Communicating genetic risk information

Theresa M Marteau

Psychology and Genetics Research Group, King's College, London, UK

It is envisaged that genetic information will be used, together with other types of information, to assess individuals' risks of developing a variety of common conditions. Such risk assessments will involve providing probabilistic information partly based upon results of genetic tests in order to facilitate behaviour change without causing excessive anxiety. The behaviours targeted for change are likely to include adherence to prescribed medication, alteration to diet, increasing levels of exercise and quitting smoking. This paper reviews research already conducted on perceptions of risk and genes, methods of facilitating behaviour change and reducing anxiety following various types of risk assessment.

Although risk assessment and interventions to reduce risks have been conducted for over 20 years, very little rigorous research exists. For the investments in the new genetics to be realised, research is now needed both in how individuals respond to risk information that involves the use of genetic information and in how to facilitate and maintain behaviour change to reduce such risks.

How best to communicate genetic information depends in part upon the objectives of providing such information. Where the information predicts the onset of an untreatable condition, such as Huntington's disease, the objective of providing such information is a good psychological adjustment: relief from uncertainty, planning for the onset of a disease, and avoidance of depression and suicide. Similarly, when genetic tests are used to diagnose fetal abnormalities, with the offer of termination of affected pregnancies, optimal outcomes are defined in terms of informed decision-making and psychological adjustment of the parents. Achieving these outcomes depends upon people receiving information and emotional support, traditionally labelled as 'genetic counselling', to help them make decisions about genetic testing and to adjust to their test results.

In the next decade, genetic information will be used in the diagnosis, treatment and prediction of common conditions, such as hypertension, diabetes and different cancers. A new taxonomy of diseases is being developed, based on genotype as opposed to phenotype, resulting in more effective and efficient development and prescribing of drugs. Genetic information will also be used as one of several risk factors in predicting disease prior to intervening to reduce risks. As the uses of genetic
information expands to include diagnosis, treatment and prediction of treatable conditions so the aims of providing such information will expand beyond psychological adjustment to include understanding of gene and environment interactions and risk, as well as facilitating behaviour change to reduce risks. This expanding array of objectives makes it clear that counselling is only one of the many skills needed when communicating genetic information about risks of common, treatable conditions.

Introducing genetics into the diagnosis, treatment and prediction of common diseases raises the question of what individuals should be told and how such information is best conveyed. There is already some evidence concerning effective ways of communicating diagnoses\(^2\), treatment\(^3\) and predictions of illness\(^4\). While little of this evidence has involved genetic information, it seems likely that many of the basic principles of communication hold good regardless of the type of information being used. Research over the next decade will be needed to investigate whether this is the case.

The focus of this paper is upon the psychological effects arising from the use of genetic testing to predict common conditions. Such predictions will involve providing probabilistic information, partly based upon the results of genetic tests, in order to facilitate behaviour change. This needs to be achieved without causing excessive anxiety. Literature relevant to each of these four aspects of predicting disease are reviewed as a starting point for the development and evaluation of effective ways of communicating genetic information in this context.

**Predicting responses to genetic information**

Responses to any information are influenced not only by the information but also by an individual's pre-existing perceptions. Individual's perceptions of risk and of genes form the basis for their responses to genetic information. Understanding these representations and how they interact with information about diagnosis, treatment and prediction provide the starting point for developing methods of communicating genetic information that result in the information being understood and which facilitate individuals acting to reduce risks, without experiencing excessive levels of anxiety.

**Perceptions of genes**

Beliefs that some illnesses, as well as an individual's constitution or vulnerability to illness 'run in families' are well rooted in Western cultures\(^5\,\,^6\). Heredity, for example, was seen as the major determinant of
health in a sample of the general population in France. In a UK study, heredity was seen as the third major cause of illness, after germs and lifestyle. Recent studies suggest that the public see many illnesses, character traits and behaviour as multifactorial in origin, encompassing genetic as well as environmental causes. A systematic review of 49 studies reporting causal beliefs for heart disease showed that the mean number of causes given was six, with the three most frequently provided being lifestyle (reported in all 49 studies), chronic stress (reported in 44/49 studies) and heredity (reported in 39/49 studies). What these studies do not tell us is how people’s causal beliefs interact. Thus, while most people perceive stress, heredity and lifestyle to play a role in the development of heart disease, how do they see the relationships between these causes?

Beliefs about the causes of a disease are important in that they affect beliefs about its controllability, thereby affecting emotional adjustment and motivation to engage in behaviour that might reduce risks. It is often assumed, erroneously, that providing individuals with information about risks to their health motivates them to change their behaviour to reduce such risks. It has been suggested that providing risk information based upon an analysis of DNA will also motivate people to engage in preventive behaviours. There is, however, preliminary evidence to suggest that emphasising genetic predispositions to illness may actually de-motivate people from engaging in risk-reducing behaviours by making them think that the condition is neither preventable nor controllable. In an analogue study, students were asked to imagine that they had received a positive screening test result showing that they were at increased risk for heart disease or arthritis. For half the participants, the screening test was described as a new genetic test which had detected a dominant gene for the condition. For the other half, the screening test was described only as a new test which had detected an increased risk for developing the condition. When risk to either disease was determined by a genetic as opposed to an unspecified test, the condition was seen as less preventable. Similarly, a recent study found that informing smokers of a genetic predisposition to lung cancer did not make them more likely to stop smoking, although it did make them more fearful.

Perceiving an illness as genetically-determined seems also to result in the condition being seen as more serious, possibly due to the perceived uncontrollability of genetically-determined conditions. Parents whose children had been identified as being at increased risk for familial hypercholesterolaemia following neonatal screening were interviewed about their experiences of the screening programme. Responses to screening seemed to vary according to perceptions of the underlying cause of the positive screening test result. When parents perceived the test as detecting raised cholesterol, the condition was perceived as
familiar, dietary in origin, controllable and not very threatening. When the test was seen as detecting a genetic problem, the condition was perceived as uncontrollable and hence more threatening, as illustrated by parents of two children who were identified as being at increased risk for FH:

_Heredity levels, as I understand it, cannot be affected by diet._
_So, you know, I feel as though this death sentence has been put on my little boy._

_It's the word heredity that sets all the alarm bells ringing – it sort of suggests that there is nothing you can do about it._

The extent to which these findings are attributable to risk assessments based upon genetic as opposed to other risk factors remains to be determined. A study is now underway in which individuals from families affected by familial hypercholesterolaemia are being randomly allocated to receive one of two risk assessments: one which incorporates genetic testing, the other of which does not (further details available from the author).

Research is needed to determine the content and structure of people’s causal beliefs about common conditions. This will serve as a basis for predicting responses to risk assessments that incorporate genetic testing. This will also provide the basis for developing ways of presenting the results of such risk assessments to minimise threat and to motivate individuals to reduce risks.

_Perceptions of risk_

Risk perception comprises two key components: (i) the likelihood of an adverse event; and (ii) the perception of the seriousness of the event itself. Given that research has concentrated almost exclusively upon the first of these components, the focus here is upon the likelihood of an adverse event. It remains to be determined the extent to which perceptions of seriousness contribute to perceptions of risk.

Individuals’ perceptions of risk affect their decisions about whether to undergo risk assessment as well as the likelihood that they engage in risk reducing behaviours\(^\text{15}\). Perceptions of risk are affected by the information an individual is given, as well as how individuals process probabilistic and threatening information.

Perceptions of risk and objective risk, although related to each other, are not synonymous. The extent to which they deviate and the reasons for this have been the subject of extensive research. The explanations can be broadly categorised as ones involving cognitive processes, and those involving emotional ones.

In making judgements about the likelihood of an event, people use a number of decision rules or heuristics not only because demands of daily
life force us all to become ‘cognitive misers’ but because the rapid answers provided by heuristics are often right. Three of the more common ones are:

- **Availability** (assessment of the probability of an event based upon the ease with which instances of the event come to mind).

- **Representativeness** (assessment of the probability of an event by judging the degree to which that event corresponds to an instance of that category).

- ** Anchoring and adjustment** (a general judgement process in which an initially generated or given response serves as an anchor and other information is used to adjust that response).

The tendency for people to overestimate the likelihood of rare events, such as plane crashes, and underestimate the likelihood of more common events, such as road traffic accidents can be explained by the availability heuristic. The rarity of plane crashes makes them newsworthy; their wide reporting makes instances easy to call to mind and hence to be judged more likely than they are. In judging the likelihood that someone may suffer from heart disease, an individual will judge how representative they seem to be of the perceived class of such individuals (e.g. overweight, under chronic stress), to the neglect of base rate information. The use of the anchoring heuristic is a likely explanation for the results of the studies reported below from Klein and Yamagishi.

In addition to using cognitive heuristics when estimating personal risks of adversity, people use other psychological processes in order to reduce the degree of threat the risk invokes. These processes result in risks being minimised. There is, for example, a well described tendency for all of us to think that misfortune is more likely to happen to others than to ourselves. When misfortune does strike, a common response is to minimise the seriousness of the event. So, for example, those found to have raised levels of cholesterol, rate having a raised cholesterol as significantly less serious than do those with levels in the normal range.

Risk information can be presented in many ways. Two key dimensions along which the presentation of risk information may vary are the quantification of the risk and the framing of the outcome.

**Quantification of risk**

Risks are presented to patients in a variety of ways: as relative and absolute risks, numerical probabilities and proportions, and using a large number of verbal descriptors. Risks presented as relative rather than as absolute risks tend to have a greater impact. For example, doctors are more willing to prescribe drugs if evidence of their effectiveness is presented in terms of relative as opposed to absolute risks.
experiment involving university students showed that emotional reactions to hypothetical risk factor test results were sensitive to different levels of relative, but not absolute, risk. Their judgements about safety were similarly influenced more by relative than by absolute risks. Students were asked to imagine that they had either a 30% chance or a 60% chance of causing an accident, and that this was either 20% higher or 20% lower than the average, same-age, same-sex person. Those told that their risk was lower than average rated themselves as safer drivers. Participants said to have a 60% chance of causing an accident when the average was 80% rated themselves as safer drivers than those told their chance of having an accident was 30% when the average was 10%.

Judgements of riskiness are also affected by the number of adverse outcomes presented, irrespective of the total number of individuals affected. In a series of experiments in which students were asked to judge the perceived riskiness of a number of events. Cancer was perceived as riskier when it was described as ‘kills 1286 out of 10,000 people’ than as ‘kills 24.14 out of 100 people’. Similar results were obtained for the other 10 causes of death. Other studies have also found that risk perceptions are influenced by the use of large numbers.

Little is known about the consequences of using numerical as opposed to verbal expressions of probability, such as ‘low risk’ or ‘high chance’. In a survey of antenatal clinics in England and Wales, the two most common ways of presenting negative or low risk test results to pregnant women who had undergone serum screening for risk of Down syndrome were as a verbal phrase (e.g. low risk, lower risk), reported by 43% of clinics, or as a verbal phrase together with a numerical probability, reported by 40% of clinics. The consequences of presenting risks in these different ways are unknown.

People prefer to receive information about probabilities of events in numerical form but prefer to express the probabilities of events to others using words, such as doubtful or likely. While some studies have found that providing numerical probabilities leads to better decision-making than the presentation of verbal probabilities (such as improbable, possible, almost certain), other studies find no difference in decision-making according to whether one or other type of probabilistic information is provided.

One of the aims of presenting risks in clinical contexts is to convey a likelihood of an adverse event, avoiding certainty either that an event will not happen (false re-assurance) or that it definitely will (fatalism), without causing undue anxiety. The relative merits of numerical and verbal probabilities in achieving these outcomes is currently being investigated in a study in which those undergoing prenatal screening for Down syndrome are randomised to receive results either using verbal or numerical probabilities (further details available from the author).
Framing of the outcome
While acknowledging the importance of probabilistic information, prospect theory states that, in making decisions, people begin by determining whether they stand to win or lose according to some reference point. Different presentations of factually equivalent information may change the reference point. Thus, when asked to state whether they would consider surgery given various probabilities of success for their hypothetical lung cancer, students, patients and physicians are more likely to choose surgery when the possible outcome is expressed as the probability of surviving as opposed to the probability of dying.

Much of this research on perceptions of risk has been conducted in laboratory settings involving judgements of events that are neither related to health nor personally threatening. Mood affects risk perception and behaviour in the face of risks. It remains to be determined how the negative mood states that often follow from informing individuals of increased risks to their health (see below) alter the processing and hence impact of risk information presented in different ways.

Facilitating behaviour change
Reducing risks identified from a risk assessment will very often require an individual to do something, such as take medication, alter their diet, increase their level of exercise or stop smoking. There is now a wealth of literature, including systematic reviews, evaluating interventions aimed at changing these behaviours using a range of methods in a variety of populations.

Multiple risk factor interventions
Many primary prevention programmes have been set up over the past 20 years with the aim of preventing heart disease through modifying cardiovascular risk factors. The interventions have mainly involved provision of information about risk and how to reduce this. The results of a systematic review and meta-analysis of 14 randomised controlled trials evaluating the effectiveness of more than one of several interventions aimed at stopping smoking, taking exercise, altering diet and taking medication to control hypertension and serum cholesterol, revealed modest changes in risk factors and no significant effects upon mortality. The authors conclude that efforts to reduce risks should be focused upon high risk groups and that health protection through fiscal and legislative measures may be more effective. More benefit may be gained from interventions targeted at just one risk factor.
Medication adherence
Low adherence to prescribed, self-administered medication is very common. In a systematic review of 13 randomised trials of interventions to increase adherence, Haynes et al. found that while adherence could be increased by certain, usually complex, interventions including various combinations of information, counselling, reminders, self-monitoring, re-inforcement and family therapy, research effort needed to be directed towards developing and evaluating new approaches to adherence so that the benefits of medication can be realised.

It may be instructive to consider the ratio of investments made in developing new medicines to the investments made in developing new approaches to ensuring that the medication is taken and hence its benefits realised. Haynes et al. comment that, with the astonishing advances in medical therapeutics during the past two decades, it is remarkable that only a handful of rigorous trials of adherence interventions have been conducted, providing little evidence that medication adherence can be improved consistently within the resources usually available in clinical settings.

Dietary change
Two recent meta-analyses of randomised controlled trials of dietary behaviour interventions show that individual dietary interventions in primary care achieve modest improvements in diet and cardiovascular risk status that are maintained for up to 18 months. While one review reports that the greater the intensity of the intervention, the greater the fall in blood cholesterol levels, authors of the other review commented that a low-intensity approach may be as effective as one involving considerable resources. Davey et al., commenting on these two reviews, consider that their most striking finding is the great variation seen in the effects produced in the different studies. There are, however, insufficient trials using similar approaches to allow sub-analyses to identify the characteristics of more successful interventions. Much might be gained by reviewing the effectiveness of behavioural, cognitive and environmental interventions effective in other contexts as a first step in developing further interventions to alter people's eating habits.

Tang et al. conclude that more research is needed to develop better methods of communicating dietary advice and maintaining adherence to such advice. Whether the changes achieved by individual dietary advice can more effectively and efficiently be achieved by population-based interventions remains a key question.

Increasing exercise
Activity levels in sedentary populations are most effectively increased by encouraging people to take brisk walks as opposed to more elaborate
forms of exercise such as working out in gymnasia or playing squash. In a systematic review of randomised controlled trials of interventions to increase exercise in healthy, free living adults, just 11 trials were identified, none conducted in the UK. Interventions that encouraged walking and did not require people to attend a facility were most likely to lead to sustainable increases in overall physical activity.

**Quitting smoking**

Law and Tang reviewed 188 randomised controlled trials of interventions designed to help people stop smoking. They found that advice and encouragement to stop smoking from a physician was as effective as behaviour modification techniques (including relaxation and the use of rewards and punishments), with about 2% stopping smoking. Advice and encouragement was particularly effective with high risk smokers including pregnant women (8% effective) and those with heart disease. Nicotine replacement therapy was effective in approximately 13% of smokers who sought help in stopping.

These systematic reviews of interventions to change behaviour make sobering reading. First it is apparent that, apart from smoking, very few rigorous trials of interventions have been conducted. From those that have been conducted, interventions have varying degrees of success, taken as a whole it is at best modest. The research portfolio is this area is thin, both theoretically and methodologically. It is, therefore, too soon to conclude that behaviour change is too difficult and costly.

**Minimizing anxiety**

Receiving information about increased risks of illness is associated with increases in anxiety. How much anxiety an individual experiences depends upon how the information is presented as well as an individual’s pre-existing level of anxiety and coping style. It is to be expected that individuals will react with concern and anxiety when informed that they are at increased risk of developing a disease. There is some evidence to suggest that a certain amount of fear facilitates behaviours aimed at reducing risks of disease, such as altering diet. By contrast, anxiety interferes with the ability to understand information and seems to inhibit behaviours that might lead to the detection of disease such as attending for clinical examinations or screening. Kash and colleagues, for example, found that the higher women’s levels of anxiety, the less likely they were to examine their own breasts or to attend for clinical examinations.
Several interventions have proved successful in reducing anxiety levels in those undergoing risk assessment, involving the provision of information and training in coping skills.

**Information**
Providing information to people before they undergo medical or surgical procedures reduces anxiety. The type of information presented is important in achieving this effect. The information needs to be sufficiently simple so that the majority of those receiving it can read and understand it and foster a sense of control over outcomes. The little research there has been on the effects of providing different types of written information given to those undergoing risk assessments, suggests that significant reductions in anxiety can be made using this relatively inexpensive intervention.

**Enhancing coping skills**
Providing cognitively-based interventions before risk assessment and after test results has been shown to be effective in reducing negative mood states in those undergoing HIV testing. Those undergoing HIV testing who received cognitive behavioural stress management were significantly less depressed after testing than those who received standard counselling. Those who tested positive for HIV and who received standard counselling plus stress prevention training were significantly less anxious than those receiving standard counselling. Stress prevention training (SPT) involved six weekly, 60 min individual sessions based on stress inoculation training and cognitive-behavioural treatments for anxiety and depression. The intervention proved acceptable to the HIV-infected cohort. In addition, relatively inexperienced psychology students were able to learn the techniques quickly and administer SPT with acceptable quality and adherence. It is possible that fewer sessions of SPT would be effective in risk assessment for conditions that are more treatable than HIV seropositivity.

**Counselling**
There are few studies of the type of counselling provided to those undergoing predictive genetic testing for uncommon conditions such as Huntington’s disease, inherited forms of breast and bowel cancer. While it seems highly likely that the pre and post test counselling provided has been important in contributing to the low levels of psychological distress reported in those undergoing such testing, there is no evidence concerning how important it has been and what have been the effective components. Lerman and colleagues evaluated the impact of individualised breast cancer risk counselling on distress about breast
cancer as well as general distress in women with a family history of the disease. Compared with women who received general health education, those who received the breast cancer risk counselling had lower levels of distress about breast cancer. Their general levels of distress were unaffected. Perhaps the most important finding from this trial was that women with lower levels of education showed the greatest reductions in distress. As in any trial, it is important to conduct subgroup analyses to determine those most in need of an intervention and amongst those, those who benefit most. This allows more efficient targeting of resources as well as avoiding what some consider a patronising policy that insists on ‘counselling for all’.

Providing a risk assessment service

It is clear from the preceding section that much research needs to be conducted to determine how best to present risk information and facilitate behaviour to reduce risks of disease. Before the results of such research are available, there are several models of population screening that can serve as the basis for risk assessments involving genetic testing. These include population-based screening for cervical and breast cancer and cardiovascular disease. The section below refers very briefly to just some of the issues that need to be considered when setting up these new risk assessment programmes.

The team

A variety of media are used to communicate risk information, including oral, written and video images presented in interactive systems. While geneticists will be part of the teams running risk assessment programmes they are unlikely to be needed to present risk assessments for common conditions to patients. Other members of the team designing risk assessments will include experts in the disease for which risk is being assessed, as well as behavioural scientists. Actual risk assessments may most effectively be delivered by individuals trained in risk assessment and behaviour change.

The training

It is essential but frequently neglected, for those presenting any form of risk assessment to be informed themselves about the meaning of the test and any possible test result. Insufficient knowledge about tests they are offering has been documented amongst midwives and obstetricians presenting screening for Down syndrome to pregnant women, and
Communicating genetic information amongst doctors and nurses working in oncology presenting predictive genetic testing for certain cancers. Telephone interviews with physicians in the US who had ordered genetic testing for FAP showed that over 30% incorrectly interpreted test results.

**The messages**

The information presented can affect interest in risk assessment and motivation to change behaviour. This raises the question of how far people should be encouraged to undergo risk assessment and to change their behaviour. There is a consensus in the genetics community that genetic information concerning prediction of conditions for which there is no treatment or for which termination is the main option should be presented in a non-directive manner, with no attempt by health professionals to influence decisions. Non-directiveness, however, is rarely defined or operationalised. Leaving aside the problems of defining non-directiveness, it is generally agreed that it is clinically appropriate to be more directive when presenting predictive tests for conditions for which there is effective treatment, such as familial adenomatous polyposis. Quite how directive is a matter of debate that cannot yet be informed by data.

Motivations of those running risk assessment programmes may affect the information that is presented, those motivated by financial profit perhaps presenting the benefits of their risk assessment in a more positive light than those not so motivated. A content analysis of 28 leaflets about CF carrier testing from commercial and non-commercial organisations in the US and the UK showed that leaflets from commercial organisations, as opposed to non-commercial ones, contained information more likely to lead to undergoing the test, e.g. less positive information was provided about life with cystic fibrosis, and the option of abortion was never mentioned. While information that is presented will need to be balanced, how should balance be judged: by opinions of health professionals, the public or by research on the consequences of providing the information, or perhaps all three?

**Concluding comment**

Expectations of the therapeutic gains from the new genetics are high. Realising these therapeutic gains depends upon how people behave when presented with genetic-related risk assessments. How can people at identified risk of disease be helped to alter their life style? How can adherence to prescribed medication be enhanced? Answering these
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questions is fundamental to the success of much that the new genetics has to offer. As this review shows, we are some way from being able to do this. As the mysteries of DNA are revealed, it is time to invest more in revealing the mysteries of people's behaviour when confronted with risks to their health.

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