

**Medical Profiling and Online Medicine:
The Ethics of 'Personalised' Healthcare in a Consumer Age
Response from the PHG Foundation**

Introduction

The Foundation for Genomics and Population Health (PHG Foundation) is the successor body to the Public Health Genetics Unit. Its overarching purpose is to foster and enable the application of biomedical science, particularly genome-based technologies, for the benefit of human health. Among its specific objectives is the promotion of a social and regulatory environment that is receptive to innovation, without imposing an undue or inequitable public burden. The Foundation has a particular interest in the way that new technologies are translated within health services, in genetic research and its impact upon clinical and public health services.

General Comments

The PHG Foundation is generally supportive of new technologies if their use can improve health care. We believe that 'personalised' medicine has an important role to play in the future of health care, particularly in supporting a paradigm shift from diagnosis and treatment to prediction and prevention. For example, novel genetic (and other) biomarkers associated with disease could be used for stratifying the population into numerous subgroups based on risk, in order to improve targeting of interventions, such as screening, at those populations at highest risk of disease. This stratified (rather than personalised) medicine could substantially reduce the harms associated with current predictive medicine and public health activities. At the level of the individual patient, early presymptomatic (or pre-dispositional) testing should promote a more tailored approach to patient care and facilitate early treatment (where available). We anticipate an increase in 'companion diagnostics' associated with new and existing treatments, and the use of pharmacogenetic testing to identify individual variations in drug metabolism genes thereby assisting physicians to select the most appropriate treatment strategy (e.g. by minimising side-effects).

We therefore welcome the advent of 'personalised' medicine within health care, but caution against its premature application in the absence of appropriate evidence of clinical benefit. Elsewhere we have argued that, in the context of laboratory diagnostic tests, the findings of any test should be robust (i.e. that they should demonstrate scientific and clinical validity), and claims made about the significance of a test should be supported by evidence¹. We have also called for structural changes such that new tests are subjected to a more responsive and proportionate assessment process as part of the pre-market review process, and that information about test performance is placed in the public domain. Some of these principles are relevant to this consultation. However, the core technologies at stake here, DNA profiling and body imaging, are in their infancy: findings are often of uncertain significance and may only become apparent over time. This seems to generate a more conceptual set of questions about the role of the state in ensuring that products and services are not just safe but also effective and useful. The majority of our comments below therefore address the types and level of evidence required to support the use of a particular test in different situations, how different

¹ PHG Foundation. The evaluation of diagnostic laboratory tests and complex biomarkers. (2008)

products and services might be regulated, and the ethical and legal implications of offering personalised medicine direct-to-consumer versus through a state-funded health care system.

Our approach to regulation

In general, we are supportive of a broadly liberal approach favouring regulation primarily through non-legislative mechanisms, provided that proposed uses can be justified as being acceptable in a democratic society and that proportionate safeguards apply². Where the health and safety of consumers is at issue, we advocate the use of statutory safeguards and, unless there are concerns about the safety of such tests, suggest there should be little restriction to making them available in the marketplace. By contrast, we recognise that where tests are offered by or funded through state providers (such as the NHS or insurers), more rigorous demands should be made of the technologies in terms of effectiveness, cost effectiveness and demonstrable clinical utility, or at the very least, parity with existing technologies. In these circumstances, a higher threshold for regulation may be appropriate.

The formulation of the consultation questions

We have found the formulation of some of the consultation questions to be unhelpful, in that they are not couched in neutral language, or have conflated a number of different propositions which require separate responses. For this reason, in some cases, we have not specifically answered the question as posed, but instead make general comments under each heading.

Consultation questions

1. Health care as a consumer good

- 1.1 The promise and development of personalised healthcare is indicative of both the rise of information technology and the increasing significance accorded to the principle of individual autonomy and consumerism in modern society. As consumers, we are encouraged to take responsibility for our health, fostered by government initiatives such as 'Choose and Book'. As a result, individuals now demand more control over their own health and greater access to information about their individual risk of disease from both genetic and environmental factors. This will necessitate a novel public health response to the promotion of health and the prevention of disease to complement existing approaches to health protection. Positive consequences include more tailored and effective treatments, better management of chronic disease³, and more targeted methods of health promotion. Negative consequences include fears about creating a group of 'worried well' who lack the information to make finely balanced judgments about the significance of information about their health, in addition to the potential ensuing burden created for national health services.
- 1.2 Whilst it is difficult to be wholly independent from, and therefore impartially either supportive or antagonistic of, such a broad and all-inclusive change in society, we believe that such a development is necessitated by the exponential rise in scientific data and knowledge. The resulting transition away from elite groups of 'experts', in whom knowledge and power is concentrated, towards a more egalitarian model, in which knowledge is distributed more widely through society is both an inevitable and welcome consequence. Ensuring that reliable and accurate

² Burke, W., Zimmern, R., Kroese, M. Defining purpose: a key step in genetic test evaluation *Genetics in Medicine* (2007) Vol 9 (10) 675-681

³ BBC News 'Self monitoring device' for HIV 6 May 2009 <http://news.bbc.co.uk/1/hi/health/8034336.stm>

information is available, understandable and easily accessible is therefore paramount, and an essential element in ensuring transparency and safeguarding public trust in science and medicine. As such, the provision of an evidence base may be seen to be a critical activity for the state in protecting the health of its population. In future decades, restricted access to information may well serve as a marker for those groups who also lack access to adequate healthcare.

2. Validity of information

- 2.1 When comparing DNA profiling services with body imaging services, there are several points to consider relating to the potential for *harm* resulting from these services. First, DNA profiling services and body imaging services should be distinguished in terms of the *testing modalities* they employ. Importantly, they differ in their potential for *direct harm* – that is, the harm potentially resulting directly from the test itself, rather than the result of the test. Whilst DNA profiling services generally use non-invasive methods to obtain an *in vitro* sample (e.g. saliva or buccal swab), where the potential for direct harm resulting from taking the sample is almost zero, body imaging services frequently use whole body CT scanning, from which the excess X-ray radiation has been linked with cancer⁴. Therefore, consumers may suffer severe harm through simply undergoing a body imaging test itself.
- 2.2 Second, the *nature of the information gained* from DNA profiling versus body imaging is different. DNA profiling services generally aim to assess the future risk of, or susceptibility to, a particular disease prior to the onset of any pathogenic process. The results are therefore expressed as a probability of whether that individual will go on to develop the disease over a set time period in the future (e.g. 10-years, or entire lifetime). In contrast, body imaging services seek to provide an early diagnosis of current disease whilst the individual is still asymptomatic, thus allowing early treatment to reduce morbidity. Therefore, although the results may still be expressed probabilistically, this simply relates to the predictive value of the test (which can be determined empirically by comparing the ratio of false positive results with those that indicate a true pathogenic process).
- 2.3 These fundamental differences in the type of information gained from each test invite a different response from those offering and taking the test: the former (DNA profiling) is a future prediction that may allow an individual to modify their lifestyle to prevent the occurrence of disease; the latter (body imaging) purports to be a screening test for early detection of disease that may allow the individual to receive treatment to prevent disease progression and the appearance of symptoms. Therefore, the potential for *indirect harm* caused by the results of a test may also be greater for body imaging than DNA profiling services, as follow-on actions may include further testing and intervention strategies in the case of false-positive results, and false reassurance and failure to monitor and treat in the case of false-negative results. (We specifically exclude from this analysis the potential for psychological harm caused by the test result, as this is a complex phenomenon that depends heavily upon individual temperament, and one which is likely to apply equally to both services.)
- 2.4 The differential risks posed by the two technologies outlined in paragraphs 2.1 and 2.2 above support a different approach to the regulation of body imaging services using radiation, for example, from DNA profiling services. In practice, different products will have different risk profiles, and the regulatory response must

⁴ Committee on Medical Aspects of Radiation in the Environment (COMARE), 12th Report, 2007

therefore be similarly nuanced. The mere presence of risk is not sufficient to justify governmental intervention in the form of regulation: there are many risky activities which individuals are exposed to on a daily basis which are not banned, such as bungee jumping or the excessive consumption of alcohol. The broader question of how these services should be regulated is addressed in section 11 on Regulation.

- 2.5 There is a further point alluded to by the title “validity of information”, which is not expanded upon in the question itself: namely, how *scientifically and clinically valid* is the information obtained from these testing services? This point is addressed in section 14 on Quality of Information.

3. Prevention

- 3.1 The extent to which governments or individuals should take responsibility for prevention of harm depends heavily upon the nature of the condition. Where there is an effective intervention (in terms of screening or treatment) which has been shown to reduce or prevent symptoms from occurring, it is arguable that individual(s) should be obliged to have such tests. However, such clear cut cases are rare.
- 3.2 With respect to DNA profiling and body imaging services, until the clinical validity and utility of each test is established, encouraging or expecting individuals to seek out and use such services under the guise of taking responsibility for their health is entirely false. Without clinical validity, these services are potentially no better than fortune telling; without proven clinical utility, these services may be ineffective and entirely inappropriate for the task of improving health and preventing disease.
- 3.3 Finally, there is an important difference between testing services provided by the state, at no cost to the individual, and consumer services for which the individual must pay. In a country with a state funded national health care system (i.e. the NHS in the UK), citizens should not be “expected, encouraged or obliged” to fund tests of proven clinical utility themselves; such tests should be provided (and in some cases, possibly mandated - see above) by the state.

4. Who pays?

- 4.1 Our view is that in a state funded national health service any follow-up tests or interventions resulting from consumer DNA profiling or imaging services should be funded by the state. Currently, there is no bar to citizens requesting medical advice from a publically funded medical practitioner in the absence of symptoms. In practice, individuals seek medical advice for a variety of reasons, including concern over future ill-health. The source of that concern - be it symptoms, family history, personal experience, or information gleaned from magazines, the internet or personal medical testing services - is not, and should not be, a factor in deciding whether to provide support to that individual.
- 4.2 With respect to DNA profiling services, it is likely that in the majority of cases, support from public services will be limited to informal counselling to help the individual understand and interpret the results, although sometimes medical practitioners may exert their clinical judgement in deciding whether to refer patients for further tests. It is therefore imperative that investment is made into training and education of general practitioners and other health care professionals about genetics. The evidence to date suggests that most medical practitioners do not have the necessary understanding to advise patients about disease risk figures that derive from access to such services.

- 4.3 With respect to body imaging services, the majority of cases are likely to include further or repeat testing in addition to counselling, in order to determine whether the individual requires treatment. Follow-on testing is crucial for detecting true positive results and instigating treatment, which clearly falls within the remit of a state funded service. Clinical experience in this area suggests that there may be empirical difficulties in distinguishing between those findings which require more substantive investigation and those which are spurious and carry no clinical significance.

5. Your experiences of online health recording systems

We have no experience of using online health record systems and our comments are confined to a few general points.

- 5.1 Clear and effective policies should be in place to safeguard the privacy of online health records. Users should be notified of any secondary uses of identifiable personal information (including for marketing or medical research) and, where appropriate, informed consent sought.
- 5.2 The adoption of online state (NHS) controlled electronic health recording systems may be difficult to implement within clinical genetics services because, more often than in other medical specialities, a family history may be an essential part of making a diagnosis. Identifiable information about family members may form part of an individual's health record and complex data protection issues may arise if one family member does not wish their personal medical information to be shared with other family members, especially if such a veto is likely to delay or prevent a clinical diagnosis being made in another family member.
- 5.3 Electronic health records systems currently piloted within the UK provide for parts of the record to be sealed and/or locked by the subject. Alternative means of record storage have been proposed that also place the individual citizen in control of their medical records. This could have significant implications for records management within clinical genetics and constitute a barrier to the future implementation of the practice of genomic medicine.

6. Your experiences of online sources for diagnostic purposes

We have no experience of using online sources for diagnostic purposes.

7. Your experiences of purchasing prescription drugs over the internet

We have no experience of purchasing prescription drugs over the internet. However, we note that the abuse of drugs accessed via the internet for onward couriership is a global problem, and in the absence of sufficient legislation, administrative regulations and cooperative mechanisms, current guidance is insufficient.⁵

8. Advertising health care products

- 8.1 Our comments are confined to the advertising of diagnostic and predictive tests rather than the direct advertising of prescription or other therapeutic drugs.

⁵ International Narcotics Control Board Recommendations 2008.

- 8.2 The key principle which we believe should govern the advertising of health care products is one of transparency, with regards to both the evidence base (or lack thereof), and the potential risks associated with the product. Current regulations provide that advertising should not make false or misleading claims. However, a higher standard is required of those products marketed directly to the consumer than via qualified professionals. These regulations have been strengthened recently by generic provisions⁶ which define more closely 'unfair commercial practices' such as misleading claims about the risks or benefits of the product (including false claims that a product 'is able to cure illnesses, dysfunction or malformations'⁷). Difficulties arise in the enforcement of regulations since many trading standards departments are poorly resourced, and staff inadequately trained in these matters⁸. In the field of medical devices and pharmaceutical products, it may be difficult to interpret the web of existing legislation to identify which regulation applies in any particular context, especially where new products are at early stages of development.
- 8.3 Where interventions carry substantial risks, such as the potential risks associated with whole body imaging, these risks should be clearly laid out in a way that is understandable to the consumer. We take the view that to restrict access to such technologies is unduly paternalistic and is not consistent with the marketing of other risky activities in other sectors (such as bungee jumping or parachuting).
- 8.4 The research linking genetic changes (or SNP's) to common complex diseases is expanding very rapidly. Our main objection to advertising DNA profiling direct-to-consumer is that (with a few notable exceptions) the claims are often not adequately substantiated⁹. For this reason, we believe (as argued elsewhere in this paper) that the key to solving this dilemma is not to restrict the availability of such tests (unless they are in some way unsafe) but for government to establish a database of evidence so that policy makers, funders of health services, physicians, patients and citizens can all be reliably informed about the evidence base behind new tests and other health related products.
- 8.5 We recognise that if many more tests are available on a direct-to-consumer basis than are available via the NHS, worries about inequity of care may lead to a lack of trust between patients and health care providers, which could have broad consequences across the whole of health care.

9. Your experiences of information technology to access individual health care expertise

We have no experience using information technology to access individual health care expertise at a distance.

10. Who pays for telemedicine?

The question of whether remote access to GP services should be provided through telemedicine is outside of our area of expertise.

⁶ The Consumer Protection from Unfair Trading Regulations(2008) No.1277

⁷ Regulation 3(4)(d), Schedule 1, paragraph 17.

⁸ However the Trading Standards Institute reports 171 enforcement actions principally in the maintenance and improvement sector over the first year of operation of these regulations.

⁹ Janssens, A.C.J.W et al. *A Critical Appraisal of the Scientific Basis of Commercial Genomic Profiles Used to Assess Health Risks and Personalise Health Interventions*. AJHG Vol. 82, 593-599.

11. Have you used the services of a body imaging or DNA profiling company?

Although we have no personal experience of using either DNA profiling or body imaging services, we have considered at length the extent of the information that should be provided both before and after DNA profiling, which is outlined in section 14.

12. Regulation

- 12.1 Our political system and marketplace economy means that there are many products and services which are promoted and sold direct-to-consumer. A complex matrix of consumer protection legislation and regulation already exists which offers protection to consumers. Since we take the view that products or services involving genetic analysis or material are not necessarily exceptional, we do not support legislation that is directed only at such products and services. Existing legislation, if properly enforced, may in our view be sufficient. However we offer the following clarifications.
- 12.2 We support the current two tier system of evaluation in which higher standards are required for the efficacy of a product when it is funded by the NHS, or a third party payer, than where the product is made available through the marketplace. This is on the basis that resources within the NHS are necessarily limited and thus only services with proven efficacy should be offered. In addition, a certain minimum level of quality is expected from a state funded health system in order to protect patients and safeguard public trust in the system. Within genetics, the UK Genetics Testing Network provides a robust process for the evaluation of genetic tests offered within the NHS prior to commissioning, including determination of clinical utility. The justification for a less stringent approval system outside the NHS is that the costs of testing are not similarly restricted.
- 12.3 However, we believe that the requirements for commercial testing services should be extended beyond their current scope. An important legislative loophole currently exists between the standards for producing testing *kits* sold over-the-counter, and offering laboratory developed tests (LDTs) as a *service* direct-to-consumer. Whilst requirements such as CE-marking and kitemarking relate to the technical reliability, accuracy and labelling of kits, the interpretation as to whether they apply directly to analytical and interpretative medical testing services offered through private laboratories is complex and would benefit from further clarification. The role of the Care Quality Commission in developing an appropriate regulatory structure which can rationalise and strengthen the applicable regulations within this sector is likely to be important in this context.
- 12.4 Medical devices such as kits are regulated by legislation, such as the In Vitro Device (IVD) Directive in Europe (98/79/EC). Whilst there may be concerns about the effectiveness of such regulation (namely that it treats most genetic tests as being low risk regardless of any predictive element), there is consensus that services are significantly less formally regulated, notwithstanding that general consumer protection and advertising standards regulations still apply. Whilst this situation may be relatively unproblematic within the context of national health care systems where both laboratory and clinical services are generally governed by professional bodies and internal controls, the regulatory framework within the private sector is much less well defined.
- 12.5 We therefore suggest as a minimum legal requirement that:
- all laboratories offering medical testing should undergo accreditation procedures and subject themselves to stringent external quality assurance

schemes (as is already the case within the NHS), such that citizens can have confidence in the assay results that are generated;

- appropriate consent procedures are put in place to ensure that clients and customers are clear about what is being offered;
- statutory regulations should also be put in place to ensure that the scientific validity of any clinical claim is real, namely that the link between the disorder and the genetic variant or biomarker has been established to be a true relationship, and thus the claimed association is valid. This is a necessary, though not sufficient, condition of clinical validity and thus should form a bare minimum evidentiary requirement;
- providers should be required to ensure that they have access to appropriately qualified professionals with the necessary competence to interpret the 'measurement' and provide advice and support to consumers regarding the interpretation of the test result. In the case of DNA testing services, this support might in some circumstances include provision for formal genetic counselling, for example, in the case of full genome sequencing, where highly penetrant diseases could be potentially be uncovered in asymptomatic individuals;
- that guidelines and regulations should be strengthened to prevent misleading claims for the product or service, including unsubstantiated and overhyped assertions concerning clinical utility that have no evidentiary basis. We are concerned that although consumer protection regulations should provide this function, in practice the fast pace of scientific development and the complexity of human genetics, coupled with the international nature of the services, means that false claims are not uncommon - suggesting that more systematic enforcement of the regulatory framework in this area is needed.

- 12.6 DNA samples that are collected by the public for private providers may be more susceptible to being contaminated or incomplete than those collected by health care professionals (since they will not be collected and posted to the laboratory by a health care professional or under their supervision). The transmission time to laboratories may be increased if public postal services are used which are more susceptible to delay. The accuracy of DNA profiling by private providers may therefore more error prone and should therefore be subjected to robust quality assurance.

13. Responsibility for harm

- 13.1 A distinction can be made between harm that arises as a direct consequence of the test itself, and indirect harms which arise as a consequence of the way in which the information arising from the test result is subsequently interpreted or used, including use by third parties. The applicable remedies and extent of regulation depend in part upon this distinction, and whether interpretation is offered as part of the testing package, or whether the service is limited to an assay (such as that offered by Illumina). Testing packages may offer a range of services from assay results and limited advice about risk (together with a recommendation to seek further advice from a suitably trained medical professional, which is the case for 23andme) to a session lasting several hours to discuss the significance of the results (such as that offered by the GENAR Institute in Istanbul).
- 13.2 There is a lack of congruence between the marketing employed by companies offering these tests (which appeals to consumers to take positive steps to improve their future health) and the contractual terms and conditions which are required to be signed by the consumer before kits are dispatched. The latter often contain

statements which seek to limit the company's liability for consequential losses, including statements that the consumer will not rely upon the results of the tests in making future decisions about their health and/or will seek further advice from a health professional if there are results that require clarification. As far as we are aware, the validity of these contractual statements have not yet been tested in court proceedings within the UK.

- 13.3 Providers undoubtedly have a duty to analyse the samples and report on the results with due care and skill. If failure to exercise such skill results in false results (such as incorrect sequencing of a particular genetic variant) that are then reported, then providers should be held responsible (since the harm is both foreseeable and proximate). There should therefore be procedures in place to validate tests where the results are either highly predictive, such as those for monogenic late-onset disorders (e.g. Huntington's disease) or where a positive finding is likely to result in treatment or intervention including finding highly penetrant susceptibility mutations (e.g. *BRCA* genes, where the test result may result in elective bilateral mastectomy).
- 13.4 With regards to tests with limited predictive value and clinical validity, such as those currently offered by DNA profiling services, we believe that normal statutory rules interpreting foreseeability and causation should apply. Providers should rarely be responsible for any consequential harm to the individual caused as a result of the test, provided that a proportionate set of consent procedures are in place such that the citizen is unambiguously informed about the nature of what he or she will receive by way of information and its possible implications. We recognise that there may be practical difficulties for providers in ensuring that those using DNA profiling via the internet are competent to consent (such as where non-competent minors seek to access such information).

14. Quality of information

- 14.1 The "quality and usefulness" of the information is related to the clarity with which it is presented to the consumer, so that he or she can understand and interpret it correctly. Probabilistic information is inherently challenging to communicate, so careful consideration must be given to presenting it in such a way that it is not only technically accurate but also understandable by the lay person. In addition to explaining the testing process and the results of the test, the limitations of the information should also be stated to avoid false reassurance.
- 14.2 The "quality" of the information is additionally related to:
- the *analytical validity* of the test itself, i.e. how accurately the assay measures or detects its target or biomarker (e.g. accuracy of gene sequencing);
 - the *clinical validity* of the test, which includes the *scientific validity* of the claimed association between the biomarker itself and the disease of interest (e.g. strength of evidence proving the link between a genetic variant and predisposition to a particular disease) and the *performance of the test* itself prospectively in the target population.
- 14.3 The "usefulness" of the information is related to its *clinical utility*, which in turn depends upon:
- the availability of proven intervention strategies that can effectively prevent or treat the disease;
 - a difference in the recommended course of action between different test results;

- the ability of the test result to empower the individual to make lifestyle changes or seek treatment to reduce his or her risk of disease (for which there is currently little evidence in relation to DNA profiling services).
- 14.4 Whilst a test without proven clinical utility should not be used by a state funded health care system, where the highest evidentiary standards are required to ensure a fair distribution of resources, there seems no reason why such a test should not be allowed on the free market. Consumers can purchase countless goods and services of questionable (or even negative) utility, and we believe that purchasing unhelpful, useless or irrelevant health care tests is no different.
- 14.5 In contrast, tests that purport to offer medically relevant information but are based on incorrect science, and have no clinical validity, should simply be viewed as fraudulent and not allowed on the market.
- 14.6 Information regarding the scientific and methodological basis for the tests should be made available to those who want it. We have previously recommended that a publically available database of tests should be provided centrally, containing evidence of clinical performance as far as that evidence is available. Where evidence is missing, particularly evidence of clinical validity and clinical utility, this should be explicitly stated¹⁰.

15. Other issues

- 15.1 We wish to highlight three further issues in relation to personalised healthcare in the consumer age: first, the testing of minors; second, the enforcement of regulations in a global market; and third, the issue of genetic exceptionalism.

Testing of Minors

- 15.2 The European Society of Human Genetics (ESHG) has recently recommended that the genetic testing of asymptomatic minors for adult onset diseases, prior to the age of consent, be restricted to those conditions where preventative actions cannot be deferred until the child is mature enough to understand the decision and its consequences¹¹. The justification for these recommendations is that delaying testing protects the autonomy of the future adult and tends to prevent parents from imposing additional pressures on the child. Where conditions arise in childhood, the recommendations distinguish between those for which an effective treatment or prevention is available (where the case for testing may be compelling) and those for which there may be no available treatment or prevention (where the harms and benefits of testing may be more finely balanced). Importantly the ESHG did not exclude such tests if a case for psychological or social benefit of the child can be made. It also recommended that that genetic counselling should be a requirement for genetic testing of all minors. We would therefore suggest that direct consumer DNA profiling companies make every effort to prevent their services being sold to minors.

Global Regulatory Enforcement

- 15.3 The difficulties of enforcing regulations regarding access to medical tests in a global market, where the consumer, the assay provider and the interpretative services may be located across multiple jurisdictions, are likely to increase as the consumer healthcare movement develops. Although some countries have tried to

¹⁰ PHG Foundation. The evaluation of diagnostic laboratory tests and complex biomarkers. (2008)

¹¹ European Journal of Human Genetics (2009), Vol. 17, pp 720-721

ban genetic tests except those obtained through a medically qualified physician¹², we believe that such regressive legislation is neither warranted (in the case of services with limited clinical validity and utility) nor sustainable. We therefore support efforts to draw up voluntary codes of practice, such as the Common Framework of Principles for Direct-to-Consumer Genetic Tests currently being drafted by the Human Genetics Commission.

Genetic exceptionalism

- 15.4 We are broadly against the notion that genetic information is sufficiently different from other kinds of health-related information to warrant special protection¹³. As the science of human genetics develops, it is becoming increasingly clear that genetic determinism is a fallacy for all but a few very rare disorders. The development, maintenance, regulation and translation of the genome is a highly complex and dynamic process, and the 'one gene, one protein, one function' idea of the late 1990s is now entirely defunct. Moreover, the 'two-bucket' theory of disease - that diseases can be dichotomised as being either genetic or not - has also been rejected as being overly simplistic, now that countless genetic predispositions have been discovered for common multifactorial diseases once thought of as being caused by environmental factors.
- 15.5 We therefore support that notion that genetic information should be treated the same as any other medically relevant information, and that it should be subjected to the same levels of protection and privacy. Like other medical information, some of it is visible and public (e.g. gender, height, ethnicity, etc.), whilst some is hidden and highly personally sensitive (e.g. *BRCA* status, diagnosis of cancer, treatment details, etc.). The information itself should therefore be regulated in proportion to the level of its sensitivity, relevance to family members and clinical utility, rather than the nature of the test analyte (i.e. DNA) dictating the degree of regulation imposed.

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¹² German Resolution of the 14th Committee, Gendiagnostikgesetz – GenDG (2009)

¹³ Thomas Murray, Genetic Exceptionalism and “Future Diaries”, in Genetic Secrets, Ed. Mark Rothstein (1999)