HeLEX
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ETHICAL, LEGAL AND SOCIAL ISSUES ARISING FROM THE USE OF GWAS IN MEDICAL RESEARCH

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INTRODUCTION

This literature review was commissioned by the Policy Unit in the Wellcome Trust to identify the key issues that have been raised by the use of genome-wide association studies (GWAS) in genomics research.

The use of GWAS in genomic research gives a new twist to the issues of consent, feedback of results, privacy, the governance of research and property rights. This is because GWAS:

- produces fine, detailed, sequence information at a resolution not possible before, and the same results potentially can be used to determine an individual's genetic variation for a range of conditions;
- the information from a GWA scan is derived from DNA (deoxyribonucleic acid) that is a unique personal identifier, which can not only provide information on the individual, but also on the individual's relatives, related groups and populations;
- this technology is becoming cheaper, more available and more powerful;
- GWA creates large amounts of individual-specific digital information that is easy to share across international borders, and can potentially be used for a number of different research purposes.

The GWAS methodology is being used within a research context where there is an increasing accumulation of samples and information being held in biobanks, or stored for use in hypothesis-led projects, and in large international consortia. This has led to the creation of 'networks within networks' where information can be compared, used and exchanged between researchers. This is necessary to support the developing scientific agenda and the recognised need for large sample sizes in order to understand the genetic basis of common diseases. Funders have been instrumental in facilitating and supporting such trends, with the creation of genomic reference libraries such as the Human Genome Project, and the introduction of data-sharing policies to facilitate the secondary use of datasets, with the aim of increasing scientific advances. It is also likely that in the future, scanning of an individual's whole genome will be possible for a small cost, due to better and more efficient technologies.

The social context in which GWAS is being used is also changing very quickly and developments in medical research are intimately bound up with current trends in society. More and more information is being placed on the web, through social networking sites such as Facebook. Individuals have greater access to their own genomes through companies such as 23andMe, deCODEme, DNA Direct and Navigenics, as well as ancestral tracing companies. In addition, there is an increasing concern in society about the way that personal information is being collected and used for surveillance purposes, but also mishandled by government agencies within the UK (Whitely, 2008). These events outside of medical research cannot be ignored, as they could potentially have an effect on the medical research context, which in the case of an unfavourable event could undermine the public trust and support that is necessary for medical research to continue and to thrive.

The scope of this literature review

We have focused on eight key issues – consent, feedback of incidental findings, data sharing, privacy, governance of research, property rights in the body, intellectual property rights and public engagement – in order to identify the ways in which GWAS and data sharing change these issues. We have identified the key papers that have discussed GWAS and attempted to bring out the ethical, legal and social issues that they raise. For the legal research, we have focused on UK law, and updated the comprehensive research that was carried out by Cara Tetlow at the end of 2008 and presented in two memos to the Trust. These are entitled 'Human Tissue Act and consent' and 'Ethical and legal considerations with respect to reporting back the results of research using DNA data'. This update reflects the UK cases decided in the first two months of 2009, as well as adding some additional key decisions that have a bearing for the use

of GWAS. The focus is on the law that relates to privacy and data protection and we have not covered the Human Tissue Act in depth, as acellular, subcellular and genetic materials – such as extracted DNA – all fall outside the definition of 'relevant material' and the scope of the Act. We have focused on the law in this way because the sharing of GWAS and secondary research is largely concerned with digital information rather than DNA. Also, we have not attempted to cover the plethora of guidelines in this area due to the time available, although we recognise that these are intrinsic to any judgment that a court would make in deciding a negligence case.

This report is not comprehensive, due to the time that we had available and the fact that this is a fast moving area. There are a number of references to unpublished work or papers in press that the authors have written in this area. This piece of research was carried out over eight weeks from the beginning of February 2009 and submitted to the Wellcome Trust on Monday the 23rd of March 2009. Dr Jane Kaye has managed and co-ordinated this research and is responsible for the final content and format. Dr Heather Gowans carried out the research to update the legal analysis of the Wellcome Trust memos and Dr Karen Melham carried out the wide-scale literature review to provide the background for the written research. A meeting was held with Dr Jane Kaye, Dr Paula Boddington, Jantina de Vries, Dr Heather Gowans, Naomi Hawkins, Dr Catherine Heeney and Dr Karen Melham to discuss these findings. Various sections were allocated to each member of the team to write up.

Our final caveat is that this paper cannot be construed as legal advice. Professional legal advice should be obtained before taking or refraining from any action as a result of the contents of this document.

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A. CONSENT

A.1 Summary

Genomic research, including genome-wide association studies (GWAS), raises a number of difficult issues in relation to informed consent. The notion that informed consent is needed from participants in research arose originally in the context of biomedical research, and in the context of protecting participants from abuse and the potential physical harms of such clinical research. It was therefore focused strongly on the protection of individual rights. Genomic research, however, tests this on at least two major counts.

First, the potential physical harms of clinical trials are less relevant for GWAS than the potential harms of infringements of privacy and related questions concerning the use of genomic information. It is questionable how the traditional framework of informed consent can accommodate these concerns. These concerns are particularly heightened by the unprecedented, detailed level of information that is produced by GWAS, the international nature of genomics research meaning that this information is shared globally; as well as the rapid developments in technological possibility in GWAS.

Second, the traditional notion of informed consent focuses on the individual whereas in genomic research there are large issues not just for the individual participant but also for family, community, and population groups. Indeed, discoveries made through GWAS giving detailed information about single nucleotide polymorphisms (SNPs) have only underlined the ways in which information about individuals is relevant to other related individuals and groups and vice versa. There is as yet no consensus of opinion about the best way forward, although there is widespread consensus that further debate and empirical and philosophical work are needed.

A.2 Context to the debate about informed consent in GWAS

In examining informed consent to genomic research and data sharing, there is much work in related fields that provides a potentially illuminating resource: in the fields of clinical medicine and biobanking, in research ethics in general, within clinical genetics, and in social science research.

There is a large amount of literature exploring problems with the notion of informed consent to medical treatment, and inherent difficulties in the understanding and application of the concept. An appreciation of these concerns is useful to place debate about genomic research and data sharing in its context (see e.g. O'Neill, 2003).

There is also a large body of literature that raises the issue of informed consent in medical research in general. Some of this examines the theoretical bases of informed consent in the clinical context and the notions of autonomy upon which it traditionally rests. Other work looks more specifically at the development of the notion of informed consent in the history of research ethics. As with much critical discussion within medical ethics, the individual basis of informed consent is one area of critique, and one that has key relevance to genomic research (see e.g. Appelbaum *et al.*, 1987; Emanuel *et al.*, 2000; Rhodes, 2005).

In the field of biobanking there has been a raging debate as to whether broad consent can be used for biobanks because of the difficulty of informing individuals about all of the uses of the data when this is not known at the time of collection. In practice, biobanks seek a broad consent and then rely on governance mechanisms, such as approval by a research ethics committee, for further uses of the samples and information. In population biobanks, the rationale has been that it would involve disproportionate effort to re-contact many thousands of participants for every new research use and that the research involves minimal risk (Caulfield and Kaye, 2009). While a broad consent may be appropriate for access to one biobank for a specific research purpose, the oversight and governance mechanisms that are required to oversee research are severely

stretched when it comes to data sharing. Issues concerning the governance of networks within networks are only just starting to be addressed by the biobanking community.

There is also by now a very large body of literature that examines communication with individuals and families in the genetics clinic and related medical contexts. This valuable body of expertise is of course highly relevant to questions of consent in GWAS, as it relates to questions concerning lay understandings of genetic concepts and of disease and health. This literature therefore could be an important resource in looking at the information provided to research participants. Additionally, some of the literature on genomic research is beginning to address the implications of research participation for family members. Some have suggested making recommendations to participants to discuss research with family members as an adjunct to the consent process. There is an extensive literature on family communication of genetic information upon which such policy recommendations could usefully draw (Forrest *et al.*, 2007, 2008; Gaff *et al.*, 2007).

Moreover, there are important questions being raised about the role of informed consent in social science research, especially in areas such as ethnography. Notwithstanding the differences between social science and genomic research and data sharing, there are notable similarities. For instance, both must address the ease with which individuals may be identified or characterised by their social or genomic information, and the ways in which both areas of research have key implications for groups and related individuals, not just for the individual who is at centre stage in all traditional discussions of informed consent. This body of social science literature therefore could also provide a resource for exploring issues within genomics (Atkinson, 2009).

A.3 Legal issues around consent

Consent Requirements

The requirement of informed consent is enshrined in the Declaration of Helsinki and now applies in relation to clinical trials carried out in the UK under the Statutory Instrument 2004 No. 1031 The Medicines for Human Use (Clinical Trials) Regulations 2004. However, in the UK there is no other legal instrument that stipulates the consent requirements for other kinds of medical research. The UK has not signed or implemented the Convention on Human Rights and Biomedicine 1997, nor does it have a Patients' Rights Act as do other countries within Europe. The Human Tissue Act, while not defining consent, lays down in the Codes of Practice the requirements of consent for the use of tissues in research. Consent is required for the disclosure of confidential information, but Common Law does not define what the nature of this consent should be – it may be implied and does not necessarily need to be in writing (*Hunter v Mann* [1974] QB 767, 772).

Under the Data Protection Act 1998, 'explicit consent' is the standard for consent to the processing of sensitive data such as health data. The Act does not stipulate, however, what 'explicit consent' is, nor what the content of explicit consent should be. Directive 95/46/EC – from which the UK Data Protection Act is derived – defines consent as 'any freely given specific and informed indication of his wishes by which the data subject signifies his agreement to personal data relating to him being processed'. Based on this, the Information Commissioner requires that:

- The consent must be informed; that is, the data subject must know what the proposed uses or disclosures of data are.
- The person must have some degree of choice and consent must not have been obtained under coercion.
- There must be some indication that the person has given consent – though this may be express (active agreement expressed orally or in writing) or implied (from the behaviour of the individual). For instance, asking the doctor about a diagnosis gives implied consent to look at the medical record.

However, explicit consent is not required for all legitimate processing.

The view of the Information Commissioner is that information given to patients should:-

- 'provide sufficient information to allow the individual to assess the risks to him or her in providing their data, in consenting to their wider use, in choosing not to object to their processing etc. This should have at least two consequences for data controllers. It should become clear that fair processing notices do not need to contain a large amount of detail about routine, administrative uses of data. It should also become clear that researchers engaged in open-ended studies are not prevented by the Act from soliciting patient data on the grounds that their fair processing notices cannot be sufficiently detailed. Fair processing notices in this case should simply need to make clear that the research in question is indeed open-ended, leaving the individual to assess the risk' (Information Commissioner, 2002).

According to the guidance, a poster in a waiting room or surgery would not be considered to meet the requirements of the Act unless it is supplemented with other forms of information such as a face-to-face consultation, a leaflet or a letter sent to the patient's home. 'Doctors may be able to easily provide specific information to patients in the course of consultations. Only where such an opportunity does not present itself will it be necessary to contact patients separately, for instance, if they are to be invited to participate in a programme of research involving the disclosure of their medical records to a researcher who may wish to interview patients with particular medical conditions.' (Information Commissioner, 2002; Kaye, 2006)).

Exceptions to consent

Under s. 33 of the Data Protection Act, consent is not required for research purposes if the data are not processed to support measures or decisions relating to particular individuals and the data are not processed in such a way that substantial damage or substantial distress is, or is likely to be, caused to any data subject. This means that data can be used for other secondary research uses without consent and can be kept indefinitely. However, there is still a requirement that individuals be told about the use of their data. Individuals do not have a right of access to the data as long as the data do not identify the individual. The Human Tissue Act also has a number of exceptions from consent as long as tissue is anonymised and has the approval of a research ethics committee.

A.4 Key challenges in informed consent in genomic research and data sharing

There are concerns raised that informed consent is being stretched to its limits in the context of genomic research; that means of enhancing informed consent should be sought to account for the particular case of genomic research; and/or that alternatives to traditional concepts of informed consent be explored. The issue of informed consent is also inextricably entwined with other issues, including privacy of research participants, ethical oversight of research, and feedback of incidental findings. Much of the literature available draws on experience of population studies that have already been conducted, for example in Iceland and in Sweden, and in the populations drawn on in the HapMap project (Annas, 2000; Arnason, 2004; HapMap, 2004; Rotimi *et al.*, 2007). The following analysis picks out some of the key questions raised by GWAS, recognising that many of these issues are interrelated.

The theoretical basis of informed consent:

There are potential tensions in the literature on informed consent in genomic research that result from fundamental conceptual differences. In some views, informed consent is chiefly a mechanism to protect participants from harms; in other views, it respects autonomy and provides a means for participants to exercise control over their data and samples (McGuire *et al.*, 2008a). These different emphases could lead to different policy outcomes in circumstances where harm could be ruled out. However, debates about the possibilities of harm, and privacy protection in GWAS, for instance, show that the assessment of potential harms is in itself contentious. It depends upon an appraisal of the technology of GWAS and information

management; an appraisal that is ongoing and also contentious. Recommendations have been made for thorough risk benefit analyses in GWAS (Caulfield *et al.*, 2008).

Future uses of data and samples:
There is general concern that the speed of technological and scientific progress in genomic research means that it is hard to anticipate future uses of data and samples, making it impossible fully to inform participants. Indeed, such is the speed of change in research protocols that even recently, where a few points of data in the genome might have been produced, GWAS are now mapping a million SNPs, and routine sequencing of entire genomes is on the horizon. This is a difference of degree of such a scale that arguably fresh approaches to the management of this information, including issues around consent, are needed.

How much information is needed for consent to be adequately informed?
This is a general issue in the informed consent literature rendered particularly pressing by genomic research. Not only is there a large body of information that could be imparted, but also there is much that is unknown: for example, about the significance of possible findings, about future directions of research, and, with data sharing, about who will be conducting research and what research questions and applications will be. GWAS have exacerbated these issues as it is now plainly apparent that future uses cannot adequately be anticipated and that the notion of 'fully informed' consent is untenable (Caulfield *et al.*, 2008). The question remains, however, of how much detailed and specific information should be given for GWAS.

Archived data and retrospective informed consent
There is concern to balance different values in research ethics; one value balanced alongside protection of participants is that of the importance of the research endeavour and maximum use of public funding and resources. This includes the use of archived data, and there is discussion of the interpretation of original consent to research and use of data and samples in future projects. The need or not to obtain re-consent from patients for such uses is also raised in the literature.

Consent as an initial step versus consent as a process and the question of re-consent:
Traditionally, informed consent has been seen as an initial step to take before embarking on research: in various contexts, the limitations of this have become apparent. The ongoing use of genetic data and samples, especially given data sharing and the context of rapidly changing possibilities and understandings in genomics, has meant that the notion of consent as a process that needs to be repeated has been raised (Fitzpatrick *et al.*, 1999; Hunter *et al.*, 2001; Ormond *et al.*, 2004; Caulfield *et al.*, 2008).

Trust
Much of the literature, both theoretical and that based upon empirical research with participants or potential participants, underscores the importance of trust in the research process and in the research community, both as a means of ensuring ongoing support and recruitment, and as an ethical value to respect the participants and the larger community. Informed consent can be a way of attaining such trust; alternatively, problems with gaining fully informed consent in the context of genomic research have led to calls for mechanisms for ensuring trust in the oversight of such research (Hoeyer *et al.*, 2004).

Broad consent/ exploration of new mechanisms to protect participants
As a response to difficulties, there have been many suggestions made that consent to genomic research including biobanking should be on the basis of blanket consent to an open set of future uses of research. However, problems have been raised with this: is this really informed consent in the original meaning of the term, and is it compatible with other requirements such as data protection legislation? There have been proposals for adjunct protection, given that the limits of informed consent in this context are recognised. Robust governance structures that may be used alongside the ethical protection of consent to management of samples and data, which can

demonstrate key virtues such as accountability, trust, fairness, and transparency, need to be explored but there is little consensus on the nature of such structures as yet (Caulfield *et al.*, 2003; Winickoff, 2007; Caulfield *et al.*, 2008; Lunshof *et al.*, 2008; McGuire *et al.*, 2008b).

Data sharing and the management of informed consent
The sharing of data raises new questions, including some that have already been raised, such as gaining informed consent where potential research partners are not specified. There are other issues raised including the distance of the users of the data from the original data collection. The possibility of a 'chain of obligation' amongst researchers has been mooted (Caulfield *et al.*, 2008), but needs further exploration given the length and geographical and temporal spread of this chain in GWAS.

Informed consent and commercial uses
Data sharing in genomics means that data may contribute to ultimate commercial applications. This can raise issues where original terms of consent either did not mention this, or more importantly, where it was explicitly excluded. The rise of personal genomics companies also raises interesting issues for informed consent, since some of these companies are using their samples in research, in contexts outside of public institutions (Merz *et al.*, 2004; Haddow *et al.*, 2007; Hawkins *et al.*, 2009).

Informed consent and inducement; informed consent and benefit sharing
It is a general principle that informed consent is made freely and without inducement. However, in different cultural contexts, different rewards for research might constitute inducement. This is important when considering genomic research which makes use of samples from many different cultural and geographical contexts. Questions of feeding back benefits to populations, especially where they are deprived, cannot be separated from questions of coercion and inducement to take part in research.

Consent and family members
The nature of genomic information means that it has potential implications for family members. The potential impact on family members has been raised in the context of informed consent, with many suggesting that participants be advised to discuss research participation with their family. However there seems to be scant attention paid as yet, to the difficulties that this might create. Some have recommended what is called a 'family centred approach' to informed consent (McGuire *et al.*, 2008b), but not only is this in tension with the individual basis of traditional notions of informed consent, but the details remain to be worked through, for example, whether this could overrule traditional informed consent.

Population issues, community consent
The nature of genomic research means that populations are implicated: for example isolated or distinctive populations may be identified by research; there are ethical issues about identifying and labelling populations; methods for obtaining consent from individuals may be culturally inappropriate in different areas; issues of group stigma have to be considered. In some contexts, obtaining community consent may be seen as an adjunct or alternative to individual consent (Beskow *et al.*, 2001; DeCamp and Sugarman, 2004; HapMap, 2004; Rotimi *et al.*, 2007).

Geographical context of informed consent
This has already been touched upon: data sharing, the nature of genomic research, and the widespread international collaboration in research, together with the need for large sample sizes, means that cultural and geographical considerations must be taken into account, including of course the legal requirements of different jurisdictions. Issues of global health justice are becoming increasingly discussed in many contexts including the bioethical literature and fairness, consistency, attention to contextual issues, and due process in the obtaining of informed consent are part and parcel of these concerns. The communication with diverse

populations, who vary widely in literacy, education, and familiarity with medical research, is a pressing concern (Chokshi and Kwiatkowski, 2005).

Informed consent and disease and patient context
As well as geographical context, genomic research on different diseases will raise different issues depending on the particular context including the nature of the disease and the context of collection. For example, research protocols that necessitate the collection of samples from a child undergoing a malaria episode raise quite different issues from the collection of samples from healthy controls. Likewise, certain patient and population groups are more vulnerable than others, such as children (Knoppers *et al.*, 2002; Chokshi and Kwiatkowski, 2005).

Informed consent and withdrawal from projects
It has traditionally been an integral part of informed consent to research that participants are informed that they can withdraw from research at any time and without penalty. However, the context of data sharing and genomic research has led many to question whether such withdrawal is realistically possible - and indeed whether it is actually fair on the research process, given the difficulty of ensuring this. Conversely, other literature has been concerned with the issue of how to ensure that such withdrawal is meaningful and practically possible (Caulfield *et al*., 2008).

Feedback of findings and informed consent:
The debate on the feedback of findings from genomic research of course has clear implications for consent, as, should feedback occur, it is arguably the case that this possibility should be part of the information given on consent.

Privacy and informed consent:
The protection of the confidentiality of patients, and likewise of research subjects, has always been a key component of informed consent, unless there are exceptional circumstances and where participants are fully and explicitly informed (e.g. in the publication of medical photographs). The extensive discussions about the potential difficulties of ensuring complete privacy in genomic data sharing are therefore crucial to informed consent. It has been suggested that it may be misleading to promise participants complete privacy (Hull *et al.*, 2008).

A.5 Points to consider
Work to address the issues related to informed consent in genomic research needs to address various key problems.

- The need to protect individuals from harms arising from wrongful or inappropriate use of genomic data may not be well served by a model of consent based on clinical medicine. There is difference of opinion on the issue of how well traditionally conceived informed consent can be protected, as well as on the appropriateness of doing so, and the need for adjunct mechanisms alongside, or in partial replacement of traditional models.
- It is virtually impossible to determine all the uses of genomic sequence data when it is deposited in a repository to be shared by many. This has led to the use of broad consent for biobanks, oversight by data access committees and research approval by research ethics committees. However, the legitimacy of these bodies to stand in the place of individual participants is an issue.
- Certain key aspects of traditional ideas of informed consent, including the right to withdraw from research, and guarantees of anonymity, are challenged by genomic research, as has been amply demonstrated in GWAS, and these challenges must be rigorously explored. Related to this, data sharing practices are changing the relations between researcher and research participant that underlie informed consent as traditionally conceptualised, and these changes must likewise be taken on board in addressing the issues.
- Whilst recognising the continuing need to provide protection for individuals, the need to consider larger social groupings, from the family up to population level, must be addressed. However, tensions between approaches focused on the individual and approaches focused

on larger groups must not be underplayed and there are important differences of practice and principle that need to be explored rigorously if manageable, equitable and effective policies are to be achieved.

- Although genomic research raises particular issues of its own in relation to informed consent, many of these issues are also being explored in medicine in general and in other areas of research and there is opportunity to utilise such thinking in the genomic context.
- The global and international nature of GWAS means that issues of informed consent must take this context into account: whilst striving for consistency, the local context may mean that detailed differences are ethically, legally and culturally appropriate and justified.
- The requirement of informed consent by research ethics committees focuses on the front-end of the research process, and little consideration is given to the rest of the research process and whether this is ethically sound. In addition, research ethics committees do not have any formal powers to ensure compliance with directions or agreements.
- New mechanisms for ensuring trust in the governance of genomic research and in its researchers, alongside the notion of informed consent, are being suggested, but there is as yet no consensus and these mechanisms need to be explored. These may include such strategies as benefit sharing, attention to issues of health care justice, and public engagement with genomic research. The pace of change in the scientific, technologic, and regulatory landscape seen with GWAS means that work on governance mechanisms is likely to be an ongoing process.
- It is important that GWAS informed consent forms explain the possible risks of breaches of privacy to individuals because of the use of the technology and global data sharing.

A.6 Key publications

The body of literature on informed consent is extremely large. For this reason, this bibliography contains only some key articles about informed consent in general, together with some of the key texts relating to genetics and to genomic research and biobanking.

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B. FEEDBACK OF INCIDENTAL FINDINGS

B.1 Summary

One of the important ethical challenges for GWA researchers concerns the accidental identification of genetic factors that reveal information that could be important to participant's health. One of the difficulties is that the results are open to interpretation, both in terms of the clinical and statistical significance, particularly for new findings. There may be a legal duty to feed back information, if a duty of care can be established, but currently we have no cases in this area that determine the scope of the duty of care for epidemiologists and secondary researchers. There appears to be a consensus in the literature that in the case of a serious condition, where the risk is significant and treatment is available, researchers or research teams have a moral obligation to feed this information back to research participants (Wolf *et al.*, 2008). However, there is greater debate about whether to feed back incidental findings where less serious conditions are identified (for instance, ones that are not life-threatening), or where no treatment is available. In these cases, the potential benefits for participants of being informed need to be balanced against the participant's right not to know. This task appears to rest currently with the researcher and his or her team and in all likelihood decisions such as those to feedback information will be discussed with, or approved by, ethics committees. There is also uncertainty as to who should be responsible for the feedback of results and how this should be done. Illes and Chin (2008) propose a pathway for managing feedback decision for incidental findings.

Types of incidental findings in GWAS

In terms of medical conditions, we can separate the following types of incidental findings for GWAS:

- Serious but not treatable.
- Serious but treatable or manageable (i.e. familial adenomatous polyposis (FAP)-related cancers, in which polyps in the colon can develop into cancers if untreated. Presence of FAP can be recognised through well-known mutations in chromosomes 1 or 5. If treated, FAP can be harmless, but the resulting cancers can be lethal).
- Not serious.
- Indicating predisposition for serious future diseases that are not preventable (i.e. Alzheimer's Disease, certain cancers).
- Indicating predisposition for serious future diseases that could be prevented (i.e. diet-related cardiovascular diseases, diabetes, BRCA-related breast cancer).
- Indicating intolerance for certain substances that could affect treatment decisions (i.e. drug intolerance).

In addition, there may be incidental findings that indicate more social or behavioural characteristics, such as for instance:

- Indicating atypical family relationships (i.e. incest).
- Indicating non-paternity.
- Indicating behavioural problems/undesired behaviour that could affect the person's quality of life.

B.2 Legal issues around incidental findings

Whether researchers have an obligation to report back incidental findings depends upon whether they have legal a duty of care. The responsibility of the funders or the institution may depend upon the obligations imposed under Art.2 of the Human Rights Act 1998. Miller (Miller et al., 2008) comment that:

> 'There is still some confusion about the basic duty at issue, between a *proactive* ethical duty to offer results to all participants and a *passive* duty to respond positively to requests for results from self-selected individual participants … These obligations are legally and ethically distinct, with privacy laws in many jurisdictions requiring that

individuals be granted access to data about themselves when they make such a request. By contrast, there is no similar obligation to *offer* results.'

Common Law duty of care

In the absence of a legally established duty of care, the *Caparo* test *(Caparo Industries plc v Dickman* [1990] 2 AC 605) must be applied to determine whether there is a duty of care, by asking the following questions:

- Is the risk of harm foreseeable?
- Is there sufficient proximity between the parties?
- Is it fair, just and reasonable to impose a duty of care?

It was established in the Creutzfeldt-Jakob Litigation (QB 54 BMLR 8, 9), that researchers do owe a duty of care to research participants and that this is 'akin to that of doctor and patient, one of close proximity.' However, this duty was found in a clinical trial which then became a general therapeutic programme. It was largely because it was a therapeutic programme that a duty of care was established without considering proximity. In other jurisdictions, such as Canada, it has been established that researchers do owe a duty of care to research participants. (*Halushka v University of Saskatchewan* [1965] 52 W.W.R. 608 (Sask. C.A.).

Establishing proximity

The crucial element in establishing proximity is whether a close relationship has been established. For doctors, this has been whether the person has been seen by the doctor and received advice on their health care *(Goodwill v British Pregnancy Advisory Service* [1996] 2 All ER 161 (CA), per Gibson LJ.) In *Everett v Griffiths* ([1921] 1 AC 631, 657-658, 680), the court held that a workhouse doctor owed a duty of care to a person whom he certified to be insane, even though this just involved signing the reception form. Also in *Re N* ([1999] Lloyd's Rep Med 257, 263) Clarke LJ thought it 'at least arguable that where a forensic medical examiner carries out an examination and discovers that the person being examined has a serious condition which needs immediate treatment, a duty is owed to the examinee to disclose those facts'. The courts make a distinction between a published report for information purposes, such as an environmental report or an academic paper, as opposed to a report that is specific to the individual or a particular transaction. As Denning LJ said: - 'when a scientist or an expert makes an investigation and report for the purpose of a particular transaction, then in my opinion, he is under a duty of care in respect of that transaction.'*(Candler v Crane, Christmas & Co* [1951] 2 KB 164). In the case of a biobank that carries out a longitudinal study on individuals over many years, constantly adding to their individual record, it has been argued that a duty of care would exist (Johnston and Kaye, 2004).

There are no decided cases on epidemiologists and statisticians, who do not take on a clinical role and may never see the person whose DNA they analyse using GWAS. While the CJD litigation cases establish that a duty is owed by researchers akin to that of doctors, this was in the case of a therapeutic clinical trial, rather than the more removed relationship of carrying out analysis of a DNA sample, and then the further removed secondary researcher who analyses the GWAS data. Therefore, proximity may be established between the researcher who has direct contact with individual participants, but it is unclear as to whether a duty of care and an obligation to feed back incidental findings could be established for secondary researchers using coded GWAS data.

Is a duty owed to other biological relatives?

Considering the current common law, it would seem that a duty of care would not be owed to other family members who had not attended the clinic, but through the use of family histories and genetic testing could be identified as at risk. The key to finding a duty would be whether the

court would regard that the other family members were sufficiently 'identifiable' and whether it would be in accordance with public policy to establish a duty, which would depend upon the facts of the case.

There is more authority in the United States that doctors can, in certain circumstances, owe a duty to third parties, such as other family members. Thus, in the case of dangerous psychiatric patients (*Tarasoff v Regents of the University of California* (1976) 131 Cal Rptr 140, infectious diseases, such as HIV (*Reisner v Regents of the University of California* 31 Cal App 4th 1195 (1995)), and genetic conditions (*Safer v Pack* 677 A 2d 1188 (1996)), doctors have been held to owe a duty to third parties. Courts in the US have established a duty of care in cases involving genetic tests where the other family member is known, but they have not attended the clinic. However the duty of care has been limited to a 'duty to warn' the patient to inform other family members of the condition *DiMarco v Lynch Homes – Chester County, Inc* (1989) 525 Pa 558 (Sup Crt of Pennsylvania); see also the Canadian decision of *Pittman Estate v Bain* (1994) 112 DLR (4th) 257), or to inform patients of the risks associated with their condition so that they can curb their behaviour in order not to harm others (*Pate v Threlkel*, 661 So2d. 278 (Fla 1995), at 282.) This would suggest that in the case of GWAS, there would not be a duty to warn family members who may have a risk of disease on the basis of the GWAS carried out on the proband.

Art.2 European Convention on Human Rights: Right to Life

Article 2 protects the right to life, and if authorities have been seriously negligent so as to create a real risk of loss of life or serious harm then, legal proceedings can be brought against them. In the case of *Osman v The United Kingdom* [1998] 29 EHRR 245 it was held that the UK is under an obligation to take 'appropriate steps' to safeguard the lives of those in its jurisdiction. Johnston and Kaye have argued that it is possible that Article 2 places a 'positive obligation' on the UK Biobank to "put in place measures to avoid a risk to the lives of the participants and this positive obligation could encompass the provision of feedback risk of a serious genetic disease if this is revealed during the course of the research project" (Johnston and Kaye, 2004, pp. 262–263). Similarly, it could be argued that Article 2 imposes a positive obligation to safeguard the lives of individuals involved in GWAS research, by providing similar feedback during the research project. This could apply to the researchers' institution or to the funders of the research.

Art. 8 European Convention on Human Rights: Right to Privacy

Personal data (particularly medical data) are protected by Article 8 as part of an individual's right to respect for private life. Domestic (national) law must provide sufficient safeguards to prevent any communication or disclosure of personal data as may be inconsistent with the provisions of Article 8. Therefore, if there were to be a disclosure of results without an individual's consent to a third party, such as a biological relative, this could result in a claim under Article 8. To establish a claim under Article 8, the claimant must show a real and immediate threat to his health and that steps could reasonably be expected to be taken to protect his physical integrity (*R (ex parte RH) v Ashworth Hospital Authority* [2002] AC 19 at para 129).

B.3 Key challenges in feeding back incidental findings

State of the art in GWAS

Particular factors complicate decisions to feedback information from GWAS. These include:

- Genetic variants associated with disease in GWAS often have unknown or uncertain predictive value, and limited information is available about, for instance, penetrance or clinical significance of the genetic variant.
- GWAS do not necessarily establish the causal genetic variant that is responsible for the association, and follow-up studies may still be in process.

- Laboratories conducting GWAS are not usually accredited diagnostic facilities, so that the results from a research laboratory are not of the same standard that as those required for diagnosis.
- To-date, there has only been a limited possibility for replication of GWAS experiments or re-analysis of results. Combined with the plethora of associations reported in literature, it may be hard to assess the validity of reported associations.

Considerations in feeding back incidental findings

Origin of samples and consent given:

- Where archived samples are used, participants may not be aware that their samples are being included in genetic studies leading to complications for decisions on feeding back information.
- Where samples are used from other countries, researchers may only have limited knowledge of, or access to, the healthcare system available to research participants. This may complicate options for validating research findings, or possibilities for ensuring that treatment or counselling are available to participants. In particular, inclusion of samples from the developing world, for instance, also raises questions about the appropriate standard of care that should be considered in decisions to feed back. For example, conditions that are 'serious but treatable' in the UK, may not in fact be treatable in other countries.
- Some guidance suggests that the duty to disclose depends on the stage of the research programme, so for example in genetic research, there may only be a limited duty to disclose because of the exploratory nature of the research, and such a duty to disclose may only apply to clinical trials research and not to fundamental research (Knoppers *et al.*, 2006).

Feedback to whom?

- Participants may not be in a position to receive or understand genetic information from researchers, and it may be inappropriate to feed back information directly to them. Physicians may be better suited to receive or interpret such information, but contact details of the physician may not be available to researchers.
- Considering the significance of genetic information for relatives of research participants, there may be an obligation on researchers to feed back significant research results to family members of participants.

Identification of significant findings and who should feed back?

- GWAS is a complex research field bringing together experts from different disciplines. Not all may be in a position to assess the relevance or utility of findings. It is not clear whether, and what pathway, research laboratories should follow to assess the validity or relevance of incidental findings.
- It has been suggested that clinicians involved in research may best be suited to liaise with participants or their physicians to report on, or to discuss incidental findings.
- Miller says that, even where the focus is on an active duty to disclose research results to research participants, differences remain in the types of results that should be returned (for example, aggregate results or individual results), and how such results should be returned. In the case of individual results, the obligation to disclose is advanced with genetic research, but it is still contentious: '[s]ome guidance frames the duty to disclose individual results so narrowly that it approximates instead to a duty to warn, or a duty of care' (Miller *et al.*, 2008).

Secondary data users:

- Common practice in GWAS means that research data are shared with researchers all over the world. Whether the duty to feed back significant information to participants should extend to secondary users is discussed, but not resolved.

Future developments:

- Participant attitudes to receiving information may change over time and it is important to keep this in mind.
- Scientific developments may mean that information that is meaningless today, becomes comprehensible and clinically relevant later on. In this case, are there any obligations to feed back information?
- An important question is whether the possible obligation to feed back (some) incidental findings expires over time.

B.4. Points to consider

Factors in deciding on feedback:

- Should a wish (not) to know be discussed in the consent process, or should this be respected.
- In deciding on feeding back information, the participants' wish not to know should be balanced against benefits for the participants of being informed. Such an evaluation is apparently straightforward where treatable conditions are identified, but more complicated in the case of untreatable conditions.
- There seems to be some consensus that there is an ethical duty that findings should only be considered for feedback if there is sufficient evidence to support their validity and if they are clinically significant.

Managing feedback:

- Illes and Chin (2008) propose a pathway for managing feedback decision for incidental findings. This includes:
    - Formulating a management pathway for incidental findings at the outset of the research project. This is to be included in the original ethics approval application so that it can be considered by the ethics committee. It is also to be mentioned in the consent documentation.
    - The consent form gives participants the option not to receive any feedback.
    - The management pathway includes a discussion of the steps to be taken to verify the validity and potential clinical utility of incidental findings, and it assigns members of the research team to this task.
    - The pathway also specifies to whom feedback will be given.
- Ethics committees have an important role to play in providing guidance on the appropriateness of feeding back research results to research participants. Part of their evaluation should take into account whether the condition is sufficiently serious to justify re-contacting of participants or their physicians, whether the disease association is reliable, and whether the participant would benefit from being informed.
- Wolf *et al.* (2008) also recommend that funding agencies should address incidental findings in their guidance documents.
- It seems sensible to propose that incidental findings are considered more prominently at the start of future GWAS projects. However, this may not need to be done by separate research groups, but is a task that could initially be discussed or prepared by centre-wide or nation-wide groups. Considering the policy work that is required in this area, a nation-wide task force may be the most promising option.
- Because research laboratories are not accredited as diagnostic labs, incidental findings can only be considered as an indication that a person may be suffering from a condition. If a decision is made to feed back incidental findings to participants, then such findings should be confirmed in accredited diagnostic laboratories, such as those of the NHS. This could not only place an additional burden on the healthcare system in the UK, but also be seen as playing a key public health role in early prevention.
- Reporting back of incidental findings could also be seen as an important way of rewarding individuals for their altruistic involvement in research, and the possible benefits of this should be considered.

- Van Ness (2008) argues that feedback of findings is an important part of building and maintaining trust. 'Realizing the predictive power of genomics requires research that, in turn, relies on public trust. That trust depends on appropriate and respectful communication between the researcher and the participant'.

B.5 Key publications

Special Issue on incidental findings of the Journal of Law, Medicine and Ethics, Summer 2008, including:

Cho, M. K. Understanding incidental findings in the context of genetics and genomics. *J Law Med Ethics* 2008;**36:**280–285.

Illes, J., Chin, V. N. Bridging philosophical and practical implications of incidental findings in brain research. *J Law Med Ethics* 2008;**36:**298–304.

Johnston, C., Kaye, J. Does the UK Biobank have a legal obligation to feedback individual findings to participants? *Med Law Rev* 2004;**12:**239–267.

Knoppers, B. M., Joly, Y., Simard, J., Durocher, F. The emergence of an ethical duty to disclose genetic research results: international perspectives. *Eur J Hum Genet* 2006;**14:**1170–1178.

Wolf, S. M., Lawrenz, F. P. *et al.* Managing incidental findings in human subjects research: Analysis and recommendations. *J Law Med Ethics* 2008;**36:**219–248.

C. DATA SHARING

C.1 Summary

There is a wide range of literature on the matter of data-sharing and access to data, highlighting the social, ethical, legal and technical aspects of data sharing. Data sharing implies by definition that data which have been collected for one purpose are being used, either by secondary users or for different purposes, or both. This can raise ethical issues relating to beliefs about the future uses of the data by study participants or data subjects (because not all data used in research are collected for research purposes). These beliefs can to some extent be formalised in consent forms, but as data are increasingly finding their way into 'resources' such as biobanks, to be used for any number of research purposes, the role of informed consent is increasingly brought into question. Legal standards for data sharing are somewhat focused on the quality of the data and whether an individual within a dataset is identifiable. Technical issues include the compatibility of systems for managing and storing data, as well as methods of coding, classification and measurement, to ensure that data are understandable outside of the context in which they were collected. The social elements of data sharing include the incentives for scientists to work together, when they may in many ways be competitors. This also brings into question the current incentives to share, such as the recognition of different contributions in publications.

C.2 Legal summary

The Data Protection Act only applies to identifiable data or data that relate to a living individual. 'Personal data' means data which relate to a living individual who can be identified (a) from those data, or (b) from those data and other information which is in the possession of, or is likely to come into the possession of, the data controller (s.1(1) DPA 1998). The research community has used coding as a means to protect the privacy of participants, so that the research team use anonymous data and the code which can identify the research participant is held by a trusted third party. The difficulty with GWAS is that 'the sample speaks for itself' and as technology improves and the availability of sequence information increases through data sharing, it may be difficult to control whether individuals are identifiable from those data and other information which is in the possession of, or is likely to come into the possession of, the researcher (data controller). In order to control this, stringent safeguards are required to prevent identity disclosure through the sharing of datasets and to ensure that access to such information remains within the hands of researchers who are bound by obligations of confidentiality.

The Data Protection Act allows the secondary use of already collected personal information for additional research purposes under the Research Exemption under s. 33 of the Act. This allows personal information to be used for research purposes not envisaged at the time of collection, as long as the data are not processed to support measures or decisions relating to particular individuals; and the data are not processed in such a way that substantial damage or substantial distress is, or is likely to be, caused to any data subject. This also allows personal data to be kept indefinitely; and individuals may not have a right of access to the research data as long as the results of the research or any resulting statistics are not made available in a form identifying the data subject. This allows personal information created through GWAS analysis to be shared with other researchers. The Data Protection Act requires that any data sharing with researchers in another country can only take place where there are equivalent standards for data protection to those within the UK. Even where the s.33 research exemption is relied upon, there is nevertheless a requirement to adhere to the remaining data protection principles.

The recent Thomas and Walport report on data sharing (Thomas and Walport, 2008) makes a number of recommendations regarding the changes to the law that should be considered in order to facilitate research within the UK. These include the establishment of safe havens for researchers (8.78) and a system of approving and accrediting researchers (8.79). They recommend looking at the governance models that have been developed for statistical research as a basis for thinking about the use of medical information in research.

C.3 Key challenges in data sharing

- The support of funders and the development of data sharing policies have led to the development of data-generating infrastructure projects and the presumption that hypothesis-led projects will also consider depositing data to be shared with others, unless good reasons can be demonstrated why this should not be so (Kaye *et al.*, 2009).
- The use of informed consent as traditionally conceived for research is difficult to obtain, as it is impossible to inform individuals of all the uses of the data at the time of collection (see section on consent).
- Kaye *et al.* (2009) make the point that scientific practice may not change overnight simply because there is a will to encourage greater data sharing, old systems of reward and recognition may sit uneasily next to a system of more open access to data. Scientific projects are often built on the trust between participants and Principal Investigators.
- Foster and Sharp (2007) talk about recognising the various stakeholders in the distribution of benefits both social and scientific. Stakeholders include those who set up genomic studies, participants, secondary users and even parties who could be affected by the secondary uses of the data.
- Chandramohan *et al.* (2008) discuss the benefits and disadvantages, not only to society in terms of potential health benefits arising from genomics research, but also of those researchers in poorer parts of the world who may not have the capabilities to exploit data in the same way as their developed world counterparts. Such researchers can become merely the producers of data and establishing and maintaining their status as the poor relatives in the field of scientific research.
- Heeney (2008) points out that even where there is legislation supporting data sharing, technical and ethical barriers remain. For example, systems which have not been designed to 'speak to each other' may get in the way of successful sharing and re-use of data by secondary users.
- Lowrance (2002) considers that traditional tools of anonymisation and consent are still useful in the realm of data sharing.
- Lunshof *et al.* (2008)argue that this system is no longer adequate to the task of protecting privacy and is therefore flawed as the ethical underpinning of data sharing in the era of GWAS.
- The Thomas and Walport report on Data Sharing makes a number of recommendations that have implications for medical research (Thomas and Walport, 2008).

C.4 Points to consider

- There needs to be recognition of the role of data sharing policies in modifying scientific practice.
- There needs to be greater sensitivity to the fact that existing relationships between those running the original study and participants may create obligations and expectations that may make data sharing difficult.
- Not all researchers will have the ability and resources to take advantage of available data, particularly if they are based in developing countries. This means that the success of data sharing policies may be limited to researchers with access to the web and other resources, as well as a recognition of the need for career development of researchers contributing to data generation in GWAS.
- Technical issues such as how data are categorised and managed need to be taken into account at an early stage in order to optimise the utility of data for external researchers, and to ensure that data are in a format that can be shared.
- The legal and ethical issues on sharing data should be considered early on in research projects.
- Careful thought needs to be given to the current reliance on anonymisation and consent as sufficient safeguards to protect participant interests.

C.5 Key publications

Bellamy, C., Perry 6, Raab, C. Joined-up government and privacy in the United Kingdom: managing tensions between data protection and social policy. Part II. *Public Admin* 2005;**83:**393–415.

Berg, M. Health information management: integrating information technology in health care work. London: Routledge, 2004.

Chandramohan, D., Shibuya, K. *et al.* Should data from demographic surveillance systems be made more widely available to researchers? *PLoS Med* 2008;**5:**e57.

Foster, M. W., Sharp, R. R. Share and share alike: deciding how to distribute the scientific and social benefits of genomic data. *Nat Rev Genet* 2007;**8:** 633–639.

Heeney, C. How to get there from here: Re-use of administrative records in the Netherlands and the UK. *SCRIPTed* 2008;**5:**294–308.

Hilgartner, S., Brandt-Rauf, S. I. Data access, ownership, and control. Toward empirical-studies of access practices. Knowledge-Creation Diffusion Utilization. *Sci Commun* 1994;**15:**355–372.

Kaye, J., Heeney, C., Hawkins, N., De Vries, J., Boddington, P. Data sharing in genomics – reshaping scientific practice. *Nat Rev Genet* 2009;**10:**331–335.

Lowrance, W. W. *Learning from Experience: Privacy and the Secondary Use of Data in Health Research*. The Nuffield Trust, 2002.

Lunshof, J. E., Chadwick, R., Vorhaus, D. B., Church, G. M. From genetic privacy to open consent. *Nat Rev Genet* 2008;**9:**406–411.

Thomas, R., Walport, M. Data sharing review report. http://www.justice.gov.uk/docs/data-sharing-review.pdf

UK Clinical Research Collaboration and Wellcome Trust. *Frontiers Meeting, Use of Electronic Patient Records for Research and Health Benefit*. 2007.

Wellcome Trust. Sharing data from large-scale biological research projects: a system of tripartite responsibility (Fort Lauderdale). 2003.

D. PRIVACY

D.1 Summary of the issues

Privacy and confidentiality have traditionally been the cornerstones of ethical medical research practice. These issues are raised anew by developments in the GWAS approach that generate rich information about individuals and populations, and by the information processing technologies, which enable this information to be managed and used. For organisations involved in research, which will ultimately benefit the public or provide vital information to be used in planning or improving services, there is often argued to be a trade off between public good and individual privacy. However, the uniquely identifiable nature of genomic data, the implications for family groups and other populations sharing genetic characteristics, and the ever increasing capacities to produce this data in large quantities has arguably shifted the parameters of this argument, with some claiming that the nature of genetic data means that it will soon be impossible to deny others access to one's genetic information due to the fact that many genetic characteristics are shared. Once data are anonymised, it is very difficult to show a breach of privacy as a matter of law. There is also a duty to ensure that participants are informed about the study they are involved in. However, these traditional methods of respecting individual participants' privacy are increasingly criticised on the grounds that due to the proliferation of data sources and the ability to triangulate them, anonymisation may not be possible.

D.2 Legal issues

The key points from the Marper judgment [*S. and Marper v The United Kingdom* 30562/04 [2008] ECHR 1581 (4 December 2008)] that apply to GWAS are that:

- given the nature and the amount of personal data that are contained in cellular samples and DNA profiles, their retention must be regarded as interfering with the right to respect for the private lives of the individuals concerned;
- the capacity of DNA profiles to provide a means of identifying genetic relationships is in itself sufficient to conclude that their retention interferes with the right for respect of the private lives of the individuals concerned;
- any State claiming a pioneering role in the development of technologies bears a special responsibility for striking the right balance in this regard.

This means that the mere retention of DNA will give rise to a right of privacy and that any interference with that right, such as use for medical research, must be justified, as in accordance with law, necessary in a democratic society, and proportionate, in that it could not have been achieved in any other way. It is unclear whether digital sequence information would be considered as coming under the provisions under the Human Tissue Act for the non-consensual processing of DNA. However, if data are anonymised then the Data Protection Act will not apply and it may be difficult to establish a breach of privacy under Art. 8 of the Human Rights Act 1998.

The Collie case (*Common Services Agency v Scottish Information Commissioner (Scotland)* which was heard by the House of Lords in April 2008 [2008] UKHL 47 is regarded as a landmark case on the compatibility of data protection and freedom of information laws. The case focused on an NHS agency's refusal of a Freedom of Information (FOI) (Scotland) request for statistics about childhood leukaemia. The agency feared that the information could identify individual children and breach their privacy, and having lost its case in the Scottish courts, it appealed to the House of Lords. Mechanisms to protect the privacy of research participants have traditionally been informed consent and anonymisation of datasets. The result of this case in respect of researchers gaining access to research data, appears to be that where a request is made for information under the Freedom of Information legislation, consideration will first have to be given as to whether the information can be sufficiently anonymised for it not to be 'personal data'. If it cannot be so anonymised, it would then have to be determined whether its disclosure would comply with the data protection principles set out in the 1998 Data Protection Act.

D.3 Key points from the literature

- The paper by Homer highlighted the possibility of separating out individuals from aggregated SNP data (Homer *et al.*, 2008). This in turn exposed problems with guarantees of anonymity, which had applied to GWAS data released in aggregated form.
- Gitschier's paper shows how the process of combining different data sources can greatly aid the process of identification of individuals (Gitschier, 2009). By using a combination of information on surnames and haplotype analysis of the Y chromosome, she was able to establish fairly accurate predictions regarding the likely relatives of certain individuals in a given population.
- Literature from the field of statistical disclosure control, such as the paper by Elliot (Elliot, 2000), shows the dangers of re-identification of individuals in a dataset given a combination of knowledge of the population they belong to and their uniqueness in that population.
- Lowrance and Collins re-state the importance of anonymisation and consent in ensuring the privacy of participants in genomic studies on the one hand and the public goods likely to arise from genomic research on the other (Lowrance and Collins, 2007).
- Lowrance and Collins also make the point that identifying individuals from sequences data is still very difficult to do and requires identifiable sequence information.
- Nissenbaum (2004) talks about the difficulties of ensuring that the protections afforded by consent and anonymisation are actually effective given the current data-rich environment and motivations to use data that may be some distance from those of the scientists originally collecting it.
- Sweeney (2001) talks about the enormous growth in the data stored and collected on individuals creating an environment where it is difficult to determine whether it is safe to release certain data.
- Tavani (2004) highlights the particular capabilities provided by information technologies, such as data mining, for aiding in the process of population segmentation and inference.
- Vedder (2000) critiques the straight dichotomy, which appears in law and is widely accepted by many in the research community, between the identifiable and non-identifiable data. He argues that both types may in certain cases support what would ordinarily be seen as privacy infringements.
- Heeney *et al.* (2009) argue that where there is the motivation to use data for decision support, in terms of consequences for individuals the distinction between aggregate or anonymised and identifiable data often breaks down.
- Lunshof *et al.* (2008) challenge the ability to guarantee confidentiality to research participants in biomedical research and this in turn undermines the traditional role of consent in these studies. They draw on the notion of 'categorical privacy' proposed by Vedder to suggest that an individual approach to privacy protection is outmoded.
- Greenbaum and Du (2008) raise the ominous prospect that it may soon be impossible to control information about one's genetic make up due both to the widespread availability of these data and the fact that one individual's genomic data reveal information about their family and the population to which they belong.

D.4 Points to consider

- An approach to data sharing is needed which takes into account the availability of other datasets and information processing technologies.
- Consent and anonymisation and their role in the protection of individuals need to be re-thought in the light of implications for populations raised by the processing and availability of genomic information.
- Reconsider the balance between public good and privacy in the light of recent indications that it may be possible to cross a point after which individuals will not be able to control what is known about their genetic characteristics.

- Acknowledge that implications of giving data for participants in genomic studies may be difficult or impossible to capture and communicate in the traditional informed consent format.
- Take into account the reasonable expectations people may have of privacy protection and future uses of data, which draw on the context in which information is given, as well as what it is possible to communicate in the consent material.
- Consider the usefulness of the traditional dichotomy between aggregated and identifiable data in the light of the nature of genomic data that have been used in statistical and census data access.
- Governance mechanisms that have been used in statistical research may be useful for GWAS.

D.5 Key publications

Elliot, M. *Disclosure Risk Assessment. Confidentiality, Disclosure and Data Access: Theory and Practical Application for Statistical Agencies*. Doyle, P., Lane, J. I., Theeuwes, J. M., Zayatzr, L. M., Eds. New York: Elsevier, pp. 135–166, 2000.

Gitschier, J. Inferential genotyping of Y chromosomes in Latter-Day Saints founders and comparison to Utah samples in the HapMap project. *Am J Hum Genet* 2009;**84:**251–258.

Greenbaum, D., Du, J., Gerstein, M. Genomic anonymity: have we already lost it? *Am J Bioeth* 2008;**8:**71–74.

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Homer, N., Szelinger, S. *et al.* Resolving individuals contributing trace amounts of DNA to highly complex mixtures using high-density SNP genotyping microarrays. *PLoS Genet* 2008;**4:**e1000167.

Kaye, J. Police access to DNA samples and information. *Genomics Soc Policy* 2006;**2:**16–27.

Lowrance, W. W. *Learning from Experience: Privacy and the Secondary Use of Data in Health Research*. The Nuffield Trust, 2002.

Lowrance, W. W., Collins, F. S. Identifiability in genomic research. *Science* 2007;**317:** 600–602.

Lunshof, J. E., Chadwick, R., Vorhaus, D. B., Church, G. M. From genetic privacy to open consent. *Nat Rev Genet* 2008;**9:**406–411.

Nissenbaum, H. Privacy as contextual integrity. *Wash Law Rev* 2004;**79:**119–157.

Sweeney, L. *Information Explosion. Confidentiality, Disclosure and Data Access: Theory and Practical Application for Statistical Agencies*. Doyle, J. P., Lane, J. I., Theeuwes, J. M., Zayatzr, L. M., Eds. Elsevier Science, pp. 43–74, 2001.

Tavani, H. T. Genomic research and data-mining technology: implications for personal privacy and informed consent. *Ethics Inf Technol* 2004;**6:**15–28.

Vedder, A. Privacy and confidentiality; medical data, new information technologies, and the need for normative principles other than privacy rules. In: Freeman, M., Lewis, A. D. E. *Law and Medicine Current Legal Issues.* Vol 3, pp. 441–459. Oxford: Oxford University Press, 2000.

E. GOVERNANCE OF RESEARCH

E.1 Summary

The use of GWAS approaches in genomic research creates a number of novel issues for research governance as large quantities of detailed data specific to individuals, can be used and shared by many researchers in many different research projects. The use of the GWAS technology itself raises particular ethical and legal issues, which are often beyond the scope of the expertise of many research ethics committees. In addition, it is not clear that research ethics committees (RECs) have the appropriate expertise to make an assessment of the data sharing risks. To combat some of these concerns, special data access committees have been established in the data-generating projects such as the Wellcome Trust Case Control Consortium (WTCCC), Genetic Association Information Network (GAIN) and Database of Genotypes and Phenotypes (dbGaP), to supervise access to these datasets. The use of GWAS in genomic research, highlights some of the perennial problems with our current governance system in the UK for medical research carried out on human beings. These are concerned mainly with the expertise of research ethics committees and whether their procedures are appropriate for the pivotal position that they have as 'gatekeepers' in the research process; and the fact that our governance structures and legal frameworks are nationally based, but our research is global. In the UK we currently have a governance framework that is complex, contradictory and confusing, with a number of different bodies with specific requirements and guidelines. The legal framework for research is also not straightforward, as it is based on general law, rather than specific legislation that lays out principles which could provide a foundation for medical research. These factors in combination create a plethora of guidelines and duplicity, as well as uncertainty, which is frustrating for researchers and runs the risk of impeding emerging technologies.

E.2 Legal issues

The governance system for medical research in the UK is a complex web of regulating bodies and guidelines that are essentially uncoordinated, leading to omissions, inconsistency, overlaps and duplication (Kaye and Gibbons, 2008). 'It is unclear how the different instruments, provisions and regulatory bodies should be ranked or prioritised where choices, conflicts or discrepancies arise. Obviously, this poses particular problems for practitioners and professionals when they try to ascertain what the law, relevant rules or applicable guidelines require of them' (Gibbons, 2009 [in press]). This regulatory framework has grown incrementally over time, with new bodies being established for specific purposes and general law being developed to apply to medical research. Unlike other jurisdictions, the UK does not have a National Bioethics Committee, nor does it have one legal instrument that applies specifically to medical research on human beings, though is does have legislation to regulate medical research on animals. The UK has not signed, or implemented the Convention on Human Rights and Biomedicine which would provide the basis for legislation. There is some concern that the current regulation of medical research would not comply with the 'good governance' principles enshrined in the Legislative and Regulatory Reform Act 2006 (LRRA), which require that regulation should be transparent, proportionate, accountable, consistent, and targeted only at cases in which action is needed. For innovations such as GWAS, this can allow flexibility, but it can also lead to uncertainty. In response, new governance mechanisms and structures, such as data access committees have been developed to deal with the deficiencies in the current regulatory framework.

The ambit of the powers of research ethics committees is laid out in the -Standard Operating Procedures for RECs, COREC, 2004; Governance Arrangements for NHS Research Ethics Committees (GAFREC), 2001;Research Governance Framework for Health and Social Care (RGF), Department of Health, 2001; Health Service Regulations 2002; Adults with Incapacity Regulations 2002 (Scotland).

Summary of UK Research Ethics Committees (Adapted from PRIVIREAL Project website: http://www.privireal.org/content/rec/uk2.php)

Type of REC	**LRECs**	**MRECs**	**RECs of particular organisations**
Where situated?	NHS	NHS	Medical Research Council UK Universities Pharmaceutical industry
Who applies to them?	Researchers	Researchers	Researchers from the institute where the committee is situated
Powers	Formal	Formal	Informal
Jurisdiction	Local	Regional	Local
Applicable laws	Health Service Regulations 2002; Adults with Incapacity Regulations 2002 (Scotland); Medicines for Human Use (Clinical Trials) Regulations 2004	Health Service Regulations 2002; Adults with Incapacity Regulations 2002 (Scotland); Medicines for Human Use (Clinical Trials) Regulations 2004	-
Guidance involved	Governance Arrangements for NHS Research Ethics Committees; Research Governance Framework for Health and Social Care; Standing Operating Procedures for Research Ethics Committees	Governance Arrangements for NHS Research Ethics Committees; Research Governance Framework for Health and Social Care; Standing Operating Procedures for Research Ethics Committees	-
Membership requirements	Except in Scotland: <18 members, of which one third must be lay members with the remainder being	Except in Scotland: <18 members of which one third must be lay members with the remainder being expert	-

	expert members. In Scotland: <18 members including doctors, a nurse, a pharmacist, a clinical pharmacologist, a person with experience in the treatment of adults with incapacity and three lay members	members. In Scotland: <18 members including doctors, a nurse, a pharmacist, a clinical pharmacologist, a person with experience in the treatment of adults with incapacity and three lay members	
Responsible/accountable to whom?	To the UKECA or the body they have nominated, e.g. COREC in England	To the UKECA or the body they have nominated, e.g. COREC in England	
Approval or advisory powers	Approval	Approval	

LRECs = local Research Ethics Committees

MRECs = multi-centre Research Ethics Committees

UKECA = United Kingdom Ethics Committee Authority

COREC = Central Office for Research Ethics Committees

There has been very little litigation concerned specifically with medical research and the bodies that oversee medical research. The only case that we have is the CJD Litigation: *The Creutzfeldt-Jakob Disease Litigation, Plaintiffs v United Kingdom Medical Research Council and another* QB 54 BMLR 8), where the court discussed the dual role of the Medical Research Council (MRC) and Department of Health staff, who were scientifically trained to service committees, and to produce and supply briefing material. On this matter Morland J. concluded:- 'They had to be alert to and aware of current medical and scientific knowledge and discoveries. In briefing the chairman and members of committees, in my judgment, they were under an obligation to alert committee members to such current knowledge and discoveries.'(*The Creutzfeldt-Jakob Disease Litigation, Plaintiffs v United Kingdom Medical Research Council and another* QB 54 BMLR 8, per Morland J., 18.)

In regard to the committee itself, Morland J. said that: 'The standard of care, in my judgment, to be imposed in respect of a committee is that of a reasonably competent and carefully inquiring group of professionals of the relevant disciplines, of sufficient standing to be entrusted with the membership of that committee, bearing in mind it is a committee which is not merely advisory but is carrying out executive and administrative functions. In my judgment, the committee is to be expected to carry out its tasks with the expedition reasonably required to deal with a matter or problem as it arises and to obtain such specialist assistance from experts as reasonably required to carry out its functions. The committee is expected to be properly briefed and supplied with relevant papers including correspondence and learned literature so that it can carry out its tasks. If the committee is not so properly briefed or supplied it cannot reasonably be faulted for not requesting the medical and scientific material from its staff but its staff can.' (*The Creutzfeldt-Jakob Disease Litigation, Plaintiffs v United Kingdom Medical Research Council and another* QB 54 BMLR 8, per Morland J, 18. This case could be applied to research ethics committees and how they carry out their functions.

In Canada, in *Weiss v Solomon (1989) 48 C.C.L.T. 280 (Que. Sup. Ct)*, 'a research ethics board was held liable for failing to adequately protect a research participant. In that case the hospital where the research was conducted was held jointly liable with the physician investigators for failing to ensure that the recruitment procedure and the consent materials were adequate to screen out participants for whom the trial posed an unacceptable risk. In this case, the materials did not reveal that the research presented a risk for patients with a certain medical condition. While the risk was remote, and would not have been a barrier to undergoing the procedure if the patient were to benefit from it, the risk was sufficient to constitute an exclusion criterion for this trial. It is therefore clear that in Canada, research ethics boards owe a direct legal duty to research participants and may be held liable for a breach of that duty. …Whether or not the research is publicly funded, a research ethics board provides the research institution with a vehicle for ensuring that the research conducted on its premises or by its personnel receives appropriate review. As the legal entity responsible for acts done on its behalf by its boards and committees, a research institution such as a hospital or university will be found to have a duty of care to those who participate in research conducted under its auspices.'(Zimmerman, 2005).

UK research ethics committees are having greater power to make decisions to exempt certain activities from further scrutiny. 'Since late 2006, UK RECs have been authorised under the NRES Standard Operating Procedures for RECs to issue 'generic' approvals for up to five years (renewable) to 'research tissue banks' (RTB) that wish to act as repositories, supplying biosamples to multiple end-user researchers for use in their own separate projects (NRES 2007). Under this new system, research tissue banks may seek and obtain generic ethical approval for their arrangements for the collection, storage, use and distribution of tissue. Where generic approval is granted, this prospectively permits a range of research to be carried out, both by the establishment responsible for the RTB and/or by other researchers to whom tissue is released by the RTB, within the conditions of the ethical approval. In such cases, neither the RTB nor other end-users need to seek any further, project-specific REC approval.'(Gibbons, unpublished, 2008).

E.3 Key challenges in governing GWAS

GWAS and data sharing challenges: traditional oversight mechanisms

'Traditionally, approval for research is obtained from a research ethics committee by a particular individual or research group. This committee holds the principal applicant responsible for monitoring the use of samples and data; however, when samples are transported across national borders, and when data are analysed by people who bear no relation to the original research project or participants, it is virtually impossible to continue to hold the original collector responsible in the same way. Therefore it is difficult for research ethics committees to exert their original mandate to ensure the ethical conduct of research.'(Kaye *et al.*, 2009).

Research ethics committees

There are a number of concerns regarding the use of research ethics committees as the main body to oversee GWAS research. These concern expertise; the nature of decision-making and the variation of decisions between committees:-

Expertise

It is doubtful whether research ethics committees have the expertise to assess adequately and understand the full extent of the science and the ethical issues raised by the use of GWAS. Research ethics committees have mainly focused on protecting research participants' interests and so in assessing genomic research tend to focus on the consent process (Mascalzoni *et al.*, 2008) and on approving the re-use of samples (Muula and Mfutso-Bengo, 2007). Under the UK Information Commissioner's Office (ICO) Framework Code of Practice for Sharing Personal Information, it is suggested that organisations undertake a 'privacy impact assessment' before sharing data; that common levels of security are put in place between organisations that are sharing; that good records management practices are evident (ICO, 2007). It is unlikely that

most RECs have the expertise to carry out such an assessment, as they are already facing increasing challenges in reviewing complex research proposals, particularly in relation to proportionality assessment (risks, benefits safeguards) informed consent and privacy protection (Hoedemaekers *et al.*, 2006). It is therefore not clear how already over-burdened committees could take on the role of monitoring and approving data access – a task that requires significant insight into the techniques used to produce and analyse data in genomics (Langat, 2005; Upshur *et al.*, 2007).

Inconsistency of decision-making

'Research ethics committees do have an authority that is recognised and respected. However, while these committees are independent, they consist of volunteer professionals and lay people, who are not necessarily required to have ethical training. One of the strengths or weaknesses, of ethics committees is that they can make tailored decisions on a case-by-case basis.'(Kaye, 2009). However, one of the problems with research ethics committees is that unlike other decision-making bodies in society, ethics committees are not required to publish the reasons for their decisions and they are not bound by precedent to follow the previous decisions of other ethics committees. Research ethics committees have attracted much criticism for ad-hoc decision-making (Tully *et al.*, 2000; Glasziou and Chalmers, 2004) because of this case-by-case decision-making approach, which has also led to a call for the process of assessing research proposals 'to be simplified and made consistent, and the reasons for decisions should be clearly argued and stated' (Souhami, 2006). The issue for new methodologies such as GWAS is that it takes some time before a critical amount of expertise is built up within the research ethics community in order to make approval for research projects an efficient and straightforward process. In the case of emerging technologies this can result in frustration for researchers and a slowing down of the research process.

Problems for collaborative projects

As the mechanisms for establishing precedents in research ethics committee decision-making are informal, there can be considerable variation in the decisions of committees. 'The result is that decisions and procedures can vary *within* regions in countries, but the differences are most acute *between* countries, where different ethical and legal frameworks exist, such as between Europe and North America. It is the area of emerging technologies and innovative, global, collaborative research proposals where there can be the most discrepancies in decision-making between ethics committees.' (Kaye, 2009).

Data access committees (DACs) - New oversight mechanisms for GWAS projects

'In the data-generating projects that provide detailed patient information, as well as sequence information, new governance models have been established to regulate access to datasets. These 'data access committees' have been established for the WTCCC, the dbGaP and GAIN projects, and determine who should have access to data and on what grounds. The basis for decision-making involves establishing whether a scientist is a '*bona fide* researcher', but the bases for assessing this are still in the process of being developed and are not uniform or publicly known. These committees are in addition to the research approval that must be obtained from a research ethics committee, as DAC approval is just for access to the dataset. However, there is no one body that is looking at disclosure when all of the genomic datasets are analysed together and the possibility of identification by using material available on the web (Kaye *et al.*, 2009).

The global nature of research

The problem with our current legal frameworks and regulatory structures is that they are nationally based, but our research activity is global. This means that we have different legal instruments that apply in different countries, so with internet searching it is possible to cross many jurisdictional boundaries by using different websites that are registered in different countries. This makes it difficult for researchers who have to conform to different legal requirements in different jurisdictions, and could place them at risk of legal liability.

Reliance on general law
Within Europe, we are moving towards more uniformity with common legal instruments to protect privacy such as the Directive 45/46/EC and case law on the Art. 8 Right to Life from the European Court of Human Rights. These would have general application to data sharing within the medical research context, but there is no legal instrument in Europe that deals specifically with this issue. Even though we have common legal instruments at a European level, there are still considerable differences between the UK and other member states, as a 'margin of appreciation' is allowed for the implementation of European Union law into national law. While we have Directive 45/46/EC, it does not apply well to data sharing and there are plans to review and update the provisions of the Directive to deal with the many breaches of privacy (outside of the medical context) currently occurring (Kaye *et al.,* 2009).

At an international level there are documents that lay down the principles for medical research, but many of these are not mandatory and are dependent on national governments signing up to them. For example, in the case of the Convention on Human Rights and Biomedicine 1997, the key document within Europe on medical research, the UK has not signed this document because of the provision on therapeutic cloning. While international documents can provide a baseline of general principles in medical research, they are not the best way to regulate emerging technologies – until the technology is well established and it is demonstrated that there is sufficient need for regulation. These higher level legal instruments provide a framework, but do not, and cannot, provide all of the details needed for the development of standards and ethical principles that can support embryonic scientific practice.

E.4 Points to consider

- It is evident that medical research would greatly benefit from an ethics review system where there was uniform, transparent and accountable decision-making. One possibility would be to make the ethics committee system based on precedent so that researchers and other research ethics committees would know how similar research issues had been decided.
- In addition, these decisions would have to be recognised by ethics committees in other jurisdictions. The ideal for researchers would be to have research approval in one country that would apply in another.
- Data sharing of GWAS results raises particular issues about how data sharing should be governed; particularly as oversight mechanisms are located at a national level whereas research is conducted globally. Therefore research ethics approval needs to be mutually recognised by other national approval systems.
- Many of the standards and requirements for research in the global contexts have been developed through initiatives such as P^3G that focus on population biobanks. We now need to think more about the protocols that are necessary for data sharing between collaborative projects, not just access to specific projects. There is a need to think more carefully about the infrastructures that might apply at a higher level.

E.5 Key publications

Caulfield, T., McGuire, A. L. *et al*. Research ethics recommendations for whole-genome research: Consensus statement. *PLoS Biol* 2008;**6:**e73.

Gibbons, S. M. C. Are UK genetic databases governed adequately? A comparative legal analysis. *Legal Studies* 2007;**27:**312–342.

Gibbons, S. M. C. Unpublished note on generic ethical approval for research tissue banks: An overview. 2008.

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Gibbons, S. M. C., Kaye, J., Smart, A., Heeney, C., Parker, M. Governing genetic databases: challenges facing research regulation and practice. *J Law Soc* 2007;**34:**163.

Glasziou, P., Chalmers, I. Ethics review roulette: what can we learn? *Br Med J* 2004;**328:**121–122.

Hoedemaekers, R., Gordijn, B., Hekster, Y., Van Agt, F. The complexities of ethical evaluation of genomics research. *HEC Forum* 2006;**18:**18–36.

Kaye, J., Gibbons, S. M. C. 'Mapping the regulatory space for genetic databases and biobanks in England and Wales'. *Med Law Int* 2008;**2:**111.

Kaye, J., Heeney, C., Hawkins, N., De Vries, J., Boddington, P. Data sharing in genomics – reshaping scientific practice. *Nat Rev Genet* 2009;**10:**331–335.

Kaye, J. Biobanking Networks – What are the Governance Challenges? In Kaye, J., Stranger, M. (Eds). *Principles and Practice in Biobank Governance*. Ashgate 2009, in press.

Langat, S. K. Reuse of samples: ethical issues encountered by two institutional ethics review committees in Kenya. *Bioethics* 2005;**19:**537–549.

Mascalzoni, D., Hicks, A., Pramstaller, P., Wjst, M. Informed consent in the genomics era. *PLoS Med* 2008;**5:**e192.

McGuire, A. L., Caulfield, T., Cho, M. K. Research ethics and the challenge of whole-genome sequencing. *Nat Rev Genet* 2008b;**9:**152–156.

Muula, A. S., Mfutso-Bengo, J. M. Responsibilities and obligations of using human research specimens transported across national boundaries. *J Med Ethics* 2007;**33:**35–38.

Souhami, R. Governance of research that uses identifiable personal data. *Br Med J* 2006;**333:**315–316.

Tully, J., Ninis, N., Booy, R., Viner, R. The new system of review by multi-centre research ethics committees: prospective study. *Br Med J* 2000;**320:**1179–1182.

Upshur, R., Lavery, J., Tindana, P. Taking tissue seriously means taking communities seriously. *BMC Med Ethics* 2007;**8:**11.

F. PERSONAL PROPERTY RIGHTS AND GWAS

F.1 Summary

Traditionally, the law has resisted recognising property rights in living or dead bodies, or parts thereof except in limited specific circumstances. However, in a recent Court of Appeal decision, property rights in human sperm have been recognised for the purposes of a cause of action in negligence and bailment. Key questions are whether such developments are relevant for GWAS, specifically:

- Do property rights exist in the samples used for GWAS?
- Do GWAS participants have rights arising from personal property law?

F.2 Legal issues

The traditional legal position has been that property does not exist in bodies except in narrow circumstances.

- A living body is incapable of being owned: *Yearworth and Others v North Bristol NHS Trust* [2009] EWCA Civ 39; [2009] WLR (D) 34 [30].
- Property can exist in a dead body (or part thereof) if sufficient work and skill has been invested in dealing with it: *Doodeward v Spence* (1908) 6 CLR 406. Thus body parts are capable of being stolen: *R v Kelly* [1999] 2 WLR 384.
- Courts have generally resisted categorising parts of live human bodies as property. However, in the recent case of *Yearworth*, the Court of Appeal recognised property rights in human sperm to found an action in negligence and in bailment. The court stated that developments in medical science now required a re-analysis of the common law's treatment of, and approach to, ownership of parts or products of the human body, whether for an action in negligence or otherwise [45].
    - The court did not go so far as to recognise an absolute property right, but elided the concepts of possessory title and legal ownership (for convenience) (at [25]). In considering whether the sperm was 'capable of being owned' for the purposes of the actions in negligence and in bailment, the relevant statutory scheme (i.e. the Human Fertilisation and Embryology Act) was relevant as it regulated and limited fundamental features of ownership. It seems likely that in considering the question of ownership of other human tissue for actions in tort or bailment, then the regulatory scheme of the Human Tissue Act would be important.

F.3 Ways forward

- Judicial recognition of property rights in human tissue may not be very significant for GWAS research. Even if a research participant has ownership of a sample which they contribute to a research study, it seems likely that participation would be construed by the courts as a gift of the tissue, creating a relationship of donor and donee rather than bailment (discussed in *Yearworth* at [48]; *Catalona*). Thus, a research participant would cede all property rights they might have in their tissue on donating it to a research study. However, a more complicated position would arise in studies where the donation is for limited purposes (for example where consent to research was limited to a particular disease). In such a circumstance there might be legal rights of redress for breach of contract, although it is difficult to see a bailment relationship arising from research unless a participant could request return of their sample (as opposed to destruction on withdrawal).
- Property rights may exist in a collection of tissue or physical DNA samples: *Washington University v Catalona* (2006) 490 F 3d 667. The ownership of this resource is likely to depend on the specific contractual (including consent) and funding arrangements for the research study.
- *R v Kelly* [1999] QB 621 raises a number of unresolved questions such as:
    - The degree of 'work and skill' required to obtain property rights.
    - Does this also apply to 'human body parts' from the living?

- Who has the property rights – the institution, the funder, or the person who carried out the work?

F.4 Key publications

Yearworth v North Bristol NHS Trust [2009] EWCA Civ 37
R v Kelly [1999] 2 WLR 384
Washington University v Catalona (2006) 490 F 3d 667

Bovenberg, J. A. *Property Rights in Blood, Genes and Data – Naturally Yours?* Leiden: Martinus Nijhoff, 2006.

Hardcastle, R. *Law and the Human Body: property rights, ownership and control*. Oxford: Hart, 2007.

Harris, J. W. Who owns my body? *Oxf J Leg Stud* 1996;**16:**55–84.

Herring, J., Chau, P. L. My body, your body and our bodies'. *Med Law Rev* 2007;**15:**34–61.

Mason, J. K., Laurie, G. T. Consent or property? Dealing with the body and its parts in the shadow of Bristol and Alder Hey. *Mod Law Rev* 2001;**64:**710–729.

Skene, L. Proprietary rights in human bodies**,** body parts and tissue: regulatory contexts and proposals for new laws. *Leg Stud* 2002;**22:**102–127.

G. INTELLECTUAL PROPERTY RIGHTS AND GWAS

G.1 Summary

Intellectual property (IP) rights in genomics provide inventors with the opportunity to protect and profit from their innovations, but can also constrain their use. IP is likely to be important for translation of GWAS studies into clinical use, although it remains to be seen just what role IP will play. As translation of GWAS is still at an early stage, there is the possibility for major stakeholders such as funders to play a role in developing innovative models of IP and translation which will optimise patient access.

G.2 Key issues

- How does traditional IP law fit with GWAS, particularly with reference to open access and data sharing?
- What types of IP may be relevant to GWAS?
- Does IP serve the role of promoting innovation or translation in GWAS?

A significant proportion of the human genome is already the subject of patents, and current genetic investigations are resulting in further patent filings (Jensen and Murray, 2005; Hopkins *et al.,* 2006). Concerns have been raised about the potential for proliferation of patents, resulting in patent thickets and a chilling effect on research and development, and as a result, reduced patient access (Caulfield *et al.*, 2006). This debate is on-going, and the question of how best to address the potential difficulties of gene patents for diagnostic testing in particular, is currently under consideration by government committees in both Australia (Parliament of Australia, Senate Community Affairs Committee, Inquiry into Gene Patents, 2009) and the USA (Secretary's Advisory Committee on Genetics, Health and Society, 2009).

Private ownership of the genome through intellectual property rights is in contrast to the model of 'open access' to the results of genetics and genomics research, which is publicised as necessary for the progression of science (see e.g. Fort Lauderdale Agreement (Wellcome Trust 2003)). This dispute between an open access and a private model has not been resolved since the publication of the human genome, and gives rise to many questions (Kaye *et al.*, 2009). However, whilst the current system of IP may not fit well with new genomics research, a good alternative system has not yet been developed or taken up (Piper and Gold, 2008).

There are various different ways that inventions arising from GWAS might be protected. There could be copyright in compilations or database rights (such as the databases of genetic information that are built from biobanks). Trade secrecy and the law of confidentiality could also be used to protect against disclosure of information kept confidential. However, the most likely means of intellectual property protection for genetic inventions such as isolated genes, SNPs or genetic tests is patents. There might also be IP in associated technologies such as software, hardware or related research tools.

Patents are traditionally seen as promoting innovation through a number of mechanisms. First, patents provide an incentive for inventors to risk the costs of innovation by providing a right to exclude use by others for a limited period. Second is the requirement of disclosure of the patented invention. A third possible mechanism is through the facilitation of investment to transform an invention into a marketable product (Secretary's Advisory Committee on Genetics, Health and Society, 2009).

How useful IP will be for the purpose of increasing innovation or translating an innovation into a useful product will depend to some extent on the nature of the innovation, whether industry is involved in the process or whether there are other drivers for innovation and translation in the circumstances. Patents can be used to block use and development of an invention, but they can also be useful for building partnerships and developing new technologies. It remains to be seen

how necessary IP will be in translational research from GWAS; much will depend on the roles and motivations of stakeholders.

Industry is increasingly involved in early stage genomics research (see for example the Procardis project: http://www.procardis.org/). Where industry is a partner in GWAS projects, then it is likely to be concerned with organising IP protection in its favour. However, research institutes and funders are in a strong bargaining position to encourage IP protection to be organised in ways that address their priorities and concerns as well (Hawkins *et al.,* 2009).

Genetic tests

Genetic tests are perhaps the most obvious outcome of GWAS research, but current models of patenting of individual genetic associations will not fit well with multiplex testing (Ray, 2008). Industry is likely to be a major player in genetic testing in the future, in contrast to how genetic testing is provided by the NHS at present. Commercial entities have already begun to offer 'life-style tests' direct to consumers and it is anticipated that genomic sequencing will shortly be available to all. Whilst it may take significant time and extra research, it is hoped that current genomics research will also lead to clinically relevant genetic tests which will allow, for example, disease stratification or early identification of common complex disorders (Ioannidis, 2009). Although the actual clinical usefulness of this technology at present is under debate, the legal framework needs to be developed.

G.3 Points to consider

- How are existing gene patents reconciled with intellectual property protection of GWAS?
- Is there a proliferation of IP rights in genomics, and does this result in a 'patent thicket'? Is royalty stacking an issue, and if so, how is it resolved?
- Are collaborative models such as patent pools being utilised?
- What role does industry play in research, and how does this change the IP models employed?
- What are the differences in IP protection between single gene and whole genome scans/multi-gene tests?
- What role do funders play in developing IP policies for innovations arising from GWAS?
- Given that much or most of this genomics research is publicly funded, should benefit sharing be mandated?

G.4 Key publications

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Committee on Intellectual Property Rights in Genomic and Protein Research and Innovation. *Reaping the Benefits of Genomics and Proteomic Research: Intellectual Property Rights, Innovation and Public Health.* Washington DC: National Academies Press, 2006.

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Hopkins, M. M. *et al. The Patenting of Human DNA: Global Trends in Public and Private Sector Activity* (The PATGEN Project). SPRU, Science and Technology Policy Research, University of Sussex, 2006.

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H. PUBLIC ENGAGEMENT

H.1 Summary

GWAS raises issues about the use of identifiable information for research purposes in a way that has not been experienced in medical research. GWAS raises issues that are challenging, such as feedback, how to protect privacy and how oversight bodies should make decisions in the public interest. The fact that GWAS creates data that are so detailed and personally identifiable raises issues about whether the current practice of anonymisation is sustainable. This in turn raises issues about the nature of the relationship between researchers and participants and how this should be framed in relation to GWAS. Therefore, it is important that a public engagement strategy is developed in relation to GWAS to ensure that policy and practice takes into account an understanding of the perspective of participants. From this understanding, appropriate governance structures and structures can be put in place to encourage public trust. To date, little public engagement has been carried out in relation to GWAS (McGuire *et al.*, 2008a).

Public engagement is now regarded as the gold standard for the introduction of new developments within genetics. Much work has been done in the field of public engagement, as it was assumed that this activity simply meant educating the public about what scientists and policymakers wished to do - the so-called 'deficit model' (Leroux *et al.*, 1998; Levine *et al.*, 2005; Rowe and Frewer, 2000). For example, the OECD has presented a three-tiered framework of public engagement (OECD, 2001). The most basic level is the one-way relationship of information-sharing with the public. The second level is public consultation including public feedback; this is more of a two-way relationship. The third level is active public participation, which encourages a partnership approach whereby citizens actively engage with decision-making and policy-making processes, and have an active role in proposing policy options and shaping policy dialogue. Active participation is seen as a way of ensuring legitimacy and accountability in public sector organisations, an example being the National Institute for Health and Clinical Excellence's (NICE) Citizen Council. While public engagement models have tended to canvass opinions, few have provided the means to reconcile differences of opinion between different stakeholders, or to come up with policy recommendations that can be translated into practice (Haimes E and Whong-Barr, 2007).

H.2 PUBLICATIONS

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3. CASE REPORTS

***Re Creutzfeldt-Jakob Disease Litigation (No.2* (2000) 54 B.M.L.R. 8 (the CJD Litigation).**

The duty of care owed by a researcher to a participant in a research project was addressed more recently in the CJD litigation. This litigation called upon the judge (Morland, J) to make a series of difficult decisions in complex human and medico-legal areas:

> 'The human growth hormone litigation is not only the first large scale medical product liability case to come to a full trial but also provides interesting guidance in a number of areas: liability in negligence of government departments and of expert committees comprised of volunteers; the efficacy of trial of generic issues, the nature of claims by those suffering psychiatric injury by reason of exposure to the risk of future injury; the proper approach to causation of injury on an uncertain date and the proper application of cost-sharing principles.
>
> Between 1959 and 1985 almost 2,000 children of very short stature were treated with growth hormone extracted from the pituitary glands of human corpses at post-mortem. The purpose was to compensate for the child's inadequate secretion of growth hormone which had resulted in dwarfism and the treatment was in many cases efficacious in bringing the child into the normal range of stature.
>
> The programme was run until July 1, 1977 by the Medical Research Council ... It was financed by the Department of Health and supervised by a Working Party of the MRC at the Institute of Child Health … The Department of Health took over the programme in 1977 although the MRC retained the responsibility for collecting and processing the pituitaries until 1980. The programme had begun as a clinical trial (thus underlining its research aspect) but, by the time of transfer in 1977, had in effect become a therapeutic programme by virtue of its extent and scientific stability.
>
> The programme came to a sudden end on May 9, 1985 after it became clear that three participants in the United States growth hormone programme had died from Creutzfeldt-Jakob disease, now recognised as the human species equivalent of BSE.' (Mildred, 1998).

In the CJD litigation, Morland, J said:

> 'Advances in medical science will inevitably not be a smooth path of progress. Without negligence discoveries, theories or advances thought to be valid or beneficial will with the passage of time prove to be ill-founded and the risks and dangers involved too great to justify the continuance of a particular therapy or the use of a particular drug. From the mistakes of the past can be learnt the successes of the future.
>
> The Courts must be very cautious in condemning a clinical trial or therapeutic programme. Too ready a labelling of an act or omission as negligent by the courts could stultify progress in medical and scientific research and render eminent experts reluctant to serve on Committees voluntarily. However during the clinical trial of a new drug or form of treatment and especially when the clinical trial is becoming a general therapeutic programme all reasonably practicable steps should be taken to minimise dangers and side effects. To discharge this duty constant alert and inquiring evaluation of the trial or programme is required. I do not accept that a Government Department or a Quasi Governmental Agency such as the MRC can discharge this duty by a lower standard of care than a commercial pharmaceutical company. In my judgment shortage and limitation of funding and the fact that the collection of pituitaries the production of

HGH [human growth hormone] and the allocation of HGH to selected approved patients were under the responsibility and supervision of Committees manned by unpaid volunteers are irrelevant. In my judgment the same duty with the same standard of care is owed to all patients who are the subjects of clinical trials or new therapeutic programmes whether the responsibility of a pharmaceutical company, Government Department or other agency….

The standard of care in my judgment to be imposed in respect of a committee is that of a reasonably competent and carefully enquiring group of professionals of the relevant disciplines of sufficient standing to be entrusted with the membership of that committee bearing in mind it is a committee which is not merely advisory but is carrying out executive and administrative functions.'

Osman v The United Kingdom [1998] 29 EHRR 245

In the case of *Osman v The United Kingdom* [1998] 29 EHRR 245 it was held that the UK is under an obligation to take 'appropriate steps to safeguard the lives of those in its jurisdiction' under Art.2 of the Human Rights Act 1998.

> Article 2:
> 'Everyone's right to life shall be protected by law. No one shall be deprived of his life intentionally save in the execution of a sentence of a court following his conviction of a crime for which this penalty is provided by law.
>
> Deprivation of life shall not be regarded as inflicted in contravention of this article when it results from the use of force which is no more than absolutely necessary:
>
> (a) in defence of any person from unlawful violence
> (b) in order to effect a lawful arrest or to prevent escape of a person lawfully detained
> (c) in action lawfully taken for the purpose of quelling a riot or insurrection.'

Article 2 protects the right to life, and if authorities have been seriously negligent so as to create a real risk of loss of life or serious harm, then legal proceedings can be brought against them.

The House of Lords ruling on UK police immunity from actions for negligence by police was overturned by the European Court of Human Rights in the Osman case in 1998. It was brought by Mulkiye Osman and her son Ahmet, who had been badly injured. His father, Ali, was killed in 1988 by a man about whom the family had warned police.

The ECtHR held that there had been no violation of Articles 2 or 8 ECHR. As regards Article 2, the majority were of the view that in the circumstances of the case the police could not be criticised for attaching weight to the presumption of innocence or failing to use powers of arrest, search and seizure, because the Applicants had failed to point to any decisive stage in the sequence of events when it could be said that the police knew or ought to have known that the lives of the Osman family were at real and immediate risk. Although the court ruled that in those circumstances the police could not be criticised, if, in future, they failed to take appropriate measures to avoid risks, the United Kingdom might be liable for that failure.

S. and Marper v The United Kingdom 30562/04 [2008] ECHR 1581

The European Court of Human Rights (ECtHR) in Strasbourg published its judgment in the case of *S. and Marper v The United Kingdom* 30562/04 [2008] ECHR 1581 on 4 December 2008. This case concerned a juvenile, aged 11 when he was arrested (and who cannot be named for legal reasons), who was charged with attempted robbery, but was acquitted; and the other was an adult, Michael Marper, who was charged with harassment, but whose case did not go to court because the charges were dropped and the case against him was discontinued. After the proceedings against the Applicants had been terminated, they both unsuccessfully requested that their fingerprints, cellular samples and DNA profiles be destroyed. The information had been stored on the basis of a law (section 64 (1A) of the Police and Criminal Evidence Act 1984, as amended by Criminal Justice and Police Act 2001 c. 16, section 82) authorising such retention. Under this Act, the police in the UK can hold fingerprints and DNA samples of persons investigated for a crime for an indefinite period of time, whereas those who volunteer fingerprints or samples in connection with an investigation, and who are not suspects in the offence, must have their samples and fingerprints destroyed (Rainey, 2008).

The Applicants applied for judicial review of the police decision, and the court at first instance upheld the police decision. The subsequent appeals to the Court of Appeal and the House of Lords were dismissed. The decision of the House of Lords (*R v Chief Constable of South Yorkshire Police* (2004) UKHL 39) overturned the ruling of the Court of Appeal, and it found that there had been no violation of Article 8 of the European Convention on Human Rights (ECHR) in these cases. First, the majority found that Article 8 itself was not engaged, because the retention of DNA profiles and fingerprints were held as neutral information to which only a modest, if any, interference with private life was attached, and the future misuse of such samples was not relevant and limited by the statute or judicial decision making. Baroness Hale disagreed with the majority on this point and noted that the holding of knowledge of genetic makeup is an intimate part of a person's private life. However, all the Lords agreed that if any interference did arise, any such interference could be justified under Article 8(2) as being necessary in a democratic society, given the benefits in preventing and prosecuting crime (Rainey, 2008).

Both Applicants in the Marper case argued that the provisions were in violation of Article 8 ECHR, because the retention of their fingerprints, cellular samples and DNA profiles was an interference with private life, which could not be justified under Article 8(2) which permits States to interfere with privacy if a measure is in accordance with the law, meets a legitimate aim and is necessary in a democratic society.

The main issue in the case, which was heard by the ECtHR, was whether the retention by the authorities of the Applicant's fingerprints, cellular samples and DNA profiles, after criminal proceedings against the Applicants were terminated, is consistent with human rights law. The ECtHR noted that fingerprints, cellular samples and DNA profiles constituted 'personal data' within the meaning of the 1981 Council of Europe Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data (CETS No.108 Strasbourg 28.1.1981). The Court further indicated that domestic (national) law had to afford appropriate safeguards to prevent any use of personal data as could be inconsistent with the guarantees of Article 8 ECHR. The Court added that the need for such safeguards was all the greater where the protection of personal data undergoing automatic processing was concerned, not least where such data were used for police purposes.

The ECtHR usefully set out the general principles that it applied to the concept of 'private life' (at para. 66):

> 66. The Court recalls that the concept of 'private life' is a broad term not susceptible to exhaustive definition. It covers the physical and psychological integrity of a person (see *Pretty v the United Kingdom*, no. 2346/02, § 61, ECHR 2002 III, 35 EHRR 1, and *Y.F. v*

Turkey, no. 24209/94, § 33, ECHR 2003 IX, 39 EHRR 34). It can therefore embrace multiple aspects of the person's physical and social identity (see *Mikulić v Croatia*, no. 53176/99, § 53, ECHR 2002-I, BAILII: [2002] ECHR 27). Elements such as, for example, gender identification, name and sexual orientation and sexual life fall within the personal sphere protected by Article 8 (see, among other authorities, *Bensaid v the United Kingdom*, no. 44599/98, § 47, ECHR 2001, 33 EHRR 10, I with further references, and *Peck v the United Kingdom*, no. 44647/98, § 57, ECHR 2003 I, 36 EHRR 41). Beyond a person's name, his or her private and family life may include other means of personal identification and of linking to a family (see *mutatis mutandis Burghartz v Switzerland*, 22 February 1994, § 24, Series A no. 280 B; and *Ünal Tekeli v Turkey*, no. 29865/96, § 42, ECHR 2004 X (extracts), 42 EHRR 53). Information about the person's health is an important element of private life (see *Z. v Finland*, 25 February 1997, § 71, *Reports of Judgments and Decisions* 1997 I, 25 EHRR 371). The Court furthermore considers that an individual's ethnic identity must be regarded as another such element (see in particular Article 6 of the Data Protection Convention quoted in paragraph 41 above, which lists personal data revealing racial origin as a special category of data along with other sensitive information about an individual). Article 8 protects in addition a right to personal development, and the right to establish and develop relationships with other human beings and the outside world (see, for example, *Burghartz*, cited above, opinion of the Commission, p. 37, § 47, and *Friedl v Austria*, judgment of 31 January 1995, Series A no. 305-B, opinion of the Commission, p. 20, § 45, 21 EHRR 83). The concept of private life moreover includes elements relating to a person's right to their image (*Sciacca v Italy*, no. 50774/99, § 29, ECHR 2005-I, 43 EHRR 20).

67. The mere storing of data relating to the private life of an individual amounts to an interference within the meaning of Article 8 (see *Leander v Sweden,* 26 March 1987, § 48, Series A no. 116, 9 EHRR 433). The subsequent use of the stored information has no bearing on that finding (*Amann v Switzerland* [GC], no. 27798/95, § 69, ECHR 2000-II, 30 EHRR 843). However, in determining whether the personal information retained by the authorities involves any of the private-life aspects mentioned above, the Court will have due regard to the specific context in which the information at issue has been recorded and retained, the nature of the records, the way in which these records are used and processed and the results that may be obtained (see, *mutatis mutandis*, *Friedl*, cited above, §§49–51, and *Peck v the United Kingdom*, cited above, § 59).

The ECtHR held unanimously that there had been a violation of Article 8 ECHR, stating:

> 'In conclusion, the Court finds that the blanket and indiscriminate nature of the powers of retention of the fingerprints, cellular samples and DNA profiles of persons suspected but not found convicted of offences, as applied in the case of the present applicants, fails to strike a fair balance between the competing public and private interests and that the respondent State has overstepped any acceptable margin of appreciation in this regard. Accordingly, the retention at issue constitutes a disproportionate interference with the applicants' right to respect for private life and cannot be regarded as necessary in a democratic society.'

In this case, the ECtHR does not dispute the legitimate retention of personal data in a database, but the court's role is to ensure that safeguards regarding privacy are established that strike the appropriate balance between protecting the individual and protecting the community.

The ECtHR also noted in the *Marper* case, that England, Wales and Northern Ireland (not Scotland) appeared to be the only jurisdictions within 47 member states of the Council of Europe to allow the indefinite retention of fingerprints and DNA material of any person of any age suspected of any recordable offence.

One of the arguments put forward by the UK Government was that as a pioneer in new techniques, it should be given a greater margin of discretion, but the ECtHR noted that pioneers of new technology have an even greater responsibility to ensure rights are protected:

> 'The Court considers that any State claiming a pioneer role in the development of new technologies bears special responsibility for striking the right balance…' (Para 112).

It would appear that the ECtHR is reminding the UK Government to take greater account of individual rights when developing the law, and it is advising the UK Government to look to other states when considering how to maintain a database that is proportionate and balanced. It therefore seems appropriate that these principles be extended to GWAS research.

The key points from this judgment insofar as this research is concerned, are that:

- given the nature and the amount of personal data that are contained in cellular samples, their retention must be regarded as interfering with the right to respect for the private lives of the individuals concerned
- the capacity of DNA profiles to provide a means of identifying genetic relationships is in itself sufficient to conclude that their retention interferes with the right to respect for the private lives of the individuals concerned
- the retention of both cellular samples and DNA profiles discloses an interference with the Applicants' right to respect for their private lives, within the meaning of Article 8 ECHR
- any State claiming a pioneer role in the development of technologies bears a special responsibility for striking the right balance in this regard.

Since the judgment in this case, 'S' and Michael Marper have had their DNA destroyed and their records removed from the DNA and fingerprint databases. Future cases in Europe determining the question of whether or not retention of an individual's DNA and fingerprints is lawful, will do so with reference to this ECtHR judgment.

Yearworth and Others v North Bristol NHS Trust* [2009] EWCA Civ 39; [2009] WLR (D) 34. *(The Yearworth case)

Another interesting development of the law concerning samples and whether they constitute 'property' was set out in the landmark decision of the Court of Appeal in the case of *Yearworth and Others v North Bristol NHS Trust* [2009] EWCA Civ 39; [2009] WLR (D) 34.
In this case, six men were diagnosed with cancer and received treatment at Southmead Hospital, Bristol, where they were told that their chemotherapy treatment might damage their fertility. They were therefore asked whether they wanted to produce samples of semen prior to the treatment. The six claimants accepted the advice of their clinicians to produce samples for frozen storage for possible future use, in case the treatment damaged their fertility. However, in June 2003 the liquid nitrogen in storage tanks where the samples were kept fell below the required level, and the semen thawed out and was lost. The men believed that their hopes of fathering children in the future were dashed, and they claimed damages for the loss, on the basis that their sperm samples were property owned by them, whose loss or damage entitled them to bring an action for negligence.

The Court of Appeal overturned the County Court decision that found that the men had no claim against the NHS Trust. Although the Court of Appeal agreed that the judge at first instance had rightly held that damage to, and consequential loss of the sperm samples did not constitute personal injury, in an introduction to the judgment Lord Judge said: 'The appeals raise interesting questions about the application of common law principles to the ever-expanding frontiers of medical science.' He said that the 'novel question' was whether the men could sue over the loss of semen samples which they had produced for possible later use and which the hospital had promised to freeze and store.

The Appeal Court judgment stated that developments in medical science now required a re-analysis of the common law's treatment of and approach to ownership of parts or products of the human body, whether for an action in negligence or otherwise. Although they found that the law had to some extent begun to be refined in relation both to ownership of a human corpse and to parts of it, it was still silent about parts or products of a living human body, probably because until recently medical science did not endow them with any value or significance (see report by Rajaratnam 2009). The Court of Appeal held that:

> 'A sample of sperm from a person undergoing chemotherapy, which a hospital stored in case he became infertile after the treatment, was that person's property and its loss or damage was capable of establishing a claim in negligence.'

***Common Services Agency v Scottish Information Commissioner (Scotland)*, [2008] UKHL 47 (The Collie case)**

On the question of how researchers gain access to different datasets for the purposes of obtaining research data (which is relevant to data-generated projects using GWAS technology in the UK), there was an interesting development in the case of *Common Services Agency v Scottish Information Commissioner (Scotland)*, [2008] UKHL 47. Mechanisms to protect the privacy of research participants have traditionally been informed consent and anonymisation of datasets. This case is regarded as a landmark case on the compatibility of data protection and freedom of information laws, which was heard by the House of Lords in April 2008. The case focused on an NHS agency's refusal of a Freedom of Information (FOI) (Scotland) request for statistics about childhood leukaemia. The agency feared that the information could identify individual children and breach their privacy, and having lost its case in the Scottish courts, it appealed to the House of Lords.

The Common Services Agency (CSA) case centred upon a request by Michael Collie, a researcher acting on behalf of a Member of the Scottish Parliament, for information regarding the incidence of childhood leukaemia for both sexes in the age range of 0–14 years, between the years 1990 and 2003, for all the Dumfries and Galloway postal area by census ward. The purpose of the request was to establish whether a nearby nuclear power station and military firing range has an effect on incidences of cancer. It was undisputed by the CSA that there was genuine public interest in the disclosure of the requested information.

The CSA (an NHS agency) refused the request on the grounds that the information requested was of such a specific nature that, if it was disclosed, there was a significant risk of indirect identification of living individuals. The CSA considered the information to be 'personal data' within the meaning of section 1(1) of the Data Protection Act 1998, and therefore exempt from disclosure under the 2002 Freedom of Information (Scotland) Act.

Collie referred the CSA's decision to withhold the requested information to the Scottish Information Commissioner (SIC), who determined that the information could be released if the personal data was 'barnardised' (i.e. a method aimed at minimising the risk of individuals being identified by modifying data which contains low numbers, by adding 0, +1, or –1 to all values where the true value lay in the range from 2 to 4 and adding 0 or +1 to cells where the value was 1). The SIC's view was that rendering personal data anonymous in such a way would enable the information to be disclosed.

The CSA appealed the SIC's decision to the Scottish Court of Session, which upheld the Commissioner's decision, stating that the 'barnardised' data were 'held' by the agency for the purposes of the 2002 Act and was sufficiently anonymous that it did not constitute personal data. The SIC was, therefore, entitled to require the CSA to disclose this information in the exercise of his supervisory powers under the 2002 Act.

The CSA appealed the decision of the Court of Session to the House of Lords. The Lords were concerned as to the potential impact of their decision, noting that, if they decided in favour of the CSA, such a decision could potentially affect the ability of individuals to use their rights under the 2002 Act to obtain anonymous statistical information from Scottish public authorities. The key issues upon which the appeal turned were: if the CSA was required to barnardise the information requested, did it 'hold' such information for the purposes of the 2002 Act; and, if the CSA was determined to hold such information, was this information 'personal data' for the purposes of the 1998 Act?

In relation to the first issue, the CSA argued that the process of barnardisation would require the production of information that was different from that originally held by it at the time of the request, and the terms of the 2002 Act did not, therefore, apply. The Lords considered that

position to be unduly restrictive, and not in compliance with the underlying principles of the 2002 Act. The Lords noted that the terms of the 2002 Act, in respect of the holding of information, should be construed as liberally as possible, and that the effect of barnardisation would be to disguise information that was already held by the CSA at the time of the request. Disclosure of the barnardised information would amount to the provision of information in an anonymised format that concealed those parts of it that have to be withheld, rather than the creation of new information not held at the time of the request

In relation to the second issue, the Lords held that information requested should not be disclosed unless it could either be anonymised so that it was not personal data, or could be released in a form which did not contravene one of the data protection principles under the 1998 Act.
The Lords noted that, in the original decision, the SIC had not properly considered the question of whether the barnardised data would constitute personal data in terms of the 1998 Act and, if so, whether its disclosure would be in compliance with the terms of that Act and the data protection principles contained therein. Accordingly, the Lords determined that the appropriate course of action was for Collie's application to be remitted to the SIC for a determination as to whether the information in question can be sufficiently anonymised for it not to constitute personal data.

The result of this case in respect of researchers gaining access to research data, appears to be that where a request is made for information under the Freedom of Information legislation, consideration will first have to be given as to whether the information can be sufficiently anonymised for it not to be 'personal data'. If it cannot be so anonymised, it would then have to be determined whether its disclosure would comply with the data protection principles set out in the 1998 Data Protection Act.

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