

Women's Issues

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Gender and the Human Genome

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ABSTRACT

Gender issues arise in relation to the human genome across a number of dimensions: the level of attention given to the nuclear genome as opposed to the mitochondrial; the level of basic scientific research; decision-making in the clinic related to both reproductive decision-making on the one hand, and diagnostic and predictive testing on the other; and wider societal implications. Feminist bioethics offers a useful perspective for addressing these issues.

Key Words: *Biobanking; Feminist bioethics; Gender; Gene therapy; Genomics; Human genome; Nuclear and mitochondrial DNA; Nutrigenetics; Pharmacogenetics; Preimplantation genetic diagnosis*

Introduction: Gender and the Human Genome; Nuclear and Mitochondrial

The Human Genome Project aimed to produce a complete map and sequence of all genes in the human genome. Following the completion of the first draft in 2001, attention has now turned to work on the implications of the information acquired in this large-scale scientific enterprise. This includes considerations of variation in the genome, which include the differences between individuals and population groups that may affect susceptibility to disease. It is important also to consider the extent to which the issues have a significant gender dimension.

The issues of gender and the human genome arise at different levels: basic scientific research, clinical applications, and wider societal implications. The

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following questions arise at those different levels:

- (1) Is the way in which basic scientific research on the genome carried out gender-neutral?
- (2) In the stage of translation to clinical applications, what are the relevant gender implications?
- (3) In what ways, if any, are men and women differentially affected in relation to the societal implications of the implementation of genomic research?

First, however, there is a question about the “human genome”—does the notion of the “human genome” itself raise gender issues? In both the public and private human genome projects, samples were taken from a number of sources, the aim being to obtain a representative picture of the genome across humanity as a whole. In the postgenome era, however, it is variation that is increasingly the focus of attention.

It is important to note, also, that when the “human genome” is discussed it is normally the nuclear genome that is at issue, rather than the mitochondrial genome, which was sequenced much earlier, without the publicity that accompanied the Human Genome Project of the 1990s. Debates about genetic reductionism and the relationship between genes and identity also tend to be carried out in relation to the nuclear genome rather than the mitochondrial. The mitochondrial genome is particularly significant in relation to gender issues because if a woman suffers from a disorder caused by mitochondrial DNA, all her children will inherit it, as mitochondrial DNA is passed down the maternal line, while nuclear DNA is inherited from both parents. From the start, then, the mitochondrial genome gives rise to issues of gender, and provides a focal point for discussion in the context of ancestry tracing. So the fact that it is the nuclear genome that has attracted most of the publicity might itself have gender implications.

But, what is a gender issue? The issue of differences of sex and gender, and questions of essentialism—to what extent there are differences in essence, and to what extent they are a social construction—remain a subject of both controversy and political correctness (Oakley, 2005). Quite apart from the feminist research that has examined the cultural basis of gender differences, it is also clear for scientific reasons that “male” and “female” as biological categories are not clear-cut, as is shown by the cases of intersex children with associated ethical issues of gender assignment. Despite cases such as these, discussion of the ethical issues relating to gender differences has focused primarily on the situation of women, and theoretical perspectives such as feminist bioethics offer a rich resource for their discussion (Sherwin, 1992; Donchin and Purdy, 1998; Tong, 1997).

Scientific Research on the Human Genome

At the level of basic scientific research on the genome, relevant questions

include how the research agenda is set; who are the powerful figures in genomic research; does the research process privilege certain values which might be regarded as “masculine” rather than “feminine”? The ways in which the research agenda is set have recently become a significant issue in relation to the need to move public engagement “upstream,” but there are also specific issues about what research is prioritized. Much has been written about “orphan diseases” in this regard: arguably there is a general problem about vulnerable populations being under served by science, but there are gender-specific issues too, such as the genetics of female sexuality (Cherkas *et al.*, 2004). There is a real question about what kind of research is possible to attract funding for.

The dominant modes of inquiry, and the extent to which women are included in scientific research or given credit for it, have also all been subject to feminist critique. As regards modes of inquiry, the ideals of objectivity and detachment have been criticized for aiming at a value-neutrality in science, which cannot be achieved, and which represents a form of masculinization (Keller, 1985). Far better, it might be claimed, to admit to the values that are shaping science.

Where exclusion is concerned, in the context of genetics, the gender issues go back to the discovery of DNA itself, relating to the controversy over credit given to the role of Rosalind Franklin alongside Watson and Crick (Maddox, 2002), but there are broader issues about the representation of women in science in general and in particular in genomics. It is in relation to decision-making in the clinic, however, that the relevant gender issues have been most high profile.

Gender Differences and the Clinic

In the context of the clinic, the gender issues include scope for decision-making; the burden of responsibility; protection of vulnerable parties; and conditions that differentially affect the sexes. Scope for decision-making is relevant both in reproductive contexts and testing of individuals, both diagnostic and predictive.

Reproductive Decision-Making

At one time, the ethics of genetics was very much concerned with distancing clinical genetics from association with old-style eugenic programmes. The latter had potential gender dimensions in so far as eugenic programmes were thought to legitimize interventions on women in particular against their will, such as sterilization. In the light of this history, clinical genetics became firmly based around the notion of choice.

The ethical implications of genetic information for some time continued to be discussed in the clinic primarily in relation to reproductive decisions—e.g.,

whether or not to opt for a termination in the case of disorder in the foetus. Although these decisions might be discussed with a couple in the context of genetic counseling, the impact of the decision falls predominantly on the woman, who has to either carry a foetus to term or undergo a termination. For this reason too, there are strong arguments for the view that it is the woman who has the right to choose: it is not the status of the foetus that is the important issue but rather the woman's choice concerning what is happening in her own body. In many societies, also, it is she who would have the majority of the responsibility for caring for a child born with or without a disorder.

The use of genetic technology for sex selection adds an extra twist to this: in some contexts, the primary reason for which a termination might be sought is the sex of the foetus—where the overwhelming preference is for a boy child. Opinions on the acceptability of sex selection differ, some seeing it just as a dimension of reproductive choice, while others take the view that sex selection should be practiced only for the avoidance of sex-linked disease. Here the burden of responsibility on women for undergoing terminations is added to by the fact of reinforcement of prevailing patterns of value attached to the sexes in society. While in some circumstances, sex selection may be for reasons of balance within the family, in others it reflects prevailing gender roles.

The advent of preimplantation genetic diagnosis (PGD) has, on the face of it, both expanded the range of choice and made possible an earlier choice in relation to the avoidance of disorder, now in an embryo rather than a foetus. It might be argued that it is advantageous to women to be able to make selection decisions that do not involve a termination. However, in principle, it may make more available the choice of a male rather than a female embryo; and in the decision-making context, more power arguably accrues to the practitioner than to the woman involved, as the object of the decision is now located outside her body. So this technology changes the circumstances of choice, but does not necessarily enhance choice for the woman.

There are also difficult reproductive decisions to be made where a woman has a disorder based in her mitochondrial DNA because, as stated above, she will pass this on to all her children, but the severity may vary to a considerable degree. One option is to avoid reproduction altogether, where the risk is perceived to be too great; another is to decide to proceed nevertheless. Either way, there are problems with blame and responsibility for the woman. Another way forward is research into nuclear transplantation techniques, so that the couple could produce an embryo using in vitro fertilization, and then transplant the nucleus into a donor egg with different mitochondria (Bredenoord *et al.*, 2008). Then the resulting children would have nuclear DNA from the couple who want to reproduce, and mitochondrial DNA from the donor egg. This idea has attracted controversy, being described as producing children with three parents (Barnett

and McKie, 2004). A quite different longer-term option would be sex selection in favour of boys in such circumstances so that the mitochondria in question would not be passed further down the maternal line.

Diagnostic and Predictive Decision-Making

Where genetic testing, rather than reproductive decision-making, is at stake, there are issues relating both to children and to adults. Genetic testing of children is particularly problematic because of consent issues and, unless there is some intervention that is possible to alleviate the situation, a diagnosis or a prediction may not be helpful to the children themselves, although parents may want to know. There are of course some conditions that affect children of one sex, such as Duchenne muscular dystrophy (an X-linked condition, but mainly affecting males). As already suggested, however, in cases of children suffering from any disorder, there are also gender issues relating to the responsibility for care, which may fall disproportionately on one sex, depending on the social context.

Some predictive genetic tests certainly have strong gender aspects, perhaps most obviously, predictive tests for alterations in genes called *BRCA1* and *BRCA2* (short for breast cancer 1 and 2), which give rise to a significantly increased risk for breast cancer. Although men do suffer from breast cancer, the issue tends to be discussed in terms that relate almost exclusively to women, with associated issues about prophylactic mastectomy: women with a strong family history of breast cancer, who are identified as being at higher risk, sometimes opt for this intervention. The significance of this as a preventive strategy is closely connected with the symbolism of the breast, which may also be related to the cultural context (Eisinger, 2007), where the breast is regarded an important aspect of a woman's femininity and identity.

Susceptibility Testing

In discussion of genomics in the postgenome era, the issue of biobanking has moved center stage. While there are different kinds of biobanks, the essential idea involves the collection and storage of DNA samples, to undertake research into genetic factors influencing susceptibility to common diseases. For this to be possible, the information in the DNA sample needs to be correlated with personal health information of the participants. In this debate, gender questions have not been prominent—attention has focused on informed consent, storage of information, confidentiality, and privacy. The purported benefits of biobanks are that they will facilitate knowledge of variation in the genome, which underlies not only susceptibility to disease but also adverse response susceptibility to drugs and foodstuffs. To do this, of course, information about genetic factors in individuals needs to be correlated with health and lifestyle information, hence the concerns about privacy.

Following developments in this sort of population genomic research, there has been considerable interest in tests of a different sort from testing for single gene disorders—pharmacogenetics and nutrigenetics. Pharmacogenetics (pgx) aims to decrease adverse drug reactions, through the facilitation of genetically informed prescribing (Chadwick, 2007). Although traditional prescribing is done on a trial and error basis, pgx aims to reduce the number of adverse drug reactions—genetic information can be relevant not only to what drug should be taken but also to the appropriate dose. One question that arises is whether there are any gender issues arising here. In principle, there might be gender issues relating to the selection of products for research: however, arguably, a more pressing one is how pgx is implemented. On the one hand, while pgx may be portrayed as being about “personalized” medicine [referred to by Daar and Singer (2005) as the “boutique model”], on the other hand, it has been argued that rather than personalized medicine, pgx could, instead, beneficially be used to target different population groups, including currently underserved ones. An example would be drug resuscitation for populations in developing countries (Daar and Singer, 2005). The fact that a drug has been taken off the market for one population group, A, because of adverse drug reactions does not mean that it could not be used successfully in another population group, B, if it can be shown that the adverse reaction is associated with a particular genetic factor that is not present in population group B. The crucial factor here, however, is the validity of the underlying research and robust association studies. Since it has been argued that in general, evidence-based medicine has the potential to discriminate against women (Rogers, 2004), because of tendency to exclude women from clinical trials for reasons connected with pregnancy, the question arises as to whether a shift to pgx has the potential to improve this situation or not, which would depend on the extent to which gender variation was taken into account in research protocols.

Nutrigenetics (ngx) raises similar issues as it deals with individual variation in response to diet. In the context, globally, of both undernourishment on the one hand and obesity on the other, the question as to the potential contribution of ngx to these phenomena needs to be considered. Where obesity is concerned, gender issues arguably relate to the fact that, apart from health concerns, issues of body image that arise are more likely to be perceived as an issue for women than for men. Gender issues are also relevant, however, to undernourishment, for a variety of possible reasons ranging from body image, in some contexts, to women’s access to scarce resources, in others.

In relation to all these types of genetic testing of individuals, there is an issue as to whether or not they are empowering. While some see access to genetic information about oneself as a mechanism of empowerment, others have argued for a right not to know, particularly where it might open the door to misuse of the information by third parties, to one’s disadvantage (Milunsky, 2001;

Chadwick *et al.*, 1997). However, this issue of empowerment, or not, cannot be resolved outside a specific social context, because whether or not the information is empowering will depend on what is possible for individuals in general and women in particular, within that context. To have information about one's predisposition to disease, for example, may or may not be helpful depending on the available scope for action, which may include availability of treatment or changes of lifestyle that may require resources not within the reach of the individuals in question.

Gene Therapy

Where gene therapy is concerned a distinction is commonly drawn between somatic and germ-line therapy, where somatic therapy aims to treat only the body cells of an individual and germ-line therapy also treats the germ-line and thus the descendants of the individual. While it might be thought that gene therapy is gender neutral, germ-line therapy, in so far as it is associated with reproductive decision-making, may share gender relevant dimensions.

There is increasing interest, however, in using the technique of gene therapy for "enhancement," for introducing improvements "beyond therapy." While the distinction between therapy and enhancement raises a number of issues, the significant question in this context is to what extent enhancement could advance the interests of women, or not; and a pertinent issue here is how an "improvement" is defined and by whom, in a social context in which gender-based discrimination exists. The general point can easily be seen by looking at a historical nongenetic example, such as the practice of foot-binding to enhance a woman's attractiveness.

Wider Social Implications

One of the ethical concerns associated with the implementation of genetic knowledge, historically, has related to discrimination of various kinds. In the contexts both of genetic testing in the clinical setting, and in relation to population genomic research, much attention has been focused on the protection of genetic information, however difficult that might be to achieve in practice. Fears of discrimination have concentrated on the insurance industry and on employment. As has been pointed out, however, by WHO (2006) in some cultural contexts, such as developing countries, it is not insurance discrimination that is likely to be the main issue: rather it may be the stigma attaching to someone identified as a carrier of a condition, or as having a predisposition to develop a disease in later life. This, of course, has gender aspects, because such information may adversely affect the marriage prospects of women in particular. It is important therefore that discussion of the possible discriminatory uses of genetic information is not confined to concerns surrounding insurance and employment, and that it has

regard to the social circumstances in which women are living their lives.

Another social issue relates to testing for paternity. Within the clinic, testing of foetuses or children, which is intended to be disease related, may yet reveal information that the husband or partner of the mother is not in fact the father. There are clearly ethical issues about the circumstances, if any, in which such information should be disclosed and to whom—for example, should it be revealed to the mother, leaving disclosure to her partner up to her? Or does her partner have a right to know?

Questions arise here about how such questions should be addressed from a theoretical ethical point of view. From the perspective of an impersonal ethic that makes no allowance for gender issues in particular, it might be a matter of weighing up the interests of each party and trying to maximize the best outcome (Kuhse and Singer, 1998, Part III). Feminist ethics, however, would urge giving attention to who is the most vulnerable in such situations, with special reference to the fact that in many cases the most vulnerable party is likely to be the woman (see, e.g., Sherwin, 1992; Wertz and Fletcher, 1991). Rather than examining the rights of individuals who are all considered in the same way, a feminist-based ethic of care urges examination of the social context in which the issue arises, which may well be one in which the woman in question or women in general have less power than men. She may even be at risk of harm if nonpaternity is disclosed. The issue is thus relational.

Beyond the issues in specific social relationships, there is a wider question about decision-making in the area of genomics. It is a field in which much has been written about ethics and governance, and a large number of committees have been established, nationally and internationally (e.g. the Ethics Committee of the Human Genome Organization, see http://www.hugo-international.org/comm_hugoethicscommittee.php). A gender issue here is the representation of women on such committees—to what extent are they participating in decision-making on these issues (Dickenson, 2006)?

Conclusion

In all three fields, from basic scientific inquiry, through decision-making in the clinic, to wider social implications, there are real or potential gender issues. One of the most important tasks for ethics is to identify ethical issues where they exist, and in this context, in particular feminist bioethics is a useful resource (Sherwin, 1992). This is so because it directs our attention to the social context in which decisions are made, and to the ways in which women have power, or not, to make decisions about genomics-related and other matters. It focuses our minds on the vulnerable, and on the relationality of decisions, rather than the abstract and universal principles attractive to some. It asks whose voices are

heard. All these questions are of the utmost importance in the context of gender and the human genome.

Take Home Message

In the postgenome era, it is not only important to look at issues of individual and population variation, in relation to susceptibility to disease, but also at gender issues. Equitable access to such benefits as accrue from the implementation of genomic knowledge must include the gender dimension.

Conflict of Interest

None declared.

Declaration

This is my original unpublished contribution, not under consideration for publication elsewhere.

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Questions That This Paper Raises

- (1) Is the term *human genome* a useful one, given the extent of variation that exists?
- (2) To what extent, if any, does the use of genetic information in the postgenome era have the potential to be to the disadvantage of women in particular?
- (3) To what extent does social context make a difference to the issues concerning gender and genomics?
- (3) In what ways do new technologies such as PGD have the potential to be empowering for women?
- (4) What are the main ethical issues surrounding the technique of nuclear transplantation to prevent mitochondrial disease?

About the Author



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