Chapter 16

Ethics and the social contract for genomics in the NHS

Chapter leads
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1. Introduction

Previous chapters in this report have illustrated the potential of genomics to lead to important improvements in our understanding of health and in the diagnosis and treatment of disease. They have also suggested that the achievement of these benefits is going to require significant changes in the ways in which healthcare is understood, organised and practised in the NHS. One of the most important of these is the need for a greater degree of integration of, and complementarity between, healthcare and medical research. A second is a growth in the importance of the collection, storage and appropriate sharing of information at scale: in the care of individuals and families, in research, and in the improvement of health systems. A third will be the importance of a faster pace of learning and a consequently greater degree of uncertainty and open-endedness in healthcare practice. Each of these is going to have profound implications for the way the NHS works and for how the obligations and responsibilities of health professionals and institutions – as well as those of patients – are to be understood. Together, however, these changes have the potential to bring important benefits to patients and their families.

The founders of the NHS were committed to two guiding principles.[1] The first of these was to the availability of healthcare on the basis of need and independent of the ability to pay. The second, which is less widely discussed than the first, was that this health care should be of a very high standard of excellence. The value of the first is clearly enhanced by that of the second. Development of genomics and its integration into day-to-day practice of the NHS has the potential to provide important improvements to the quality of care provided equitably by the NHS, but the pace of development and the need for large scale data interpretation mean that research and innovation will need to become more integral to routine NHS practice than we have seen to date.

In this chapter, we reassert the importance of these important founding principles of the NHS and argue that if the potential benefits of genomics are to be realised, there is a need for a rethinking of the wider 'social contract' for medical practice and research in the UK.* After introducing and discussing the social contract and its importance, we go on to identify and discuss four areas to which we believe particular attention needs to be paid. The first of these is consent. The second is the use of information in the care and treatment of both patients and their relatives. The third is the need to rethink the duties and responsibilities of health professionals, and the fourth concerns the responsibilities of health systems in the context of rapid developments outlined in previous chapters.

* In this chapter we use the term 'social contract'. In similar discussions others have sometimes used the term 'social license'. We prefer the term social contract because of its helpful implication of the location of healthcare and medical research in a broader context of social arrangements, practices and institutions.

2. Rethinking the social contract for medical care and research

The NHS Constitution reminds us that our health service is founded on a common set of principles and values that bind together patients, the public and staff in order to ensure that it can be effective and equitable. It recognises that each party has important rights that must be respected, but also that each owes each other responsibilities. Through this combination of reciprocal rights and obligations the NHS aims to operate fairly and effectively for mutual benefit. The NHS Constitution is thus the expression of a form of 'social contract' which aims to bring the highest levels of human knowledge and skill to save lives and improve health. We need to understand and agree how those rights and responsibilities work in genomics if we are to harness its potential to fulfill the promise that the NHS "is there to improve our health and well-being, supporting us to keep mentally and physically well, to get better when we are ill and, when we cannot fully recover, to stay as well as we can to the end of our lives".[2]

Under this social contract, the health service has important responsibilities. We feel safe in entrusting our bodies and intimate personal information to health professionals in part because of the rights we have to protect our ourselves through the giving or withholding of consent, in part because we have confidence that those staff will act with integrity in our interests – for example in maintaining high standards of confidentiality – and in part because there are systems in place that protect our interests and hold professionals to account. In the era of genomic medicine the basis of this contract needs to be revisited. Most obviously, this is because linking up of large data-systems containing personal identifiable data,† on a scale not previously necessary (or possible), is a prerequisite for success, but also because genomics will provide both diagnoses and predictions and will affect patients, families, the general public in different ways over time.

Whilst it is clear from earlier chapters in this report that genomics has the potential to bring great benefits to patients, there has been considerable, and understandable, public concern over the handling of personal data by the NHS,[3] coupled with suspicion over the involvement of commercial organisations in the handling of 'big data'.[4] A recent survey commissioned by the Wellcome Trust on commercial access to data suggests that the public see genomics as both of great potential benefit and as presenting important risks.[5] The success of genomic medicine will depend on patients having confidence that the way genomic information is generated, held and used will properly protect their interests. This requires re-examining the traditional rules around confidentiality, which focused on secrecy and the keeping of information as separate and private. Such a rigid separation cannot operate in genomics, which requires clinicians to consider the patient's specific genetic situation in comparison with knowledge gleaned from others. The most important structural implications of the move to genomic, big data-driven medicine is the requirement for a greater degree of interdependence between the care and treatment of individual patients on the one hand and the collection and analysis of data relating to the care of very large numbers of other patients, often in real time, on the other. The clinical interpretation of genome findings requires information about clinical features in others with similar findings: the genotype-phenotype association remains an important clinical tool. Genomic medicine will require use of patient level information to support better clinical decisions in the future and for others. For this to be ethical, and acceptable to patients, a stronger focus on information security, data analysis and decision-support will be required as will greater clarity about and broad agreement on the relevant obligations of health professionals and systems.‡

† In the past, research studies providing the data for health care improvement would either have been collected using specific consent, and or a non-identifiable form. Genomics raises problems with anonymization because the data cannot be completely anonymised and can result in identification of individuals. Attempts to deal with this issue through consent are problematic because of the wide range of possible outcomes, over time, from genomic research.

‡ In addition to the benefits to individuals of interpreting their information in the context of a larger dataset, there will also be situations in which information arising in the direct care of one person, can prove vital in informing the care of others e.g. where it leads to the identification of an infectious disease, or where it identifies others at high risk of a condition. Provision will also need to be made for dealing with such situations.

As the Nuffield Council on Bioethics has suggested,[6] the successful and appropriate use of data-driven approaches to healthcare and research will require the NHS to provide the public with a mutually acceptable statement of the expectations that they can have of the use of data. This would need to include a realistic explanation of the ways in which genomic medicine can personalise care, addressing the sometimes excessive assumptions about the predictive or diagnostic power of genetics. It would require an explanation of how the management of data protects privacy, but necessarily uses information that cannot be completely anonymous because of the uniqueness of our genetic identities.[§] It would need to recognise that one of the consequences of advances in genomic medicine is that initial consents to the use of genomic information cannot be fully informed about future uses or interpretations, and so there is a need for high standards of broad consent to be complemented by continuing oversight of the way genomic data is handled.

This needs to be coupled with assurances of the competence of the NHS to deliver on the information governance requirements of genomic medicine. People need to be able to trust it to hold data securely, make it available to clinicians reliably when needed for patient care, and to patients themselves in an intelligible manner. Current information systems in the NHS are unlikely to be sufficient to earn this level of trust since they often seem to operate on a tacit permission to continue until the public withdraws their support due to mistrust. Instead, people need to be satisfied that genomic medicine operates in their common interests, whilst protecting their individual privacy, and does not exploit some to benefit others. Protection of individual privacy cannot be absolute,[**] nor can data ever be guaranteed as entirely secure, but there needs to be an understanding of the associated risks and reassurance that breaches are appropriately prosecuted.

The basis for greater trust and confidence created by such a social contract could encourage the growth of "genomic citizenship" or the genetic altruism and solidarity described by the Human Genetics Commission[7]: Genomics offers benefits and responsibilities for the individual, the family, the broader community and globally that cannot be realized by keeping the secrets revealed from one genome separate from others.

This requires a mutuality that is not captured by current systems. The idea of a social contract provides a basis for such an arrangement because it endures over time, brings benefits (and obligations) for both patients and the professionals (and services) who offer care. To achieve this, processes for creating common understanding are required, as well as mechanisms for revising the agreement when necessary.[8]

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§ Even though we differ in only roughly 0.1% of our genetic codes, this still equates to some 3 million variants.

** We will also need to address how protection of privacy is related to identifiability of genomic information, i.e. just because a sequence is potentially identifiable because of its uniqueness, does not mean that the privacy of a person is more invaded than were the data truly anonymous.

3. Renegotiation of the social contract - reasons

3.1 Overview

We began this chapter by highlighting three key requirements for the achievement of the benefits of genomics in the NHS for patients and families. These were: a greater degree of integration and complementarity between research, innovation, and clinical practice; the collection, storage and more effective use of health data; and, a more central role for learning and open-endedness in day-to-day clinical practice. In what follows, and against this background, we discuss a selection of some of the key areas of medical practice and research in which new ways of thinking about and practising medicine – each an important element of the social contract - are going to need to be considered in the renegotiation of the social contract. They are: (1) consent, (2) confidentiality and caring for families, (3) the obligations of health professionals and researchers, (4) the appropriate uses of data and samples, and a range of governance and system responsibilities.

3.2 Valid consent

The obtaining of valid consent is an important part of good ethical practice in healthcare and research. Whilst consent is an important component of ethical practice, it is not in itself, however, a guarantor of high ethical standards. In genomics as elsewhere, consent needs to be understood as an important component of an ethics ecosystem along with, for example, the duties and obligations of health professionals to treat patients with respect and care, and the requirement for health systems to provide protections to ensure that those who provide their consent are not exploited, discriminated against or unfairly treated.

Consent is nonetheless an important part of good medical practice and high standards of consent are essential. In genomic medicine both the importance and the limits of consent become increasingly apparent. The wider introduction of genomics into medical practice will present significant challenges for the achievement of understanding. Many of the key concepts in genomics are both complex and likely to be new to many patients (as well as the health professionals offering them) and may present problems of explanation and understanding as will many of the features the healthcare system in which genomics plays a central role: the close relationship between research and clinical practice; the collection, storage and use of health data; and, the open-endedness and uncertainty at the heart of a learning healthcare system. These latter two factors will be important both at the time of consent and at the time of communication of results which may be revised over time as new evidence accumulates. There will often be a degree of uncertainty about findings and their current or future implications as well as uncertainty about the potential future research uses to which data may be put and what additional (or incidental) findings this may produce in the future. This uncertainty may be at the level of evidence available; more big data is needed to ensure that findings are reproducible, and that confidence limits are minimised. It may also be reflected in the fact that even where good evidence exists, the chances that the finding will result in a particular symptom or condition is uncertain because it may be just one factor in amongst several that determine whether the condition manifests. NHS health professionals will also need to improve their acknowledgement of such uncertainty, as too often the language of single highly penetrant gene mutations is used for susceptibility factors that may never manifest as signs or symptoms.

Case study 1

Results of genomic investigations in patients investigated for neuro-developmental delay (whose samples have been collected and stored in a national resource) reveal a mutation in a gene that increases the chance of a brain tumour. The relative risk that this mutation appears to confer is very high, but the absolute risk less than 15%. Records of the consent taken at the time of testing reveal no mention of the possibility of tumour/ cancer risks being found. Health professionals are concerned about disclosing this finding since no specific consent was given to find it. Some argue that the patients have a right to know about their increased risk, but others argue that the lack of clear evidence based interventions and the 85% chance of not developing such a tumour would go against disclosure. However, had consent been explicit about this possibility the health professionals would have disclosed regardless of 'actionability'.

The quality of consent needs to be sufficient to reflect the importance of respecting patients' autonomy, but what kind of understanding of genomics is good enough for a decision to be seen as autonomous? This problem is sometimes presented as a problem about 'broad consent'. To what extent can consent to participation can be thought to be genuine 'consent' where significant implications of the decision are unknown, or unknowable at the time? Can such consent meet the requirements for validity? How broad can consent be and still be valid, or indeed prevent claims of insufficient information, to make a decision?

In this context, a key question is going to be how might the validity of consent be judged in contexts of such complexity and uncertainty? It is clearly not reasonable for the answer to be that consent to genomic testing, storage of the sample and communication of the data is only valid with 'full understanding'[††]. This would mean that the benefits of genomics could not be realised. It would also mean the imposition of a highly paternalistic approach to consent in which patients were not allowed to come to the conclusion – in the real world, against a background of significant uncertainty, that this is something they would like to pursue.

All of this suggests a need not only for the development of new evidence-based approaches to best practice in consent but also a clearer statement of the complementary roles of consent and of other protections. It is our view that an important question should be what protections and controls need to be in place such that when people do give their valid consent – inevitably on the basis of a degree of uncertainty and open-endedness - they are not exploited, discriminated against or unfairly treated.

It is also going to be important to consider what function consent is required to play, given the familial aspects of some genetic findings. Unlike an operation or procedure where there is a physical intrusion for which the operator requires consent, consent to genomic testing may perhaps be better seen as being explicit about entering a relationship with agreed ground rules about mutual responsibilities and rights. These mutual responsibilities and rights extend to the individual, their relatives who may be unwitting 'stakeholders' in the outcome of genomic testing and to the population as a whole who stand to benefit from largescale geno[me]type/ pheno[me]type correlations.

Finally, it may be that the challenges presented by the uncertainties and open-endedness of genomic medicine require a rethinking of some aspects of the role of non-directedness in the doctor-patient relationship. It may, for example, turn out to be the case that patients are more likely to be content with the decisions they have made where the process of decision making involves a greater degree of clinician involvement/deliberation than is the case elsewhere. So whilst the genome 'sequencing' is a technical step, that can be undertaken with minimal medical intervention (spitting into a pot and sending it through the post) the complexity of the possible outcomes of analysis may require an extended clinical interaction to ensure that different types of outcomes (clear/ uncertain, mild/ severe, current or future) are assimilated in the consent process in a more clinician directed way than would normally be expected. Might this, perhaps, be a place where the evidence reveals joint decision-making comes to be seen as 'better' (by patients) than one that is more 'non-directive'?

[††] This is of course also true for existing care outside of genomics. The difficulties in promoting 'full understanding' are rarely acknowledged in policy documents, but have been examined by N. Manson and O. O'Neill 'Rethinking Informed Consent in Bioethics' (2007).

3.3 Confidentiality and the availability of the best care for patients and families

High standards of confidentiality and the securing of potentially sensitive health care information are going to be at the heart of good genomic medicine practice. However, there are at least two important ways in which patients and the public are likely to be supportive of new practices in the use of patient information, each of which suggests the need for new thinking on the appropriate uses of health-related information and their limits:

The first of these relates to the potential benefits for individual patients of having at least some of their clinical data analysed together with genetic findings from others in population-scale (secure) data bases so that evidence can be acquired on the nature of the link between genotype and phenotype. This might improve their own care, now, in the future and improve the care of others.

Case study 2

A mutation in the BRCA1 gene was thought for many years to confer a high risk of breast and ovarian cancer. More recently, evidence suggests that it is a benign variant and that the surveillance and interventions offered to those with the variant were therefore wrongly directed. This evidence has only come to light through international efforts and database linkages of family history details and segregation of the variant with disease in families. Although those with the variant have previously been advised they are at high risk, they, their relatives and future individuals can now receive more up to date clinical advice.

Source See for example, E.T. Rosenthal, et al, 'Exceptions to the rule: Case studies in the prediction of pathogenicity for genetic variants in hereditary cancer genes', Clinical Genetics 2015

The second situation when the sharing of patient information in new ways might be expected to command support is to distinguish individual clinical information about a disease or condition from the inherited mutation(s) that led to the clinical findings. Whilst professional guidelines such as those from the GMC[9] specifically list genetic information as one possible reason for breaching individual confidence (if doing so would protect people from serious harm), it might in certain cases also be possible to share relevant information without any breach of confidence. In practice, it is not always necessary to disclose to relatives (existing or future ones) that a specific patient has been diagnosed with, say, inherited breast cancer. They can be informed that in a particular family there is an inherited tendency to cancer that could be usefully tested for in family members who are worried about their risk. On some occasions, the second approach might raise concerns that discussing the test would identify a particular family member and constitute a breach of her or his confidentiality but this need not always, or even often, be the case – particularly in large or multi-generational families where others have the familial disease in question. For example, a woman who is concerned about her family history might simply be offered an appropriate genetic test without this raising any confidentiality concerns about the individual in whom the familial cause was first identified. We would argue that where this is the case, a social contract that would allow such information to be available for use by clinicians in the appropriate care of family members (for example, testing for the particular familial mutation to determine if extra surveillance is warranted) would be publicly acceptable.[‡‡] Although this involves the use of information beyond the individual care of the person, this approach would only use familial information and not disclose any individual details thus maintaining confidentiality.

[‡‡] See for example, Confidentiality and sharing genetic information with relatives. Lucassen A, Parker M. Lancet 2010 May 1;375(9725):1507-9. Genetic information: a joint account? Parker M, Lucassen AM.BMJ. 2004 Jul 17;329(7458):165-.

Case study 3

A man with a mutation in a mismatch repair gene resulting in a high lifetime chance of bowel and other cancers steadfastly refuses to inform his siblings, or allow his doctor to do so, of the risk they might be at. The health care professionals know that one sister has had bowel cancer and is therefore likely to harbour the same mutation. This sister is at increased lifetime risk of endometrial cancer and might therefore benefit from a risk reducing hysterectomy. The heritable aspect of the cancer is insufficiently common to justify testing unless there is a family connection. Unless we allow the use of the familial information, we have to choose between either testing everyone, for little clinical utility in most cases, and at a cost to the NHS, or not testing at all.

The health professionals have 3 options (1) to respect the man's wishes (2) to breach his confidence on the basis that it is justified by the opportunity to prevent harm to relatives who might unknowingly have the mutation as per GMC guidelines (3) Use NHS tracing to contact the sister's GP and tell him/her that a referral to a genomic service is recommended because she might be at increased risk. Option (3) does not need to breach the man's confidence because only information that is familial is communicated.

One way forward therefore is for the boundaries of confidentiality in genomics to be seen, at least in some situations, at a familial rather than individual level. Taking a recent court case (ABC vs St Georges[10]) as an example, it may indeed not have been good practice for clinicians to tell the daughter that her father had the genetic condition, Huntington's disease without his consent, but it might have been perfectly good practice to tell her that the facts of his case, and his family history – both which were in the public domain - indicated a potential familial risk about which she could seek independent advice and treatment. This separation of approaches to confidentiality of individual clinical information from familial genetic information has not yet gained widespread traction in practice, in part because of the limited situations to date where it was required, and in part because in some situations communication about familial information could lead to inference about individual clinical information. Evidence from qualitative research on the topic suggests that although health professionals find thinking in a familial way about genetic information difficult, many patients assume such an approach is happening and are surprised to hear that sharing and familial use are not standard practice.[11] This suggests that there may be patient and public support for an approach adapting the default position on confidentiality such that: Instead of breaching confidence only if one can prove they are preventing serious harm in specific others (current GMC guidance), the default becomes that relevant information that might prevent harm is communicated unless there are good reasons not to.

There will, inevitably be some situations of a different kind in which the question of using of properly confidential patient information in the care of family members will need to be considered. Confidentiality is an extremely important part of good medical practice. The provision of evidence based medical advice and treatment requires patients to undergo tests and to entrust confidential information to professionals. Confidentiality also shields patients from embarrassments and intrusions into their private lives. Protecting patients' confidences is as important in genomics as any other area of medicine. However, since certain genomic information may also be relevant for others, for example, biological relatives, or be dependent on data from them before it becomes information, there is a higher degree of interdependence in the generation of information than most areas of medicine. The principles of confidentiality and data protection therefore require special attention in this context, especially as the scope of genetic and genomic testing increases.

On the one hand genomic findings may convey or predict sensitive, potentially stigmatising facts, on the other hand much genomic information is common to many, and particularly common to that of biological relatives. The information generated in circumstances of confidence to one person, may allow inferences to be drawn about its significance to another, whose views may, or may not be known. Conversely, inferences about a particular genomic output may only be possible if confidential information is first obtained from others. These aspects of genomics can mean that health care professionals do not know whether they are balancing their duties of confidentiality with the rights and freedoms of others appropriately. This needs to be borne in mind when calls for better data sharing between laboratories and countries are made. Data is most useful if linked to some clinical information and those submitting the data need to be clear what is acceptable within the rules of data protection and confidentiality. This is captured in the idea of 'fair processing principles' which can be developed through the elaboration of a new social contract for genomic medicine.

Case study 4

Communication is not good between the different family members of one family at risk of sudden cardiac death through a pathogenic gene mutation that alters cardiac repolarization. Although information letters about the condition, the risks and the surveillance and treatments available have been given to the index patient in whom the mutation was found, it is clear that these have not been passed on. When another member of the family is referred for assessment of his family history of sudden cardiac death, health professionals are unsure whether utilizing the genetic result of the index patient in this assessment would breach his confidence. Whilst it would be inappropriate to reveal the clinical details of this index patient, telling the family member that there is reason to believe he might be at risk of a heritable mutation, is not.

One way in which such situations might be preempted is through the obtaining of consent at an early stage for the use of such information for the caring of family members as well as for submission to (inter)national databases for the benefit of wider family or other families. Through consent, patients can authorize the use of their confidential information for other purposes, including research and the treatment of others, and it may be appropriate to take steps to encourage patients to adopt this form of altruism or 'genetic solidarity' as a routine step in genomic medicine, or a social contract for confidentiality.[12] Whilst much clarity can be achieved by such encouragement and explicit 'up-front' statements, there will be times when consideration needs to be given to whether it might be legitimate to use the information in question without specific consent. GMC guidelines on the limits of confidentiality with respect to genetic information[9] are helpful in clarifying this possibility, but increasingly genomic testing is creating situations where the wishes of an individual are not known, and not easy to obtain, yet a result - perhaps not anticipatable at the time of testing - is relevant to others. As genomics reaches into many more areas of routine medical practice, consideration will need to be given as to whether conventional notions of a duty of confidentiality are realistic or appropriate and how genomic findings of different kinds ought to be dealt with.

3.4 The obligations of health professionals, laboratory staff and researchers

Genomic medicine will have implications for what it means to be a good and ethical health professional in the NHS. It is likely that careful thought is going to be required on the question of how are we understand the obligations of doctors to their patients in this new world e.g. in the context of greater uncertainty, evolving knowledge and ongoing feedback, and a greater concern for the care of families. The clinical use of genomics is likely to take place in the context of a greater degree of interdependence between clinical practice and research, the collection and use of large datasets, and a much greater emphasis on ways of improving understanding and interpretation through the on-going refreshing of datasets with new data in real time. It will be increasingly difficult to argue that research and clinical ethics involve separate sets of principles, a distinction on which many current professional guidelines are based. Originally, the Declaration of Helsinki denied the acceptability of 'therapeutic' or 'clinical' research by professionals on their patients unless direct benefit to them was expected. Since, 2000 there has been a slight relaxation, with additional safeguards applied in the category of research combined with care, but maintaining a clear separation between the two.[13] Article 14 now recognises that research may be permissible provided that harm is avoided. It states that 'Physicians who combine medical research with medical care should involve their patients in research only to the extent that this is justified by its potential preventive, diagnostic or therapeutic value and if the physician has good reason to believe that participation in the research study will not adversely affect the health of the patients who serve as research subjects'.[14] However, rapidly developing medical practices, and particularly genomics, will force us to revisit these positions since these areas involve research and care being alloyed together so that each activity is dependent on the other.

This also works the other way around. It means that researchers and data managers may increasingly come to be seen to have responsibilities that cannot easily or completely be divorced from clinical care. As the clinical predictions from genomics become clearer, it will become the norm rather than the exception that research will produce information that has potential clinical significance. It will be important to clarify when researchers are expected to liaise with clinicians, and which clinicians they should contact. Novel ways of linking research laboratories with clinical teams and quality assurance approved laboratories for validation will be important for effective interaction between research findings and clinical practice.

In relation to clinical care, the level of detail of stratified and personalised medicine means that the resources to support decision-making and underpin evidence-based care will be different. Randomised controlled trials the gold standard evidence generating tool in many areas of medicine, will be more difficult to employ. Evidence about a particular genomic finding will require large scale (often across national boundaries) phenotype-genotype correlation that take ancestral genomic background and environmental factors into account. Nuanced yet uncertain diagnoses or predictions will remain the norm in genomics for the bulk of clinical practice over the next few years. Yet this is in the face of a discourse about genomics that often mixes appropriate claims of technological accuracy with claims about their clinical predictions which remain far less deterministic than commonly perceived. This will require judgment to play a greater role, something that is consistent with the origins of the evidence based medicine (EBM) movement but has become less prominent.[15] This recognition of the importance of the subjective views of patients on what is material to their decisions in a 2015 UK Supreme Court decision in Montgomery vs Lanarkshire (2015) will need to be examined in the wake of genomic medicine's possibilities including its uncertain predictions.[16]

3.5 System responsibilities

These issues cannot be resolved by individual clinicians, but need to be addressed collectively through the appropriate design of health systems.

Case study 5

A large study of whole genome sequences reveals a series of 'pathogenic' mutations in a sample of well individuals who took part in sequencing in order to make the search for a diagnosis in a relative more effective. Questions are raised about whose responsibility it is to communicate this information, how the downstream implications for the NHS in terms of clinical follow up, surveillance and treatment are managed, and how the evolving evidence about the predictive value of these mutations in terms of disease can best be communicated and by whom.

Decisions on whether such contact should be made require complex analysis and awareness of the ethics of risk communication. It seems unreasonable to place that burden on primary care alone. Provision needs to be made by the health service for analytical capacity and ethics support to advise researchers on when contact might be appropriate and clinicians on the significance of the information and how best to communicate it without causing confusion. It will also be important to establish when contact should be expected, and the NHS could be held liable for failing to seek to achieve it. For example, what level of risk, certainty or medical interventions would need to be available for lack of contact to be negligent and who would judge? When might contact be a discretionary matter and how could this respect potential rights not to know? It would be inappropriate to create legal obligations beyond those of fair and non-discriminatory processes. If we are to achieve a consistent service, these issues will need to be tackled at a health system level not on individual clinical or research responsibilities. In any event, NHS clinical services and research studies are not currently resourced to be able to take on this role.

Further, if we recognise both the importance of consent and its limitations as a guarantor of ethical practice, and that genomic medicine challenges conventional approaches to confidentiality in significant ways, then a key question becomes what complementary protections and controls need to be in place such that when people do give their valid (but inevitably imperfect) consent, they are not exploited, discriminated against, unfairly treated and have their privacy unacceptably encroached upon. If, furthermore, we acknowledge that even where health professionals and laboratory staff perform their duties to the best of their ability there may be structural or institutional factors affecting the care of patients and the protection of their interests, this suggests a need to think carefully about the responsibilities of systems. That is, the responsibilities beyond those of individual health professionals, research groups or hospitals.

Such responsibilities are likely to include questions relating to appropriate and accountable governance, oversight, data-security and where required, regulation. The ability of the NHS to show that it can be trusted on these issues will be an important foundation for the reasonableness of the new social contract that we propose. It will need to create systems that ensure widespread sharing of linked genomic data that helps interpret the patient's specific information. This will necessarily originally be derived from individuals, but will need to be (a) available in a way that obscures identities where possible, (b) be subject to information governance safeguards. This is unlikely to be achievable in a fragmented provider system without national co-ordination. It may also require specific legislative authority.

However, it will also be important to establish clear responsibilities for ensuring equitable, access to the benefits of genomic medicine In clinical guidelines and national commissioning standards. It will not be reasonable to expect people to accept the new social contract unless the health service accepts responsibilities for ensuring that the benefits will be available to all.

Finally, protections against unfair discrimination will need to be enhanced. The Human Genetics Commission recommended on a number of occasions that specific provision providing protection against discrimination on the basis of genetic characteristics should be introduced. This would play an important role in making the new social contract a reasonable one to propose.

4. Conclusion

In this chapter we have outlined some of the ethical challenges presented by the greater use of genomics in the NHS. We began by noting that the realisation of the important benefits of developments in genomics for patients is going to require significant changes in the ways in which health care is understood, organised and delivered. We picked out three particular aspects as having particular significance: the greater integration and complementarity between research and clinical practice; the central importance of data collection and analysis; and, the increasing role of uncertainty and open-endedness in genomic medicine. Against this backdrop, we have argued that the sustainable achievement of the benefits of genomics requires a broad renegotiation of the social contract for medical research and medical practice in the NHS. We picked out four areas in which this is likely to be particularly important: (1) consent; (2) confidentiality and the care of family members; (3) the duties and obligations of health professionals including laboratory staff; and, (4) system responsibilities and governance. There are a number of other important issues we could have discussed. Perhaps the most important of these concern the use of health data for research, and the potential importance of commercial companies in such research. Beyond the immediate clinical uses of data, the quality of care and the quality of knowledge about disease and treatments will be greatly improved by encouraging research activity on the data. Much progress is going to require the involvement of commercial and technology partners. If it is accepted that such activities are in the public interest and are a necessary condition for the NHS to meet its commitment to improvements in the diagnoses and treatments available to patients, careful thought is going to need to be given to the question of how this can be achieved in a way that commands public trust and contributes to, rather than undermine, higher standards of equitably available health care. Despite their importance, we have not discussed these issues at great length in this chapter because they are already the subject of a great deal of academic and policy debate.

The working out and agreement of the terms of any such contract requires the active involvement of many stakeholders including patients, health professionals, researchers, policy-makers, and wider society. This suggests a key role for public engagement and involvement. Evidence suggests that members of the public are aware that genomics has the potential for great benefit but that its use presents a number of risks and challenges. Whilst the risks cannot be entirely eradicated it is reasonable to expect that given certain safeguards and adequate oversight there will be strong public support for the development of a health service with dynamic genomics and the effective use of health data at its heart.

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