

## Developing a consensus on data sharing to support NHS clinical genetics and genomics services

### Overview

The National Data Guardian for Health and Care (NDG) advises and challenges the health and care system to ensure that citizens' confidential information is safeguarded securely and used properly. Dame Fiona Caldicott was appointed as the first National Data Guardian for health and care by the Secretary of State for Health, Jeremy Hunt, in November 2014<sup>1</sup>.

The NDG's role is to help make sure the public can trust their confidential information is securely safeguarded and make sure that it is used to support citizens' care and to achieve better outcomes from health and care services. Dame Fiona believes that an enhanced public dialogue about the way that such data is used, by whom and for what purposes is an important element of building public trust.

The National Data Guardian's Review of Data Security, Consent and Opt-Outs published in July 2016<sup>2</sup> did not cover the sharing of genetic and genomic data in any detail. However it did refer to some of the issues that have been central to this piece of work:

*“Genomics offers huge potential for personalised medicine to improve the effectiveness of healthcare while reducing or eliminating side-effects. However, the lines between direct care and secondary use of data<sup>3</sup> are blurred: interpreting the clinical significance of an individual's genomic variants is reliant on the data of [a] larger cohort of patients with similar disorders. The timescales of the Review have not enabled a detailed consideration of this area. Useful work has taken place on these issues, for example a recent joint report from the Public Health Genomics (PHG) Foundation and the Association for Clinical Genetic Science (ACGS) makes a number of commendable recommendations.”*

After the publication of the 2016 Review, the National Data Guardian discussed with the PHG Foundation and ACGS undertaking further work to explore developing a consensus on data sharing to support NHS clinical genetics and genomics services.

The background to the discussions was a workshop held in June 2015 by ACGS and PHG Foundation to examine the legal, practical, regulatory and technical factors that impede genomic data sharing, to which Dame Fiona Caldicott, the National Data Guardian contributed. The workshop and resulting report<sup>4</sup> identified inconsistent practices in the sharing of data to support patient care.

In order to explore whether the provision of advice from the NDG, or a further process involving the NDG, might help to address concerns about the legitimacy of genomic data sharing, which contribute to this variance in practice, an evidence session was held in October 2016 by the NDG

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<sup>1</sup> <https://www.gov.uk/government/news/national-data-guardian-appointed-to-safeguard-patients-healthcare-information>

<sup>2</sup> <https://www.gov.uk/government/publications/review-of-data-security-consent-and-opt-outs>

<sup>3</sup> See Annex B for explanations of terms 'direct care' and 'secondary uses'

<sup>4</sup> Data sharing to support UK clinical genetics & genomics services - workshop report. Raza S, Hall A, Rands C et al. PHG Foundation / ACGS (2015). ISBN 978-1-907198-20-5. <http://www.phgfoundation.org/file/17089/>

Office, with collaboration from the PHG Foundation and the ACGS. The scope of the evidence session was limited to the sharing of data within the NHS for direct care and routine service delivery.

Clinical genetics professionals and clinical scientists present at that session described and provided many examples of the “iterative process” needed to interpret genetic variants resulting from a genetic test in order to give a patient the most accurate diagnosis, to provide them with the best possible care and to avoid unnecessary or even harmful treatment or surveillance.

Interpreting the significance of a genetic variant can require access to information about other people, to assess, for instance, how often the variant is linked with disease or not. This may well involve access to data from other patients under the care of different health professionals, potentially living in different parts of the country or even the world.

A strong message was conveyed during the session by clinical genetics professionals and clinical scientists that the sharing of data to support this process is necessary and urgent for there to be confidence that the best and safest care is provided to patients. There was discussion about, but not agreement on, the extent to which this can be carried out with anonymised data or the extent to which the data would be identifiable, since this relies heavily upon context. Where the data about one or more patients needed to care for another is considered to be identifiable, participants discussed that this challenges conventional understandings of medical confidentiality, echoing the point made also in the NDG Review of July 2016 that “*the lines between direct care and secondary use of data are blurred*”.

At the session three key issues were identified as important for the NDG to explore and consider; how far the iterative process described requires information which potentially identifies individuals, what sort of consent would be appropriate to underpin the sharing of information that identifies individuals, and whether the data sharing described could be considered to be for the ‘direct care’ of all the individuals involved.

Following that session the National Data Guardian, her panel and team had further engagement with the ACGS, PHG Foundation and other stakeholders about possible next steps.

## Evidence session

The scope of the evidence session held in October 2016 was limited to the sharing of data within the NHS for direct care and routine service delivery, with partners accepting that further work may well be necessary to examine the issues surrounding wider data sharing, including data sharing for research or other secondary purposes.

The evidence session was attended by 27 participants who included clinical geneticists, genetic counsellors and clinical scientists, legal and health policy experts, government officials, members of the NDG panel<sup>5</sup> and representatives of the NDG’s office.

A summary of the evidence provided and the discussions that took place at that session is provided below.

The evidence session was split into three parts. First, presentations were given about why and how data is required to be processed in order to provide high quality and safe clinical care in the field of genetics and genomics. The second session examined the legitimacy of data sharing and some of the important legal and ethical principles that pertain, and attendees heard about

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<sup>5</sup> The National Data Guardian appoints an independent group of experts - the NDG panel - to advise and support her work: <https://www.gov.uk/government/organisations/national-data-guardian/about>

a data sharing tool which is currently being used to share data to support clinical care. The third part turned to the question of whether and how the NDG could be constructively involved in progressing challenges identified around data sharing.

### **Presentation: Data sharing to support UK clinical genetics and genomics services**

**Alison Hall, Head of Humanities and Sobia Raza, Policy Analyst - Data Science, PHG Foundation**

In June 2015 the PHG Foundation and UK Association for Clinical Genetic Science co-hosted a workshop to examine the legal, practical, regulatory and technical factors that impede genomic data sharing. The impetus for this meeting was concern that unsatisfactory and inconsistent data sharing practices cause significant differences in patient care and compromise quality and safety. Key recommendations from the workshop highlighted an urgent need for national agreement to optimise sharing within the NHS and to develop consensus on the legitimacy of data sharing in order to deliver high-quality, safe and effective genetics / genomics diagnostic services. These recommendations formed the starting point for the evidence session.

#### **1. Establishing the clinical requirements**

### **Presentation: Data sharing to support NHS genomic laboratory services - the laboratory perspective**

**Professor Sian Ellard, Consultant Clinical Scientist**

Professor Ellard described the process that a clinical scientist will undertake in order to reach an accurate interpretation of a patient's genetic test. Testing may identify one or multiple rare genetic variants or mutations<sup>6</sup> present in a patient's DNA sequence, depending on how many genes are analysed. The clinical scientist will assess whether any of the variants identified are likely to be disease causing

The clinical scientist will go through a process of assessing evidence that includes the type of variant, the predicted effect upon protein function, published literature or databases of variants previously identified as disease causing, family history and clinical information relating to the patient and others who have the variant.

An example of how the process might work in practice was described:

Professor Ellard's laboratory was referred a couple who had two pregnancies terminated because antenatal scans had shown severe brain abnormalities. Sequencing all the protein-coding parts of more than 20,000 genes in the parents' DNA samples identified a rare gene variant carried by both of them. A DNA sample has been stored from one of the affected pregnancies and further testing showed that the fetus had inherited the variant from both parents. Neither of the couple's healthy children had inherited the variant from both parents. Then the laboratory went through the process of assimilating other evidence to determine whether this variant was likely to be the cause of the brain abnormality in the two affected pregnancies.

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<sup>6</sup> Throughout this document, the term genetic 'variant' is used to include disease causing, or potentially disease causing genetic variants, and to include what is also known as a 'mutation'.

In an international database they found one entry of this variant in the US which was marked as of 'uncertain significance'. On contacting the US clinic, the UK laboratory received confirmation that they had seen this variant in a patient that had the same very rare phenotype (clinical presentation). This meant that a pre-natal test could be offered to the couple early in their next pregnancy to ascertain if the fetus had also inherited these two variants.

Professor Ellard described the importance of having a national database into which laboratories can deposit details of genetic variants and their classification as likely to be disease causing or not, together with relevant phenotypic data, and for NHS laboratories to be mandated to deposit data into it.

## Presentation: Data sharing to support NHS genomic laboratory services - the genetic counsellor's perspective

### Dr Vishakha Tripathi, Consultant Genetic Counsellor

Dr Tripathi described the role that genetic counsellors and geneticists play in helping patients through the genetic diagnostic process and in particular, the scope and role of consent within clinical genetics and the choices there may be around the sharing of data.

Dr Tripathi described how diagnosing and treating patients may require data from family members and involve family members in investigations which may have implications for them as well. Where relationships have broken down or relatives have died, obtaining this information presents challenges. Additional challenges can be that investigations reveal unexpected facts about relationships or raise questions about the responsibilities clinicians may feel to alert other family members about their risk of genetic conditions, which may conflict with their duty to preserve confidentiality.

An example was given of a 35 year-old woman who comes into a clinic worried about her risk of breast cancer. She reports that her mother, maternal aunt and cousin had the condition. It emerges that relationships with her family have broken down and she does not think her relatives will consent to sharing their information to help her clinician ascertain her risk.

Dr Tripathi also presented a typical consent form for genetic testing, which informs patients that tests may enable relatives to benefit from genetic testing, may reveal unexpected information (for instance including information about a child's biological parents), that the sample may be sent to another laboratory for testing and that a leftover sample may be used to help develop new tests.

She said that in her experience, most patients view genetic information to be much like any other medical information once they have adjusted to the implications of it. She described patients as generally having trust in their clinical team that information will be managed securely and proportionately.

Dr Tripathi also raised the concept of 'familial consent' in the context of the fact that many of the genetic variants found in one person are the same in close family members, and that therefore a finding in one person might be very important to the risks of others. A question was raised as to whether there are circumstances where genetic information could be considered to be familial rather than individual? Dr Tripathi suggested that this could be presumed unless a patient objects, as currently takes place in Iceland and Japan and in some genetic services in the UK.

## Presentation: Data sharing to support NHS genetic/genomic practice - the clinical perspective

### Professor Anneke Lucassen, Consultant Clinical Geneticist

Professor Lucassen underlined how vital it is for clinical geneticists and laboratory scientists to use linked genotypic and phenotypic information in order to diagnose and treat patients. Such data cannot be fully anonymised. She also highlighted the importance of data sharing not just to determine an individual's management, but also for the appropriate care of relatives.

An example was given of a two-year old boy with non-specific symptoms where clinicians had ordered a genetic test to 'rule-out' certain diagnoses. Results revealed a variant in a particular gene that plays a role in heart rhythm abnormalities. In order to identify whether this variant was pathogenic a search of databases was made to see if it had previously been identified as disease causing. Because the variant theoretically affected the gene's message production and it had previously been described in an adult with long QT syndrome (a heart condition) it was labelled by the laboratory as 'likely pathogenic'.

One way of being more certain about pathogenicity was to investigate other family members and see whether they also carried the variant and had symptoms or signs that could be attributed to the variant. Many were tested and found to have the same variant, but no heart rhythm problems, even on specialised testing, suggesting that the gene finding was not as disease causing as first suspected. She highlighted some of the downstream consequences of this uncertainty: investigation of the child's family was good clinical practice to (a) ensure they were not at risk and at the same time (b) help determine the significance of the finding, but a consequence was that many NHS resources were directed at cardiological review of the boy and his family over many years and a diagnosis that was now far from certain. Such review is important as additional evidence over the years may mean the findings can be more clearly labelled as disease predisposing, or not.

The example underlines that accessing information about others is necessary to interpret information about an individual. This means the iterative process necessary in genetic medicine may well involve more than one patient and more than one health professional, potentially living in different parts of the country or the world. This challenges conventional understandings of medical confidentiality.

Professor Lucassen also gave an example of a patient who was identified as having a variant in a high risk 'cancer' gene resulting in her particular rare cancer. The patient had siblings living locally but she was not in regular contact with them. She was given a letter from the clinical genetics service to pass on to her siblings describing their risks and the availability of testing. (Each had a 50 per cent chance of having the cancer-associated gene variant, so this is standard practice in genetic services). The GP looked after all of the siblings and knew that the siblings could undergo preventative treatment if they have the gene variant, but did not feel able to raise this with them as he/she considered that this would be breaking the patient's confidence.

This case highlighted the question of whether it could be good practice for the GP to separate clinical and genetic information. In this way, the siblings under the care of the GP could be told 'you are at risk of condition X, this is what you can do about it' without breaching the clinical confidence of the patient in whom the familial inheritance was first discovered. Could this be regarded as a form of contact tracing for genetics?

Professor Lucassen also referred to professional guidelines on consent and confidentiality in clinical genetic practice [‘Consent and confidentiality in clinical genetic practice: Guidance on genetic testing and sharing genetic information’ Joint Committee on Medical Genetics<sup>7</sup>]. This guidance is currently being revised and a third edition is due in the autumn of 2017. The current (2011) guidelines state that it is appropriate for genetic medicine services to be explicit about a presumption of sharing familial information appropriately: Good clinical practice in genetics requires a degree of sharing of familial information. The standard consent form contained within the guidance includes a statement to this effect and highlights that any test results might be used to benefit other members of the family. Whilst some health professionals worry that this could appear to suggest they were not taking consent and confidentiality seriously, in fact most patients arrived at a genetic service in part to help other family members and so did not see a problem with such an approach. In fact out of about 10,000 uses of the form in her service this presumption had led to problems in about five cases.

Professor Lucassen said that clinical genetic practice has always had a close relationship with research, such that for any particular patient it might be difficult for them to know whether their test or screen was done through the NHS or a research study (see work of Hallowell et al<sup>8</sup>) and that in the genomic age, this was perhaps even more so. Nevertheless she argued that the sharing of linked genotypic and phenotypic data of individuals and families in national databases was crucial to good NHS clinical practice to understand the relevance of particular findings. Such sharing could not be considered research in her perspective as they were very much required for good clinical practice so therefore were not secondary uses of the data.

## 2. Establishing the legitimacy of data sharing

### Presentation: Genomic data sharing and the law

Dr Jon Fistein

Dr Fistein gave a high level description of some of the key legal principles which underpin the way data may and must be shared. He referred to guidance from the Human Genetics Commission report of 2002, *Inside Information: Balancing interests in the use of personal genetic data*<sup>9</sup>.

The report looked at how interests in genetic privacy and confidentiality should be protected in a way that does not harm comparably important interests of others. It set out a principle of confidentiality: “*Private personal genetic information should generally be treated as being of a confidential nature and should not be communicated to others without consent except for the weightiest of reasons*”; a principle of privacy: “*In the absence of justification based on overwhelming moral considerations, a person should generally not be obliged to disclose information about his or her genetic characteristics*”; and a principle of consent: “*Private genetic information about a person should generally not be obtained, held or communicated without that person’s free and informed consent.*”

He also referred to the principles in the Data Protection Act, picking out principle 2 under which data should only be used for the purposes for which it was collected. He asked whether data

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<sup>7</sup> [www.rcplondon.ac.uk/sites/default/files/consent\\_and\\_confidentiality\\_2011.pdf](http://www.rcplondon.ac.uk/sites/default/files/consent_and_confidentiality_2011.pdf)

<sup>8</sup> <http://jme.bmj.com/content/35/2/113.short>

<sup>9</sup> [http://webarchive.nationalarchives.gov.uk/20061023110946/http://www.hgc.gov.uk/UploadDocs/DocPub/Document/insideinformation\\_summary.pdf](http://webarchive.nationalarchives.gov.uk/20061023110946/http://www.hgc.gov.uk/UploadDocs/DocPub/Document/insideinformation_summary.pdf)

collected for the treatment of a patient can be used for other purposes, such as informing the treatment of another patient.

Dr Fistein discussed how the concept of confidentiality might be considered within a ‘distributed health system’ where many professionals, not necessarily in the same organisation, might be involved with a patient’s treatment. He suggested that the concept of ‘reasonable expectations’ might be important and that this would need to be tested outside the consulting room.

Dr Fistein referred to the “culture of anxiety [that] permeates the health and social care sector” referred to in Dame Fiona’s Information Governance Review<sup>10</sup> report published in 2013, and suggested that this culture of anxiety might be present around clinical genetics services. He quoted the definition of direct care defined in the same report. Dr Fistein asked whether the use of information about one person to directly affect the quality of care that someone else receives could or should be included under this definition.

Looking at the possible legal bases for allowing the use of personal confidential information, Dr Fistein described the four key routes for this:

- Consent - impliedly or explicitly; for example, gained in a direct care setting
- Legal requirements to disclose (e.g. statutory or as ordered by a court)
- Public interest justification for disclosure (e.g. prevention of significant harm to a named person, prevention/detection of serious crime)
- Applications under s251 of the NHS Act 2006 to set aside the duty of confidentiality for particular purposes.

There was discussion about whether each of these routes might apply to clinical genetics services.

Dr Fistein examined how consent might be used as the basis for the sharing of personal confidential information in the context of clinical genetics. For consent to be valid, it must be informed and voluntarily given by someone with capacity. There was discussion about what meeting this test would mean practically in terms of the information patients should be given, and the expectations patients might have about the way that data about them would be shared.

There was discussion about the need for the use of identifiable information for clinical genetics services, in particular asking whether ‘anonymised’ data<sup>11</sup> could deliver desired benefits for clinical genetics services, and more generally asking to what extent a risk-benefit judgement could be applied.

Dr Fistein suggested that the role for the NDG in this area could be to provide more detail on the definition of direct care, to provide examples of good practice for data sharing within distributed clinical services, to give recommendations for promoting transparency about data uses in the NHS (including clinical genetics services) and to say more about patient expectations e.g. what the requirement for ‘no surprises’ means.

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<sup>10</sup> <https://www.gov.uk/government/publications/the-information-governance-review>

<sup>11</sup> See Annex B for description of anonymisation

## Presentation: DECIPHER - A data sharing platform used to share genetic and genomic data to support clinical care

Dr Helen V Firth, Consultant Clinical Geneticist

Dr Firth started by dispelling some false assumptions about genetics, underlining that there are still a great many unknowns around genetics; that gene variants are by no means rare and do not always cause disease; and that literature and existing databases are littered with mistakes as this strand of medicine and science continues to develop.

Dr Firth explained that most genetic disorders are rare or ultra-rare, which means that finding other patients with variants in the same gene and comparing their clinical features (phenotypes) is essential in order to provide safe care to patients.

She described the Deciphering Developmental Disorders project<sup>12</sup> (the DDD project), a project between 23 NHS genetics services to which 13,500 families were recruited. All patients consented for genetic and clinical data to be submitted to a central database creating a “virtual” unified service. Dr Firth gave an example of the way the project enabled working as a collective genetic service. In one case, three patients with undiagnosed developmental disorders (in Newcastle, Manchester and Liverpool) were found to have the same genetic variant. Pooling data from these patients facilitated their diagnosis, which would have otherwise been impossible without data on other similar patients.

When considering the legitimacy of data sharing, Dr Firth recommended that a principle of proportionality be applied which balances the depth of data shared with the breadth of data shared.

She described an urgent need for a dynamic, flexible, comprehensive and scalable way to share the information needed to support NHS clinical genetics services, which should build on the current and the rapidly evolving knowledge of the human genome. The data needs to be captured and structured in a way that it can be shared appropriately.

Dr Firth explained how the DECIPHER<sup>13</sup> system has been developed over 12 years and explained how those involved in the DDD project saw the benefits for NHS patients of clinical genetics services working as a combined distributed network of expertise across the country.

Dr Firth and NHS colleagues have built DECIPHER working with genomic scientists and computer scientists at the Sanger Institute. Its main purpose is to enable links to be made between clinical features and the gene, to map which bits of the genome are relevant for clinical practice.

The system has been used internationally to share data since 2004. Since 2011, data has been shared between a consortium of 23 NHS genetic services in the DDD study and for more than a year, NHS genetics services have been contributing data, as part of the standard treatment of their patients, to an NHS consortium that currently has 15 members. The genetic services submit information about genetic variants they have observed into the database, annotated with their opinion regarding pathogenicity. Some genetic services also submit some very high level clinical data (e.g. a handful of clinical features using terms such as ‘cataract’ or ‘short stature’). Participation of each NHS genetics service in the NHS consortium has been approved by local Caldicott Guardians and the infrastructure has Research Ethics Committee approval.

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<sup>12</sup> [www.ddduk.org](http://www.ddduk.org)

<sup>13</sup> [hops://decipher.sanger.ac.uk](https://decipher.sanger.ac.uk)

There are more than 36,000 patient records which can be accessed by NHS clinical genetics services in the consortium, together with a further 22,000 open records (for which patients have given explicit consent for data sharing) to look up genetic variants and identify if there are other cases of the same variant that have been identified by other participating genetics services and whether the other case(s) share clinical features.

Where there is no match in the database that helps to resolve their case, the clinician may also seek explicit consent from patients to share their information more widely beyond the NHS genetics services within the consortium, so that international comparisons can be made.

Matches between the genetic variants shared by unrelated individuals made through the DECIPHER system, facilitating diagnosis and gene discovery, have resulted in more than 1,000 discoveries leading to publications in the scientific literature.

Dr Firth said that the system only stores the data needed, it does not for example store names, dates of birth or addresses. The local identifier is visible only to the submitting centre and can only be decrypted to identify an individual by clinicians and clinical laboratory staff working in that centre. She stated that access is only given as required and is password protected and encrypted and there is a secure data centre that is regularly penetration tested. If deemed appropriate, technical solutions that would enable the hosting of NHS Consortium data in DECIPHER behind an N3 firewall<sup>14</sup> could be explored.

### 3. How can the National Data Guardian be constructively involved?

In the third session, Dr Mark Taylor of the NDG panel chaired a discussion about how the NDG might be constructively involved. In this session there was some discussion about the scope of the work. Some felt that it was important to keep the defined brief of developing a consensus around data sharing within the NHS to support direct care in order to focus on the most immediate problems and those that potentially might be resolved most quickly. While questions of sharing information with family members had been raised on the day, it appeared that the more pressing matter to be resolved was the legitimacy of the deposition of variants and relevant clinical (phenotypic) information in databases to which the NHS has access in order to inform the treatment of patients receiving care.

Others felt it might be helpful to extend the scope of the work, pointing to the supra-national aspect of care in this field currently. Affected individuals can live anywhere and some participants in the workshop described collaboration with geneticists internationally in order to diagnose patients in the UK.

Dr Taylor laid out a number of considerations that he felt were relevant for the National Data Guardian to consider and understand. These considerations included:

**Identifiability of the data:** Some in the room argued that one should assume that the combination of genotypic and phenotypic data that needs to be shared to underpin care for patients of clinical services will be identifiable or potentially identifiable. Proponents of this approach argued that although identifying a name or address of a person from such data may be

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<sup>14</sup> See Annex G for explanation of N3

very difficult, this is possible now and may become easier in the future; anonymisation as a means to deal with sharing was therefore not sufficient.

Others argued that the risk of identification from the data was very small or nil, saying that the data that needed to be shared to enable good clinical care would, in many cases, only be identifiable to the patient and their clinician.

It was suggested that a Privacy Impact Assessment could be used to assess if the data required to support clinical genetic services is, or could be, anonymised in line with the ICO Code on Anonymisation<sup>15</sup>. This would then remove the need to use consent or other legal gateways for sharing identifiable information.

**Direct and indirect care:** It has become established practice within the NHS that where direct care is being delivered, identifiable data may be shared to support this care on the basis of implied consent with those with whom the patient has a legitimate relationship.

The Information Governance Review of 2013, also known as Caldicott 2 explores this and provides a working definition for when information that identifies individuals could be shared on the basis of implied consent<sup>16</sup>. This review strongly emphasised the need for there to be a legitimate relationship between the person looking at the information and the individual receiving care and that the data sharing should meet the reasonable expectations of that individual. In other words, there should be no surprises for individuals about how their information is used.

During the evidence session, members of Dame Fiona's panel did not reach a point of agreeing that all the uses of patient data that were described as necessary to support clinical genetic services could necessarily be aligned with the definition of direct care as described in Caldicott 2 for all of the individuals involved.

In particular there were challenges around the legitimate relationship requirement in the Caldicott 2 definition in the scenario where a clinician or clinical scientist would be using information about patients who were not under their care to potentially advance the diagnoses and treatment of a patient who was under their care.

Some participants argued that the iterative nature of variant interpretation in clinical genetics, which is integral to reaching an accurate diagnosis, meant that the activity of sharing data about many patients in order to deliver clinical care should all be understood as falling under the direct care definition.

An idea was put forward that 'legitimate relationship' might be redefined as 'legitimate interest' or a separate definition of 'legitimate interest' developed. The proposal was that clinicians and healthcare professionals governed by professional standards and delivering care through clinical genetic services might be said to have a legitimate interest with all the patients whose data is involved in their work. Another suggestion was that clinical genetics professionals working in the NHS could be understood to be within a distributed care team delivering care.

The question of whether this sharing of data would be within the 'reasonable expectations' of patients was also raised by NDG panel members. Several geneticists who have routine contact with patients put forward the view that their patients expect and want them to discuss their case with other clinicians across the NHS and, in some cases, internationally.

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<sup>15</sup> <https://ico.org.uk/for-organisations/guide-to-data-protection/anonymisation/>

<sup>16</sup> <https://www.gov.uk/government/publications/the-information-governance-review>

**Consent:** If the data needed to underpin clinical genetics services were understood to be identifiable, the question arises - what is the appropriate legal basis for sharing it? In the absence of a statutory basis, the most appropriate basis would appear to be consent; either implied or explicit.

Participants described differences in consent and information provision practices across clinical genetic services. Patients who undergo a genetic test are sometimes asked to sign a written form, which may refer to DNA being stored, used to diagnose family members or sent to a lab. Some trusts rely on implied consent for using and sharing patient data and no written consent is taken relating to using and sharing patient data.

The consent process adopted by the 100,000 Genomes Project was also raised as potentially informative, albeit that it is primarily focused on taking consent for research (indirect care) uses. The explicit consent taken by the 100,000 Genomes Project could be described as a 'hybrid consent', combining research and care aims and which covers both direct and indirect care purposes<sup>17</sup>.

The consent is explicit and is sought by eligible NHS patients' clinical teams. It outlines, for example, that participants' de-identified data donated for research purposes, could have tests and research done on it which have not been invented yet. The consent process has also been designed to make clear to patients that not all the uses of the data, and possible results and risks, are foreseeable at the moment of consent being taken. The project also offers participants various options beyond cancer or rare disease diagnostic care - including receiving further health-related 'additional findings' from their genomic results. These can provide participants, via their NHS teams, with information relating to increased risk found of a limited number of rare and serious but treatable conditions.

Some participants felt that explicit consent could be used consistently in NHS clinical genetics and genomics service in order to provide a clear legal basis for the sharing of identifiable or potentially identifiable data, without the need to reach consensus around the applicability of the definition of direct care to the pooling of data. However, others queried the feasibility of obtaining explicit consent especially as genetic medicine becomes more mainstream in the NHS, with patients more routinely undergoing genetic testing, not just under the care of genetic specialists. They argued that a requirement for explicit consent to be taken was unrealistic and could negatively impact upon service delivery and patient care.

Some participants also strongly resisted the suggestion that explicit consent would be necessary as they felt that all of the data sharing described falls under direct care and that implied consent can be used as a basis for data sharing and usage. Others reiterated their position that genetic variant data could be shared in anonymous form and therefore that a legal basis to share the data, such as consent, was not required.

Some participants at the evidence session suggested that there might be some contexts in which patients' reasonable expectations would allow sharing on the basis of implied consent even if the activity did not fall under the definition of direct care given in the Information Governance Review. For instance, it might be that given the contextual features of genetics, patients' reasonable expectations are somewhat different from those in other clinical interventions, thus allowing information to be shared more widely on a basis of implied consent than it might be for

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<sup>17</sup> <https://www.genomicsengland.co.uk/taking-part/>

other types of care. NDG panel members felt that in order to justify this, a strong case would need to be made that this would be in line with patients' reasonable expectations, and there would need to be extensive information giving to ensure 'no surprises'.

There was agreement that whatever the approach, any advice should take account of broader challenges (such as the forthcoming EU General Data Protection Regulation) and of the pace of technological change, so that it does not make resolving these difficult at a later date.

## Other issues

Participants noted that a number of initiatives led by other stakeholders will be important in helping to develop optimal data sharing practices and in building wider public trust. These include further development of the infrastructure required for managing and sharing genetic and genomic data in the NHS and identifying proportionate and appropriate security measures to mitigate and minimise risk. Revisions to professional guidance will also inform and educate a wider group of clinical specialties and facilitate greater consistency of practice.

## Conclusion and next steps

During the October 2016 evidence session, a number of possible actions and next steps were raised. Additional discussions between the NDG, PHG Foundation and the ACGS refined these further. Suggested next steps were also shaped by the attendance by the National Data Guardian and some of her panel members at a December 2016 meeting with Genomics England's Ethics Advisory Committee, engagement with the work on genetics that has been undertaken to support the Chief Medical Officer's 2017 annual report<sup>18</sup>, discussion about NHS England plans to reconfigure NHS genomic laboratory services and consideration of the General Medical Council's revised Confidentiality Guidance<sup>19</sup>.

Constructive feedback was also received from the Association of Genetic Nurses and Counsellors, the British Society for Genetic Medicine and many of the individual participants at the October 2016 evidence session. As a result of this, the National Data Guardian would suggest two key next steps.

### 1. Further work to explore appropriate consent for routine NHS clinical genetics and genomics services

Consent was a key issue raised at the evidence session and in further discussions.

The NDG believes there should be further work to explore how the consent process might cover both direct and indirect care purposes as genetic and genomic medicine become a more routine part of care for a greater number of NHS patients. The NDG is aware of a number of pieces of work that may be able to support this.

The professional guidance for clinical genetics and genomics services on consent and confidentiality<sup>20</sup> is currently being revised, due for publication in the autumn of 2017. It would be useful to explore how this guidance might address these issues.

Consent was an important part of the discussion with the Genomics England's Ethics Advisory Committee in December 2016 and the suggestion was raised of further testing with patients of 'hybrid consent'.

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<sup>18</sup> Insert online reference once published

<sup>19</sup> [http://www.gmc-uk.org/guidance/ethical\\_guidance/confidentiality.asp](http://www.gmc-uk.org/guidance/ethical_guidance/confidentiality.asp)

<sup>20</sup> [www.rcplondon.ac.uk/sites/default/files/consent\\_and\\_confidentiality\\_2011.pdf](http://www.rcplondon.ac.uk/sites/default/files/consent_and_confidentiality_2011.pdf)

We understand that Genomics England and NHS England are now taking forward joint work to explore how a consent model which would have elements covering both clinical and research activities could be tested after the planned reconfiguration of genetic laboratories in the NHS brings an NHS genomics service closer to mainstream clinical use, outside the 100,000 Genomes Project.

It has also been helpful for the NDG to engage with the thinking that has been undertaken to support the Chief Medical Officer's (CMO) 2016 annual report Generation Genome<sup>21</sup>, which was published in July 2017. It examines the potential for genetics to improve care for NHS patients and looks at what this might mean for the way data is used and for the conversation that should be had with the public. Dame Fiona fully supports the call from the CMO for an open dialogue with the public and notes the CMO and Genomics England's work to support this in relation to the 100,000 Genomes Project.<sup>22</sup>

As work in this area develops, key issues for consideration would be how far patients' reasonable expectations of data use would or could align to consents, such as hybrid consents. These might make an offer to NHS patients of much greater options for data sharing to inform future research activity than is currently standard as part of NHS care, but also (in the case of genomic medicine) tend to require a patient to consent to a greater level of information sharing to provide the most accurate diagnosis than is typical in other more traditional diagnostic approaches. Genomics in the NHS challenges the traditional boundaries between research and clinical care and, given the extent of specialist information that may be required to achieve a diagnosis, also challenges traditional understandings of who might be within the patient's clinical team.

It will also be important to consider what information might be necessary to support patients' understanding during and after the consent process, (including for children and young people and their families), and whether and how such a consent might be reconcilable with the Common Law of Confidentiality, the Data Protection Act and the General Data Protection Regulation and the Human Rights Act.

As many of the participants in the evidence session highlighted, genomics will become increasingly relevant to all of medicine and these issues therefore will need to be considered more widely than only in relation to clinical genetic and genomics services. The challenges posed by this increasing blurring of the traditional boundaries between 'care' and 'research' - or perhaps the emergence of a third category, the 'hybrid' model will require robust consultation and feedback from research participants and NHS patients and their families and the wider public, as well as with professionals and policymakers to ensure acceptability and feasibility in routine NHS care.

One option to gain this feedback could be to explicitly test the model as a pilot within the 100,000 genomes project research framework.

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<sup>21</sup> <https://www.gov.uk/government/publications/chief-medical-officer-annual-report-2016-generation-genome>

<sup>22</sup> <http://www.progress.org.uk/genomics>

## 2. Use one or more privacy impact assessments (PIA) to examine suitable arrangements for the sharing of data within the NHS where genomic testing could be used as part of routine clinical care.

The Privacy Impact Assessment (PIA)<sup>23</sup> process could be used to examine issues such as the level of identifiability of the data, whether mitigating steps can be taken so that some or all data needed could be considered anonymised under the ICO code, what the available legal bases are for existing and proposed flows and linked to a revised consent model, what information and options could be provided to patients and the public.

NHS England is reconfiguring NHS genomic laboratory services, an important step in building a genomic medicine service in the NHS.

The National Data Guardian suggests that this is an excellent opportunity to use a PIA to support the NHS to examine the privacy implications of services sharing data anonymised in line with the ICO Code and potentially identifiable information, such as a combination of variant data and phenotypic data.

### August 2017

This paper has been prepared by the Office of the National Data Guardian, with significant support from the ACGS and PHG Foundation. Constructive feedback was also received from the Association of Genetic Nurses and Counsellors, the British Society for Genetic Medicine, Genomics England, NHS England, the CMO Office and many of the individual participants at the October 2016 evidence session.

We are extremely grateful to all who have made such valuable contributions.

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<sup>23</sup> <https://ico.org.uk/for-organisations/guide-to-data-protection/privacy-by-design/>

**Annex A: List of attendees at the joint evidence session held in October 2016**

Name	Job title and organisation
Dr Mark Bale	Deputy Director, Science Research and Evidence Directorate Department of Health
Lindsey Blake	Senior Delivery Manager Office of the National Data Guardian
John Carvel	Member of the National Data Guardian's Panel
Professor Angus Clarke	Clinical Professor, Institute Of Medical Genetics, Cardiff University
Louise Coleman	Genetic Alliance
Professor Ann Dalton	Director, Sheffield Diagnostic Genetics Service and representing ACGS
Professor Sian Ellard	Director of the South West NHS Genomic Medicine Centre and Professor of Genomic Medicine at the University of Exeter Medical School
Kathy Farndon	Head of Data and Informatics, Genomics, NHS England
Professor Frances Flinter	Consultant Clinical Geneticist and Caldicott Guardian, Guy's & St Thomas NHS Foundation Trust
Dr Helen Firth	Consultant Clinical Geneticist, Cambridge Hon Faculty Member, Wellcome Trust Sanger Institute, Hinxton
Dr Jon Fistein	Clinical Research Fellow, University of Leeds
Simon Gray	Director, Office of the National Data Guardian
Alison Hall	Head of Humanities, PHG Foundation
Dr Alan Hassey	Member of the National Data Guardian's Panel

Lorraine Jackson	Deputy Director, Data Sharing & Cyber security, Department of Health
Professor Anneke Lucassen	Professor of Clinical Genetics University of Southampton and consultant in clinical genetics UHS
Dawn Monaghan	Head of Data Sharing and Privacy, NHS England
Professor Willem Ouwehand	Professor of Experimental Haematology, University of Cambridge
Professor Jem Rashbass	National Director for Disease Registration and Cancer Analysis, Public Health England
Dr Sobia Raza	Data Science Policy Analyst, PHG Foundation
Dr Beverly Searle	Unique
Karen Swift	Business Support Manager Office of the National Data Guardian
Dr Mark Taylor	Member of the National Data Guardian's Panel
Dr Vishakha Tripathi	Consultant Genetic Counsellor, Guys Regional Genetics Service
Jenny Westaway	Senior Communications Manager, Office of the National Data Guardian
Dr Jo Whittaker	UK Genetic Testing Network Scientific Development Advisor
Dr James Wilson	Member of the National Data Guardian's Panel

## Annex B. Summary of terms used in the paper

The terms and definitions used in this paper are based on those used in the Information Governance Review 2013<sup>24</sup> and National Data Guardian Review of Data Security, Consent and Opt-Outs 2016<sup>25</sup>.

**Anonymisation:** The process of rendering data into a form which does not identify individuals, or which makes the risk of re-identification sufficiently low in a particular context that it does not constitute personal data.

**Consent:** The informed agreement for something to happen after consideration by the individual. For consent to be legally valid, the individual must be informed, must have the capacity to make the decision in question and must give consent voluntarily. In the context of consent to share confidential information, this means individuals should be aware and understand how their information is to be used and shared (there should be ‘no surprises’), and they should understand the implications of their decision, particularly where their refusal to allow information to be shared is likely to affect the care they receive. This applies to both explicit and implied consent. See the Information Governance Review for definitions of explicit and implied consent.

**Data Protection Act 1998 (DPA):** The Act of Parliament which regulates the processing of information relating to living individuals, including the obtaining, holding, use or disclosure of such information.

**Data sharing:** The disclosure of data from one or more organisations to a third party organisation or organisations, or the sharing of data between different parts of an organisation. This can take the form of systematic, routine data sharing where the same data sets are shared between the same organisations for an established purpose or for exceptional, one-off decisions to share data for any of a range of purposes.

**Direct care:** Defined in the Information Governance Review as a clinical, social or public health activity concerned with the prevention, investigation and treatment of illness and the alleviation of suffering of individuals. It includes supporting individuals’ ability to function and improve their participation in life and society. It includes the assurance of safe and high quality care and treatment through local audit, the management of untoward or adverse incidents, and person satisfaction including measurement of outcomes undertaken by one or more registered and regulated health or social care professionals and their team, with whom the individual has a legitimate relationship for their care.

**General Data Protection Regulation (GDPR):** The General Data Protection Regulation (GDPR) is the new EU Regulation 2016/679 adopted by the European Parliament and Council, which is intended to strengthen and unify data protection for individuals within the European Union.

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<sup>24</sup> <https://www.gov.uk/government/publications/the-information-governance-review>

<sup>25</sup> <https://www.gov.uk/government/publications/review-of-data-security-consent-and-opt-outs>

**ICO:** The Information Commissioner's Office, established as the UK's independent authority to uphold information rights in the public interest, promoting openness by public bodies and data privacy for individuals.

**Identifiable information or data:** See 'Personal confidential data'.

**Indirect care or secondary uses:** Defined in the Information Governance Review as activities that contribute to the overall provision of services to a population as a whole or a group of patients with a particular condition, but which fall outside the scope of direct care. It covers health services management, preventative medicine, and medical research. Examples of activities are risk prediction and stratification, service evaluation, needs assessment, and financial audit.

**Information Governance Review:** Following a request from the Secretary of State for Health, Dame Fiona Caldicott carried out this independent review of information sharing to ensure that there is an appropriate balance between the protection of patient information and the use and sharing of information to improve patient care. It is available here:

<https://www.gov.uk/government/publications/the-information-governance-review>

**N3:** N3 was a private network used by NHS and other health and care organisations. Security protocols set by NHS Digital required firewalls between local networks and computers and the N3 network, to control what can go back and forth. N3 network connectivity was replaced by the Health and Social Care Network in March 2017: <https://digital.nhs.uk/health-social-care-network>

**Personal Confidential Data (PCD):** Personal information about identified or identifiable individuals, which should be kept private or secret. For the purposes of this paper 'Personal' includes the DPA definition of personal data, but it is adapted to include dead as well as living people and 'confidential' includes both information 'given in confidence' and 'that which is owed a duty of confidence' and is adapted to include 'sensitive' as defined in the Data Protection Act.

**Secondary uses:** See 'indirect care'