Communication of Genetic Information within Families: The Case for Familial Comity

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Abstract

Advances in genetic technologies raise a multitude of ethical issues, some of which give rise to novel dilemmas for medical practice. One of the most controversial problems arising in clinical genetics is that of confidentiality and who may disclose genetic health information. This paper considers the question of when it is appropriate for health professionals to disclose clinically significant genetic information without patient consent. Existing ethical principles offer little guidance in relation to this issue. We build on suggestions that genetic information may be viewed as collective or shared information, and we introduce the concept of 'familial comity' as a fresh way to consider the issues.

Keywords: Genetics, Ethics, clinical, Confidentiality, Family, Genetic privacy

Introduction

Over the last half-century, genetic technologies have played an increasingly prominent role in the provision of health-care. From the introduction of newborn screening programmes in the 1960s to the sophisticated molecular genetic diagnosis and predictive testing available today, genetics may be characterised by its potential to offer otherwise unattainable medical opportunities to at-risk individuals and couples.

As well as attracting significant clinical interest, this now-entrenched area of medicine has raised many social and ethical questions. Debates have arisen about the legitimacy of pre-natal diagnosis, predictive testing in young people and the delivery of services within multi-ethnic populations. The debates have involved professionals, policy-makers and academics in a range of different disciplines. Although this literature addresses numerous ethical problems arising in genetic medicine, much discussion is issue-specific. Few contributions have proposed ethical constructs that are different from – and complementary to – those typically employed in other areas of clinical practice. In this paper, we use the particular ethical problem of non-disclosure of genetic information between family members to consider the merits of a new and complementary ethical concept for genetic

medicine: familial comity. We build on previous suggestions that genetic information may be viewed as collective or shared information, and we demonstrate how the concept may have broader appeal.

After presenting two case-studies illustrating the problem of non-disclosure in clinical practice, we briefly review some of the laws and guidelines applicable to this issue. We then discuss the need for, and justification of, a consideration such as familial comity. We argue that family comity offers a novel and intuitive way of framing the place of genetic information within families. The concept might be articulated as one of the 'tenets' of practice in clinical genetics, that is, an over-arching concept that enables one to reason through micro-issues that arise in individual clinical consultations. We conclude by responding to the criticism that family comity exemplifies "genetic exceptionalism," and by suggesting some other problems in genetic medicine to which the concept may be usefully applied.

An Array of Challenges

Clinical genetics raises many issues that prompt a close examination of the ethical principles that inform practice in this area of medicine. In addition to the issues raised above, ethical and policy challenges have also arisen in relation to the storage of population-specific genetic information in biobanks, and whether it is ever acceptable to report genetic test results under a research protocol [8]. One property of genetic information underpinning the emergence of concerns such as these is its ability to yield predictive health information not only about an individual but also his or her genetic relatives. Whether properties such as this render genetic information exceptional is contested, and we discuss this further below.

One of the most controversial problems arising in clinical genetics is that of confidentiality and who may disclose genetic health information. Specifically, one question concerns under what circumstances, if any, it is appropriate for health professionals to disclose clinically significant genetic information without patient consent. Consider the following actual cases, which were reported in the popular press:

A dying Melbourne man in his 20s could not be warned that he might be carrying a genetic mutation that causes bowel cancer. A second man from another Melbourne family faces a 50–50 prospect of survival after developing the same disease. The second man's parents knew of the family's history of the disease but did not tell their son. Both men were not told of their disease risk [by their respective health professionals] because under Australian laws only relatives have the right to pass on sensitive family information. In both cases, the cancers could have been cured if diagnosed early enough [23].

[Caroline, 28,] learnt last February that the lump she found in her right breast was cancerous.. Upsetting as the news was for someone so young, it was not as shocking as what happened next. She discovered that what had caused her cancer had been known to doctors and other family members for some years. But no one had thought to tell her. Had...she had access to her family's medical history, there is every reason to think she would have been diagnosed earlier. The missing detail from Caroline's medical history was only revealed after her consultant recommended she should have a genetic test. It was while waiting for the results that she learnt that a similar test had been carried out on another branch of the family several years earlier, which had established that her grandmother carried a faulty gene [17].

In both of these cases, those affected by non-disclosure have inherited genetic mutations which predispose them to cancer. The men in the first scenario have inherited a mutation in the gene responsible for familial adenomatous polyposis (FAP), which predisposes individuals to bowel cancer. If the genetic mutation is known, appropriate surveillance or clinical intervention will increase the chances of early detection and treatment (or avoidance) of the cancer [11, 21, 27]. Withholding this information may be detrimental to a genetic relative's health because if they do carry the mutation and do not undergo regular surveillance or elect to have a colectomy, they will almost certainly develop bowel cancer.

In the second scenario, Caroline has inherited a mutation in a breast cancer gene (either BRCA1 or BRCA2). Carriers of mutations in these genes have an up to 80% risk of developing breast cancer by age 80 [10]. Whilst surveillance is not as effective as for FAP, Caroline could have reduced her risk of developing breast cancer through regular mammography, clinical breast examination or prophylactic mastectomy.

Non-disclosure between family members seldom occurs. Situations in which a health professional contemplates a breach of confidentiality are even rarer (they are often due to practical constraints, e.g., lack of contact details for relatives). This issue is nevertheless ethically significant [6]. Non-disclosure challenges the central ethical principles guiding practice in clinical genetics and prompts a re-evaluation of the interface between clinical practice and dominant theories of medical ethics as they apply to genetics. As we will discuss below, scenarios like these might be avoided if the ethical principle of family comity were observed in the practice of genetic medicine.

Confidentiality: Regulation and Clinical Practice

Confidentiality is a cornerstone of all medical practice. Respecting confidentiality promotes patient autonomy, upholds an implied promise and demonstrates virtuous professional conduct. Breaches of confidentiality may give rise to serious consequences, including a loss of trust in the professional–patient relationship, and potential legal liability.

In most jurisdictions, clinical best practice guidelines and legislation designed to protect privacy or data stipulate that when genetic information is to be shared with family members, the most appropriate person to do the sharing is the individual who has undergone the genetic test [20]. But as the above case-studies illustrate, the information is not always passed on. Communication of genetic information is a complex issue and appears to be mediated by a variety of factors including the following: family members may have lost touch with each other; they may not be communicating with each other because of a dispute; or there may be other complications, such as a suspicion of misattributed paternity [19]. An individual's duty of disclosure may conflict with a desire to protect kin from distress, and from reluctance to convey bad news.

Strategies for managing these issues vary throughout clinical practice but have been found to include the following: placing the responsibility of disclosure onto a third party; delaying disclosure temporarily or permanently; waiting until 'the time is right,' and disclosing only to selected family members [6, 13, 28]. Some have argued that given the potential difficulties and inevitable inefficiency associated with disclosure by patients, health professionals should be free to convey critically important health information to the patient's relatives without the patient's consent [7]. Recent literature in clinical genetics has questioned the current paradigm of individual control over

genetic information [4, 15]. Yet many instances of non-disclosure may be due simply to a break in the chain of communication rather than to an explicit refusal to pass on genetic information.

The Australian Law Reform Commission Report (ALRC) on the protection of genetic information recommends liberalisation of the current restrictive approach. Recommendation 21-1 in the ALRC Report Essentially Yours: The Protection of Human Genetic Information in Australia states that amendments to the Privacy Act 1988 should be made to permit health disclosure of genetic information by professionals without consent in circumstances where there is a threat to life or health [2]. The Australian National Health Privacy Code is also currently being drafted. In its present form, the Code allows for disclosure of an individual's health information which is or could be predictive at any time of the health of another individual. It proposes that disclosure should occur only in situations where an organisation reasonably believes that this is necessary to lessen or prevent a serious threat to that other individual's health or life, and where all reasonable steps to obtain consent of the first individual have been taken. Further, the individual to whom the information directly pertains will need to be given adequate warning that such a disclosure will take place [2].

The American Society for Human Genetics has published comprehensive guidance on confidentiality in clinical genetics practice [1]. It recommends a permissive and discretionary approach to disclosure, but stresses that it should be exercised only in exceptional circumstances. Certain conditions should be met before disclosure, including the following: there should be a serious, imminent, foreseeable and serious harm to an identifiable relative, and the disease must be preventable, treatable or able to be monitored.

In the United Kingdom, the Joint Committee on Medical Genetics (a committee of the British Society of Human Genetics and the Royal Colleges of Physicians and Pathologists) is soon to publish a report examining issues of confidentiality in clinical genetics. Until then, health professionals working in genetics must conform to the same guidelines as other health professionals; namely the Data Protection Act (1998) and confidentiality guidelines published by the General Medical Council [7]. Anecdotally, the experience of one of this paper's authors (AN) suggests that clinicians are in practice reticent to breach confidentiality in the absence of clear legal precedent.

The Need for Familial Comity

If the recommendations of the draft Australian National Health Privacy Code are adopted, health professionals in Australian clinical genetics services will have a greater range of options. As well as facilitating their consultands' autonomous decision to share information with other family members, they will gain the discretion to disclose information to the patient's relatives if the patient does not do so.

This development may not be greeted with universal enthusiasm, given the increase in responsibility it occasions. Encouraging autonomous decision-making by consultands occupies a central role in doctor–patient relationships and underpins confidentiality; it is a necessary foundation for open dialogue between clinician and patient [12]. In genetics this is evidenced by the centrality of non-directive counselling in which the genetic counsellor's role is to provide information and support that will allow the patient to make decisions that they feel are right for themselves and their family

[29]. At the same time, it is commonly recognised in clinical genetics that strict adherence to the principle of respect for patient autonomy may not always be appropriate.

In response to these tensions, clinical genetics should balance an individualistic conception of autonomy with a more relational and communitarian one that encourages people to take account of their responsibilities to others [3]. This relational approach already informs practice to the extent that clinicians may encourage disclosure, and in response, many patients choose to share important genetic information with family members out of a sense of familial responsibility [13].

A related, broader view is gaining in popularity, namely, that information gained through genetic testing should be seen to belong to a family rather than to an individual alone [27]. Parker and Lucassen suggest that genetic information may be viewed either as 'personal' information or information which belongs to a 'joint account' [24]. A 'joint account' assumes that information should be available to all genetic relatives unless there is good reason assume otherwise. This places the ethical principles of justice and reciprocity at the heart of decision making [24]. Managing a 'joint account' in clinical practice may be difficult at first, but this is not sufficient reason to dismiss the idea.

If genetic information is to be viewed as common or shared, then it may be useful to articulate a new concept to capture this notion. In 2002, The Human Genetics Commission in the United Kingdom proposed a concept of "genetic solidarity and altruism" [15]. This recognises that genetic information binds people together, and that this common bond requires altruistic behaviour on the part of each individual to ensure that common good is promoted.

Taking solidarity and altruism further, we propose the supplementary concept of familial comity. 'Comity' is considerate behaviour towards others. This principle is intended to reflect the notion of relational autonomy (i.e., the notion that an individual's personal autonomy should be considered in relation to their responsibilities to others [3]), and the social responsibility that should be attached to genetic information, which is hereditary in nature. Comity provides a useful counterbalance to the principle of autonomy, and supports a 'joint account' approach to the practice of clinical genetics [24].

Whilst clinicians should respect individual patient autonomy to the greatest extent possible, the principle of familial comity demands that the implications of genetic information for genetic relatives should also be routinely considered. In circumstances such as that illustrated by the cases above, individual patients should still be encouraged to disclose information where this is warranted. If they do not do so, the principle of familial comity could be used to justify further disclosure of that information by health professionals. Studies already reveal that whilst genetic counsellors currently uphold patient autonomy above other ethical obligations to genetic relatives [9], they will often take further steps to ensure that significant information is disclosed in situations where they believe this may not happen [18]. Professional codes also encourage clinicians to strike a balance between upholding confidentiality and addressing the needs of genetic relatives [16]. Practicalities may interfere with the observance of this principle. The clinical team may simply have no means of identifying the proband's relatives, for example.

Familial comity does not merely state that people should be considerate to one another. The concept is intended to provide an over-arching tenet of clinical genetics one which can be useful

over a number of domains. It offers room for new approaches to familiar problems, without requiring weighing of principles, and it applies to both professionals and families. It will be easy to translate this principle into everyday clinical practice, and it will shift some of the ethical debates in genetics beyond classical principle-driven analyses.

Familial Comity in Practice

Confidentiality is not the only issue in clinical practice to which the principle of familial comity may be usefully applied. It may also inform decision making in controversial cases of genetic testing of younger children. Recommendations generally restrict genetic testing in younger children to circumstances where there is direct medical benefit to the child [14]. Purely predictive genetic testing (such as testing for Becker Muscular Dystrophy in young boys) and carrier testing (such as fragile-X carrier screening in young girls) is therefore generally discouraged [5]. Some have argued, however, that there may be other benefits to the individual and family which make some such testing desirable, including reducing what may sometimes be a virtually incapacitating state of uncertainty for the child and parents, or preparing the child for his or her future [26]. Again this scenario presents challenges to an individual's autonomy, and issues relating to the psychological and other kinds of harm that may arise from this testing are paramount. Nevertheless, if we see the issue in terms of striking a careful balance between respecting individual autonomy and acting with familial comity in mind, this goes some way towards addressing the ethical conflict arising out of this issue.

Does Familial Comity Represent Unwarranted Exceptionalism?

Discussions of whether the features of genetic information and technologies are unique enough to warrant a separate ethical analysis has been ongoing for as long as people have been publishing on the implications of genetic technology [25]. Genetic information has several interesting attributes: it uniquely identifies most people; it is ubiquitous; it can be obtained from a very small biological sample; it can be predictive of future health, and it will have implications for genetically related family members.

These attributes are not exclusive to genetics. Contact-tracing of partners in cases where a patient is found to be infected with HIV (or another sexually transmitted disease) presents similar confidentiality problems to the case studies we have discussed. Similar issue also arise in cases where people are informed that a particular individual living in close proximity is infected with tuberculosis.

Notwithstanding these similarities, genetic information is unique in that it combines all of the attributes mentioned above [22]; other kinds of health information do not possess this combination of properties. Furthermore, it might be argued that the principle of familial comity might be usefully applied in the context of communicable disease control, as well as genetic information.

Conclusion

The practice of genetics presents challenges to the centrality of patient autonomy. This means that we need to consider alternative concepts to accommodate the ethical issues brought about by new

developments in genetic technologies. The principle of familial comity complements existing ethical principles, and provides a way to balance the best interests of the individual with the interests of their genetic relatives.

Genetics is an area of medicine where new developments frequently arise. This forces us to constantly re-evaluate what should be considered ethical practice. Ongoing debate about ethical principles will help guide practice in this new area of clinical medicine.

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