with current practices around tick-
boxing Terms and Conditions. Even a signed consent form – which some companies use – does not
establish a legally or ethically valid consent and the best way to protect consumers is to ensure
individual contact with a trained professional (13).

c) General concerns

We accept that there may be testing services available that are of very low risk. These tests will
disclose less significant individual genetic information, for example nutrigenomics or lifestyle
genetics, and or information related to ancestry. What is important, is that the consumer
understands the distinction between these lower risk results and results which provide health-
related genomic information. However, DTC companies offering genomic testing regularly blur
these lines, in fact these lower risk tests contribute to blurring the boundary between the
recreational and the clinical. For example the market leader, 23andMe, offer a test whose name
(Ancestry+) implies that the primary purpose of the test is ancestry tracing. Nevertheless, this same
package includes testing for predisposition and carrier status relating to more than 50 health
conditions, including serious ones such as predisposition to breast cancer indicated by BRCA+ status,
or to Alzheimer’s disease. We are concerned that the boundaries between trivial and non-trivial test
results are deliberately blurred here, so that health testing is presented as an action as relatively
inconsequential as finding out that some of your ancestors came from Scandinavia. We would
endorse differential regulation for tests with different purposes, with an ‘in principle’ restriction of some tests DTC.

8. the potential benefits and risks, for individuals and for the NHS, and the ethical implications of the NHS offering genomic testing to healthy individuals willing to pay and share their data anonymously

If the NHS were to offer genomic testing to healthy individuals for payment, this effectively turns the NHS (or the part of it offering this service) into a commercial genomic testing company whose primary aim is to acquire data for research purposes. The advantage for the individual is that they have access to genomic information that they may find useful or interesting, in the same way as from a DTC company, but from a provider in whom they can have more confidence that the appropriate laws or ethical guidelines are being followed. There are potential benefits to society in the same way the NHS benefits from data collected from healthy volunteers through for example biobanks or longitudinal health surveys. It is possible that a large data set of ‘healthy’ volunteers could be used as a comparator for data from people with genetic disease and could therefore contribute to understanding causes of genetic disease.

However, people who contribute to say, biobanks, are volunteers gifting their biological resources for the benefit of society, not consumers purchasing a service. If the NHS offers paid-for genomic testing, the individual’s biological matter and data become monetised, creating a completely different relationship with the NHS. As such we can not recommend that the NHS offers genomic testing to consumers who pay. Even in terms of the NHS offering genomic testing for free to healthy volunteers, it is unclear whether the benefits currently outweigh the risks and costs. The additional costs of providing support and counselling to healthy volunteers may outweigh any benefits.

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