PHG Foundation

# Give and take?

# Human Bodies in medicine and research

# Response from the PHG Foundation

## Introduction

The Foundation for Genomics and Population Health (PHG Foundation) is an independent international non-profit organisation working to achieve the responsible evidence based application of bio-medical 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

*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 apply[1]. Thus in general we favour a regulatory environment that recognises the utility of human bodily material (and in particular the secondary use of data derived from human bodily material). Such data is crucial in public health epidemiology, where the obligation to obtain consent from every potential participant or anonymise would otherwise be impractical, prohibitively costly, exclude vulnerable groups, and likely to result in flawed results.

*The importance of human bodily material in secondary epidemiological research*

The systematic collection of human tissue samples through disease registries or biobanks has had a profound impact on our understanding of health and disease. Over the next few years, the integration of environmental and lifestyle information in biobanks and cohort studies has the potential to generate new knowledge about the risk factors and course of common complex diseases. The importance of secondary research for these purposes should not be underrated.

*The scope of this consultation response*

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

Whilst there are many consultation questions which are potentially relevant to our work, we wish to confine most of our comments to those questions which relate to the analysis of genetic material from tissue. Although the consultation paper explicitly rejects a legalistic interpretation of the term 'human bodily material', the glossary clarifies that the terms of reference of the consultation only relate to cellular material (and not extracted genetic material). However, it is clear that where tissue is held for the purpose of genetic testing to assist relatives that this will be included within the scope of the consultation. (Indeed one of the examples given in section 2 of the Consultation response is the provision of human material for genetic testing to benefit relatives).

It is significant that the supporting text in the consultation paper does not acknowledge the complexity of the legal framework that currently applies to the genetic testing within the UK. Indeed, section 45 (and schedule 4) of the Human Tissue Act 2004 provides for a regulatory framework to apply to tissue which is held for the purpose of analysing human DNA in the material with qualifying consent (unless those results are used for an excepted purpose).[2]

**Consultation questions**

**1. Are there any additional types of human bodily material that could raise ethical concerns?**

The retention, storage and use of human bodily material specifically for the purpose of genetic and genomic analysis might raise potential ethical concerns, particularly if genetic information generated from that test is sensitive or predictive, and can be used to diagnose existing disease or predict future ill health.

**2. Should any particular type(s) of human bodily material be singled out as 'special' in some way?**

Genetic exceptionalist arguments are commonplace. However, we do not subscribe to the view that all genetic material and information is deserving of special protection. Indeed the vast majority of genetic information is uninformative and not predictive, given our current state of knowledge. Indeed our current state of ignorance about the vast majority of the genome confers additional problems (see Q30).

**3. Are there significant differences between providing human bodily material during life and after death?**

Cultural and ethnic groups may place differing values on human bodily material depending upon whether it is derived from the living or the dead. We support the existing regulatory framework that allows a competent adult to make an autonomous decision about the fate of his or her body after death (as codified in the Human Tissue Act 2004). Whilst we recognise that in practice, family members may oppose these

[2] Human Tissue Act 2004 Schedule 1 Part 1 paragraph 4.

decisions, we consider that, subject to limited exceptions, the view of an individual about the fate of their own body should be pre-eminent.

**4. What do you consider the costs, risks or benefits (to the individual concerned, their relatives or others close to them) of providing bodily material?**

4.1 The genetic testing of individuals within families can sometimes raise difficult ethical issues, particularly where family members disagree about whether they wish to have a genetic test that might reveal predictive or diagnostic information. This is particularly problematic where the identification of a genetic mutation in one family member effectively confirms the presence of that condition in another family member (who has refused testing). Balancing the autonomous interests of family members is an important part of clinical genetics services. Thus the genetic testing of human bodily material can carry significant costs and benefits if this is used to make a clinical diagnosis or predictive risk assessments about future disease status.

4.2 The experience of clinicians suggests that in many cases, family members would wish relevant genetic samples and data to be available to their family members, if that would assist in the process of making a diagnosis or clarifying the risk status of other family members. Some genetics units make a point of discussing these issues with their patients as a matter of routine, and documenting these discussions in a pro-forma consent form which records the willingness of the patient for their tissues and data to be used for the benefit of family members.

**6. Are there any additional purposes for which human bodily material may be provided that raise ethical concerns for the person providing the material?**

As outlined above, the use of human bodily material for the purpose of predictive genetic testing might lead to fears of discrimination or stigmatisation. There is a need for ongoing empirical research to substantiate these claims.

**15. Should different forms of incentive, compensation or recognition be used to encourage people to provide different forms of bodily material or to participate in a first-in-human trial?**

See Q4 above.

**16. Are there forms of incentive that are unethical in themselves, even if they are effective? Does it make any difference if the incentive is offered by family or friends, rather than on an 'official' basis?**

Where an incentive is offered by family, depending upon the circumstances of the case, it may be more, rather than less difficult for an individual to refuse. Where health care professionals are involved, they should use their clinical judgement to ensure, as far as they can, that the decision is freely made.

**21. In your opinion are there any forms of encouragement or incentive to provide bodily material or participate in first-in-human research that could invalidate a person's consent?**

Where the incentive is deemed to be 'undue influence' it could be considered to have invalidated the nature of the consent given. There are legal precedents which are helpful, but ultimately many cases have to be determined on their merits.

**22. How can coercion within the family be distinguished from the voluntary acceptance of some form of duty to help another family member?**

The Human Tissue Act has established a system of independent assessors whose role is to independently assess potential tissue donors and recipients to guard against the possibility of coercion or undue influence. This provides some degree of protection against exploitation.

**23. Are there circumstances in which it is ethically acceptable to use human bodily material for additional purposes for which explicit consent was not given?**

23.1 Our health care system is predicated upon evidence based research - yet obtaining evidence about the clinical and scientific validity and utility of the natural history of diseases, and thus the efficacy of an intervention is often problematic. Thus we support the use of human bodily material for purposes such as public health monitoring, clinical audit and certain types of epidemiological research, the results of which guide the development of more effective health services, particularly public health interventions. This is particularly important where the use of an opt-out or opt-in system of consent might otherwise exclude marginal groups or undermine the quality of data.

23.2 We are supportive of initiatives that combine the use of technological devices (to de-anonymise personal data) together with obtaining implied consent from individuals at GP practice level for example, to widen the scope of data that can be accessed within existing regulatory frameworks.

**24. Is there a difference between making a decision on behalf of yourself and making a decision on behalf of somebody else: for example for your child, or for an adult who lacks the capacity to make the decision for themselves?**

Within clinical genetics, the prevailing view is that predictive genetic testing of asymptomatic minors for adult onset diseases, prior to the age of consent should be restricted to those conditions where preventative actions cannot be deferred until the child is mature enough to understand the decision and its consequences[3]. This is justified on the basis 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, most professional 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). Where decisions are made on the part of an adult who lacks capacity to make a

[3] European Society of Human Genetics, European Journal of Human Genetics (2009), Vol. 17, pp 720-721

decision for themselves, part of the family's role might be to provide a 'substituted judgement' about what that person might have decided for themselves had they been able to.

**25. What part should family members play in deciding whether bodily material may be used after death (a) when the deceased person's wishes are known and (b) where they are unknown? Should family members have a right of veto?**

25.1 Our view is that when the wishes of a deceased person are known that these should prevail over the preferences of family members. Where the wishes of a deceased person are not made explicit, family members may play an important role in clarifying what they believe the preferences of the deceased person might have been (a form of substituted judgement).

25.2 Family members have a statutory right under the codes of practice to the Human Tissue Act 2004 to provide a continued consent to retention of human bodily material from the deceased after Coroner's post-mortem[4]. In the absence of consent from family members (including those who have a social rather than a genetic relationship with the deceased), the default position is that human bodily material is destroyed rather than being retained, which could compromise the interests of other genetically related family members who might rely upon testing of samples from the deceased for mutation analysis. We are hopeful that the introduction of medical examiners pursuant to the Coroners and Justice Act 2009 might in time provide an expert genetics resource to coroners, and might prevent valuable samples from being needlessly destroyed as a result of the disjunction between Coroners' and Human Tissue legislation.

**26. To whom, if anyone, should a dead body or its parts belong?**

Our preference is for the existing situation (i.e. that there is no property in a dead body) to continue - with the caveat that samples of human bodily material taken for diagnostic purposes - should be regarded as part of the deceased person's medical record after death, as the Scottish human tissue legislation provides.

**28. Should companies who benefit commercially from others' willingness to donate human bodily material or volunteer in a trial share the proceeds of those gains in any way?**

The ethical principle of equity suggests that in some contexts it may be appropriate to acknowledge the significance of donations of human bodily material, for example in a cohort study or clinical trial in a low or middle income country, by devising innovative ways of providing some recompense for the time spent participating in the

[4] See for example Human Tissue Authority. (2009) Code of Practice Disposal of Tissue, paragraphs 50-55
http://www.hta.gov.uk/legislationpoliciesandcodesofpractice/codesofpractice/code5disposal.cfm?FaArea1=customwidgets.content_view_1&cit_id=721&cit_parent_cit_id=713

research. However, if those proceeds are distributed in the form of payments to individuals, care is needed to avoid the charge that they are potentially coercive.

**29. What degree of control should a person providing bodily material (either during life or after death) have over its future use?**

We continue to believe, contrary to current legislative policy, that it is not always appropriate for a living individual to retain an absolute veto over the use of their tissue, if the analysis of genetic material in that tissue might be used to avoid serious illness or death in a family member, for example for mutation analysis. In such circumstances, we advocate the use of a civil court to decide whether genetic testing should proceed against the wishes of the individual. We envisage that such a procedure should only be used as a last resort, where no other sources of tissue or data are available, and when the analysis of material from a relative might yield important information that might guide treatment and avoid serious illness or death.

**30. Are there any other issues, connected with our Terms of Reference, that you would like to draw to our attention?**

The combination of new technologies (next generation sequencing machines) together with new research methodologies which involve systematic data sharing, integrating genomic, environmental and lifestyle information, and making whole genome sequences publically available, has a number of implications. It might be more difficult to predict what the harms and benefits associated with research or whole genome sequencing might be, and as a consequence of this, the use of consent as a tool to legitimate the entire process might be increasingly problematic. Similarly novel data mining techniques may render conventional forms of anonymisation less secure. These advances suggest that new approaches may be needed to respect the interests of research participants in addition to the existing use of consent and/or anonymisation.

PHG Foundation
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