Knowledge of No Return

Getting and Giving Information About Genetic Risk

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New genetic information can provide constructive preventive possibilities for individuals and for society but can also create new dilemmas for them. In consultations dealing with the risk of inheriting cancer, many problems connected with the notion of information exchange come to the surface. Individuals have to deal not only with the information given by the doctor, or how to give information to the doctor, but also with the problem of informing others, close kin with a similar risk potential, or getting information from them. In all these information exchanges concerning risk of cancer, different notions of 'information' are being invoked and used as resources in the talks, implying communicative problems at different levels. Some of these problems are discussed in this article.

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Through research advances in medicine and genetics, individuals in Sweden, as in other parts of the Western world, may now learn that they carry a genetic predisposition to diseases such as cancer. Technology is rapidly making such genetic information accessible, but we do not yet know much about how it is interpreted and understood by the recipients. Individuals who believe themselves to be healthy learn that they may be bearers of genetic characteristics that entail health risks not only for themselves and their children, but also for siblings or other close kin. The family tree, so familiar to professional and amateur genealogists, acquires a new significance when family experiences of sickness and health are linked with genetic information about individual risks.

Information about genetic predisposition implies that individuals are confronted with the responsibility of, first, deciding what to do in order to avoid possible future disease and, secondly, understanding genetic assessments and medical facts so as to be able to communicate with significant others in the family. While new genetic information can contribute to individual and general prevention, it can also pose dilemmas. Who should be informed about new research results? Should people be told about the risk of diseases for which there is no cure? Should the relatives of someone who has actively sought and received information about the genetic risks of cancer also be told —even if they have not asked for any information? Who should be responsible for informing the families as a whole about genetic risk—the researcher, the clinician, the individual immediately at risk?

People in Sweden who fear they are at risk of inheriting cancer and want information can consult doctors at a few specialized university clinics. These consultations raise many problems connected with genetic knowledge and related information exchange. The interactions exemplify situations where risk has to be communicated, interpreted and understood initially in the form of probabilities, based on a particular family's history of disease. The person seeking information must play an active part, setting the scene from memory or by consulting relatives. As a result, he/she often possesses information about other relatives—and has to reflect on what this implies. Besides considering information provided by the doctor, or on how to give information to the doctor and what the doctor should be told, he/she has the problem of informing others and getting information from them. In all these exchanges of information about the risk of hereditary cancer, different notions of scientific information are invoked and used as resources in the talks. This leads to communicative problems at different levels. Here I propose to take up a few of these problems.

THE STUDY

In order to understand and analyse aspects of information exchange about genetic risk, I have collaborated with Carol Tishelman, doctor of medical science and researcher

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in nursing at the Karolinska Institute, Stockholm. We studied the interactions between people who, through a variety of sources, had been referred to a special clinic for hereditary cancer and the doctors involved in this work. The clinic provided these people with information about their individual 'risk profile', often calculated in terms of risk percentages and primarily based on genealogical mapping for hereditary cancer. This meant that risk had to be discussed in relatively general terms and could not be assessed more exactly until mutations had been found and a blood sample analysed. Both these possibilities were at hand in the latter part of our research; in the early stage, however, risk profiles had to be constructed from genealogical mapping of the family's disease history. Assessments of this type seem to be the standard procedure when the familial disease situation suggests that heredity and genetic mutations have not yet been found.

In the practice of genetic information of which I speak here, the doctor's role is to assess what he/she believes to be a particular individual's risk of hereditary cancer and to inform about the options for future action. In order to assess the individual's risk, the doctor needs as much pertinent information as possible about the family's history of disease (who died of what and when), including access to documents such as death certificates, diagnostic tests, stored blood samples, and so on.

In the course of the study, which was conducted during 1993 and 1994, consultations between clinicians and 31 information seekers were observed and audio-taped by CT and myself. The consultations lasted for an average of about 60 min. The diseases discussed were mainly breast, ovarian and colon cancer. Most persons were interviewed by us before and after the information sessions with the clinicians.

Ten of these persons and their families also took part in a close anthropological follow-up study, which I conducted over a period of four years. It is mainly from this study that the present examples are drawn. Five are women who have accepted prophylactic surgery and have given me their narrative accounts of the whole process: from the presymptomatic risk diagnosis, through the difficult decisions about how to deal with the risk, to the time of the operation and afterwards when they had to learn how to deal with the loss of bodily parts (breasts or ovaries) and adjust to newly constructed breasts, for example. They also described the period after the operation, when learning how to live without risk in close familial relationships. This anthropological study has resulted in a book, *Living with risk—five women, genetic tests and the fruits of knowledge*, published in Swedish (1). From an anthropological perspective it is essential to describe how advances in medical/genetic technology have cultural implications in that they gradually change people's perceptions of their bodies, sickness and health and ultimately of life and death.

NEW KNOWLEDGE AND THE NOTION OF INFORMATION

In everyday communication, the notion of information tends to be used simplistically. Information is, however, never simply the neutral transmission of facts, not even when dealing with scientific knowledge and genetic research. It is always interpreted and evaluated from a particular perspective in a specific context (2–4).

That information can be neutral is thus a myth. In consultations about genetic risk, the process of communication is not just a matter of transmitting information from one who knows to one who does not know. People do not accept facts at their face value, but interpret them according to their own understanding, biases, and emotions (4). In no sense do people simply assimilate neutral information. They learn facts that concern them. These are facts-for-them, facts loaded with emotions, and their acceptance and interpretation may lead to anxiety and feelings of helplessness or relief. In one way or another the person who is informed has to deal with, interpret and live with what has been said. Knowledge created and formulated in a scientific context is thus recontextualized when it is expressed, first, in a clinical situation as information about the risk of hereditary cancer and then as an interpreted version in people's real lives (3).

From the point of view of professional–lay interactions in genetic information, one can say that abstract scientific knowledge is gradually decontextualized from its location in research and accommodated—by being extracted, appropriated and reported—to individuals, thereby detaching it from whatever meanings prevailed at the time of its original discourse and redefining it in a new context (5). This makes information about genetic knowledge risky because its content never remains exactly the same over the different phases of recontextualization and reinterpretation of new meanings. This is a problem in that the new meanings form the foundation for important decisions that have to be made by women seeking information about their risk situation.

GETTING INFORMATION

One of the women in the study, who later had prophylactic surgery, was surprised and moved by the existence of this option of which she had been ignorant. She was nine years old when her mother died of breast cancer and since then she had been afraid of the same thing happening to her. After studying the genogram with her family's disease history, the doctor calculated her risk as being about 40%. Concerning the possibility of having her healthy breasts removed by prophylactic surgery, she exclaims:

'That is rather … drastic …'

'Yes, it is drastic, but I just want you to know that the possibility exists.'

'I could just say that I want to do this and then you would fix it … or? Is that how it happens?'

'Yes.'

'Without you knowing more?'

'Yes, it is up to the patient to consider, understand and perceive this risk. Often there is a strong anxiety behind one not wanting to live with this risk.'

'Well, my risk … or, my anxiety has come from my having recently become a mother … I have two girls. Just that, it is two girls … and since I was nine years old when my own mother died, I am afraid that … imagine that I die from them, too. I know how awful it is to lose your mother when you are a child, so that's where my anxiety started.'

'Yes, that is how it has been for others as well.'

'But now there is at the same time this about risk. Now I feel that … well, a little as you said … there is so much information that just goes round and round in my head.'

'But this is not something that you have to decide now.'

'No, I understand that. But can you go through this thing about risk again.'

'Well, it also depends on whether you find other relatives who are or have been ill with breast cancer.'

'So, you advise me to … look further into that … what they have died from?'

This short extract from a two-hour dialogue between one clinician and the woman illustrates women's core dilemma in such situations. They have to assess and understand their own risk situation while also deciding which close relatives they should talk to and how they should tell their children about the risks. It is clear from the sequence that the woman is relating her understanding of risk to her whole life situation and wants to talk about that. She has difficulties in understanding the abstract risk calculations she has been given earlier in the form of 35–40% risk. Risk is not transferable from population research to an individual life (6, 7).

The following sequence—from the end of a consultation between one doctor and one of the women—starts with the woman's reaction to the doctor's presentation of her options. These are either to take regular tests or have prophylactic surgery, i.e. removal of her ovaries. The doctor tries to explain that it is not the doctor who can help the woman decide; she has to make up her own mind whether or not she wants to have her ovaries removed.

'Certainly … do I have to choose that myself?'

'No … yes, yes, in some way I have to say yes because I cannot choose for you, nobody can choose for you, we can simply give you the information, this is the risk you run, this big risk of developing ovarian cancer, one can... inform you about the risks and the advantages with an operation and then you have to make the decision yourself.'

'Yes of course.'

The doctor claims that it is possible to 'simply give' this woman information. The word 'simply' seems to imply a notion of information focusing on the cognitive aspect of a report of some indisputable facts. Even if the doctor does succeed in expressing this neutrally, as though the woman faces a completely open choice, she may well interpret the neutrally formulated information as a meta-message: 'You should have an operation.' This was the case with this particular woman; afterwards she claimed she had heard the doctor express that she should have prophylactic surgery. It is also common knowledge that when someone informs us of a possible solution to a problem, we often interpret this as a piece of advice or an instruction—as a plan of action (3).

Another problem connected with the misconception that it is possible 'simply' to give information is that the facts talked about, or the information exchanged, in a dialogue may have ramifications for people who are not present; for instance for a sister or a female cousin of one of the women. Information about oneself may entail information about someone else. Talks such as these can thus be said to involve more people than the actual participants.

Giving information—responsibility for the family

The doctor tries to construct the family tree—the genogram—by interviewing the women about their close relatives and their possible history of disease. The focus is on information about the woman's family and the doctor asks for the family's disease history, narrowing down family history to disease and cause of death.

One woman had shown me a photograph from a family wedding, a picture of a memorable day with happy faces in a close kinship group. When genetic researchers, seeking hereditary relationships behind various forms of cancer transform this group into a genealogy, they also extend it to include additional generations. Moreover, whereas the woman sees her family as real people of flesh and blood, on the genogram the doctor is talking about the family group in terms of abstract symbols; squares for males, circles for females and lines that connect them with their family of orientation (ancestors, grandparents and parents) and their family of procreation (children) (1). Although the wedding photograph of happy relatives and the genogram describe the same family, they differ greatly in what they convey to those who belong to the family on the one hand and to those who are studying the heredity of a cancer disease and its course within the family on the other. The woman is aware of the family's history in all its phases—births, weddings, deaths—and it is the real indi-

viduals that she knows, likes or dislikes, is related to in various ways, and what they have meant and mean to her in her life that constitute the source of her memories and her ultimate choices and reactions.

The topic of contacting other members of the family tends to be raised by the doctor for one of two reasons. The person is asked either to inform his/her siblings that they may also be at risk, or to contact family members for a blood sample. In order to analyse genetic mutations, researchers need blood from 'risk families'. In the next extract the doctor is informing the woman that her siblings share her situation.

> 'Did you ever talk about this? Because they are in the same risk situation as you, one could say.'
>
> 'Ehh, are they?'
>
> 'Yes, because you are all siblings the three of you, so to speak, and it is exactly the same thing there, couldn't you? … I don't know what sort of contact you have with them but why don't you mention that you've been … to them.'
>
> 'I could talk to them, if they are interested too.'
>
> 'Yes, now you know what it means and can explain it, and if it is the case that they … some don't want to hear anything about it but think it is best … "I don't have any symptoms, I don't want to do anything", and others feel that it would be a good thing.'

In that she has received certain medical information, the young woman cannot escape responsibility. She has to decide whether to give or withhold information. It is of course difficult to claim that we know she expresses resistance to the idea of informing her sisters. But one can say that a close analysis of the sequence in question indicates that she will go along with the doctor's suggestion, albeit reluctantly, without explicitly embracing the idea. What is not explicit in these informative talks is the nature of the relationships between family members.

One woman accounts for not knowing her family's health history by referring to family feuds, breaks between siblings or branches. She finds it embarrassing and difficult to ask relatives for information. She presents a story from a different perspective to the one suggested by the doctor's questions. The doctor is focusing on the distribution of cancer in the family rather than on reactions to the disease. The doctor wants to hear the story of the disease, while she wants to tell the story of a family.

This is not just a problem because of different needs of the parties involved when seeking to gain and give information in genetic research. There is also the dilemma in that many persons at risk never learn that such an option exists. The recontextualization of genetic knowledge can be likened to a continuous chain of interpreted meanings that the genetic researchers do not get to know about since this is a matter for each woman who seeks information. At the same time the information may never reach other women who are in the same risk situation. In one case, the woman asked her maternal aunt how she should use her new knowledge of being at risk of breast cancer.

> 'But the reaction I got was that my aunt says "you can absolutely not take this to the rest of the family, because everyone is feeling so bad about my sister being ill and perhaps dying of breast cancer. We cannot come out and say that there is also a risk for the grandchildren, and so on …" So because of that, the information had to stay there for now. And my brother reacted with "I don't believe in that.' He thought somehow that I was exaggerating but my sister-in-law got involved and she has talked to her daughters, who think that their father should have a test when that is possible.'

This woman became so absorbed by the fact that she was at risk of breast cancer that she decided not to do anything about others in her family at that time. She had to concentrate on her own prophylactic operation, her thoughts about the future and her own children.

> 'No, I don't think any further than my brother … it is us, isn't it. I don't think any further away or what happens on the other side where we have not had any contact. I don't think of them. It is enough to think about oneself and those closer to one … there isn't enough time and feelings to think about others. Moreover, a good many on mother's side are already dead. Mother has one sister and then there are … the children of grandmother's siblings and they are not so many either. I don't know our kin group all that well. We have met father's family a great deal but not with mother's family over the years, so we do not know a lot about them. We haven't met or seen each other at all.'

THE BOUNDARIES OF KINSHIP AND THE GENETIC FAMILY

A woman confronted by her genogram can hardly deliver an objective account of her kinship relations that will give the doctor a complete picture of the genealogy and her disease history. Moreover, other individuals who perhaps run the same risk as the woman in question may be left in the dark about this because they are not contacted or informed.

Information about potentially fatal diseases raises many questions about life and death. An individual's decision to give or withhold information about cancer can be interpreted in terms of either insufficient knowledge about the risk situation or bad or non-existing relations with the relatives in question. The women are aware that relatives may not know about the hereditary aspect of the disease at the time, and thus of the danger of withholding information. So there is no explicit allocation of blame or respon-

sibility when it comes to withholding information. But if information about hereditary cancer risk is to be transmitted to others, the responsibility for doing this is placed on the individual woman, not on the doctors. This is also what makes the whole question of imparting information dependent on individual kin relationships.

Consultations accordingly involve an exchange of information about cancer, for two purposes. One is to obtain information about the family's past and present health in order to assess risk; the other relates to future action and deals with whom to inform and to what extent once the risk has been assessed. For both purposes it is necessary to contact and inform unwitting relatives—to obtain information about the past or to obtain permission for a blood test. These two aspects are interrelated. What one sees on looking back can influence one's view of the future. The family's past health is a trajectory for future health risks. In the context of genetic diseases, members of a particular individual's family are automatically involved through the genetic network—they, too, may be potentially at risk of cancer. Asking for information about past diseases conveys something about possible future disease.

This is where the question of responsibility comes in. The woman's implicit responsibility for informing those who are close to her or others with whom she has no contact or whom she dislikes is a difficult dilemma. Who is likely to become diseased besides those who already are diseased or dead? Who runs the same risk as those already diseased and should they therefore be informed?

When the women obtain information through the genogram about their risk of inheriting a cancer disease, they begin to see their family in a new light. The consultations clarify the situation and the women realize who runs the same risk as they do, who are not at risk and that in this context, close and distant relationships refer only to blood ties. In this way the genogram can come as a shock, changing the women's appraisal of their own place in the family and conveying unexpected implications for close kin and other relatives.

The genetic knowledge colours emotional relationships in ways that may be deeply upsetting for the women. It also alters relationships with people whom they must consider and be responsible for in their personal decisions. This in turn may problematize the conception of the family and close kin (8). The knowledge interpreted by women about their genetic risks may also influence their relationships with persons with whom they previously had no contact, deliberately or otherwise, as well as with persons who have yet to be born.

As I understand it, at the time and place of this particular study, genetic information is intended, among other things, to give persons with a high risk a chance to deal with their situation in a rational, preventive way. For this purpose, the genealogies should be mapped as perfectly as possible. At the same time, research is being conducted with, it seems, the goal of documenting enough families with a high incidence of cancer of a certain kind in order to be able to trace the mutants on the particular genes. Since it is up to the particular woman to contact the kin group for this purpose, as well as for the purpose of prevention, the mapping is dependent on personal choices and emotional relationships.

The real-life complexity of such family relationships is evident from the descriptions I obtained from the women in this study. They illustrate the dilemmas that the genetic researchers, in their search for familial cancer disease, have to live with, since the information is sought by each particular woman, not by the researcher, the clinician or the medical institution.

While the new genetic information can provide individuals and society with constructive opportunities for preventing disease, it can also pose new problems. The project for communicating information draws the informed person into something from which there is no return; he or she can never return to the state of being non-informed. This may make people reluctant to assume responsibility for informing their loved ones, but at the same time they do not wish to withhold information that might save lives. It may also entail emotional dilemmas about people to whom they cannot relate because of feuds, divorces or other familial complications. Once imparted, information about genetic risks is there to stay and for many individuals it may be a catch-22 situation.

THE DECISION

The woman's decision on whether or not to have prophylactic surgery must be based on her interpretation of the information. There is no other way. Once the risk has been stated, the women have to live with this major concern. A couple of the women were exposed to conflicting medical opinions: the geneticists were in favour of a particular operation, while the surgeons recommended something more radical. One woman who tries to understand her options and to decide on whether to have an operation to reduce her personal risk is caught in a huge dilemma:

> 'The surgeons want to take away everything, womb, colon, everything … But am I free from risk then? And the genetic doctors got quite upset because they did not think that was necessary, that it was too drastic. So, it seems as if they are … that they don't agree … I had decided but now it is very difficult … I don't think the doctors understand what it is like to live with these thoughts. I think it feels very heavy to … this with my children and not knowing, and I feel very anxious … My eldest girl will start her first year in school this autumn and I think that if I cannot be there and think if … What happens, imagine if I soon get cancer.'

This very condensed extract from a series of long talks between this woman and me shows how the risk decision she has to make is virtually impossible. This has to do with her perception that the experts disagree, which prevents her from fully understanding and judging which alternative—surgery or not—is the more risky. Since she feels that not even the doctors can decide what is best for her, the information she receives becomes contradictory, unreliable and incomprehensible.

Despite such dilemmas in relation to the genetic information and tests to which they are exposed, women do choose prophylactic surgery and have healthy breasts and/or ovaries removed in order to eliminate risk (9).

Women who undergo genetic assessments or tests have to make a choice and need a strategy for continuing to live with risk. The main alternatives available today are frequent check-ups, and prophylactic surgery. The women relate to their options in different ways and their reactions can vary from autonomy to resistance, revolt, or surrender. The process they go through can be seen as a transition from high to lowered risk or from a belief that 'this is not happening to me' towards a belief that 'this is going to happen to me'. Two of the women became trapped in a vacuum where this transition become difficult.

STARTING TO LIVE WITHOUT RISK

The women gradually find a way of minimizing the difficulties of a situation in which new knowledge has entrapped them with no possibility of finding a way out. The genetic knowledge establishes emotional relationships for the women that cause upheavals in their lives and may be perceived as deeply disturbing. Their interpretations of the situation involve an invisible or tacit process of embracing a view that our society is slowly coming to accept. Besides yielding new knowledge about the physiology of man, scientific discoveries create new images of what a life is worth, how the body can be manipulated and how society can survey or control the health of its members. The medical advances seem to transmit a meta-message, a promise of eternal life in health, with all disease eliminated. The reactions of the women in this study and their way of coping with the situation suggest a tendency to regard surgery as a natural means of dealing with health risk. There is a gradual cultural change in the way we view the boundaries of the body and a human life. The women I have followed are a case in point.

However, the transition from regarding prophylactic surgery as something absurd or drastic to the acceptance of such an operation as natural takes a long time. Submitting to such a process as having healthy breasts or other parts of your body removed is scarcely feasible unless you are convinced that, in some sense, the choice is the right one. The women use various means to control their personal risk but in doing so they expose themselves to further dependence and medical supervision. At a time when assessments of risk in relation to disease are so uncertain, it may seem better to act than to wait for disaster to strike.

In this way the women's perceptions of personal risk incorporate the uncertainty felt by the doctors. The woman decides to remove the part of the body that is causing the uncertainty. The doctor can then feel that he/she is in charge of the situation despite the uncertainty about the capability of these operations to prevent cancer. For the women, however, the operations transform the earlier risks into a new state of illness, involving pain, scars that have to heal, complications of various kinds and years of adapting to something alien to the body.

One of the most important questions for these women is how genetic information and surgery can help them live without risk in the future. It is by no means certain, however, that their strongest feeling is one of relief. Here, finally, is what one of the women has to say on the subject.

> 'There is a risk that one transforms the risk into something else. That one cannot live without risk. That one does not dare to live life to its possible one hundred percent. It is like a coding of some sort, I think. It is extremely difficult if it has been instilled into you very early. I'm constantly thinking about my girls and whether they will have to go through this and whether they have this gene. So, there is this continuous thinking about other people, in a way. So, sometimes I think that if I had not been told this, then the risk would still be within me subconsciously, but not with them.'

Because the information is directed at persons who have had traumatic experiences in their lives, who have seen severe disease, suffering and death in their loved ones, their memories are incorporated in their experiences and in how they interpret the genetic information. They are healthy but are diagnosed as being at risk of future disease. As the last extract shows, the risk diagnosis imparts knowledge from which the woman cannot escape. She may have had an intrinsic fear of cancer for many years, but now that risk has been confirmed.

The consultations described in this study are examples of very complex communicative situations. Researchers may feel they can treat certain information in a very specialized, neutral and value-free manner. In clinical practice, knowledge emanating from genetic research has to be adapted by the doctor to a particular person's life. This information becomes embedded in the person's whole life situation; she cannot treat it simply as a piece of information, cannot 'just be informed'—it locks her into a risk situation. Deciding on how to deal with risk is a lengthy process for an individual and research on this process is urgently needed. This longitudinal study on the seeking and giving of information about genetic risk on the part of a few women raises questions that call for further ethical discussion among those involved in the practice.

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