

Balancing autonomy and responsibility: the ethics of generating and disclosing genetic information

N Hallowell, C Foster, R Eeles, A Ardern-Jones, V Murday, M Watson

See end of article for authors' affiliations

J Med Ethics 2003;29:74–83

Correspondence to:

Dr N Hallowell,
Department of
Psychological Medicine,
The Royal Marsden NHS
Trust, Downs Road, Sutton,
Surrey SM2 5PT, UK;
ninah@icr.ac.uk

Revised version received
10 August 2002

Accepted for publication
26 September 2002

Using data obtained during a retrospective interview study of 30 women who had undergone genetic testing—*BRCA1/2* mutation searching—this paper describes how women, previously diagnosed with breast/ovarian cancer, perceive their role in generating genetic information about themselves and their families. It observes that when describing their motivations for undergoing DNA testing and their experiences of disclosing genetic information within the family these women provide care based ethical justifications for their actions. Finally, it argues that generating genetic information and disclosing this information to kin raise different types of ethical issues. The implications of these findings for ethical debates about informed choice in the context of genetic testing are discussed.

Many commentators have noted that, in contrast to other types of medical information, which pertain primarily to individuals, the information derived from molecular genetic testing and/or pedigree analysis (hereafter referred to as genetic information) necessarily has implications for biologically related kin.^{1–3} It is generally accepted that the familial nature of genetic information distinguishes it from other types of medical information. As many authors have noted, this observation raises questions about the adequacy of current informed consent procedures in the context of genetic testing and the legitimacy of the disclosure/non disclosure of genetic information to biological kin.^{1–4}

THE FAMILIAL NATURE OF GENETIC INFORMATION: IMPLICATIONS FOR INFORMED CONSENT

The model of informed consent used within clinical medicine is based upon the principle of respect for autonomy.⁵ Informed consent procedures exist to protect the individual's right to self determination; more specifically, their right to make autonomous decisions about their health care—to voluntarily accept or refuse treatment and to be informed of the risks they may incur through medical procedures.

It has been noted that "... in recent years the primary justification advanced for requirements of informed consent has been the protection of autonomous choice ..."⁶ For many clinical purposes this model of consent is perceived as acceptable, or at least workable, in practice. When it comes to genetic testing, however, basing consent upon a model of autonomous decision making can be seen as a little more problematic,^{1,2} for it ignores the fact that the results of genetic tests have implications not only for the patient, but also for their biological kin. The extent to which the familial nature of genetic information creates an ethical problem for our current conceptions of informed consent is explored below.

Using data collected in a study of women who have undergone *BRCA1/2* mutation searching, this paper demonstrates that, when accounting for their role in the testing process and disclosing genetic information within the family, women draw upon discourses of self determination and responsibility. It observes that, while these women view their role in generating

genetic information for their relatives as less ethically contentious than disclosing this information to their kin, their accounts raise important questions about the adequacy of current conceptions of autonomous choice within the context of genetic testing. It argues that if we are to develop a rigorous ethical justification for informed choice in this context, then we need to acknowledge that the individuals who consent to this procedure conceive of themselves as selves in relation. As such, they regard themselves as under an obligation to undergo genetic testing to provide information which will advance other family members' autonomy, often at the expense of their own.

HEREDITARY BREAST AND OVARIAN CANCER (HBOC) AND THE ORGANISATION OF *BRCA1/2* MUTATION TESTING IN THE UK

Between 5 and 10% of cases of breast/ovarian cancer are inherited, approximately 50% of these are caused by a mutation in one of two genes, *BRCA1* and *2*.^{7–8} Mutation carriers have increased risks of developing these cancers, which may be as high as 80–85% in the case of breast cancer and 30–60% in the case of ovarian cancer.^{9–10} The medical management of high risk women includes chemoprevention, breast/ovarian screening, lifestyle modification, or prophylactic breast/ovarian surgery.^{11–12}

At the present time predictive genetic testing for HBOC is generally only available to at risk men¹³ and women in the United Kingdom once it has been established that a *BRCA1/2* mutation is present within the family. At risk family members' access to predictive testing is usually dependent, therefore, on relatives who have already had breast/ovarian cancer undergoing mutation searching, because it is only after it is confirmed that an affected relative carries a *BRCA1/2* mutation, that predictive testing for family specific mutations can be offered to at risk relatives. Thus, individuals who have previously been diagnosed with cancer assume a pivotal role in generating genetic information for their biological kin. They can be seen as providing a gateway to genetic testing for others, for without their consent to this procedure, their relatives are prevented from establishing their carrier status. Furthermore, because the law of confidentiality in the UK effectively

prohibits clinicians from disclosing genetic test results to other interested parties, including other family members, the index case initially bears the responsibility for informing their relatives about the result.^{14–17} Thus, for technological and legal reasons, affected women (and men) are positioned as responsible for both generating genetic information and disclosing it to their kin. The aim of the present study was to determine women's understanding of the ethical issues generated by mutation searching, so as to gain insight into their information and support needs.

Finally, it must be noted that affected individuals who are identified as mutation carriers have an increased risk of developing a second primary cancer.¹⁸ Thus, affected women and men, like their at risk relatives, may need to make decisions about prophylactic surgery or surveillance, following confirmation of their carrier status.

METHODS¹⁹

This study involved in depth interviews with 30 women who had previously been diagnosed with breast/ovarian cancer and subsequently undergone *BRCA1/2* mutation searching. Ten women had been identified as *BRCA1/2* mutation carriers (carrier group), 12 had not been found to carry a known *BRCA1/2* mutation, (inconclusive group), and eight were awaiting DNA test results (waiting group).

Recruitment

Participants were recruited by a genetics nurse specialist when they attended the cancer genetics clinic or by letter from the consultant in charge. The project was carried out according to the principles of grounded theory research and thus, recruitment ceased once theoretical saturation was reached in the data set.²⁰

Sample characteristics

The median age of the sample at the time of interview was 54 years (range 39–71 years). Twenty seven women had previously been treated for breast cancer, two for ovarian cancer and one for an unspecified gynaecological cancer. Time since the most recent diagnosis of cancer ranged between six months and 31 years (median five years). Twenty three women had children. Twelve women were educated until the age of sixteen, six until they were eighteen years, four had further education or professional qualifications and ten had a graduate or postgraduate degree. Three women had worked in a medically related occupation at some time in their lives.

Family history of cancer

Eighteen women had a maternal family history of breast and/or ovarian cancer, seven a paternal family history and in five cases it was not clear whether the (putative) mutation had been inherited via the paternal or maternal line. A total of 119 relatives were reported as affected with cancer (range 1–10, median four per family). Twenty six women had at least one first degree relative affected with either breast/ovarian/endometrial/prostate cancer.

Data collection and analysis

In depth interviews (between one and two and a half hours) were carried out at a location of the participant's choice between November 2000 and June 2001. The interviews explored the following themes: diagnosis and treatment of cancer; family support during illness and testing; experience of DNA testing; decision making about testing; communication of results within the family, and the impact of genetic testing on their lives. All interviews were tape recorded with consent and transcribed. The transcripts were coded using the method of constant comparison.²¹ The data were initially indexed on a case by case basis, which allowed patterns and

relationships between the codes to emerge within the data set, leading to the development of second order categories. Between interview comparisons were drawn and deviant cases were taken into consideration. The analysis revealed that there were no differences in the women's responses when stratified by either disease or prognosis. Any differences that were observed in the responses of women receiving different types of test results are discussed below.

FINDINGS

When describing their experiences of mutation searching these women drew upon two potentially competing ethical discourses: one in which selves are seen as relational entities that exist within a network of relationships that carry with them obligations of care and another in which selves are seen as autonomous individuals—self governing agents. The women drew on these discourses in different ways when describing their role in generating and disclosing genetic information. Indeed, the following analysis suggests that these women experienced their role in generating and disclosing genetic information very differently.

Generating genetic information for others

Most of the women in this study did not see mutation searching as having any direct health benefits for themselves, in terms of providing them with access to medical services. They were either already engaged in annual surveillance, or had previously undergone therapeutic/preventive surgery on their breasts and/or ovaries, or had decided to forgo preventive surgery for the present. That does not mean, however, that they failed to derive any personal benefits from mutation searching, for their accounts revealed that this was clearly not the case.

Carol²²: "It [testing] made me feel as though, because I have had the experience (breast cancer), I am helping and perhaps saving a life. But that is important to me because if any of my sisters' girls had, or is about to have, breast cancer and they were not aware and didn't do anything to test themselves and didn't know anything about it, it would have been a waste, a complete waste of all I have gone through, at least I am doing something with it now which is helpful." (Carrier)

These women constructed mutation searching as primarily affording them the opportunity to help their relatives: their children, siblings, grandchildren, and other family members.

Interviewer: "You said that one of the reasons was . . . to find out information for your daughters, were there any other reasons that you decided to proceed with testing?"

Oona: "Oh no, really just to help my daughters and any further family. I mean to me it's immaterial now I know that I have got breast cancer, or had breast cancer." (Waiting)

Like the participants in a recent study of *BRCA1/2* predictive testing,²³ all the women in this study said they had undergone testing to obtain genetic information for others, indeed, for 90% of the sample this was reported as being the most important, or the only, reason for undergoing this procedure. Mutation searching was thus perceived as enabling one to demonstrably care for others—to act as a moral agent and fulfil one's obligations to care for other family members.

Jane: “. . . I asked if it was possible my sister might be at risk, because my cousin had died from cancer at the age of 38 [unclear]. So I was worried about my sister, because she’s not as strong willed as myself, and if I could prevent her going through the same kind of treatment, then I’d do anything for her, to help her, basically. So that’s how I got on to the genetic testing part of it.” (Inconclusive)

In constructing their role as generator of genetic information as acting responsibly, these women can be seen as drawing upon a care based ethic.²⁴⁻²⁶ In justifying why their obligations of care were satisfied by generating this information, these women drew upon discourses that construct genetic information as empowering because it provides individuals with choices.²⁷ Accordingly, these women stressed that their relatives should have access to information that could affect their future health.

Verity: “Well, I think in lots of ways it’s nice to know because almost forewarned is forearmed isn’t it, at least now, um . . . the girls will be well watched . . . if they have any problems they’ll be given priority because this is known, if they have the gene”. (Carrier)

As far as these women were concerned, to be in receipt of genetic information which confirms one’s risk or carrier status is to be forewarned, and to be forewarned presents individuals with choices, most importantly, with the possibility of forearming oneself against disease. Thus, in addition to alleviating their relatives’ anxiety about their potential risks, the participants were concerned to provide their relatives with information that was deemed necessary for them to make informed risk management decisions.

Cherie: “I thought, oh, well, that [testing] would be good, because I’ll know whether I’ve got it or not, which then means that I will know whether my daughter has got it or not, and my nieces wouldn’t have this big axe hanging over their head—because that’s how you sort of view it really. That’s how I view it, more for them rather than how I viewed it for myself . . . We’d hoped they could find this gene, and then it could take the worry off their shoulders.” (Inconclusive)

Therefore, these women saw their role in generating genetic information for their relatives as not only a responsible thing to do, but also as the right thing to do.

Angela: “I’ve got two nieces and obviously I felt a responsibility to, towards them and to their parents. So I thought for them also it would be useful and that I should do my bit really.” (Waiting)

Thus, generating genetic information about themselves, and, as a consequence, producing information for other family members, enabled these women to act, or to be seen as acting, as a moral agent, as caring for their relatives.

In justifying their participation in mutation searching these women frequently cited the need to preserve others’ autonomy, often at the expense of their own, and as such, presented themselves as altruistic testers par excellence. This observation raises questions about the nature of their consent in this instance. As was noted above, the information gained from genetic testing pertains to families, groups of people who stand in both biological and social relationships with each other. This study suggests that those who undergo *BRCA1/2*

mutation searching are not only aware of other family members’ interests in the information derived from this procedure, but primarily undergo genetic testing with these interests in mind. As far as most of the women in this study were concerned, mutation searching was not egocentrically motivated, it was not about self determination, in the accepted sense, but was an altruistic act. Ultimately, they saw themselves as acting for the benefit of others; they were consenting to testing on the behalf of their kin. Thus, whilst all voluntarily agreed to undergo mutation searching one can question the limit of their autonomy. Although there was no evidence that testing was explicitly coerced by medical staff, or in most cases other family members, arguably their choices were implicitly constrained by their obligations to care for, or help, their kin. The women who took part in this study were acting as selves in relation, not as autonomous agents as such. As selves in relation they perceived themselves as having an obligation to other family members to undergo mutation searching so that they could provide their kin with the genetic information they needed to make informed decisions about their risk management. Thus, as far as most women were concerned, they did not make an explicit decision concerning whether they underwent mutation searching, because, in a fundamental sense, they felt they had no real choice. While, in theory, these women could have refused to undergo this procedure, thus, preserving their right not to know²⁸ their genetic status,^{4, 29} in practice, they perceived their actions as constrained by their need to care for other family members. These observations lead us to question the adequacy of the concept of autonomous choice in the context of genetic testing.

Disclosing genetic information to others

While, however, these women did not necessarily see their role in generating information for others as ethically problematic, the disclosure of genetic information to others was described as raising unforeseen moral dilemmas,^{20, 30, 31} particularly by those identified as mutation carriers. Although all the women conceived of themselves as having a moral obligation to inform their family members³² of their carrier status, for after all this is why they had undergone genetic testing in the first place, many regarded the disclosure of genetic information to their relatives as (potentially) difficult or ethically burdensome. First, many women indicated that they had not really reflected on which members of their family they might need to tell if, and when, they received a result. While they were all prepared to disclose information to sisters and offspring, many had not considered that they might need to inform their brothers or other members of their family of origin, and commented that that had been, or would be, difficult.

Verity: “I didn’t think backwards I only thought forwards. I only thought about my offspring and their offspring. I really didn’t consider my brother or, no I didn’t . . . I didn’t give any thought to that really until I spoke to Dr X and they started explaining . . . and then suddenly I thought ‘oh crumbs’. That’s why I say it’s like throwing a stone into a pond and the circles start coming out.” (Carrier)

Second, some women said they were uncertain about how or when they should disclose information to their kin—for example, Isobel, who had been confirmed as a mutation carrier two years previously, talked about her feelings about informing her lateral kin, a task she had not yet undertaken. She said she felt “a great burden,” when her doctor had told her that she would have to tell her cousins about their risk.

Third, many women acknowledged that disclosing genetic information to their family potentially conflicted with their obligation to care for their relatives, because the disclosure of this information could result in increasing their relatives' anxiety about developing cancer.

Natalie: "Well I think the important thing is how you deal with imparting it to the relatives and um as I say I don't really remember how I dealt with my daughters, it was very tricky. And especially when they are young you don't want to burden them with information, but then they have a right to know I think." (Carrier)

Consequently, some women no longer constructed themselves as the providers of information, which would necessarily foster others' autonomy, but reconstructed themselves as the bearers of "bad news".

Mary: "Other members of the family I haven't done anything with, because how do you approach them? Because [to] my mother, her sisters, and brothers "cancer" is a death word . . . the ones who are further away I haven't approached. One, because I don't see them very often. Two, how do I do it? This is the dilemma. Do you ring people up, write to the people, go and see them and say 'look there is this chance'? I think if it was me, I would want to know . . . because then you can do bits and pieces. But if you are the bearer of this news, I think you are torn. You don't know what to do for the best." (Inconclusive)

Thus, depending upon the test outcome, many of these women perceived themselves as potentially forced into a situation in which they would be responsible for causing others harm. In this sense, having genetic information to impart to their relatives generated an ethical dilemma for these women.

In articulating the ways in which they approached and negotiated this dilemma, many women questioned their previously stated assumptions about the status of genetic information. From being empowering in and of itself, they considered whether having access to genetic information was universally good. Thus, many questioned whether having genetic information about oneself necessarily promoted autonomy at all times and in all circumstances. Some women commented that it was more difficult to receive this type of information at particular points in the life course—for example, when young or already incapacitated by illness. Others described having access to genetic information about the family and oneself as potentially harmful, because it reminded one of the risks of cancer occurring in others or recurring in oneself, and they acknowledged that this was not necessarily a good thing.

Beth: ". . . you might be lucky and you haven't got it [a mutation] and you are free, but if you are unlucky you are sort of doomed to a life of check ups going on and on and on, and always worried that it's going to come out at any moment or the next year or the year after, and [thinking] would I have to plan for the future because you may not have one, and all this." (Carrier)

Many women also commented that giving their relatives genetic information meant that their kin would then have to make difficult risk management decisions—for example, decisions about prophylactic oophorectomy or mastectomy; decisions that could be avoided or postponed whilst uncertainty about their carrier status remained.

Interviewer: "Will your sister have testing if they find the mutation?"

Sally: "Yes, yes everybody will. And then that's when it becomes difficult I think, when it will become a big issue." (Ivr: "Why?") "Because then people will have to make decisions as to what they are going to do about it." (Waiting)

Thus, when considering the implications of receiving genetic information about oneself, many women questioned whether, in some circumstances and for some individuals, ignorance may, indeed, be bliss.

Caroline: ". . . There must be quite a lot of people who are terrified of the whole topic [testing]. And to have to go further into it with no knowledge of whether you are doing a good thing, or it may be bad, you know people don't want to know that they have a gene or genes." (Ivr: ". . . Why do you think that some people might not want to know that?") Well, a lot of people prefer to be ignorant about all sorts of things don't they? I mean if you know that there is something unpleasant in your, either you make up or in what's going to happen, it affects your whole personality and how you are going to behave." (Waiting)

Balancing responsibility and autonomy

These women are faced with an ethical dilemma. On the one hand, they do not wish to harm others by giving them genetic information which may negatively affect their lives, while on the other, they regard others as having a right to information which may facilitate their health management decisions. In an effort to uphold their obligations of care and yet preserve others' autonomy, these women adopted different rhetorical and behavioural strategies.

First, they invested in discourses of hope; namely, the view that we as a society are currently on the brink of a technological breakthrough in the treatment or prevention of cancer.

Isobel: "The odds are that even if, that even if the girls develop perhaps the cancer, by the time that they do, there may be means of zapping it before it starts being a problem or screening so precisely that you know the rogue cell will be sorted. I mean it's just moving towards all these things all the time." (Carrier)

Arguably, drawing upon discourses of scientific progress enabled them to justify transmitting what they regarded as potentially "bad news" to younger relatives. Second, many described how they waited until an appropriate time to tell their relatives, namely, a time when they perceived the news would cause them less anxiety or distress—for example, following the birth of a child rather than during a pregnancy. Third, they delegated the responsibility of disclosure to others, either unconnected third parties, such as solicitors, who held the information for family members to access at their convenience, or female married in kin, for example, daughters/sisters in law.

Other women, particularly those who were either awaiting a test result or had received an inconclusive result, reported that they had intentionally avoided causing their relatives worry by not telling them they had undergone mutation searching. These women reasoned that as they had no conclusive results at present, they did not feel a need to inform their kin. Indeed, some were of the opinion that telling their relatives that they had undergone genetic testing, prior to receiving a conclusive result, would cause them needless

worry about cancer risks; risks, which may, or may not, exist. Others, who had received inconclusive results, had informed their relatives about their involvement in mutation searching from the outset, because they felt they had an obligation to inform their relatives of the possibility of surveillance. They said they had experienced no problems in disclosing this information to their kin, as Cheryl put it: “no news is good news until it is bad”.

In summary, this study indicates that some individuals who undergo genetic testing experience the disclosure of test results to other family members as ethically contentious. Whilst, however, ethicolegal debates^{1 2 4 15} frequently present the disclosure of genetic information within the family as a coerced choice—to tell or not to tell—in practice, there appears to be no such clear cut distinction. The data presented above suggest that the ethical dilemma faced by women in the present study was not whether they should disclose genetic information to their relatives—indeed, all the women felt that all implicated family members should receive this information—but how they should effect this in practice. Thus, while all the women intended to disclose the information they received from testing to relevant parties, they frequently expressed uncertainty about who they should disclose information to, how they should disclose it, and when disclosure should occur.^{26 31} Thus, as far as these women were concerned, disclosure itself was not perceived as ethically contentious, but rather the practice of disclosing genetic information to particular individuals was experienced as generating particular moral dilemmas; dilemmas they had not considered prior to testing. Such observations raise questions about the type of information these women had received, or understood, prior to consenting to testing, and thus, the degree to which their consent was in fact “informed”.

CONCLUSIONS

To summarise, when describing their experiences of mutation searching the women who took part in this study described their actions as motivated by their obligations to care for their relatives. At one and the same time, they saw themselves as having the responsibility of providing their kin with genetic information that would foster autonomous decision making about their health risk management and as having a responsibility to protect their relatives from the harms that this information might cause.³² In an effort to uphold their obligations of care and yet preserve others’ autonomy, these women adopted different strategies. They invested in discourses of hope, they delegated the responsibility for disclosure to others or they postponed informing their kin about their risk status until either their relatives’ personal circumstances had changed or they had more conclusive information to give. By acting in this way these women constructed themselves as moral agents, as doing the right thing, even when, by their own admission, the right thing was not always easy to discern.

This study suggests that generating genetic information to give to others, and disclosing this information to others, raise different types of ethical issues. In this final section of the paper we briefly look at the implications of these empirical findings for ongoing ethical debates and clinical practice.

It was argued above that this study raises questions about the suitability of conceptualising consent in the context of genetic testing as involving autonomous and informed choices. Although we are not claiming that we can reach conclusions about what consent to genetic testing should be on the basis of these observations, these data suggest that the current model, which is based upon a conception of autonomous decision making, may be inadequate.

First, it necessary to distinguish between two ways in which the term “choice” is used in discussions of consent. In the first, “choice” is used to refer to the act of “choosing” between two or more options—that is, the exercise of choice, whilst in the

second, it is frequently used to mean the “options” that are available. Whilst these uses can be seen as distinct they are interdependent, in so far as the act of choosing presupposes the existence of viable choices. As far as the women in this study were concerned, the choices that were on offer: (a) undergoing testing to generate information that may increase family members’ health care options or (b) refusing testing and, as a consequence, limiting others’ options, were not morally neutral. Thus, while all voluntarily consented to mutation searching—that is, technically *chose* to undergo testing, many felt that, given their obligations to their kin, their actions were constrained by a lack of viable choices, and therefore, the act of choosing in this context can be seen as more chimerical than real.³³ This observation raises more general questions about the nature of consent in the context of genetic testing.

It has been argued that the freedom to choose medical treatment is not absolute, but subject to many different types of constraints, and thus, the best we can hope for is that treatment decisions are “substantially autonomous”—that is, that there is at least a “substantial degree” of freedom of choice or independence of decision making.³⁴ As Beauchamp and Childress note,³⁵ it is impossible to generalise about the amount of freedom of choice needed for a decision to be regarded as “substantially autonomous”, thus, autonomous choice can only be defined according to context specific criteria. While we agree that a fully autonomous choice is a theoretical ideal, we question whether “substantial autonomy” is achievable when consenting to genetic testing. Arguably, the familial nature of genetic information compromises the possibility of making an autonomous decision about genetic testing on two counts. First, an individual’s DNA test results have direct implications for biologically related kin and second, the persons who undergo testing have social obligations towards these kin. For these reasons we would argue that the decision to undergo testing cannot be entirely egocentric, but may be influenced not only by one’s awareness of others’ interests in obtaining genetic information, but also by one’s own interests in maintaining relationships with these interested parties.

Thus, whilst the current practice of obtaining informed consent from individual patients satisfies legal requirements, thereby allowing clinicians to circumvent an array of legal actions, it can be argued that it fails to acknowledge that those persons who undergo testing are relational entities in both the biological and the social sense. The findings presented above suggest that we need to ground consent upon an ethic that takes into account the social nature of human beings. Such an ethic would acknowledge that human beings are social beings whose actions and choices are constrained by virtue of the fact that they exist within a network of relationships. Human genetics is about relationships—biological and social relationships—and it can be argued that any rigorous ethical justification of informed consent to genetic testing needs to take this into account.

The finding that some women experienced the disclosure of genetic information as generating unforeseen ethical dilemmas suggests there was a lack of awareness of the familial implications of accessing this information in some instances. Thus, on a more practical level, this research suggests that if consent to genetic testing is to be seen as “informed”, then those persons who undergo genetic testing need to be made aware of the extent of their role in disseminating this information within their family. First, they need to understand that they will bear the initial responsibility for disclosing this information to their kin. Second, they need more information about which members of their kinship may be at risk of carrying a mutation. Third, they need advice about how and when to go about informing family members about the possibility that they may carry a genetic mutation. Finally, there may be individuals who refuse to disclose genetic information to other family members. Given the legal guidelines on patient confidentiality and the non directive nature of genetic

consultations, such instances raise ethical problems for practitioners. While explicitly encouraging disclosure to implicated family members would compromise the non-directive nature of genetic consultations, providing advice and information about the implications and (potential) problems of living with a test result that they do not, or cannot, share with their relatives can be seen as legitimate.

Finally, if we are to talk of informed decision making in the context of genetic testing for *BRCA1* and *2* we not only need to ensure that clinicians provide information about the implications of mutation searching, but also that those who seek testing are given time to reflect upon their actions. At the present time, predictive *BRCA1/2* testing protocols in the UK incorporate a "cool off" period allowing women (and men) to consider the implications of testing, for themselves, and their family before proceeding. Arguably, the introduction of a similar period for reflection prior to mutation searching can be seen as warranted.

In conclusion, this study suggests that generating and disclosing genetic information raise different types of ethical issues. Although the women who took part in this study may not have perceived their role in generating genetic information as particularly ethically contentious, their accounts suggest that whilst the decision to undergo genetic testing was not perceived as ethically problematic, it was not a morally neutral choice. If we are to develop a rigorous ethical justification for informed consent to genetic testing, we need to take into account not only individuals' information needs, but also the relational constraints on their actions. We need to acknowledge that those who consent to genetic testing may be less interested in furthering their own autonomy, than in enabling others to make autonomous decisions.³⁶

ACKNOWLEDGEMENTS

We would like to thank all the women who participated in this research. Thanks are also due to Sue Davolls, Gillian Mitchell, Richard Houlston, and Fiona Lennard for their support during the recruitment and data collection phases of this research. In addition, we are very grateful to Louise Irving, Julia Lawton, Richard Ashcroft, and Elizabeth Chapman. This research was funded by a Cancer Research UK Grant No P1026.0501 to M Watson, C Foster, R Eeles, A Ardern-Jones, R Houlston, and V Murday.

Ethical approval for the study was obtained from the Royal Marsden NHS Trust LREC

Authors' affiliations

N Hallowell, C Foster, The Institute of Cancer Research, London, UK
R Eeles, Translational Cancer Genetics Team, Institute of Cancer Research and Royal Marsden NHS Trust, London, UK
A Ardern-Jones, Cancer Genetics Clinic, The Royal Marsden NHS Trust, London, UK
V Murday, St George's Hospital, London, UK
M Watson, Department of Psychological Medicine, Royal Marsden NHS Trust, London, UK

REFERENCES AND NOTES

- Skene L. Patients' rights or family responsibilities? *Med Law Rev* 1998;**6**:1–41.
- Bell D, Bennett B. Genetic secrets and the family. *Med Law Rev* 2001;**9**:130–61.
- Richards MPM. How distinctive is genetic information? *Studies in History and Philosophy of Biological and Biomedical Sciences* 2001;**32**:663–87.
- Laurie GT. In defence of ignorance: genetic information and the right to not know. *Eur J Health Law* 1999;**6**:119–32.
- Beauchamp TL, Childress JF. *Principles of biomedical ethics* [4th ed]. New York: Oxford University Press, 1994.
- See reference 5: 124.
- Miki J, Swensen J, Shattuck-Eidens D, et al. A strong candidate for the breast ovarian cancer susceptibility gene *BRCA1*. *Science* 1994;**266**:66–71.
- Wooster R, Bignell G, Lancaster J, et al. Identification of the breast cancer susceptibility gene *BRCA2*. *Science* 1995;**265**:2088–90.
- Ford D, Easton DF, Stratton M, et al. Genetic heterogeneity and penetrance analysis of the *BRCA1* and *BRCA2* genes in breast cancer families. *Am J Hum Genet* 1998;**62**:676–89.
- Whittemore AS, Gong G, Iltis J. Prevalence and contribution of *BRCA1* mutations in breast and ovarian cancer: results from three US population based case control studies of ovarian cancer. *Am J Hum Genet* 1998;**60**:496–504.
- Burke W, Daly M, Garber J, et al. Recommendations for follow-up care of individuals with an inherited predisposition to cancer. II. *BRCA1* and *BRCA2*. Cancer Genetics Studies Consortium. *JAMA* 1997;**277**:997–1003.
- Eeles RA. Future possibilities in the prevention of breast cancer: intervention strategies in *BRCA1* and *BRCA2* mutation carriers. *Breast Cancer Res* 2000;**2**:283–90.
- While men can also carry *BRCA1/2* mutations which increase their risk of developing breast (*BRCA2*) and other types of cancer, at the present time most of patients undergoing mutation searching are women.
- Nuffield Council of Bioethics. *Genetic screening: ethical issues*. London: Nuffield Council on Bioethics, 1993.
- General Medical Council. *Confidentiality: guidance from the General Medical Council*. London: GMC, 1995.
- Genetic Interest Group. *Confidentiality guidelines*. London: Genetic Interest Group, 1998.
- Harper PS, Clarke AJ. *Genetics, society and clinical practice*. Oxford: BIOS Scientific Publishers, 1997.
- Ford D, Easton DF, Peto J. Estimates of gene frequency of *BRCA1* and its contribution to breast and ovarian cancer incidence. *Am J Hum Genet* 1995;**57**:1457–62.
- Ethical approval for the study was obtained from the Royal Marsden NHS Trust LREC.
- Glaser B, Strauss A. *The discovery of grounded theory*. Chicago: Aldine, 1967.
- Strauss A, Corbin J. *Basics of qualitative research*. London: Sage, 1990.
- All names are self chosen pseudonyms.
- Goelen G, Rigo A, Bonduelle M, et al. Moral concerns of different types of patients in clinical *BRCA1/2* gene mutation testing. *J Clin Oncol* 1999;**17**:1595–1600.
- This study raises questions about the extent to which the altruism voiced by the participants is (a) gender specific or (b) related to the biosocial nature of genetic information. In response to the first question, previous research on men and women's motivations for undergoing predictive genetic testing for breast and colon cancer suggests that altruism in this context may not be gender specific. See McAllister M. *Predictive testing for hereditary non-polyposis colorectal cancer (HNPCC): a theory of engagement* [PhD thesis]. Cambridge: University of Cambridge, 1999. See reference: 26. These researchers also note, however, that when it comes to disclosing this information to their kin, women are more likely to shoulder this responsibility than men are. Whether the altruism witnessed in the present study is specifically related to the biosocial nature of genetic information is less clear. Arguably similar care based justifications could be given for live organ donation—for example, bone marrow or kidney donation or the generation of HIV test results for dissemination to sexual partners. While HIV testing may also result in personal health benefits for those who are tested, live organ donation can be seen as primarily other orientated rather than self motivated, at least in the sense that these procedures, like mutation searching, have few health related benefits for the donor.
- Hallowell N. Doing the right thing: genetic risk and responsibility. *Social Health Illn* 1999;**21**:597–621.
- D'Agincourt-Canning L. Experiences of genetic risk: disclosure and the gendering of responsibility. *Bioethics* 2001;**15**:231–47.
- Petersen A. Counselling the genetically "at risk": the poetics and politics of "non directiveness". *Health, Risk & Society* 1999;**1**:253–66.
- Harris J. *The value of life*. London: Routledge, 1985. Harris notes that the existence of a positive right to ignorance is not self evident. Whilst he acknowledges that it may be the case that individuals may not want to know certain information, he argues that it is difficult to see this as a right to "not know", or indeed, that it imposes a duty on others to withhold disclosure. Furthermore, he observes that it is not clear on what principles such a right is founded, although he suggests it may be "parasitic" upon the principle of respect for autonomy.
- Chadwick R, Leviit M, Shickle D. *The right to know and the right not to know*. Aldershot: Avebury, 1997.
- Daly MB, Iltis M, Babb J, et al. Communication of *BRCA1* and *BRCA2* results to at risk relatives. *J Law Med Ethics* 2001;**29**:30.
- Foster C, Watson M, Moynihan C, et al. *To tell or not to tell* [mimeograph]. London: Institute of Cancer Research, 2002.
- In contrast to other writers who have suggested that the geneticisation of disease has resulted in a biologisation of "the family" (see Finkler K. *Experiencing the new genetics*. Philadelphia: University of Pennsylvania Press, 2000) it is not our suggestion that "family" should be interpreted to mean biologically related kin. The women in the present study, whilst acknowledging their obligations to disclose genetic information to implicated kin, did not define "family" solely in biological terms.
- Strathern M. *After nature: English kinship in the late twentieth century*. Cambridge: Cambridge University Press, 1992 for an in depth discussion of this issue.
- See reference 5: 123.
- See reference 5: 124.
- It must be borne in mind that this study only included women who had undergone DNA testing, whether those who decline testing perceive their choices as constrained is an empirical question.

..... COMMENTARY

In this commentary, I wish to address an important issue that is raised in a number of contexts in contemporary clinical genetics: the tension between a respect for individual autonomy, especially a desire for privacy or a desire not to undergo genetic testing, and the obligation to generate and pass on genetic information to those other persons in an individual's family to whom it may be important and relevant. This tension arises because genetic information is at once both individual and familial, and can be felt by professionals—by clinical geneticists and genetic counsellors, and also by general practitioners and other specialists—as well as by their clients. There is no way of avoiding this tension without leaving the field—of clinical genetics, family practice, or obstetrics, among other specialties.

The paper by Parker and Lucassen¹ argues that this tension is necessary and unavoidable in that the complete victory or dominance of either pole—of the autonomy of the individual client, or of the obligation for clients or professionals to pass on to the wider family every possibly relevant genetic fact—would lead to serious problems. I agree wholeheartedly with this, but the point under discussion then becomes how to strike an appropriate (wise?) balance between these competing claims. Parker and Lucassen express their dissatisfaction with the traditional primacy of the professional's concern for the patient physically present in the clinic. Helpfully, they distinguish between issues that arise before and those that arise after a genetic test result has been generated. They examine these issues in three case scenarios.

In the paper by Parker and Lucassen, Box 1 contains a case outline of a boy diagnosed with the sex linked disorder, Duchenne muscular dystrophy (DMD), whose mother—Alison—is a carrier of the condition. She withholds this information from her pregnant sister—Sue—because she feels it would be wrong for Sue to terminate a pregnancy even if she was carrying an affected male fetus and Alison “knows” that Sue would disagree with her on this. There are several respects in which this scenario does not do justice to the issues. First, the reproductive risk to Sue is exaggerated—there is only a one in four chance of an affected child if Sue is a carrier, and there is only a 25% (one in four) chance of that (making a few simplifying assumptions) so that the actual risk in Sue's current pregnancy is one in 16 (just over six per cent). But then one wants to know just how much information was passed to Sue about her nephew before the diagnosis was made, and whether the diagnosis and its implications really have been withheld from the rest of the family too. If a muscle biopsy was performed, it would be difficult to imagine the wider family being kept completely in the dark. Or was it the fact that DMD is a genetic disorder that was withheld? Or the nature of sex linked inheritance? Or the precise nature of the mutation present in Alison and her affected son? Unless Alison and Sue have been out of contact for years, at least some of this information is likely to be known to Sue, through their parents or other relatives if not directly from Alison. How does Alison come to be so certain about Sue's responses to genetic risk information, if they are out of contact or have not talked about the condition affecting Alison's son? So just what is it that has been withheld? I am not saying the scenario is impossible, but in the space allotted to it in the paper it is simply not feasible to present a fully nuanced presentation of the problem, so that it is difficult to develop an appropriate and realistic counter-argument.

In my experience, a more likely reason for Alison not passing on information about reproductive risks to Sue is that the pregnancy is advanced, that Alison feels certain that Sue

would not go through prenatal diagnosis at this stage and that she wishes to protect her from worry until the child has been born. At that stage, there comes the difficult decision as to when to discuss the DMD with Sue, if she has a son (it is a bit easier if she has a daughter), but she is likely to bite the bullet and do so if Sue does not ask about it herself once she has her child in her arms. She may well have deliberately shut her mind to such concerns during the pregnancy but then become able to face them after the birth. A tacit collusion between the sisters, avoiding the issue, is more likely than a judgmental desire on the part of one sister to restrict the reproductive options open to the other.

If the sisters are not in close touch, however, and if Sue has no idea about the problem with her nephew, then Alison's reluctance to pass on the information to her sister may reflect her own (Alison's) emotional turmoil. She may fear that her sister—if she chose the path of prenatal diagnosis—would be devaluing her (Alison's) affected child, and that might be difficult to cope with. Alison may be struggling with her own feelings about what to do in a future pregnancy, or perhaps she thinks she may actually be pregnant. What would it say about her son and her love for him—let us call him Duncan—if she or her sister had a termination of his affected brother or cousin? She needs the opportunity to acknowledge and reflect upon these turbulent emotions. Genetic counselling professionals can play a very helpful role here, but that role would be completely undermined by any sense of threat in the relationship between Alison and her clinician or counsellor. Any sense of, “You had better tell her now because otherwise we will”, would be damaging and likely to lead to further blocks in family communication. The professionals could frankly recommend—discarding any facade of non-directiveness—that the Alison pass on information to Sue about diagnosis, and could discuss the possible impact on family relationships of her failure to do so, especially if Sue had an affected boy and came to realise that Alison had actively withheld the information from her.

The second case scenario discussed, relating to non-paternity, does raise difficult issues that I will not address in detail. If there is a carrier test available, whether by direct DNA analysis or biochemical testing, then the clinician can suggest that both parents be tested to confirm whether the recurrence risk is one in four or not. If accurate carrier testing is not available, and the doubt about paternity has been raised solely by Polly in her phone call, then the professionals could make it very clear that the one in four risk applied only if Richard was the father, without necessarily suggesting they had any reason to doubt that. It would be difficult for them to do much more than that unless they had more substantial grounds for doubting paternity.

Problems arising during discussion before a test is performed can be difficult to resolve, as in the third boxed case scenario of a request for prenatal diagnosis for Huntington's disease when the at risk father does not want predictive testing and is not being told about the prenatal diagnosis. In such a case, as Parker and Lucassen make clear, the pregnant woman's decision to undergo prenatal testing would almost certainly be respected. The issues raised in discussing this case, however, are important: when should consent for testing one individual be required from other family members for whom the test result may have implications? When should testing at the request of one individual be made conditional upon their prior agreement to inform others of the test result? Would such prior agreement be of any ethical or legal weight if the tested individual later withdrew consent for information to be shared with their relatives? These issues are also raised in the discussion of an apparently similar case scenario in the paper by Tassicker and colleagues.²

Consent for testing must clearly be obtained from the individual to be tested, but it would not be feasible or appropriate

to demand consent from other family members as well, although open family discussion of the disease and of the genetic testing process may certainly be recommended. Prior agreement to inform others of the test results could hardly carry any legal weight if the consent were later withdrawn, but a discussion of how the test results may be communicated to other family members would usually form a part of pretest genetic counselling. In this prenatal diagnosis scenario, pretest counselling could raise for discussion with the mother such issues as whether she could in practice keep the fact of the prenatal diagnostic testing and the test result from her partner; would this be blurted out, most hurtfully, if the couple were arguing and she thought he was behaving unreasonably? Would this knowledge itself distort their relationship and condemn it to negativity?

On balance, the arguments in favour of a narrower sense of the clinical geneticist's or genetic counsellor's role and duty are stronger than those in favour of the broader set of professional obligations to many parties. While truly exceptional circumstances may lead us to behave otherwise, we almost always will have to attend above all else to the needs and wishes of the person in front of us in the clinic, although it can be perfectly proper for us to challenge their behaviour in a number of ways, such as seeking to persuade them to pass on relevant information to others.³ This will often entail a radical departure from strict adherence to the ethos of non-directiveness, which is itself, and very appropriately, a central feature of genetic counselling practice. In the two contexts of prenatal diagnosis and predictive testing for late onset, essentially incurable disorders, an active and engaged form of non-directiveness is essential, to be distinguished from professional indifference/withdrawal or the abandonment of the client. In other contexts, however, it is entirely proper for a genetic counsellor to recommend and work towards open family communication and even to persuade a client to undergo genetic testing if this is likely to yield practical, medical benefits—such as the improved medical management of risk from familial cancers or heart disease.^{4,5}

While agreeing with much of the conclusion of the paper by Parker and Lucassen, I would like to encourage thought and discussion around the question of support for genetics professionals from clinical ethicists or ethics committees. Support from colleagues and from others interested in the issues can be very helpful in scrutinising the arguments for making one particular clinical decision rather than another in a particular case, and more formalised discussion among clinicians and others can be helpful in considering general situations and perhaps drawing up recommendations about good practice in the general case. I would argue that the role of the "others" in such discussions can be very helpful indeed, but I would suggest that the disciplinary background and training of these "others" should be kept as broad as possible. I would not single out ethicists as the primary professional group to bring into this role. While there are ethicists with all the attributes required to facilitate constructive discussions among clinical professionals about difficult cases, this will have as much to do with the personality and previous life experiences of these particular individuals as with their training as ethicists. Other individuals, equally capable of a constructive role in assisting professionals to tease through some of these issues, may come from backgrounds such as clinical psychology, psychotherapy or counselling, communication and discourse studies, pastoral theology, the law, or management studies. The "success" of a clinical ethics consultation may be seen as the consideration given to the issues, leading to more refined or circumspect views rather than reversing decisions⁶ and there may be many different perspectives that can usefully be engaged in such discussions.

While wanting to encourage the interest of ethicists in the area of genetics, I would not want to see an outbreak of territorial wars among the colonialists of clinical decision

making—between ethicists and other interested parties. Instead, I would welcome the active engagement of appropriate individuals from any of these backgrounds or others in constructive discussions with genetics professionals, focused on strategies to help resolve the difficult clinical or family issues that arise in relation to genetic disorders and genetic testing. The diffusion of genetic testing for Mendelian disorders into other areas of medicine, to the extent that it happens, may raise similar concerns in these other areas. Those with experience of handling such issues—whether working as clinical geneticists or genetic counsellors or in support of these groups—will have an important role to play in the development and training of those professionals who have not previously come across these issues in practice.

One important issue that is touched upon in Parker and Lucassen's discussion, but is not addressed in detail, is that of testing carried out on one family member specifically for the benefit, or at the request, of another family member. This happens regularly in families with apparently inherited forms of breast or breast/ovarian cancer, as is considered in detail by Hallowell and colleagues.⁶ Defining the mutation present in an affected family member enables their relatives to be offered accurate genetic testing to see if they are at increased risk of the same form of cancer. Finding a mutation in one of the breast cancer susceptibility genes, however, is not without clinical, practical, or emotional consequences for the affected individual. It indicates that they are at increased risk of recurrence (in the form of a second tumour in the same tissue) and at increased risk of a primary tumour in other organs, as well as making it clear that other family members are definitely at increased risk—the family history has not arisen purely out of bad luck.

Hallowell and colleagues address these issues through an interview study of women affected by, and with a family history of, breast (and/or ovarian) cancer. These women gave consent for their genetic testing to go ahead—for an attempt to be made to find a mutation in *BRCA1* or *2*—and they usually did so out of a sense of family obligation and altruism, despite the anxiety this might cause them, so as to make it possible for their relatives to undergo predictive testing if a mutation were found. When mutations were found, they were then put in the position of having information to pass on. This information might not always be received by their relatives as useful or helpful, so some of them found themselves cast in the role of the giver of bad news, who was thereby imposing the burden of difficult decisions on close kin. Not surprisingly, the women sometimes found it difficult to find the right moment to pass on this information and a number only informed their relatives about the genetic testing once the mutation had been found, and a few delegated the task of informing others to a third party.

The principal concern of the authors is expressed as the threat to autonomy from the women's sense of obligation to their kin, which is the force driving them to undergo testing. The women's autonomy is regarded as "substantially compromised" by the biological implications of the genetic testing for their kin and by their social obligations towards these relatives. There are two strong grounds for challenging this representation of autonomy as if it were under threat. First, there is the excessively "abstract" notion that one should be able to exercise autonomy as if living in a social vacuum—that other people get in the way of one's own justified desires—which denies the essence of our humanity as social beings. Second, there is the implicit decision to accept the current UK model of genetic testing for familial breast cancer, without making it clear that this collective policy could be regarded as the ethical issue or as setting up the ethical problem for family members.

People do feel constrained in their actions by their network of social relations. This is only to be expected, and to reject the importance of such influences is to adopt the morality of a

solipsist. Indeed, there is something absurd in the idea that one's behaviour should not be influenced by a consideration of other people's interests—it amounts to adopting (or at least recommending) the ethics of the amoral sociopath. We humans only exist as persons in relation. This mode of existence may, at times, feel inconvenient, but—as observations of feral children only confirm—*Homo sapiens* is essentially a social being. Social obligations do not prevent those who experience them from making real choices—from exercising their autonomy. Rather these obligations, sometimes mutually incompatible, provide the framework within which (difficult) choices have to be made. To express resentment at these constraints or difficulties may be understandable but does not constitute grounds for rejecting the obligations. That would be the equivalent in morality of the response to the traveller's question, "How do I get to X?"—the famous but unhelpful response being, "I wouldn't start from here."

A reasonable conclusion to the discussion of Hallowell and colleagues is to recommend that professionals should raise for discussion some of these family issues before going ahead with mutation searching in the *BRCA1* or 2 genes of women with breast cancer. A "cooling off" period before the test goes ahead is reasonable, as with predictive testing for the same disorder and for Huntington's disease. The pretest discussion should certainly include difficult topics such as the obligations of the woman and of the professionals towards other members of the woman's family. The professionals should also offer assistance with the process of disclosure and support for the woman and her family after testing. As mentioned above, the expectation that professionals should be expected to practise "non-directively" in this counselling context may not be appropriate as we should be able frankly to recommend family openness and an active but caring approach to the disclosure of relevant information to other members of the family.

Finally, there are the policy decisions about how genetic testing for familial risk of breast cancer should be made available. There are at least three ways in which such testing can be made available. The current UK model is fairly efficient in that those at risk of cancer only gain access to testing if they actively seek it, if their pretest risk is substantially higher than the population risk and if a relevant gene mutation has been found in an affected relative. The known mutation can then be sought in those at risk, and the interpretation of the test result is usually clear—the individual tested is either at high risk of cancer or at general population risk. Another approach is to test affected women with a family history suggestive of an inherited predisposition and then to offer testing to their relatives once their family's mutation is known. This maximises efficiency but entails actively suggesting mutation searching in families where there may not have previously been much concern. Finally, there is the individualistic approach of testing those who seek testing, regardless of whether a mutation is known in their family. A positive result—finding a mutation—is then of relevance to the individual and their relatives, but the interpretation of some gene variants may not be straightforward and the value of a negative test result may be rather limited—the failure to find a mutation could result either from the absence of a detectable mutation in the family or from the absence of such a mutation in the individual when there is such a mutation in the family. The two situations differ greatly in significance.

Professionals and health services in the UK have chosen to adopt the first of these three strategies, a thoroughly reasonable but not inevitable or unchallengeable approach. It is this strategic decision that requires affected women to undergo mutation searching before their relatives can gain access to predictive genetic testing. This collective decision by professionals may well be the best policy from a public health and resource consumption perspective—maximising the informativeness of the testing that is performed on at risk individuals,

and minimising the number of tests performed overall—but it comes at a price. This price is the burden of a sense of obligation to be tested so as to make predictive testing available within their families. We should not shut our eyes to such structural, or policy based, constraints on genetic services. We should guard against the tendency to view such structural constraints as fixed and inevitable while individual decisions are seen as raising difficult ethical issues; the social structures within which we make our individual decisions are also open to challenge.

A Clarke

Department of Medical Genetics, University of Wales
College of Medicine, Heath Park, Cardiff CF14 4XN, Wales,
UK; ClarkeAJ@Cardiff.ac.uk

REFERENCES

- 1 **Parker M**, Lucassen A. Concern for families and individuals in clinical genetics. *J Med Ethics* 2003;**29**:70–3.
- 2 **Tassicker R**, Savulescu J, Skene L, *et al*. Prenatal diagnosis requests for Huntington's disease where the at risk father does not want to know his genetic status: a clinical, legal and ethical viewpoint. *BMJ* 2003;**326**:331–3.
- 3 **Nuffield Council on Bioethics**. *Genetic screening: ethical issues*. London: Nuffield Council on Bioethics, 1993.
- 4 **Clarke A**. The process of genetic counselling: beyond non-directiveness. In: Harper PS, Clarke A. *Genetics, society and clinical practice*. Oxford: Bios Scientific Publishers, 1997: 179–200.
- 5 **Elwyn G**, Gray J, Clarke A. Shared decision making and non-directiveness in genetic counselling. *J Med Genet* 2000;**37**:135–8.
- 6 **Hallowell N**, Foster C, Eeles R, *et al*. Balancing autonomy and responsibility: the ethics of generating and disclosing genetic information. *J Med Ethics* 2003;**29**:74–83.

AUTHOR'S REPLY

We read Clarke's commentary with interest and were pleased to see that he appears to concur with most of the key points raised in our article. Like Clarke, we are of the opinion that the individualistic notion of autonomy that underpins current medical models of informed consent overlooks the fact that human beings are social beings. The data reported within our paper suggest that those who are faced with making decisions about mutation searching are influenced by their social obligations to biological kin. To regard medical decisions in general, and decisions about genetic testing in particular, as taking place within a social vacuum, as based entirely upon individualistic preferences and choices, ignores the myriad of differing (social, economic, political etc) constraints on the decisions that human beings have to make. Genetic testing raises a range of ethical issues for both patients and professionals alike. The taking and giving of consent to such procedures, like all human practices, is a complicated and messy affair, which arguably, defies an easy theoretical solution. As moral agents we act within the real world, thus like Clarke, we believe that these real world constraints on our behaviour should be acknowledged within our ethical theorising. Whilst our paper concentrates on the structural constraints of kinship on testing decisions, we are mindful of the macro socioeconomic structures that influence policy decisions in this area, which, as Clarke so elegantly points out, raise ethical issues for both the wider society and for family members.

In conclusion, we would agree with Clarke that while current policy on genetic testing for breast and ovarian cancer may raise ethical issues for those undergoing mutation searching, as demonstrated in our paper, the alternative models of service provision he describes raise equally contentious ethical issues. Leaving aside the question of resource allocation, the first alternative Clarke discusses raises the

spectre of implicit coercion to undergo genetic testing in families where this is not perceived as an issue and therefore, could be seen as impinging upon individuals' right to not know genetic information about themselves. Model three, while allowing individuals to make seemingly "independent" decisions about undergoing testing, does not overcome the ethical dilemmas they may then face with regard to disclosing a positive result to their kin. Furthermore, as Clarke rightly points out, the probability of receiving an uninformative result in such a scenario is high. Individuals may misinterpret a "clear" test result as indicating that the cancers in the family are definitely not inherited and that they and their kin are not

at risk,¹ with the result that they and their relatives decide to forgo medical interventions which have been proven to reduce cancer risk in mutation carriers.

N Hallowell

Institute of Cancer Research, Royal Marsden NHS Trust,
Downs Road, Sutton, Surrey SM2 5PT; nh113@cam.ac.uk
M Watson, Department of Psychological Medicine, Royal Marsden NHS
Trust, London, UK

REFERENCE

- 1 **Hallowell N**, Foster C, Eeles R, *et al.* Genetic testing for women previously diagnosed with breast/ovarian cancer: examining the impact of *BRCA1* and *BRCA2* mutation searching. *Genet Test* 2002;**6**:79–87.



Find out what's in the latest issue
the moment it's published

Email Alerts

Sign up to receive the table of contents by email every month. You can select from three alerts: Table of Contents (full), TOC Awareness (notice only); *Journal of Medical Ethics* related announcements.

www.jmedethics.com