"It's your right - but do you really want to know?"

Dr Yvonne Skinner is a consultant psychiatrist in private practice. She has held numerous positions on Government boards and committees in the mental health area. She was a member of the Pituitary Hormones Trust Account Advisory Board set up by the Commonwealth Government. You wouldn't know it from the title — with words like "trust account" in it, you think of other things — but the task of that committee was to consider claims in relation to psychiatric illness contracted due to patients' being informed of their increased risk of contracting Cruetzfeld-Jacob disease as a result of injections of pituitary hormones, which they had received in the 1970s. It was a pretty hot topic a few years ago.

Dr Skinner's practice includes assessing and treating patients who have suffered psychological reactions to being informed of the risk of contracting diseases or experiencing medical problems. This has led to her developing an interest in the ethical issues surrounding the disclosure of information to patients, balancing the duty to inform against the psychological impact on the patient of bad news.

Before we start, I wonder if you could all help me with a survey I am conducting. I just want to ask members of the audience to try to use their imagination, because I am going to give you a hypothetical case of something and would like you to try to imagine yourself in that position.

Try to imagine that you are about 22 or 23 years old, or if you really are 22 or 23 years old, you don't have to imagine that, but imagine the rest of the scenario. So just imagine that the things I say will affect you in your life. You have an unfortunate relative, John, now 42 years old. Your memories of him are very happy memories of an intelligent man whom you used to enjoy being with, who was fun and to whom you were quite devoted. He is a close relative, but your friends don't know about him because he has been in care in an institution for the last few years, because he is not able to look after himself. You went to visit him last year, but haven't been back because it was quite upsetting for you to see him dribbling and unable to walk. You are not even sure whether or not he knew who you were.

Then you go to your GP, who tells you that John's disease has been found to be hereditary. There is no treatment or cure for the condition, but they have found that it is hereditary. There is a test for which your GP can write you a referral to find out whether your risk of having the condition that John has is extremely high or quite low.

Please tell me: just put your hand up if you think that you would have the tests, and particularly if you think you would then go back to get the result. Thank you very much. That is the overwhelming majority. Is there anybody who wouldn't go? Thank you, a handful of people. That's very helpful.

The full paper, as Stephen Barnes said, will be on the website. I thought I might depart from that a little and talk about aspects of this topic that might be interesting to draw out. The title is "It's your right—but do you really want to know?"

Changing views about informing patients

Over recent years, social change, legal decisions and technical advances have brought about quite significant changes in provision of information regarding health status and the risk of contracting or developing a disease. Years ago, doctors generally thought that they should provide the patient with sufficient information to make a decision about treatment and about procedures they might undergo, but it was also thought that they should be careful not to give their patients too much information to avoid the patient's becoming too alarmed about the procedure or too disturbed by the information.

In 1993, the High Court dealt with the issue of informed consent to medical procedures in the case of *Rogers v Whittaker*, with which most of you are familiar. The Court's judgment altered the previously accepted view of the need to inform patients of risk. In addition, there has been a change in way health workers regard disclosure of information; there is now a trend away from paternalism, towards the belief that people need to be informed so that they are better equipped

to make decisions. A study of health professionals suggested that a sizeable minority would disclose information, including genetic information, whether the person wanted it or not. So it is possible that people who might choose not to undergo genetic testing might be persuaded to be tested, or might have the results thrust upon them whether they wanted them or not.

I think we have to be careful because informed consent is quite a complex concept. It must be considered along with a whole lot of things, including the patient's educational level, their capacity to understand and a number of psychological factors which influence decision-making.

The psychological impact of receiving genetic information

It has been shown that people generally over-estimate their coping skills and minimise the threat to themselves that a particular result might pose. In the case of genetic testing, people generally approach genetic testing with an over-optimistic view of the probable result. They think, "Well, it won't be me", often to the point of being quite unrealistic.

It is generally considered and accepted that people under the age of 18 should not be tested because they would be too immature to make a decision and to cope with possible adverse results. Some psychiatrists have been warning of the possible adverse consequences of genetic testing. Some scientists have responded by stating that the psychological problems are over-stated and that warning about psychological problems might reduce the use of what could be a very valuable scientific tool and might deprive people of needed information. Cultural factors also need to be taken into account because there is a very firm belief, in some cultures, that giving people adverse results might cause a deterioration in their condition.

Other people working in this area, statisticians and mathematicians, have suggested that the problem might be in the way we present the statistics. What does it mean to be told you have a one-in-twenty chance of having a child with a particular disease or that your risk of dying from cancer was 10 percent, but that, with this new test we have done, it is now 20 percent? Should we say that you have a risk of dying of 20 percent or that your survival probability is 80 percent? My difficulty with the problem is not mathematical, but rather about the way people interpret the figures and how people are going to make use of information about statistical probabilities.

Coping behaviour

When people receive bad news, they react by adopting psychological defence mechanisms. That is a way of coping with something. We try to protect ourselves emotionally and not get too stressed. So that we don't go into the spiral of depression, we have ways of thinking about things that make it better for us. These mechanisms depend on a number of factors, including personality type, family background and the dynamics within the family, especially a family which has this illness, and background factors that a health worker might not know anything about, for example that the person's friend died five years ago or went through a terrible lot of suffering before they got better. Each person's overall life experience is quite an important factor in the way they cope with bad news. So we can't predict about any individual person; even one who might seem to be strong and coping, we can't be sure that he is going to react in a certain way. Usually the immediate reaction to bad news is shock, denial, "No, this can't be right", and anger, "Why me, why does it have to be me?" However, there is a considerable range of reaction. The distress varies from mild to quite severe and some people do suffer continuing psychiatric disturbance.

Provision of information regarding health risks

Health providers often do need to advise about the risk to people when there has been some problem, like a contaminated blood transfusion or vaccine, where equipment has been inadequately sterilised or if a health worker is suffering from some kind of communicable disease. They would be expected to provide advice on what a person needs to do in checking for the disease and about any available treatment.

The New South Wales Health Department has notified people of the risk of Hepatitis-C from blood products and HIV infection from inadequately sterilised equipment. In 1994, the New South Wales Chief Health Officer notified 149 women who had been assisted in childbirth by a

particular obstetric registrar who was later found to be HIV-positive. None of the women tested positive. These women went through a lot of anxiety, although the risk of contracting HIV infection would have been very low. In fact, it would have been the first time in the world that such a thing had happened.

Pituitary hormone replacement

I would like to speak a bit about pituitary hormones because I was involved with so many people whom we dealt with as a board. I will tell you a bit of the background.

In 1985, the Commonwealth health authorities suspended the pituitary hormones program in operation since 1963. Although the department didn't notify the relevant patients directly, the Minister for Health issued a press release. He stated that the program had been suspended because some overseas recipients of pituitary hormones had died from Creutzfeldt-Jakob Disease (CJD). CJD is a neurological disorder characterised by rapidly progressing dementia, with muscle spasms, involuntary muscle movement and death.

In 1993, after a few deaths from this disease in Australia, the department became worried. They began to actively trace recipients to inform them that they were at increased risk of CJD because they had been treated with pituitary hormones, mostly in the 1970s and early 1980s. As a member of the pituitary hormones board, I talked to a number of these people, who formed a support group. We also received applications from people who felt that they had been very badly treated and that they were suffering psychiatric disability because of being informed of this risk.

The first problem was that there was a lot of confusion about the information they received and about the way they received it. They didn't know what was going on, they didn't know much about the disease, and they were very worried about the risk of transmission to other members of their families. If there was a risk, how do you stop it if they hadn't already transmitted it? They tried to get information but just couldn't find it and could not understand what was happening.

Many families went through quite a lot of disruption at the time. There were all sorts of problems: people who had not told a partner, perhaps a partner they intended to marry or a new marriage partner, that they had ever received the hormone; and there were some parents who hadn't told their children that they had received the hormones as children. Some of the children felt guilty because they felt that their parents had used this treatment to help conceive them. Some of the parents felt very bad because they had requested the treatment for their children for short stature. They felt very bad because they had thought they were doing their best for their child and now the child was faced with this terrible risk.

Other things they complained about included the embarrassment of being excluded from being blood donors. Some who had actually been blood donors thought that they might have transmitted the disease. They complained about a lot of other problems as well. For these people there was no investigation. You couldn't go and have a test to see if you had it. They were just told that, at some point in the future, they might suddenly develop this disease. Most people reported a degree of anger, depression and rumination about the possibility of contracting this terrible disease. Some were diagnosed with psychiatric illnesses: depression, anxiety and obsessive compulsive disorder being the commonest. Some got better, some had episodic illness and some had continuing illness. As I said, some felt fear when this news was made available because of all sorts of family complications.

It seemed to me also that some people who complained about not receiving these services were people whose treatment had been unsuccessful. I remember one man who had had treatment for short stature as a young adolescent and then again later in life for infertility. He was in fact quite short and had no children. So he had had all this treatment with a poor outcome and was then he told that he might develop this terrible disease later in life. Talking to some of these people in the original group who were coping and who didn't actually put their application to the board, they say that even now, many years later, they are still thinking, "Well, is this a possibility? Is this going to be the final outcome for me?" So there is still a threat there and every time they get some symptom or they stumble, they think, "Oh no, is this a sign that I am getting this disease?"

Genetic testing

I would like to talk a little about genetic testing. I will talk about Huntington's disease for two reasons: it is a disease for which genetic testing has been available longer than for any other genetically inherited disease; it is also an autosomal dominant disorder. That means that, if you have the mutant gene for Huntington's, then you will get the disease, and if you don't have the gene, then you won't. The onset is usually in the fourth decade of life. There is no treatment. That is the sad point.

The disease is linked with mild involuntary muscle spasms. The person usually presents with fidgeting, twitching sort of movements; accompanying that is a loss of judgment and sometimes mental changes. Over time, the movements becomes quite bizarre, wildly throwing out an arm, knocking things over or punching somebody. As they try to walk, they sometimes seem to suddenly dance off in another direction. It is quite bizarre, and is accompanied by gradual dementia, leading to deterioration where the person can't look after himself, and eventually death.

I remember when I was working at one of the hospitals in the late 1990s with ten patients with Huntington's disease. All the relatives came to visit them, but no relatives, zero percent, would agree to have the test. A lot of staff were very surprised and shocked that they didn't want to.

Generally in the most developed countries, about ten percent of those who have been offered the test have accepted it.

Psychological studies have been conducted on a self-selected group out of this ten percent. Part of the reason for this is the importance of people's psychological reactions and the risk of suicide. Were people becoming very depressed or suicidal after testing? Anyone in the group who had suffered depression or had suicidal thoughts was excluded, and the group was followed up for a maximum of 12 months. Personally, I think that we should be doing longer term studies to study the long-term effects.

The reasons given by the relatives who decided to be tested were that they wanted certainty in their lives, they thought they would like to plan for their future and that they could inform their children. If they didn't have it, then the chance of their children having it might be reduced.

Reasons given by the 90 percent who decided not to undergo testing were the emotional and psychological consequences they feared should they have a positive result; they feared that they would be checking all the time for symptoms and interpreting any symptoms as an indication of early stages of the disease; and they feared losing hope – "Well, there is no effective treatment, so basically what is the point?"

They were concerned about insurance. I read about one person who had been checked immediately and who was arguing with an insurance company because they wouldn't offer insurance. Family considerations are important. You might think that, if you had a family with three children and two of them had negative results, then that would be very good and everybody would be very happy, but in fact that can cause quite a lot of problems because if one of them is positive and the other two are negative, you get all sorts of conflicts between siblings and the parents feel guilty. This can lead to all sorts of family disruption.

There was a fear of increased risk to children and how they would cope if they were found to be positive and could be a carrier. A lot of people thought, "Well, I probably am. Even if I didn't have it, I would just go into a downward spiral". One of the things that surprised the investigators was that non-carriers became quite depressed. There were all sorts of reasons, like guilt and all sorts of family dynamics and other factors that might have been in play, but some of the people you would expect not to be depressed were.

Genetic testing for assessment of risk of common diseases

The other type of genetic testing we do is for risk of a common disease. These are not usually done unless there is a family history and a concern that, because of the family history, there is increased risk. For some conditions, like bowel cancer and breast cancer, we can test to see

whether or not a person is in the group with the increased risk.

However, this is where the difficulties of applying statistics to individuals come in. Although some will be in the increased risk group, it doesn't mean they are necessarily going to get the disease. If there is a strong family history, for example, of breast cancer, the test might show that the risk is 80 or 85 percent that that individual will get the disease, but there is still a 20 percent chance that they won't. If the result is negative, it doesn't mean they won't get it, it just means that the risk for them is the same as for the rest of the population, which is still a risk, so they still might get it. The people who undergo testing are, of course, already aware that there is the possibility of increased risk because of the family history.

The people who are offering the test hope that the person - if they are willing to do this test and are sensible - would say, "I have got this increased risk, therefore I will follow all the medical advice." Advice offered includes increased screening, taking hormones or having preventive surgery. Again, the research is interesting. Although 80 percent of people who had these strong family histories and who were offered a test and had said, "Yes, we will have a test", less than fifty percent actually went ahead with the test. The other interesting thing is that none of the studies has shown that the people at greater risk have gone into increased screening and testing or have changed their lifestyle as recommended, like stopping smoking, going on a high fibre diet or doing more exercise, lifestyle things you would think a person with this increased risk might consider. The other finding was that some people who were found not to be at increased risk thought that they were not at any risk at all, and so didn't have to worry about checks. That was another of the worries.

The researchers found that a lot of people aren't going to help themselves and change, even people who were at high risk, and they found that family dynamics were important in this context because families would argue with one another about who was at high risk and "Why should you have this special attention?" There was a lot of argument and fighting.

Before concluding, I just want to ask you the question I asked in the beginning. Has anybody changed their mind about whether or not they would have testing? None.

I have given you an outline of some of the psychological reactions people might have when informed about their risk of contracting disease. The current practice of health providers' notifying about disease suggests that they will continue to advise. I think that, if treatment is possible, then they should know the results of testing, so that they can determine what to do. I still question whether, with something like the Creutzfeldt-Jakob problem, which we hope won't come up again, but if something like that came up again, how should that be handled and, if no treatment is available, whether advising everybody who might be at risk is going to be helpful, and what should be done?

Future developments

Further developments in genetic testing will allow determination of risk for many diseases. It is a very rapidly developing area. There are all sorts of diseases being investigated so that genetic tests can be done, including some others like Huntington's disease.

Of more concern, I think, is the development of tests which are actually already available, for behavioural or personality traits. There are tests that can determine, for example, the genetic trait of aggressiveness, and for psychiatric conditions such as depression or anxiety, which can be inherited.

At present, it is very widely believed that people under the age of 18 should not be tested, for all sorts of reason. But I wonder whether, in the future, people are going to consider testing children and what they are going to test them for, and how parents are going to react to proposed treatment when the children have tested positive.

If found to be positive to some disease they will acquire in the future, will they be treated differently from their siblings? Will parents want to have their children checked for traits such as aggression? If they are aware of these kinds of personality traits, how will they treat their

children? How will it affect their attitude to the child's upbringing? I wonder, if someone was told that he had this genetic trait of aggression, if this would be a good excuse for bad behaviour and become a self-fulfilling prophecy. I think the problems in relation to children need to be thought about.

I think we need to balance the provision of information against the possible risk of psychological harm to the person. If we find it impossible to predict those who will be adversely affected, we can't just say that because this hasn't happened before, that this is not going to bother them. We have to think more and do more studies about the long-term psychological consequences of providing information.