

# Background document for revision of guidance: consent and confidentiality in clinical genetic practice

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June 2017

The BSGM commissioned Dr Sandi Dheensa to write this background paper to highlight areas that may need revision, or where gaps have arisen due to changes in practice, since the 2011 edition of Consent and Confidentiality in Clinical Genetic Practice guidance. It also attempts to recognise that clinical practice now extends well beyond the specialty of clinical genetics.

The document aligns each section with the previous guidance from 2011, discusses recent developments and research, and makes suggestions for issues that the new guidance may need to incorporate.

The BSGM convened a workshop on 26/06/17 to discuss the document at. The aim of that workshop was to consider:

1. In which areas is the 2011 guidance now out of date? Which aspects are still useful and can be retained?
2. What developments have there been since 2011 that need to be considered in new guidance? Are there relevant research literatures, other professional guidance, or developments in the law to inform this?
3. How do we ensure new guidance is relevant to, and usable by, mainstream specialties dealing with genomic medicine, especially as the 100,000 genome project finishes recruiting and becomes embedded in NHS practice?
4. Do we need a revision or a re-write of the guidance?
5. Are the case histories still useful? What additional examples might be helpful?

This work is being led by the ethics and policy committee of the BSGM in conjunction with the Joint Committee on Medical Genomics. Please consult these committees for further updates.

# British Society Genetic Medicine (BSGM) and Joint Committee on Genomics in Medicine (JCGM) consent & confidentiality in clinical genetic practice: background paper

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## Summary

**A non-exhaustive list of the issues that the JCGM/ BSGM might wish to consider for the new Consent and Confidentiality guidance are as follows:**

1. What issues the so-called mainstreaming of genetics/ genomics raises for consent and confidentiality practices. How can guidance address?
2. What recommendations to make about facilitating communication within families, especially how mainstream specialties might do so, and what role adjuncts to communication such as 'to whom it may concern' letters and web-tools (such as electronic health records with additional functionality that allow for sharing information) should play.
3. What recommendations to make regarding confidentiality, the value of adopting a familial approach (i.e. treating genetic information as confidential to families, not individuals), and ways to address concerns about separating personal and familial genetic information in practice.
4. What recommendations to make about responsibilities to re-contact in the light of new genomic interpretations and other potential 're-contact triggers'.
5. Regarding the template 'record of discussion' form used for genetic testing- often called DNA consent form:
  - whether a distinction between personal clinical and familial genetic information should be made explicit.
  - whether we should recommend that the template "consent form" for genetic/ genomic testing is used more widely (without ad hoc unworkable riders) to facilitate the appropriate sharing of information between different genetic services.
  - What amendments to the form would be helpful- eg the current statement about offering patients the choice to be re-contacted before further tests are done on their sample may need amending (it is now less likely that labs will want to run more sensitive tests on groupings of patients without further specific requests from the clinicians involved).
  - how it can be made clearer that professional judgement (rather than deference to specific details on consent forms) remains important when making decisions, for example about communicating to specific relatives after the death of testee.
6. Regarding consent in whole genome analysis, what recommendations to make given that:
  - consent can only ever be broad, because of the range of possible findings and (eg in 'hybrid' practices, where clinical care and research are integrated) because the range of research studies for which genetic information might be used is often unknown at the outset.
  - ways to operationalise consent as a *process*, rather than as a one-off event, are needed, i.e. to allow patients to update their preferences, and to enable HCPs to ensure consent remains valid, over time. In particular because evidence about particular genomic variants may change.

7. Regarding communication of results with patients:
  - Which, if any, potential incidental findings (IFs) patients should be told about, and how, if at all, this should be broached in the consent process for genomic testing.
  - Whether, in future, certain findings should routinely be offered to patients/families undergoing genome tests (as per offer of 'additional findings' in the 100,000 Genomes Project).
  - Whether and when the terms 'incidental', 'secondary', 'unsolicited', and 'additional' are appropriate to use.
  - What should happen if an adult onset risk is discovered in a child from an already sequenced genome (e.g., from a panel test, whole-exome, or whole-genome sequence) as opposed to in response to a parental/ guardian request (covered by existing 2010 British Society for Human Genetics guidance).
  - Whether alternative forms of data storage and access may mitigate disclosure about future risks for fear of otherwise 'losing' a result
8. What recommendations to make regarding hybrid practice that combines research and clinical care (as per the 100,000 Genomes Project) and what 'counts' as ethical practice, given that conventionally, there are differences in approaches between, and governance of, research and clinical practice.
9. Regarding patient databases and increasing digitization:
  - whether and how the use of patient held electronic health records (such as "My Medical Record" or "Patients Know Best") would affect the 2011 Consent and Confidentiality guidance on the use of medical records to confirm diagnoses
  - the role of existing and lapsed patient registries in genetic services and whether these create professional obligations about, for example, (re)contacting
  - how the biorepository generated by the 100,000 genome project ought to be governed and used once the project ends, and what, if any, role UK genetic services might play in these decisions
10. Considering discussion-based activities (e.g., UK Genethics Forum, Practical ethics GeCIP, clinical ethics committees) as effective routes to implementation of new guidance.
11. How key legal issues might affect future practices e.g., cases such as ABC vs St George's Healthcare NHS Trust and the passing of the General Data Protection Regulation (GDPR). Regarding the GDPR, what counts as 'personal' information is relevant in two contexts: (a) collecting information about patients' relatives for the purposes of drawing a pedigree and (b) using genetic information generated through one person's test to test that person's relatives. Any consideration should also take into account that General Medical Council (GMC) guidance states genetic information is both "about your patient [and]...about others", and that this information can be disclosed to benefit others (i.e. protect them from death or serious harm).

## Introduction

This background paper highlights aspects of the 2011 Consent and Confidentiality guidance that merit consideration in light of recent and upcoming developments. These include:

### 1. Clinical practice developments:

- the introduction of 'broad' genome tests into routine and mainstream practice (ie the move away from targeted testing for specific genes done mainly in specialist services)
- the launch of the 100,000 Genomes Project—a venture that will expedite the translation of whole-genome sequencing (WGS) and genomics research into clinical practice.

2. Pertinent international debate and scholarship on developments in genomic medicine, including about the ethical, social, and legal aspects of emerging genetic and genomic practices will be summarised. This includes relevant research by the Clinical Ethics and Law Unit at Southampton (CELS), summarised in Box 1 .

### 3. Legislation/guidance developments and forthcoming revisions:

- Relevant court cases that have involved nondisclosure of a relevant risk to a relative (ABC vs St George's Healthcare NHS Trust 2015 and Smith & Anor vs University of Leicester NHS Trust 2016)
- Revised Caldicott guidance (2016)
- Revised GMC confidentiality guidance (2017)
- Forthcoming consultation (begins Autumn 2017) on the GMC consent guidance revision (last published 2009)
- New European General Data Protection Regulation (GDPR). Guidance on how this applies to NHS data will be published shortly we understand.
- Forthcoming revision of the Association of British Insurers (ABI) and UK government concordat and moratorium on the use of predictive genetic tests (due for review November 2017)
- New codes of practice (2017) for the Human Tissue Act (2004)
- New edition of the Royal College of Pathologists 'Retention and storage of pathological records and specimens' guidance (2015)

Each section of the 2011 guidance is discussed below with reference to the above developments.

## **Box 1: CELS research referred to in this background paper**

### **“Consent and confidentiality in genetic medicine”**

This was a research project funded by a medical charity grant, awarded to investigate the 2011 Consent and Confidentiality guidance and the gaps highlighted in that edition. It involved several phases. Firstly, there was a systematic review of international studies to explore patients' and healthcare professionals' (HCPs) views and practices around confidentiality in clinical genetics. Secondly, there were qualitative focus groups with UK healthcare professionals (incl laboratory scientists) and interviews with NHS patients (who had received targeted genetic testing) to explore their views about the familial approach (or 'joint account' approach) to confidentiality (see Section 5.6 and Appendix D Box 5 for more details). An analysis of all UK genetic service consent forms was also undertaken to investigate how they compared with a consent form suggested in the 2011 Consent and Confidentiality guidance (see Section 3.10 and Appendix D Box 3 for more details). This research moreover explored current practices around the way HCPs facilitate communication between relatives. (See Section 1 and Appendix D Box 2 for more details).

### **“Mainstreaming genetics: re-contacting patients in a dynamic healthcare environment”**

This was an ESRC-funded collaboration between the Universities of Exeter, Southampton, and Cardiff that aimed to explore the ethical and legal challenges surrounding the possible obligation to re-contact former patients when information becomes available about new tests, treatments, surveillance options, or about the clinical significance/pathogenicity of a particular gene variant. The project explored patients and HCPs' views, practices, and expectations using a UK-wide HCP survey, a Europe-wide HCP survey, a patient survey, and interviews with UK HCPs and patients. This led to a meeting with the European Society of Human Genetics (ESHG) Public and Professional Policy Committee (PPPC) at the ESHG's annual conference (May 2017). (See Section 3.4 and Appendix D Box 4 for more details) and a policy workshop is imminent.

### **“Incidental Findings (IFs) from genetic tests”**

This NIHR-funded research aimed to gain an empirical and ethical insight into the experiences and attitudes of healthcare users and professionals towards such IFs from genetic testing, using both quantitative and qualitative research methods. It involved 27 clinic observations and 48 interviews with patients and HCPs and included several examples of IFs found in NHS practice- largely from comparative microarray testing. It highlighted that patients want to know about the possibility of 'other' findings but that they did not think consent (or more importantly, refusal of consent) would always be a workable option (See Section 3.7 for more details on IFs).

### **“Views and experiences of participants in 100,000 Genomes Project”**

Through a combination of observations, questionnaires, interviews, and focus groups, this Wellcome Trust-funded research has explored the challenges and opportunities raised by WGS and the 100,000 Genomes Project more generally. One aim of this project has been to help determine good practice for consent and feedback of the so-called 'additional findings' as well as to explore an appropriate ethical framework for 'hybrid' activity that combines clinical care and broad research by academic, commercial, and industry actors. The questionnaire arm asks about primary motivations for taking part (eg research or diagnosis) and has nearly 1000 responses. The data is currently being evaluated. This work contributes to the Practical Ethics strand of the Ethics and Social Science Genomics England Clinical Interpretation Partnership (GECIP).

### “Using digital tools to facilitate the clinical integration of WGS”

This Health Education England-funded pilot intervention is exploring the acceptability and feasibility of using web-tools (electronic health records with integrated apps) to facilitate the communication of genetic risk information among families. (See Section 1 for more details about communication).

Publications from these research studies available on request (some included in reference list)

## 1 Fundamentals of clinical genetic practice

Much of section 1 of the 2011 Consent and Confidentiality guidance still holds, but fundamental practice has already seen a more widespread shift from targeted testing of individual genes<sup>1</sup> to whole genome tests and their incorporation into mainstream medical practice<sup>2</sup>, so that some nuanced aspects of practice have changed. Much of this document will discuss the way that recent developments affect this fundamental practice. In this section, two issues are highlighted: (1) the definition of ‘genetic information’ (which is useful to discuss here because it frames the rest of the discussion in the document) and (2) the way HCPs facilitate communication in the family.

### 1.1 The definition of 'genetic information'

The 2011 guidance states that genetic ‘information’ covers “a wide range of different types of information...variations in the genetic code that are simply part of normal human variation and that carry no known health consequences ...[and] for example, the inferences about a person’s genetic code that can be made from their appearance or from a particular family history of a condition”.

A development since 2011 is the passing of the General data protection regulation (GDPR), which comes into effect on 25 May 2018. Like the Data Protection Act (DPA), the GDPR applies to ‘personal data’. However, “the GDPR’s definition is more detailed and makes it clear that information such as an online identifier – e.g. an IP address – can be personal data. The more expansive definition provides for a wide range of personal identifiers to constitute personal data.” (ICO, 2016)<sup>3</sup>. Although “specific derogations are defined for data concerning health”, according to the European Society of Radiology (ESR, 2017), HCPs will have to prepare for “[increased] data access for patients, [new] rules for data processing including explicit consent of the data subject in the absence of derogations, or technical and organisational safeguards” (p295).

Unlike the DPA, the GDPR specifically defines genetic information: “personal data relating to the inherited or acquired genetic characteristics of a natural person which result from the analysis of a biological sample from the natural person in question, in particular chromosomal, deoxyribonucleic acid (DNA) or ribonucleic acid (RNA) analysis, or from the analysis of another

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<sup>1</sup> Acknowledging of course that karyotype analysis was an early form of broad genome testing, just at a much lower resolution than available through WGS/ WES/CMH

<sup>2</sup> The work of the PHG Foundation on Genomics plays an important role in facilitating the integration of appropriate consent and confidentiality practices into non-genetics specialties.

<sup>3</sup> <https://ico.org.uk/for-organisations/data-protection-reform/>

element enabling equivalent information to be obtained” (rec 34). It classifies genetic data as sensitive.

In light of the GDPR's definitions on what counts as 'personal' information, the new guidance might wish to comment on the practice of collecting information about patients' relatives for the purposes of drawing a pedigree, and using genetic information generated through one person's test in order to test that person's relatives. During previous iterations of the guidance it was held (following advice from the ICO) that holding family history information in medical records was acceptable, as long as consent was sought if details other than identifiers were collected. New guidance may want to consider how this new definition of personal information affects current practice of using familial information to target appropriate testing in relatives, and should also take into account that GMC guidance states that genetic information is both 'about your patient [and]...about others', and that this information can be disclosed to benefit others (i.e. protect them from death or serious harm). Further exploration of the distinction between 'use' of and 'disclosure' of genetic information might be useful. These considerations are relevant to any approaches that distinguish personal clinical and familial genetic approaches to confidentiality, discussed further in Section 5.

## 1.2 Sharing information in the family

A fundamental aspect of genetics practice, as mentioned in Section 1 of the original guidance, is that after making a genetic diagnosis, HCPs will usually ask the individual patient to share their results with the relevant relatives.<sup>4</sup> This practice is in line with Europe-wide genetic testing guidance published since the JCGM guidance. These recommended that HCPs discuss the familial implications of any test with a patient, as well as a strategy on how to tell relatives, and that HCPs offer “written material to help the counsellee to spread the information in the family.” (Eurogentest, 2011). This written information is often in the form of 'dear relative' or 'to whom it may concern' letters to alert them of their risk and help relatives to seek a referral and may be more or less targeted, and followed –up to see whether dissemination has taken place- depending on local practices

Since 2011, there has been some relevant research about sharing information in the family. Hodgson and Gaff (2016) looked at the proportions of relatives told about a genetic risk across several (albeit older and non-UK) studies and pointed out that 15-20% of relatives do not find out about their risk (Clayton et al., 2005; Gaff et al., 2007; Landsbergen et al., 2005). Our research highlights that such figures demonstrate only the known non-disclosure rates. It is often not possible to measure, for example, whether relatives—especially those living in other catchment areas or countries—get to hear about their risk. Across 17 focus groups, around 30 cases where HCPs thought disclosure had not happened were highlighted (see Appendix D Box 5 for more detail).

Wiens et al. (2013) systematically reviewed the literature about patient experience of family communication. They found that the most often cited barriers were 'a desire to protect oneself and relatives' (e.g., against harmful information and against feelings of guilt), 'family dynamics' (e.g., having a poor relationship, or no relationship at all, with the relative), and a perception that

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<sup>4</sup> There was little exploration of when, if ever, direct contact between HCPs and relatives was justified—more on this in Section 5.

the information is not relevant. This suggests patients need some support communicating 'bad news', navigating difficult family relationships, and understanding the information.

CELS' research has shown that communication by 'to whom it may concern' letters may not always improve communication. Patients may find it difficult to hand out these letters—not just because the message is 'bad news', but also because it can be difficult for them to know to whom they should be handed out. Moreover, this research highlighted that there was a lack of clarity among HCPs and patients about whether simply passing on the letter discharged responsibilities sufficiently. HCPs thought there ought to be clearer recommendations on when they should be more involved or for patients to contact them for help if they were struggling with communication.

**>>See Appendix D Box 2 for more details about these research findings.**

Interestingly, in the Netherlands, following a particular case where communication did not happen, HCPs specifically name relatives to whom patients should give letters and check whether they have received them a month later. However, this has never been properly evaluated and there is no standardisation (personal communication).

The new Consent and Confidentiality guidance might wish to consider what recommendations to make about facilitating communication within families, especially how mainstream specialties might do so, and whether alternatives to the letter ought to be considered. CELS' research as well as research by Goodman et al. (2016) might help to identify alternatives that use web-tools. Family communication is a thread that will run throughout the rest of this document and is discussed more in Section 5.

## 2 Issues of confidentiality in clinical practice

The 2011 Consent and Confidentiality guidance pointed out that HCPs are under an ethical and a legal duty to keep patients' personal information confidential, but that this duty is not absolute. Since the 2011 guidance, GMC confidentiality guidance has been revised (GMC, 2017). The message regarding disclosure of information is largely the same as in the previous version: HCPs may disclose personal information without breaching their duty of confidentiality if (1) the patient consents ('whether implicitly or explicitly'—though these terms are not defined); (2) the disclosure is of overall benefit to a patient who lacks the capacity to consent; (3) the disclosure is required by law or is permitted/approved under a statutory process; and (4) when disclosure can be justified in the public interest (para 9).

The aspect of the revised GMC guidance pertaining to genetic information remains unchanged:

“If a patient refuses to consent to information being disclosed that would benefit others, disclosure might still be justified in the public interest if failure to disclose the information leaves others at risk of death or serious harm...you will need to balance your duty to make the care of your patient your first concern against your duty to help protect the other person from serious harm.” (para 75).

What is new about the 2017 guidance is that it helpfully clarifies public interest as sometimes being the private interests of one person. Section 5 returns to the issues of family disclosure.

### 3 Issues of consent in clinical practice

This section is broken down into subheadings corresponding to subsections of the 2011 Consent and Confidentiality guidance. First, however, it is worth considering terminology: the new guidance might wish to highlight that use of qualifiers before the term 'consent' can cause ambivalence (for example, is 'consent' different to 'informed consent'?) If so, what are the differences and what implications does this have for the validity of consent, as well as the differences between consent and permission?

It is also worth mentioning here that consent for eg whole genome analysis can only ever be broad because of the wide range of potential findings a test can produce. Moreover, if genomic information is collected in the context of a hybrid venture such as the 100,000 Genomes Project, consent might be broad in the sense that the research studies for which the data can be used span a broad range of topics. The 2011 guidance did not explicitly discuss broad consent in these two regards, although it did consider "a broad consent to allow communication of potentially beneficial relevant information to any relative...appropriate."

What might concern HCPs is that taking a broad consent approach falls outside the scope of their training (Bernhardt et al., 2015) and that a recent UK Supreme Court judgement, *Montgomery vs Lanarkshire HB*, might add further difficulties for practice. The judgement requires HCPs to tailor information-provision based on the expectations of a 'reasonable' patient and the circumstances of their situation, rather than according to professional standards. This might suggest that HCPs should explain certain possible results or research studies to particular patients. Broad consent for IFs and research is a thread that will run throughout the following sections, and is discussed in more detail in Sections 3.5 and 3.7.

#### 3.1 Enabling clinicians from a range of backgrounds to take consent

This aspect of the 2011 guidance remains relevant today. The Health Education England Genomics Education Programme has developed an online course called 'Preparing for the consent conversation'. Although aimed at training HCPs to seek consent for the 100,000 Genomes Project, aspects might be useful for the seeking of consent for broad genome tests more generally and the 'conversation' usefully moves the concept to an ongoing issue rather than a 'yes'/'no' toggle. The European 'Gen-Equip' project has launched and involves online modules to educate primary care professionals in genetics (see [primarycaregenetics.org](http://primarycaregenetics.org)). Moreover Genomic Medicine MSc courses are now run by several centres around the UK and include modules on ethical, legal, and social issues and genomic counselling.

#### 3.2 Ensuring that colleagues involved in the care of the patient are kept informed

Sometimes patients do not want, for example, their GPs to know about particular genetic test results because they are worried it could increase premiums on insurance. While the ABI and UK government concordat and moratorium is currently in place, a forthcoming revision is due for review November 2017. A recent BSGM survey enquired about known transgressions of this moratorium in practice, and further research is awaited. CELS' research has shown that current consent forms can act as a barrier to ensuring colleagues involved in care are kept informed: several consent forms modified from the JCMG guidance template ask patients if they agree or

disagree for results to be passed to their GP. Offering patients the choice might mask the problematic nature of 'disagrees' (Dheensa et al., 2017b).

**>>See Appendix D Box 3 for more details about these research findings.**

### 3.3 Consent for continuing and further investigations and

### 3.4 Consent for samples taken for storage only

As the 2011 guidance pointed out, consent should be viewed as a process, not simply a one-off event, and consent can be assumed valid if a further investigation remains in the scope of the original consent and the patient has been told that further investigation might take place.

CELS' research about the consent forms that different genetic services in the UK use has shown that some asked for the name of the condition or gene to be tested and as a result, HCPs are concerned about the level of detail to write on the forms—i.e. whether to name the gene they were going to test, or the suspected diagnosis. They are concerned that if they write something specific, consent might be valid only for that specific test, but if they write something broad (e.g. 'tests for my child's presenting condition'), patients worry that they could test for anything (Dheensa et al., 2017b)

**>>See Appendix D Box 3 for more details about these research findings.**

There is moreover a question of how long consent is valid for, and whether it can be assumed that a patient has not changed their mind about decisions made during the initial consent process. One way around this would be to re-contact patients before further tests are done on their stored sample—and indeed the JCGM recommend that HCPs should ask patients at the time of initial consent whether they want to be re-contacted. However, and as the JCGM also pointed out, re-contact might not always be possible, or practically difficult/ resource onerous.

As well as when new tests are available, re-contact of a former patient might be warranted if a finding (e.g., a variant of uncertain significance, or VUS) is reclassified or when new symptoms are associated with a diagnosis. CELS collaborative research about re-contact showed that patients thought re-contact in general was desirable. Patients who were re-contacted long after being discharged were pleased to have been (Carrieri et al., 2017c). However, the majority of services do not routinely ask patients about their re-contacting preferences but almost all re-contacted patients and relevant family members if *ad-hoc* significant new information arose.

Methods of re-contact varied across services as did the re-contact 'triggers'. Some genetic HCPs expressed concern towards the patients potentially missed due to their *ad-hoc* practices. HCPs mentioned the lack of databases to keep track of patients and easily retrieve information about those affected by particular conditions (see Section 5.5 for more on patient registers). Indeed, a common theme across all HCPs and patients interviews was that there was a tension between the desirability in principle of re-contacting, and its practical feasibility.

Another issue was the lack of clarity about roles, including who ought to re-contact (genetic HCPs, multi-disciplinary teams, patients) and who ought to keep up-to-date with reclassifications of variants (genetic HCPs or lab scientists) (Carrieri et al., 2017a; 2017b; 2017c; 2016; Dheensa et al., 2017c).

>>See Appendix D Box 4 for details about these research findings.

The new guidelines could include a case to illustrate the question about whether/ how HCPs should recontact patients. An example case is given below.

Meera has undiagnosed developmental delay and several signs and symptoms that are not explained by any particular diagnosis. Her parents have spent a long time trying to get a diagnosis to understand her prognosis. She has had many inconclusive genetic tests. Five years later, after discharging her from his service, a paediatrician is working with a geneticist using a large biorepository, and identifies a handful of people across the world who have similar signs and symptoms to Meera. These patients have received a molecular diagnosis of a particular rare condition. Remembering Meera and her parents, he considers this to be a trigger to re-contact the family. He can now offer her a test, which might facilitate a diagnosis and better understanding of her condition. (Dheensa et al.,2017c).

A recommendation, grounded in this research, is that HCPs should discuss re-contact with patients, including issues that might trigger it, routinely in the consent process for testing or whenever they collect and record patient. At May 2017's Public and Professional Policy Committee meeting, the European Society of Human Genetics endorsed this as a recommendation. The new Consent and Confidentiality guidance might therefore wish to emphasise their 2011 recommendation about mentioning re-contact in the consent process in revised guidance- not necessarily as a choice but as a point of information for patients that this might be necessary in the future.

The use of web-based approaches to help patients manage their preferences might help to ensure their consent stays valid over time and might enable patients to instigate a re-contact, although this would widen the so-called 'digital divide.' In fact, any solution that puts some responsibility to act on patients/parents might lead to inequity, but one that uses ICT might make the divide greater. This reflects Tudor Hart's inverse care law: those who need healthcare less will use it more, while those whose needs are greater will not engage as effectively, and in turn, their needs will be relatively neglected. (Dheensa et al., 2017c).

### 3.5 Consent for samples tested in laboratories [and for research in 'hybrid' activity<sup>5</sup>]

The 2011 guidance stated that in addition to consent for the test itself, it might be appropriate to include discussion of the following issues as part of the consent process, particularly since these issues are not routine for other types of tests or procedures:

1. Disclosure of relevant genetic information to relatives
2. Consent to research
3. Consent to the storage and future use of the sample, and the information derived from it.

As genetic testing adopts an increasingly hybrid model such as the 100,000 Genomes Project, with its involvement of academic, commercial, and industry researchers, this second bullet point in

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<sup>5</sup> Section 3.5 in the 2011 guidance was not about research in hybrid activity but the issues discussed are relevant here.

particular needs to be discussed with patients. In fact, the consent process as a whole needs discussion in hybrid ventures. The reason is that in clinical practice, consent is integrated into clinical discussions and is not always recorded separately. Sometimes patients may give their consent because they trust the HCP offering the test and trust that appropriate actions will be taken. Consent in the research setting is by contrast more formalised, detailed, and written, and a high level of understanding of risks and benefits is required for consent to be valid (although of course participants might still take part because they trust the researchers, or because their researcher is their/a HCP) (Hallowell et al., 2010; Ponder et al., 2008).

A 2012 Human Genetics Commission report suggested that introducing a national system for requesting generic consent for use of genetic and clinical data from patient records would significantly accelerate the development of new treatments and increase the attractiveness of the NHS as a research body (HGC, 2012). In her 2016 recommendations about confidentiality and information governance, Dame Fiona Caldicott<sup>6</sup> appears to endorse this view. Although she states that “people should be able to opt out of their personal confidential data being used for purposes beyond their direct care unless there is a mandatory legal requirement or an overriding public interest” (42 and 6), she also states that “data that has been *de-identified* according to the [Information Commissioner’s Office, ICO’s] anonymisation code should not be subject to the opt-out” (section 3.2.34). [emphasis added]. It is worth noting here that anonymised (stripped of all identifiers) is not the same as de-identified (stripped of some). In fact, whole-genomic sequences often cannot be truly anonymised (Gymrek et al., 2013). There is a balance to be struck between promoting the openness of data (which allows it to be linked to other data, in turn leading to new clinical insights) and the privacy of those whose data it is (Vayena and Gasser, 2016).

Regarding the use of health (NHS) data for research, the Wellcome Trust/Ipsos Mori (2016) conducted in-depth questionnaire and focus group research about patients/the public's views and found that participants thought that de-identified data was still 'information about them.' Fifty-four percent said they supported commercial access to their health data for health research (leaving 46% who did not say they supported it). Most wanted the NHS to seek their permission before allowing access to companies. Genomic sequencing was considered the most ‘risky’ type of health information because it was deemed inherently personal and because the ‘full extent of what might be possible with this type of data is as yet unknown’. Nevertheless, it was concurrently deemed the type of information with the most potential value.

A huge amount of research has explored similar questions in a biobanking context. (Like 100kGP, biobanks collect data for research but can—and do—provide some clinical results). In sum, these studies suggest that patients/the public are supportive of research and of broad consent but have concerns about privacy. A survey of 13,000 members of the public from the USA found that 66% stated they would be willing to participate in a biobank that used broad consent for the types of research for which their data would be used. Most (86%) participants would want to know what would happen if a researcher misused their health information (Sanderson et al., 2016). Similarly, in a UK study, McCormack et al. found that families affected by rare diseases are supportive of

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<sup>6</sup> Caldicott published her report in the wake of the care.data venture, which was abandoned after facing much criticism (data controllers ought to have made it possible to opt out of the processing of data earlier, to have made these options clearer, and to have been more transparent about the uses to which data would be put (Carter et al., 2015; Sterckx et al. 2016; Vezyridis and Timmons, 2016)). The UK government has now said as part of its ‘UK digital strategy’ policy that it will “introduce stronger sanctions for deliberate and negligent re-identification of anonymised data” (ICO, 2017b, para 10). It remains to be seen whether this applies to de-identified data.

research endeavors and of the use, and international sharing, of their data and biosamples, but they have concerns about data security and misuse.

Recently, Prainsack and Buyx (2016) have argued that there ought to be a 'harm mitigation fund' for anyone affected by willful re-identification of data. This fund would be

“an institution governed by people who are independent of the organisations using data and who review appeals from people who claim to have been harmed by data use. Harm mitigation funds could make positive decisions on appeal even when no laws or rules are broken...They would exist to complement, not replace, legal provisions, and would not affect any statutory rights of individuals” (p.498).

On the topic of hybrid practice, Kass and Faden et al. argued in 2013 that the maintenance of a sharp distinction between research and clinical care, and the adoption of different oversight mechanisms, causes conceptual, moral, and empirical problems. It masks similarities between the activities, and overemphasises the differences between them, which “leads to overprotection of the rights and interests of patients in some cases and to underprotection in others.” (Kass et al., 2013 pS5). They advocate a move towards a 'learning healthcare system', a system in which “science, informatics, incentives, and culture are aligned for continuous improvement and innovation, with best practices seamlessly embedded in the delivery process and new knowledge captured as an integral by-product of the delivery experience.” (Institute of Medicine, 2015). It has recently been proposed that the NHS should move more towards a learning healthcare system, where genomic data, and data from electronic health records, and digital health tools (such as wearables and apps), would be pooled and pseudonymised and identifiable data would be made available for research and analytics (Smart, 2016). According to members of the Genomics England board and associated staff, the 100,000 Genomes Project could lay the groundwork for this approach to healthcare in the UK (Dheensa and Samuel et al., forthcoming). Some crucial considerations are whether a hybrid model is feasible, desirable, and workable. The new Consent and Confidentiality guidance might consider adopting some aspects of Faden et al.'s (2013) ethical framework if genetic/genomic practice moves towards a hybrid model.

### 3.6. Covering the possibility of death in the consent process

An analysis of consent forms (Dheensa et al., 2017b) used in genetic services showed that several services ask patients on the consent form for genetic testing to name someone to receive their result in the event of their death. Doing so can introduce difficulties—for example if HCPs feel they should use the result for a different relative to the one named or the named person is unreachable. The inclusion of such an option contradicts the 2011 guidance that “a broad consent to allow communication of potentially beneficial relevant information to any relative is... more appropriate” (p9 section 3.6) than asking for names and contact details of particular relatives. Using the suggested 2011 guidance form, which does not ask for a named relative, would avoid this problem.

**>>See Appendix D Box 3 for more details of these research findings.**

### 3.7 Incidental or unexpected information [and 'additional' findings]

Since the 2011 guidance, and in the wake of increased chances of identifying incidental findings- lfs- with next-generation sequencing (NGS) technologies, there has been extensive research about patient preferences for receiving such findings. These generally show that people want results

based on their treatability and perceived utility (e.g., preventable life-threatening conditions), although many also want to know about findings that have 'personal utility' (i.e. not clinically actionable but with personal value). In these studies, participants have said that if HCPs have information about them, then they would want to be told, but that HCPs should not have a 'duty to hunt' for findings (Middleton et al., 2016; Shkedi-Rafid et al., 2014).

This hints at two 'types' of findings that have both been referred to as IFs – those that are not actively sought- they are stumbled upon in the search for something else, and those that are (sometimes called 'additional findings' or 'opportunistic testing'<sup>7</sup>). The below discussion focuses on the former.

The new Consent and Confidentiality guidance might wish to consider the international debates about which, if any, IFs—or potential IFs—HCPs should tell patients about, and how, if at all, they should broach IFs in the consent process for genomic testing. Berg et al. (2011) contend that to discuss all possible IFs, the consent process could require six hours of face-to-face discussion over the course of several sessions and talking about results would take up to five hours. They suggest that to help HCPs discuss and manage IFs, and patients understand them, potential IFs should be categorised into three bins, and each bin managed differently:

- Bin 1: clearly pathogenic results with immediate clinical utility (although the definition of this is ambiguous)--should be reported to patients
- Bin 2: variants with a known or presumed association with disease but not medically actionable--potential disclosure should be discussed by the patient and HCP at the time of consent
- Bin 3: variants of unknown or no clinical significance--should not be reported.

Others have similarly suggested categorising IFs (Ayuso et al., 2013; Bredenoord et al. 2011). This approach has been criticised because IFs cannot easily be categorised: the pathogenicity of results is not always clear at the time of reporting, and the HCP might need to disclose a finding, whichever bin Berg et al. (2011) would claim it falls into, to test family members in order to help ascertain which bin it belongs to. Moreover, the suggestion that bin 1 findings always ought to be disclosed arguably constrains patient autonomy (Crawford et al., 2016)

The new Consent and Confidentiality guidance might also wish to consider the different approaches to consent proposed in the literature about IFs. Suggestions have included open/blanket consent at one end of the spectrum (e.g., consent to all IFs), broad/tiered consent in the middle (e.g., to reflect the binning approach), and specific consent at the other end (giving patients a list of specific IFs) (e.g., Grady et al., 2015). Interestingly Crawford et al. (2016) found that HCPs thought Ifs should (and could only) be discussed in a broad and general way in the consent process, both due to time constraints and because too much detailed information would be overwhelming. Patients, including those who had received an IF, did not remember whether and how HCPs mentioned IFs in the consent process for genetic testing but assumed—or trusted—that HCPs probably had done so.

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7 The ACMG (Green et al., 2013) recommended that a set of additional findings should be sought whenever a genome is sequenced in clinic, while the ESHG (Hehir-Kwa et al., 2015) recommended that analysis specifically target a particular health problem. The 100,000 Genomes Project offers participants a search for a list of 'additional findings'. There is a question of whether the new Consent and Confidentiality guidance should recommend that these findings be offered routinely to patients/families undergoing genome tests.

Another question is whether new guidance should recommend a standard term for findings that are unrelated to the clinical diagnosis (Christenhusz et al., 2014). Such a finding can be significant for patients and the term 'incidental' may not do justice to this (Tan et al. 2016; Shkedi Rafid et al., 2014). The term 'secondary' has been used, but is problematic because it does not capture situations where the so-called secondary variant is the only (clinically significant) variant found. The term might furthermore inaccurately suggest a temporal relationship where one finding is found first and the other second. Tan et al. (2016) surveyed patients and found that the term additional findings had was most preferable for survey participants, followed by 'secondary' findings, 'incidental' findings, and 'ancillary' findings. In focus groups, most participants preferred the term 'additional' findings because it seemed non-threatening while not downplaying potential seriousness as per the term incidental, however, the 100kGP has adopted this term for findings that are actively sought as part of an opportunistic screen.

### **3.8 Use of samples of lab clinical control, quality assurance, audit, education and training**

No changes are needed here.

### **3.9 Counselling as part of the consent procedure for a genetic test**

No changes are needed here, except that the degree of counselling, and the qualifications of those undertaking will like need to be more varied in mainstream genomic testing

### **3.10 How should consent be recorded?**

The 2011 guidance refers to GMC consent guidance which states that patients can indicate their consent either orally or (if it is for an intervention or test that is complex) in writing. The JCGM argued that documenting consent for genetic tests in writing can be useful to record key aspects of the consent discussion and decisions made (about sharing information etc.) in case other HCPs see the patient or their relative in future. The GMC guidance remains unchanged. However, the new revision will be out for public consultation in autumn 2017.

The 2011 guidance also suggested that HCPs use a form such as that in Appendix C2 of the 2011 guidance to document these discussions. The guidance notably calls the form a 'record of discussion' in a bid to move away from the idea that the act of completing the form itself (as opposed to the discussion about the procedure) is 'consent' in and of itself.

The form features two statements- or presumptions made- (that notably have no response options--e.g. agree/disagree options) that correspond to aspects of clinical genetic practice and are necessary for the effective running of such services. These statements are

1. I acknowledge that my results may sometimes be used to inform the appropriate healthcare of members of my family.
2. Occasionally leftover samples may be useful in checking laboratory techniques and my sample might be used as a 'quality control' for other testing, for example, that of family members.

A third statement does in fact require a response: whether the patient wants to be contacted before further relevant tests are done on their stored sample or whether tests should go ahead

without re-contact. Here, an issue that the new Consent and Confidentiality guidance might need to consider in the revision is whether re-contact is practically possible—if it is not, it should not be offered. As the 2011 guidance pointed out, “a genetic service may not be able to give an assurance that [re-contact] will happen automatically”. More generally, the new guidance might wish to consider whether this option still reflects practice appropriately: it is taken from a time when labs might have wanted to run increasingly sensitive tests on groupings of patients, which is now less likely.

The form does not mention IFs or VUS explicitly- because at that time these were extremely rare findings—but these have been covered in emerging practice in the box for free-text where HCPs can record discussion of these findings.

Use of the form was a suggestion and new guidance might consider recommending wider unadulterated adoption. As part of an empirical research study, CELS audited whether genetic services were using the form and what amendments- if any- had been made to it. This showed that UK genetic services were largely using forms that asked patients to agree/disagree to statements about sharing information to benefit family members etc., and that included several- often unworkable- additional statements. The lack of standardisation of forms made it difficult to share information between genetic services; that patients were sometimes frustrated by the forms; and that, as mentioned earlier, HCPs worried about the level of specific detail to write on the forms (e.g., about which genes to test), and whether they would have to stick to these specifics in practice (Dheensa et al., 2017b). The revised guidance might thus wish to consider greater harmonization of forms used. This might facilitate the sharing of information between services, which aligns with the recommendation in Section 5.3 of the 2011 guidance that “It is appropriate for good clinical care that technical information and laboratory reports should be shared between the different laboratories undertaking the testing, and be available to clinical staff in genetics units.”

Some other changes that could improve the suggested consent form are:

- making the potential differences between personal clinical and familial genetic information clearer
- making clear on the form itself that sometimes professional judgement will need to be utilized when specific details on consent forms are unclear or contradictory, for example about benefitting relatives.
- checking the reading age of forms, which HCPs in our research worried was too high for many patients.

**>>See Appendix D Box 3 for more details of these research findings.**

In 2013 Ayuso et al. made recommendations (see box below) for what consent forms should feature. These recommendations were based on a systematic review of papers published by expert authors on consent and international guidance (including the 2011 guidance) by scientific societies and ethical boards:

1. The scope of the test
2. Brief description of the test process
3. Benefits to expect
4. Possible disadvantages, risks, or complications
5. Voluntary nature of the test

6. Possibility of refusal at any time without consequences
7. Description of alternative diagnostic methods, if any
8. Description of the measures taken to ensure confidentiality and privacy of the results at present and in the future
9. Destination of the biological samples when the study ends (storage, encryption, anonymisation, or destruction) or future use of samples
10. Management of IFs that may appear in the study, and the right not to know

Although the new Consent and Confidentiality guidance might wish to consider integrating Ayuso et al.'s recommendations, doing so could lead to a longer consent form. However, the recommendations could be useful to inform the discussion around consent or patient information leaflets.

The new Consent and Confidentiality guidance might need to consider the suggested consent form in light of the GDPR. On this topic, the ICO (2017a) states that:

“consent [for processing data] under the GDPR requires some form of clear affirmative action. *Silence, pre-ticked boxes or inactivity does not constitute consent.* Consent must be verifiable. This means that some form of record must be kept of how and when consent was given...Where you already rely on consent that was sought under the DPA or the EC Data Protection Directive (95/46/EC), you will not be required to obtain fresh consent from individuals if the standard of that consent meets the new requirements under the GDPR.” (Rec 25) [emphasis added].

### 3.11 Consent to clinical photography and video recording

No change is needed here except that HCPs might wish to amend their consent forms if they will be using apps such as Face2Gene, which uses 'Facial Dysmorphology Novel Analysis' (algorithmic facial recognition technology) to diagnose genetic conditions. The app allows HCPs to share photos of patients on online forums to seek help with diagnoses, and to use the photos for training/education. Several genetics services in the UK are using Face2Gene. See <https://www.face2gene.com/>

## 4 Consent by others

The 2011 Consent and Confidentiality guidance (and the 2010 BSHG childhood testing guidance) suggested that HCPs should not test children for adult-onset conditions. Research since then suggests that parents and HCPs worry about record keeping—that is, if a child is not tested now, then information about their risk would be lost. Fenwick et al. (2016) have encouraged HCPs to frame parental requests for such testing from 'whether to test' to 'when to test'. The revised JCGM guidance section on children might include a section on what happens if an adult-onset risk is discovered in a child from an already sequenced genome (e.g., from a panel test, whole-exome, or whole-genome sequence) as opposed to in response to a parental/guardian request, which is covered by BSHG guidance. The new guidelines could include a case to this 'newer' example.

## 5 The use of familial information in clinical genetic practice

### 5.1 The family history as an aid to genetic diagnosis

No change is needed here except to consider the way the GDPR might affect family history documentation.

## **5.2 Use of medical records to confirm diagnosis**

This section might need to be revised if electronic medical records, such as My Medical Record or Patients Know Best, are rolled out (as is the NHS's aim for 2020) in clinical genetics.

## **5.3 The use of existing genetic information to facilitate accurate genetic testing**

This section might need to be revisited in light of the GDPR, if only to update the paragraph that states "If information is recorded about one family member in another person's records, the Data Protection Act gives specific rights for that relative to obtain copies of the information that is held about them". Although this was an assumption in the 1<sup>st</sup> and 2<sup>nd</sup> edition of the guidance, an explicit statement might be helpful. It is hoped that the GDPR will not be interpreted as requiring specific consent from each relative in a family pedigree for documentation, but rather than a reactive duty to let those enquiring know, is appropriate. Documentation of detailed family history is an important part of phenotypic information gathering that facilitates genomic data interpretation.

## **5.4 Retention and storage of pathology and genetic samples in laboratories**

This section cites an older version of the Royal College of Pathologists' (RCPATH) guidance on the retention and storage of pathological records and archives. An updated version was published in 2015 (RCPATH, 2015). The advice about optimal periods of storage and retention remains unchanged (and in fact, the RCPATH guidance recommends that consent should be sought for retaining diagnostic specimens according to the 2011 Consent and Confidentiality guidance). However, it contains specific advice about data storage of electronic outputs from genome-wide sequencing technologies:

"Immediate needs [for data storage] can be met by transcription of specific results into conventional diagnostic reports. However, much of the efficiency of these technologies will be compromised if the raw data files are discarded, requiring repeat sampling and re-sequencing when analysis of new biomarkers is required. Personalised data storage strategies will be needed as NGS methods become routine unless massive data storage capacity can be assured for collective holdings" (para 162).

## **5.5. National or international disease registers**

The 2011 guidance supported the sending of relevant information to (inter)national disease registers for service evaluation and gathering evidence for future healthcare. However, many genetic disease registers across the country have now ceased to exist due to lack of funds and a related concern that keeping a register would lead to greater legal obligation to ensure regular updates of those on a register. The new Consent and Confidentiality guidance might wish to comment on the different statuses of registers and clinical databases

Related to this, the new Consent and Confidentiality guidance might wish to comment on the responsibilities engendered- and by whom- through the biorepository generated by the 100,000 Genomes Project once it enters the NHS. About this, Genomics England state that:

“The priorities of the government and Genomics England are to ensure that the identity of all participants is protected, the data storage and access to the dataset is secure, and researchers and clinicians have confidence in the dataset’s accuracy. To ensure public confidence in matters of confidentiality and access, the Chief Medical Officer for England, who also sits on the board of Genomics England, monitors this work. Genomics England’s dataset is a national asset and it is this government’s clear policy that the dataset, as protected by its owner the Secretary of State for Health, will not be sold or distributed to third parties and that any income generated will be reinvested in health care and health research.”

It is not clear here what 'protected' means regarding the identities of participants (e.g., whether it means the data will be de-identified, what this means for privacy/confidentiality) and how participants' identities will be protected.

## 5.6 Disclosure of information to others

While Section 1 discussed the way HCPs facilitate family communication, this section focuses more specifically on situations where a patient has been seen many years previously, or by a different HCP, and it is unclear if or what consent they gave regarding the use of their test result to benefit the health of relatives. The 2011 guidance recommended that when explicit consent is missing or unclear, using a patient's mutation details to direct testing of a relative can be justifiable because it can mean the HCP can offer a more accurate test to the relative, which might alter their treatment or surveillance, for example. As per GMC guidance it pointed out that relevant information can sometimes be disclosed to relatives in an anonymised way, without breaching the original patient’s confidence.

Although the 2011 guidance did not go into detail about the conceptual framework behind this recommendation, it is worth pointing out that these recommendations are based on a view of confidentiality in which genetic information is conceptualised as confidential at the familial rather than individual level (Lucassen and Parker, 2010; Parker and Lucassen, 2004). This approach proposes that genetic information should be available to all family members unless there are good reasons not to, and so, HCPs should consider taking disclosure of relevant familial information to relatives as their default starting position. The question, ‘can we share the patient’s information?’ is reconsidered as, ‘can we share the familial genetic information with those to whom it might be relevant?’ The interest of all those who might have inherited the same finding- albeit discovered in one individual- is the primary consideration. The 2011 guidance suggests that if a HCP realises that intrafamilial communication has not happened, and there is an overall benefit to a family member to be had, lack of explicit consent to sharing should not prohibit use of relevant information to test a relative (Dheensa et al., 2017a).

Since the 2011 guidance, comparable Australian policy has been published about genomic research: rather than treating all results as confidential to one person by default, researchers must make clear that the participant cannot prevent the sharing of information that could benefit family members at risk of a serious illness for which treatment is available or pending (Branum et al., 2015). Australian regulations for clinical practice, provided by statute, give HCPs permission (but do not enforce a duty) to disclose information to relatives.

CELS' research (Dheensa et al., 2017a) has explored views and practices around the approach to confidentiality suggested in the 2011 guidance. HCPs perceived several problems with a familial

approach to confidentiality but these mainly arose from concern that clinical confidences would thereby be breached. HCPs had not considered separating familial genetic information (e.g. a BRCA1 mutation) from personal clinical information (a patient's breast cancer diagnosis) but when they did, some thought that if they shared genetic results these might point to a clinical diagnosis in a particular person, who could then complain their confidence had been breached.

HCPs worried that if they had not discussed sharing information with family members at the outset of their interactions with patients, sharing information later down the line would jeopardise trust in them and the NHS. Indeed the 2011 guidance recommended such discussions were held routinely with patients before testing, but our research showed that this had not entered widespread practice. HCPs also thought that taking on a responsibility for some relatives could change the standard of care and lead to them having to take responsibility for more relatives and they lacked the resources to do so. Many claimed they would alter their practice if there was a nationwide consensus, even though arguably the 2011 guidance was exactly this.

The new Consent and Confidentiality guidance might wish to consider encouraging HCPs to discuss familial implications at the earliest opportunity, and making clear that HCPs taking a familial approach would have a bounded or reactive responsibility, i.e. not to tell all possible at-risk relatives about risk, but limit this to specific relatives, known about (see ABC case). Having a default position of sharing genetic information, rather than keeping confidence, would then change practice only in narrow circumstances.

Interestingly, interviews with patients (Dheensa et al., 2016) showed that they saw genetic information as essentially familial, with several saying something akin to “this isn't my information, I don't own the gene”. They had some concerns about the familial approach: they worried that if HCPs could share genetic information without consent, they could share other (more stigmatising) medical information without consent as well—and they preferred to be asked permission, or at least told, how genetic information would be used, at the outset of the clinical encounter.

The new guidance might also wish to consider emphasising the potential of a familial approach in revised guidance and devising additional, discussion-based, activities to explore and integrate the revised approach into practice (e.g., using UK Genethics Forum).

**>>See Appendix D Box 5 for more details of these research findings**

## Appendix A: Key legal issues

The JCGM may wish to explore whether Appendix A1 (details of the Human Tissue Act 2004) needs to be updated in light of the 2017 codes of practice. In Appendix A2 details of the Data Protection Act will need to be updated in light of the GDPR. The new guidance should consider the way ABC vs St George's and Smith and Anor vs Leicester might affect practice. These cases are detailed below.

## Relevant case law examples:

### A.1 ABC vs St George's Healthcare NHS Trust

A man diagnosed with Huntington's disease (HD) in November 2009, and detained in a psychiatric unit after a conviction of manslaughter two years earlier, refused consent for the professionals involved in his care to tell his pregnant daughter about her risk (50% chance of having inherited it). His reasoning was that if she were told, she would become upset, kill herself, or terminate the pregnancy.

The multidisciplinary team looking after him were in regular contact with the daughter and queried whether they should tell her about the familial risk. Ultimately, they decided that they could not do so in the presence of his explicit refusal. He was deemed to have the capacity to consent, and withhold consent.

In April 2010, the daughter gave birth, and in August 2010, one of the professionals involved in her father's care accidentally disclosed the diagnosis and its familial implications. She decided to have a test and in January 2013 which showed that she had inherited the Huntington's predisposition from her father.

The daughter sued in negligence, claiming that the health professionals involved should have told her sooner so that she could choose whether to continue her pregnancy based on her inheritance (she argued that already in her late 30s, she would not want to bring up a child as a single parent if she were soon to become ill with Huntington's disease). Her lawyers relied on the 1<sup>st</sup> edition of JCMG Consent and Confidentiality guidance, particularly the aspect that stated sharing the genetic information derived from one patient's test can be justifiable to benefit others.

Any professional negligence claim needs to meet the so-called Caparo test, which states that the harm caused by the negligent actions must be reasonably foreseeable; the relationship between the parties must have reasonable proximity; and it must be fair, reasonable, and just to impose liability. The lawyers of St George's healthcare trust outlined nine policy reasons a duty of care to the daughter would not be 'fair, just, or reasonable'. One such reason was that doctors would be placed under conflicting obligations—liable to be sued by their patients for disclosing 'confidential' information, or sued by third parties for failing to do so. Mr. Justice Nicol of the High Court struck out her claim, because he considered that establishing such a duty would be too big an incremental change in the law.

However, in May 2017, three appeal court judges overturned the High Court's decision, meaning the case may now go to trial. Lord Justice Irwin, who gave the leading judgement, cited two US cases (*Pate vs Threlkel*, *Safer vs Pack*) in which the courts held that relatives had the right to be told about a genetic risk in the family. He furthermore cited the 2015 case of *Montgomery vs Lanarkshire*, as evidence of the increasing trend in law to emphasise and recognise the autonomy of potential patients.

Lord Irwin said:

"On the face of it, it seems to me this policy reason ["(viii) Doctors receive a very great deal of confidential information. It would be burdensome to place on them a duty to consider whether any of it needs to be disclosed to third parties. The time and resources committed to this will be a distraction from treating patients."] lacks any bite when applied to geneticists. As will already be clear from the [Consent and Confidentiality]

professional guidance to which I have referred, and indeed from the inherent nature of genetic medicine, geneticists frequently acquire definite, reliable and critical facts of clinical significance about their patients' relatives." (para 40).

The courts recognised that professional guidance already acknowledge the difficulties that HCPs might experience balancing their duties, and that there is already a professional obligation on HCPs to consider their responsibility to relatives and balance this with their duty to patients. A question for the court is whether this professional obligation to consider relatives' interests ought to become a legal duty, and when this legal duty ought to apply (e.g. should it apply only in cases where there is risk of serious harm? And, in light of the Montgomery ruling, who should decide what constitutes serious harm?) (Uzoigwe, 2017).

## **A.2 Smith & Anor v University of Leicester NHS Trust**

Connor and Callum Smith were born with adrenoleukodystrophy (ALD). Callum was diagnosed at age 6 in 2006 and died aged 13 after his condition deteriorated. Connor was not showing any signs or symptoms at this stage but was tested and shown also to have inherited the condition. He received treatment, including a bone marrow transplant, although he does suffer significant intellectual and neuropsychological problems.

The boys' mother sought to sue the University Hospitals of Leicester NHS Trust, because it transpired that in 2003, a consultant there had seen the boys' second cousin, Neil Caven. Neil has a mild version of Adrenomyeloneuropathy (ALD). The consultant requested a blood test for very long chain fatty acids which might have diagnosed the condition, but Mr Caven did not attend for blood tests. Had consultant pursued testing more actively, a result would have confirmed ALD, which would have led to testing of the wider family, it was argued. Thus, Callum and Connor could have been tested and diagnosed sooner. As it happened, Neil was tested in 2006, only after Callum and Connor themselves were diagnosed.

Like ABC vs St George's, this claim was struck out of court. This time, when applying the Caparo test, it was held that while the harm was foreseeable, there was insufficient proximity, and again, that it would not be 'fair, just and reasonable' to impose a duty of care. The defendant had just been treating the patient and not his wider family and that where the scope of the alleged duty had effectively been to inform a third party of a diagnosis reached in respect of a patient, there was insufficient proximity between the parties for such a duty to be imposed.

The family's lawyers argued that the hospital had arguably assumed a responsibility to Caven's relatives by virtue of providing genetic services and investigating pedigrees—"the class of those who might be injured by a failure to diagnose such a genetic condition is defined and ascertainable and the scope of the duty alleged is to take reasonable steps to provide the patient with an accurate diagnosis that would enable relatives to seek genetic testing" (para 28). They moreover argued that the ABC case was distinct because the patient had refused consent for the information to be shared with relatives—while in the ALD case, there had been no such refusal. However, the judge was not convinced and pointed out that regardless of the patients' refusal to give consent in the ABC case, the decision to strike the case out of court was also grounded in the principle of duty of care, rather than confidentiality.

## Appendix B: Flowcharts

## Appendix C: Suggested consent forms

These sections should be changed in light of preceding changes.

## Appendix D: CELS research

### Box 2: Sharing information in the family with 'to whom it may concern' letters: findings from the consent and confidentiality study

This research has shown that the letter's 'journey' can meet obstacles. HCPs writing these letters were unsure about how directive to be and about how to strike the balance between informing the recipient, encouraging them to seek a referral, and not causing them alarm. Patients said that being given the letters created pressure upon them to talk to their relatives, because sending them the letter 'out of the blue', without accompanying discussion, was unacceptable. Such discussion was difficult because sometimes patients were estranged from their relatives or had a poor relationship with them.

There were examples where the letter became an uncontrolled form of communication, e.g., where family members disseminated a patient's letter without their permission or where a patient disseminated the letter to many more relatives than just those at risk.

There was also a lack of clarity among HCPs and patients about whether their duty or responsibility was discharged by providing a patient with such a letter. Interestingly, HCPs were reluctant to write to relatives directly even in situations where their patient had specifically asked them to, as they thought the information would be better received if coming from a family member or were worried about contacting people who had not been referred to them

HCPs reflected that patients need a pathway to seek help with communication especially at times where follow-up appointments were less likely. Even where help was offered patients did not always accept it, or know how to do so. HCPs realised they could make ways of accessing help clearer to patients.

**>>Taken from Dheensa et al., forthcoming.**

### Box 3 : Consent—how it should be recorded and what should be discussed: taken from the consent and confidentiality study

#### Content of consent forms

This research showed that all UK genetic services (and the one cancer service) had a consent form they could use for genetic testing, but only one used the JCGM template without changes. None called the form a 'record of discussion'.

All 24 mentioned a statement about benefit to relatives, but 16 asked patients to tick agree/disagree, or to initial or tick against the statement (the absence of which could be

intended or interpreted as disagreement). Regarding the statement about 'quality control', four forms gave patients an initial/tick or agree/disagree option.

23/24 of forms used in practice featured up to 10 statements additional to those on the JCGM form, the most common being to ask the patient to name someone to receive their result if they were unable to do so (e.g., if they died). Some of these statements were problematic. For example, asking for a named person might create difficulties if HCPs feel they should use the result for a different relative to the one named. It might furthermore be unworkable if the named person is unreachable. The inclusion of such an option contradicts the 2011 Consent and Confidentiality guidance that “a broad consent to allow communication of potentially beneficial relevant information to any relative is... more appropriate” (p9) than asking for names and contact details of particular relatives.

Some forms gave patients the choice about sharing their result with their GP. This could impede their care, since the GP is most likely to be involved in ongoing clinical management following the result. This is relevant to Section 3.2 above and the 2011 guidance recommendation that “reasons why the usual process [i.e. notifying GP] is not followed should be carefully documented in the clinical notes” (p7).

Other forms asked patients to confirm that they had received information and had the option to ask questions. These items are directly drawn from research consent forms and relate to standard parts of clinical genetic practice. It is thus questionable whether there is any benefit in including them on consent forms. Arguably, the items are superfluous.

These findings reflect that UK genetic services were mostly using a 'tick-box' method on consent forms. Moreover, it reflects that most services' approach to confidentiality was individual, in that their forms gave individual patients control over genetic information, even though it might have familial implications (more detail on this in Section 5 above).

### **Qualitative research about the use of consent forms**

HCPs thought consent forms were useful for prompting them to discuss key topics, providing evidence of consent, which could facilitate information-sharing, and providing patients with a sense of control.

Notably, however, consent forms constrained ethical practice in several ways. While forms could help to make clear whether HCPs could use a test result from one patient to benefit family members later seen in their service, the forms were less helpful when the family member was seen elsewhere. The reason for this was that different services had different expectations and standards about the specific wording against which the patient should have signed, and thus whether they considered a completed form valid.

HCPs found that the forms themselves, especially ones made long and complex by the number of items to read and tick, frustrated patients and were unacceptably burdensome. Some worried about the high reading age on patient-facing literature generally. This weighed against the perceived benefit that forms might provide patients with a sense of control.

Regarding IFs and VUS, participants wondered whether and how forms should integrate discussion, and decisions made, about these findings and whether they should incorporate

categories of IFs. Generally, HCPs thought forms that attempted to integrate IFs/VUS in detail would become too long and too complicated, such that completing forms would cut into the more important and tailored consent discussion, thereby constraining ethical practice.

Another related question about the consent process was how specific to be on forms, for example regarding what genes to test. Participants explained that being too broad, e.g. around what they would be testing for, caused patients to worry that HCPs might test for something else or check other parts of their medical record. However, writing something specific made HCPs question whether they had to seek fresh consent, for example, to test a different but potentially relevant gene. HCPs furthermore worried that patients might mistakenly perceive a named gene as a diagnosis. Questions about whether to be specific or broad related to HCPs' perceptions about which findings they could feed back to patients—some worried they could not tell patients about findings unless they were written on the consent forms. This issue relates to the recommendation from Section 3.3 above and the 2011 guidance—that “good liaison between laboratory staff and clinical staff is required so that clinicians can consider whether further testing falls within the scope of original consent, or whether further consent ought to be obtained” and that “one way to determine preferences is to ask at the time of initial consent” (p8). In this study, HCPs and lab scientists' concerns about whether they morally ought to or legally needed to stick rigidly to what is written on the consent form made it difficult for HCPs and lab staff to determine whether further testing “falls within the scope of original consent”.

As for all of the research presented here, the findings and subsequent recommendations need to be considered in light of the increasing use of genetic tests in mainstream specialties.

>>Taken from Dheensa et al., 2017b

#### Box 4: Practices, expectations, and preferences about re-contact: taken from the re-contact study

##### Survey of current practices

Twenty genetic services responded to a survey about current re-contacting practices. The majority of services (12/20) indicated they do not routinely ask patients about their re-contacting preferences during the consent process. Of these, 6/12 recorded patients' re-contacting preferences, 6/12 did so only occasionally, and 8/12 did not record them at all. Despite not always asking patients about preferences, 19/20 services reported re-contacting patients and relevant family members because of significant new information. However, only 3/20 reported that they *routinely* re-contact former patients and 16/20 services reported that they re-contact on an occasional basis.

Reasons for re-contact were the availability of new genetic tests/results; family follow up (a new member of the family being referred prompting a review of their file); reclassification of a VUS; new clinical guidance; or reproductive relevance (a patient reaching reproductive age).

Regarding practical methods of re-contacting, only 7/20 services responded that they have developed these (3 of which were the centres that reportedly re-contact routinely). Methods

varied: re-contacting via a patient's GP or paediatrician; informing the patient directly via a letter or telephone call; using NHS clinical databases.

Regarding 'triggers' for re-contact, 14/20 said a 'clinically actionable' finding would be a trigger. Another commonly reported trigger was the publication of new or revised guidance or a new laboratory report. One service added that they would re-contact only if new information related to a small group or an individual because only then would the workload be manageable.

Most services (14/20) said they would re-contact a patient (or their GP) about information that had potential clinical implications for them/their family, even if the patient had said they would not want to be re-contacted. Interestingly patients too thought that HCPs sometimes ought to override patients' initial wishes to not be re-contacted.

### Qualitative research about re-contacting

HCPs had developed diverse (sometimes inefficient and unsustainable) personal methods for re-contact, often reliant on their own memory. For some the inefficiency was perceived to be exacerbated by expanding services (as more patients are referred to genetic services, remembering each one becomes less possible). Some genetic HCPs expressed concern towards the patients potentially missed due to their *ad hoc* practices. Databases, or the lack thereof, to keep track of patients and easily retrieve information about those affected by particular conditions, were mentioned frequently. Further issues raised were:

- whose responsibility it would be to initiate any re-contact (which was complicated by the increasing use of genetic testing by mainstream medical specialties).
- whether and what responsibility patients should have
- who ought to keep up-to-date with the reclassification of variants—genetic HCPs or lab scientists—with some lab scientists suggesting there should be a two-way responsibility (the lab responding to genetic HCPs' requests, or a lab scientist who happens to be a specialist in a certain area alerting HCPs to new developments in that area).
- whether re-contacting could be collaborative between different HCPs and patients. This could get around some of the perceived resource constraints in the NHS. Some patients pointed out that it would be useful to involve GPs since they would likely have the most up-to-date patient demographic and clinical information. Some patients proposed an ICT-approach involving an electronic health record that automatically alerts them to potentially relevant updates.

Overall HCPs emphasised the importance of reaching a consensus. Based on this research we have suggested that that re-contacting, including issues that might trigger it, should be discussed routinely in the consent process for testing, or whenever patient data are collected and recorded. This discussion would help to clarify expectations. Patients should be informed that the clinical genetics team holds their records and provides the best information available at the time, but that the patient is welcomed to re-contact the team (1) when a potentially relevant family event occurs, such as a death or birth, or a child reaching reproductive age, and (2) at regular intervals (if agreed by both parties depending on the specific condition). Future contact could trigger HCPs to review the patient's file to check whether any new information is relevant to them. If the patient does not agree – for example, if they or the HCP feel they do not have the capacity, willingness, and/or time to contact the team and/or to be kept up-to-date regarding

their condition, the discussion would still help to clarify patient preferences, and the balance of responsibility between patients and HCPs. The patient should always be able to notify a change of preference to the clinician (this could be in itself a trigger for HCPs to review files). The ESHG PPPC has endorsed this recommendation.

>>Taken from Carrieri et al 2017a; 2017b; 2017c; and 2016; and Dheensa et al., 2017c.

### Box 5: Approaches to confidentiality and facilitating communication: taken from the consent and confidentiality study

#### Qualitative research about confidentiality: HCPs' views

Regarding the individual vs familial approach ('joint account' approach) to confidentiality, the project showed that HCPs questioned their responsibility to their patient's relative(s) when consent was ambiguous or when the patient had refused consent to share information.

Although participants claimed they rarely encountered situations where patients explicitly refused to tell relatives about a relevant genetic risk, (and they assumed that if a relative had not been told about a risk straightaway, they eventually would be told) they mentioned around 30 different cases where disclosure had not happened. Furthermore, they recognised that patients who said they would tell family members but did not actually do so might limit their awareness of the actual number of incidences of non-disclosure.

When a patient refused consent or was reluctant to share information with a relative who was their patient too, HCPs felt more compelled to tell the relative about their risk even if the original patient's explicit consent was unclear or absent. If the relative was another service's patient or had not been referred, the HCPs preferred to encourage their patient to disclose (although they might also discuss such cases at eg the UK Genetics Forum).

They perceived several barriers to taking a familial approach to confidentiality, i.e., using the familial mutation to test the relative. Most HCPs had not considered the possibility (in many cases) of separating familial genetic information (e.g. a BRCA1 mutation) from personal clinical information (a patient's breast cancer diagnosis). Notably, a few participants thought that if the relative inferred the patient's identity it was still not a breach of confidentiality on their part, as long as they themselves had not included any identifying information. Other reasons for not putting the familial approach into practice fell into two categories:

#### (1) Barriers about relationships

- Sharing any information could 'interfere' with family relationships and make relationships that were potentially already dysfunctional even worse.
- Patients and relatives pre-existing relationship made them better-placed at sharing the information.
- It could be shocking and invasive of the relative's privacy even if a patient told them about a risk, but less shocking and less invasive compared with if a HCP told them.
- Sharing information later down the line would jeopardise trust in them and in the NHS if they had not told patients they could share information at the outset of the clinical encounter. (Indeed the 2011 Consent and Confidentiality guidance stated, "If such

information is relayed to other family members without appropriate consent, the trust between the patient and health professional may be undermined.” (p4). However HCPs did not routinely discuss sharing information to benefit family members at the first clinical encounter.)

- Assumed that relatives would eventually find out about their risk.
- If patients did not tell family members about a risk straightaway, they could be convinced to do so eventually in follow-up appointments. (At the same time, they commented that follow-up appointments were less common in routine practice and so were opportunities to convince patients to talk to relatives.)

## (2) Barriers about structural factors

- Taking on a narrow responsibility for some relatives could change the standard of care and lead to them having to take responsibility for more relatives. They felt they lacked the resources to do this.
- Legal consequences: they could be liable for not sharing information with all at-risk family members as well as for breaching individual confidentiality. Thus, the ‘litigation culture’ in the health service made them reluctant to share information, even when such sharing might be appropriate.
- Taking responsibility for relatives of patients would be a substantial shift in genetics practice, keeping individual patient confidence was the norm, and communicating with relatives, or their GPs for example, was not yet “culturally acceptable.” They said they wanted nationwide consensus, even though the 2011 guidance were meant as a consensus statement.

## Views on 2011 guidance

Many participants said they were aware of JCGM guidance and the familial approach to confidentiality advocated within, but thought the guidance had not shifted routine practice. Some argued that they would be more comfortable taking the sharing of genetic information as a default position if doing so was widespread practice, as then (they thought) there would be safety in numbers, better protection from litigation, and equality in care for patients and families nationwide.

At the same time, participants perceived guidance to have limited utility, because they typically do not cover every situation and ultimately require professional judgement in the very situations where they felt ill equipped to do so.

>>Taken from Dheensa et al. 2017a

## Qualitative research about confidentiality: patients' views

In line with the familial approach to confidentiality, interviews with patients showed

- they saw genetic information as essentially familial, with several saying something akin to “this isn’t my information, I don’t own the gene”.
- they perceived at-risk relatives as having a right to know about their potential risk and that this view would be unchanged even if they were estranged from these relatives.
- they thought information was often best coming from a family member but that patients needed support talking to relatives.

- they found it generally acceptable for HCPs to share information without the tested person's explicit consent (Importantly, they considered any breach of confidence in so doing as trivial compared with the benefit of knowing about risk in a timely fashion)
- they thought relatives should be tested and offered treatment/intervention as soon as their risk was apparent because risk was a 'ticking time bomb'.
- they thought patients' consent should not be sought before each time genetic information was shared because delays to sharing could cause risk to relatives.
- they identified non-medical harms and benefits that they thought it important HCPs consider, such as the damage that delayed diagnosis could have on family relationships and psychological well-being. HCPs did not consider such issues in any depth in focus groups.
- none thought HCPs should respect patients' refusals on the basis that the information was private and personal to them.

Patients thought there could be good arguments to also disclose conditions not amenable to medical intervention, such as early onset Alzheimer disease, although they perceived such situations to be more complicated because of the distress disclosure could cause.

Crucially, patients did have some concerns about the familial approach:

- if HCPs could share genetic information without consent, they could share other (more stigmatising) medical information without consent as well.
- they preferred to be asked permission, or at least told, how genetic information would be used, at the outset of the clinical encounter.
- patients thought genetic information was personal as well as familial and were unsure how, in practice, HCPs could use information from one patient to test another.

>>Taken from Dheensa et al., 2016

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2017

Dr Sandi Dheensa

Clinical Ethics and Law

University of Southampton

Disclaimer: this document is intended to act as a guide for revision of the consent and confidentiality guidance in 2017. While every effort has been made to include relevant research, it is not intended to be a systematic review of all relevant papers.