GENETICS

The routinisation of genomics and genetics: implications for ethical practices

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Among bioethicists and members of the public, genetics is often regarded as unique in its ethical challenges. As medical researchers and clinicians increasingly combine genetic information with a range of non-genetic information in the study and clinical management of patients with common diseases, the unique ethical challenges attributed to genetics must be re-examined. A process of genetic routinisation that will have implications for research and clinical ethics, as well as for public conceptions of genetic information, is constituted by the emergence of new forms of genetic medicine, in which genetic information is interpreted in a multifactorial frame of reference. Although the integration of genetics in medical research and treatment may be a helpful corrective to the mistaken assumptions of genetic essentialism or determinism, the routinisation of genetics may have unintended consequences for the protection of genetic information, perceptions of non-genetic information and the loss of genetic research as a laboratory for exploring issues in research and clinical ethics. Consequently, new ethical challenges are presented by the increasing routinisation of genetic information in both biomedical and public spheres.

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Fifteen years after the inauguration of the Human Genome Project, genetic medicine has evidenced neither the great benefits nor the great harms predicted. The sequencing of the human genome, once thought to be the key to unlocking the discovery of common genetic contributors to cancer, heart disease, diabetes and other complex diseases,[1 2] has turned out to be only a first step along a much longer path. Only a handful of common susceptibility alleles for common diseases have been confirmed to date. This is probably because those that have not yet been identified have modest or weak effects and contribute to disease through interaction with other alleles and environmental factors, the environmental factors often conferring the greater relative risk.[3] Thus, clearly, traversing the path to improved understanding of inherited contributions to complex disease will require other, non-genetic kinds of information.[4 5] Genetic information will likely advance clinical medicine, but only within a multifactorial framework for the investigation and clinical management of common disease (or, more broadly, of complex traits).

Within this multifactorial frame of reference, the generation of genetic data will continue to offer potential benefits and entail some risks. Increasingly, however, these benefits and risks will be set in a broader context in which genetic information is one among a range of different kinds of information related to health. Consequently, genetic information may no longer be treated as exceptional in comparison to other kinds of biomedical information. This shift from being regarded as unique and exceptional to being regarded as an ordinary aspect of routine medical research and care might best be described as the routinisation of genetics. We believe that in much the same way as the conceptual shift that continues to take place around HIV infection—in which HIV or AIDS is viewed increasingly as non-exceptional in the provision of medical services—radically changed the moral landscape of HIV or AIDS research and care,[6] the routinisation of genetics will transform a number of persistent ethical challenges associated with the production of genetic information. Thus, as the routinisation of genetics begins, it is important to examine both the continuing need for special protections for genetic information and the potential lessons to be learnt from the era of genetic exceptionalism.

The potential effects of the routinisation of genetic information on ethical protections for human subjects and scholarship in bioethics have not been examined to date. At the same time, routinisation has been ongoing among many segments of the public, as genetic information often has been interpreted in the context of pre-existing cultural frameworks and social issues rather than as constituting a new or separate topic of discourse. We examine how the intersection of the emerging biomedical routinisation of genetics and the pluralistic public interpretations will shape future challenges in research and clinical ethics.

''GENETHIC'' EXCEPTIONALISM

The development of special ethical protections and clinical practices for genetic research and diagnostic technologies was prompted by an assumption that the production of genetic information was of potentially greater clinical importance than other kinds of medical information. Fuelled by early success in identifying specific alleles associated with markedly increased risk of diseases such as breast cancer and Alzheimer's disease, many bioethicists concluded that researchers would soon discover

Abbreviation: IRB, institutional review board

alleles that accurately predict the likelihood of disease, and that many such alleles would be discovered after the sequencing of the human genome.[7 8]

The special ethical status afforded to genetic information is evident in both research protections and clinical practices. With regard to research protections, when a biomedical study includes genetic analyses, the genetic components of the study typically are the primary frames of reference for assessing ethical challenges. For example, special regulations have been proposed to protect genetic information generated in research[9] and additional standards have been developed for ethical review of the protection of subjects in research studies requiring individual genetic information.[10] In the US, the Office of Human Research Protections suggests that institutional review boards (IRBs) regard genetic information as entailing potentially greater social and psychological risks than other kinds of biomedical information, such that genetics studies ''warrant careful IRB review and discussion'', under the assumption that the risks they pose are different from those of other kinds of biomedical studies.[11] In addition, many IRB and research ethics committees mandate specific language for informed consent when DNA samples need to be collected or genetic information produced. High-profile genetics studies, such as the International HapMap Project, require extensive community consultations before samples are requested from people and follow-up discussions with members of those communities after results are generated.[12]

With regard to clinical services, genetic tests are often accompanied by pretest and post-test counselling. Such counselling is often non-directive, with special emphasis on the meaning of the genetic test within the broader spectrum of values and interests that are relevant to patients and their families.[13] Unlike most clinical diagnostic services carried out by medical professionals, genetic tests are typically offered to patients, not recommended. Even in circumstances where the potential for clinical benefit is clear, ethical controversies persist on the legitimacy of genetic testing, as evident, in recent debates about the testing of adolescents for genetic conditions and the expansion of mandatory newborn screening programmes.

As genetic information is interpreted in the context of other kinds of biomedical information, these special ethical protections and clinical practices will probably diminish, both because the medical assumptions that gave rise to those protections and practices are increasingly viewed as problematic and because of the higher costs associated with their maintenance. This is not to say that concerns about privacy and medical confidentiality will diminish—indeed, these may increase as more health-related databases become available and public awareness of these resources increases—but genetic information will no longer be treated as different from other kinds of medical information (with the exception of highly penetrant mendelian and familial contributors to disease, for which genetic information alone can be predictive of substantially higher individual risk).

Although reductions in genetic determinism and exceptionalism may be viewed as positive developments to the extent that these have led scientists and bioethicists to overestimate the promises and perils of genetic research and medicine, we may take the opportunity of integrating genetic information with other kinds of information to learn some important lessons from the period of genetic exceptionalism. One such lesson is that it is possible to inform study participants and patients about highly individualistic psychosocial risks associated with complex diagnostic procedures and participation in biomedical research. Another is that innovative practices can help place patients and potential research subjects in a position where they can appreciate the meaning of uncertain, ambiguous or complex test results—for example, through expanded informed consent, pretest and post-test counselling and other communication strategies.[14]

In addition to these lessons, emphasis on ethical challenges in genetic research and medicine has also provided opportunities for broader moral reflection on bioethical issues.[15] For example, genetic information has been a useful context in which to examine the interests of third-party relatives in patient diagnoses and the interests of socially identifiable groups in research studies.[16] Despite third-party risks and group interests not being unique to genetic information, the special emphasis on genetic information and unique funding opportunities to study various ethical aspects of genetic research have enhanced opportunities to explore these topics in a sustained way. Although this special bioethical focus on genetic information has, perhaps, inadvertently contributed to increased ''genethic'' exceptionalism, the routinisation of genetic information may reduce opportunities for bioethicists and others to use the special case of genetics to highlight and examine other moral issues in biomedical research and clinical care.

PUBLIC FRAMES OF REFERENCE

Although bioethicists and other health professionals have tended to treat genetic information as a special case in research and clinical ethics, in many ways members of the public have taken the opposite strategy by incorporating genetics into existing frames of reference. Although it has been assumed by some that the general public interprets genetics primarily in a deterministic, exceptionalist frame, empirical research has shown that public conceptions of genetic information are not monolithic.[17] Indeed, only limited segments of the public privilege genetic information in how they perceive common disease, and those perceptions tend to be tied to pre-existing cultural world views.[18] Thus, in a sense, members of the public have routinised genetic information already, albeit in diverse ways. By interpreting genetic information in their own cultural and social contexts, debates on genetics have highlighted issues important to many members of the public, such as concerns about the increasing costs of care and access to advanced medical services.

In our studies on cultural perceptions of genetic research, members of African-American communities in which we have conducted interviews, focus groups and public meetings have consistently questioned the value of investing public funds in genetic research when their everyday experiences of common diseases such as diabetes, heart disease and cancer suggest more immediate behavioural and ambient causes that can be remediated with less expensive, low-tech interventions. Similar concerns have been found in other contexts.[19 20] In historically underserved communities, genetic research and genetic medicine have served as high-profile examples of biomedical research, but the most pervasive concern is not that genetics is different but that it is more of the same: the perpetuation of research with little immediate community relevance, racialised science and medicine, and yet another set of clinical services to which economically disadvantaged populations will have limited access.

Members of some African-American focus groups we facilitated acknowledged that both inheritance and environment have roles in common diseases, although the role of environment is often viewed as more salient. For those who take this perspective, the routinisation of genetics will accord with their everyday sense of disease aetiology. Nonetheless, members of those groups were concerned that the use of racial and ethnic classifications in biomedical research and medicine could lead to the stereotyping of monolithic racial or ethnic identities as indicating particular genetic

frequencies and environmental exposures that gloss over diverse individual ancestries and local, community-specific variations in environmental exposures.

Members of other African-American focus groups, by contrast, articulated the view that genetics has little or no role in causing disease, but an inappropriately large part in reinforcing discrimination, by appearing to give race a biological basis in humans. For those who take this view, the biomedical routinisation of genetics will represent the continued geneticisation of race and diversion of biomedical resources from what they believe to be the root causes of disease—namely, disparities in economic and political resources that increase psychological stress, exposure to toxicants and limited access to care for many members of minority communities. Issues of community relevance and access will remain, even as biomedicine increasingly interprets genetic information within a multifactorial framework. These issues are linked to the relationship between social identity and health status, in which genetics has become a particularly potent proxy for public perceptions of differences between groups.

The concerns expressed by both those who value and those who discount the relevance of genetic information in common diseases relate primarily to the geneticisation of social identities, in which particular patterns of genetic variation are associated with specific groups of people.[21] The use of genetics to define groups is not exceptional, however; it is merely the latest instantiation of a long history of using biological characteristics to reinforce social boundaries between people. As such, many members of the majority population continue to routinise genetic and other biomedical information according to a racialised framework.

CHALLENGES

For ethicists, the routinisation of genetics raises an important challenge: Can the potential benefits associated with the routinisation of genetic information (eg, reduction in misplaced genetic determinism and exceptionalism) be realised without a corresponding loss in the special ethical protections and clinical practices developed largely because of privileging genetic information in medical research and care?

An answer to this challenge may be found in the ongoing culturally specific routinisation of genetic information taking place outside the professional precincts of biomedicine. The public interpretations have occurred as members of different communities encounter genetics in everyday life and extend their existing world views to ''make sense'' of it. This sense-making effort, like most extensions of the existing cultural logic to novel problems, is rarely explicit.[22] If we conceptualise bioethics as comprising a culturally specific community, then the problem becomes one of how to re-interpret the innovations of ''genetic ethics'' in the context of the broader discipline of bioethics in a more explicit manner that anticipates and reflects various options available. How, for example, do pretest and post-test counselling or other efforts to reduce psychosocial harms to affected people make sense with respect to non-genetic information and multifactorial frames of reference for understanding the aetiologies of complex disease? The sense that is made in the broader context will differ from the original meaning in the more specialised context of genetics. Nonetheless, the contrast with a genetic frame of reference can tell us a great deal about the new challenges presented by multifactorial and multidisciplinary research and care.

Special cases, such as the one made for genetics, often make useful contrasts for considering the more complicated matter of general cases. The era of exceptional treatment of HIV infection, for example, highlighted several ways in which standards for the protection of medical confidentiality were more problematic than widely believed.[6] By making the ethical trade-offs of regarding genetic information as either unique or a routine element of medical research and care more explicit, we have the potential for more informed self-reflection about the ethics of integrative approaches to medicine that do not privilege any one type of information above the others.

Although much attention has been paid to what makes genetic information potentially different from other kinds of information, and the implications of those differences for ethics, little consideration has been given to the ethical implications of working with multiple kinds of information. The special case of genetics can serve as a useful measure of the costs and benefits of various approaches for obtaining and communicating individual and aggregate biomedical information. Genetics can also serve as an example of the extent to which the risks and benefits of one kind of information may be overstated or interpreted in isolation from other kinds of data. Perhaps the most interesting insight from such an explicit comparison is the possibility that the ethical challenges associated with the study and care of people with rare diseases differ in some important respects from those associated with the study and care of people with common complex diseases for which multiple kinds of information are required to produce a research or clinical finding. This general line of thought preserves genetic exceptionalism for some limited purposes (including continued ethical innovation), but also uses the contrast with exceptionalism to characterise a central, relatively little-explored issue in the larger domain—that is, the problem of working with multiple kinds of information that contribute to disease and health status.

Dealing with culturally specific views on genetics presents a different challenge: How do we accommodate diverse views on the ethical challenges posed by genetics, and yet facilitate a cross-cultural consensus that rejects the use of biological information to define social identities?

This is more difficult because it goes far beyond the scope of the bioethics community itself. Perhaps the best bioethicists can do is to continue to develop review processes mandating the participation of members of communities affected by the information that biomedical activities generate as researchers and practitioners plan the studies and the provision of care from which that information flows.[23] Taking account of the differing cultural perspectives of study participants is an area in which ethics researchers in genetics have been particularly innovative, one that has interesting potential links with parallel work carried out in community-based participatory research, which has become another specialised venue for ethical innovation.[24]

Reducing risks of group stigmatisation and discrimination will require the development of broad cross-cultural consensus on the social relevance of purported associations between genetic and other biomedical information and social identities. The association of biomedical data with social identities is less risky when differences in genetic frequencies or disease incidences are viewed as consequences of the ways in which people are grouped rather than as biological confirmations of perceived differences between those groups. Although bioethicists probably cannot alter the larger socio-cultural processes by which different audiences interpret genetic and other biomedical information, they can help researchers in framing research findings in ways that draw attention to existing health disparities between groups and in designing protocols that limit opportunities for others to define those groups on the basis of perceived biological differences.

Many of the innovations in the ethics of genetic research and medicine may be interpreted as attempts to anticipate

and reduce public concerns about genetic information, particularly where those concerns reflect differing group perspectives because of the ways in which that information is routinised in existing cultural frameworks.[25] The discourse of bioethics, however, has tended to avoid engaging diverse populations in efforts to integrate alternative frameworks that members of those publics use to interpret the relevance of biomedical information in their everyday lives. That bioethical exceptionalism, however, similar to genetic exceptionalism, may be appropriate in only certain limited, special cases, such as the acute moral dilemmas that are often used as illustrations in the teaching of medical ethics. To make sense of more common encounters with biomedical practices and information, however, including finding guidance for incorporating diverse perspectives on health-related information into general standards for research and clinical ethics, we may have to routinise bioethics in the larger context of other culturally specific frameworks.

HUMAN SUBJECTS

Research described in this publication was approved by the appropriate IRB (University of Oklahoma Health Sciences Center, Oklahoma City, Oklahoma, USA).

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