INNOVATIVE HEALTH TECHNOLOGIES PROGRAMME

The Construction of Risk Estimates in a Cancer Genetics Clinic

KEY FINDINGS

The application of modern genetic knowledge in health services has transformed the way we think about health and illness. For example, new technologies of predictive testing have served to underline the importance of future states of health rather than current pathology. They have shifted the clinical 'gaze' from a study of symptoms to an analysis of risk, and they have given emphasis to methods of long-term surveillance rather than short-term medical intervention. The consequences of these and other developments - for health professionals, for patients, the general public and the NHS as a whole - are not fully understood. This project sought to investigate some of the pertinent issues.

RESEARCH FINDINGS

The 'field' for the research was an NHS regional cancer genetics service (CGS). We examined the ways in which patients got referred to the service, how clinicians decided what level of risk a patient might be at, how they communicated relevant information to those who were at moderate or high risk, and what 'being-at risk' meant to the patients-clients of the service. We found that:

- Many GPs who refer patients to the genetics service routinely omit from their letters standard information that is required in order to assess risk. The most common errors were omissions of relatives' age of death and diagnosis and omissions of whether relatives were from the maternal or paternal side of the family. At the same time GPs often include data on other aspects of a patient's background especially references to psychological states. This suggests, perhaps, that the GP use of referral is best understood in terms of 'situated actions' than general principles, and it may be that GPs do refer 'appropriately' in terms of their own systems of relevance.
- Professionals within the CGS may also set aside or re-interpret published guidelines in their deliberations on risk. This can be done for a variety of reasons including recognition of interesting cases, borderline cases, patient anxieties, or because of a feeling that the guidelines simply did not fit a particular case. Why such actions occur again demands a study of situated actions rather than general principles.
- The above considerations have implications for the deployment of computerised decision-support devices in general practice. For whilst such devices might sharpen the process of risk referral it is clear that they will not be able to substitute for the expert knowledge that is routinely called upon in clinic deliberations.
- People deemed to be 'at risk' often feel unsure about their status within the health care system, worrying that they are neither sick nor healthy. Such a state of 'liminality' can have profound effects on how they seek to position themselves within the health care system.
- People deemed to be at 'low-risk' of inheriting a familial cancer often seek to get such assessments revised (upward), whilst people deemed to be at high or moderate risk often accept such news with equanimity.
- Why low-risk patient experience difficulty in coming to terms with their risk assessments is not clear. However, it seems likely that differences between the ways in which professional and lay people 'calculate' risk, differences in their understanding of what the genetic causation of disease implies, and differences in the assessment of familial health needs are implicated in the acceptance of risk estimates.

RESEARCH TEAM

Professor Lindsay Prior, Cardiff University & Queen's University, Belfast
Dr Jonathon Gray, University Hospital of Wales
Professor David Hughes & Dr Stephanie Adams, University of Wales, Swansea
Professor Roisin Pill, University of Wales College of Medicine
Dr Fiona Wood & Dr Susie Scott, Cardiff University

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Patients 'at risk'

Genetic knowledge has transformed the way many health care services are delivered. It is no longer only those who are already suffering from disease who fall into the orbit of NHS care, but also those who are deemed to be 'at risk' of developing heritable diseases including some breast, ovarian and colorectal cancers. The patients in our study had been referred to a regional cancer genetics service by GPs or other specialists because a close relative or because several successive generations of relatives had suffered from (breast, ovarian, colorectal or other) cancers.

While social scientists have written a great deal about discourses of risk in the context of health, the question as to how professionals assemble risk estimates and what the consequences of 'being-at-risk' might be are issues that have been relatively under-researched. Our study focused on these both sets of issues, and our results demonstrate how health professionals construct beings-at-risk, and how patients or clients integrate knowledge of risk into their everyday worlds.

Identifying the at-risk population:

Identifying people at 'low', 'moderate' or 'high' levels of risk of inheriting one of the familial cancers is a complex process. It begins with a referral. Referrals have more than tripled since the CGS was first opened in 1999. To study specific aspects of the referral process we analysed 100 GP referral letters, undertook interviews with around a dozen GPs, and gathered 'naturally occurring' data from CGS referral meetings.

Written guidelines containing criteria for recognising the 'at-risk' are available to health professionals. Such guidelines emphasise the importance of noting the ages and number of family members with cancer, rather than the social, psychological, or biological problems of the presenting patient. However, we noted that referring GPs often tended to place more emphasis on the latter than the former, and even within CGS itself reference to the psychological states of people being referred could form an occasion for retaining an apparently 'low-risk' patient in the clinical service for further investigation. We further noted that health professionals in both secondary and primary care often referred to their 'clinical experience' as the foundation for practice, irrespective of what guidelines might recommend. In fact, the deployment of this and other forms of 'personal' knowledge emerged frequently throughout the service. (See, Box 1).

Identifying the at-risk person

One of the key tasks for health professionals is to translate data about risk derived from populations - epidemiological data - to individuals. In order to complete such a task, professionals commonly call upon a wide range of low-tech and high-tech devices and implements. These include, for example, computer drawn family histories (pedigrees) and computer calculated risk estimates. (See, Box 1).

Box 1: THE DEPLOYMENT OF EXPERT KNOWLEDGE AND THE ROLE OF DECISION-SUPPORT DEVICES IN THE CONSTRUCTION OF CANCER GENETIC RISK ESTIMATES
CG = Clinical Geneticist NC = Genetic Nurse Counsellor

CG2: Yes, the interesting thing is if you really start to tease it apart there are lots of black lines all over the place, they are all on different sides of the family. This is her grand-maternal's, er (1.0) niece. 40s

NC1: That's 3rd degree

Referring to family history (pedigree)

CG2: Well that is 3rd degree, yeah. And then her (0.4) well, her mother's grandfather's sister at 67, so I think we can discount that one. This is the one that is of more concern. She has a sister at 35 and then somebody else at 38 over here. So there are two young people and I suspect that puts her into a high oh! 24.6 percent. (1.0) Mm.

Computerised decision -support device suggests moderate risk. High risk > 25%

NC1: What did you think, because you had some good thoughts about this one?
NG2: (2.0) Erm.
CG2: This is one that I would put into a high-risk group. Can you think why?

Reject the support device. Use expert knowledge

It is sometimes argued that, given the existence of such programmes it may be possible to 'download' the early stages of cancer risk assessment to primary care. Such downloading may indeed serve to sharpen the use of referral criteria (see above), but it is clear from our work that the interpretation of pedigree data often necessitates the use of tacit (specialist) knowledge that cannot easily be codified. (For example, different risk assessment programmes integrate key variables into their calculations in very different ways ways that call for expert interpretation).

For people at a suspected high-risk of inheriting mutations, the possibility of blood testing and mutation identification arises. In the case of breast and ovarian cancer two highly penetrant genes have so far been identified (BRCA1 and 2). Their use in NHS lab investigations raises interesting issues of (global) intellectual property rights. At a practical level the exploitation of the patents raise interesting issues about the uncertainty of current genetic knowledge. For example, the difficulty in identifying which if any of the hundreds of BRCA1 mutations may be present in an individual sample. Further, it is clear that these 2 genes cannot possibly account for the known extent of familial cancer risk. Consequently, identifying risk via a family history can often be more reliable than identifying risk via a laboratory sample. However, it seems that this is neither an easy nor a popular message to 'sell' to certain audiences including sections of the general public.

Box 2

Lay notions of inheritance often can differ from those of professionals

P07: I think you inherit most of your genes from your mother, definitely… because most children, most daughters, well most children have the same blood group as their mothers don't they. So obviously that is a big factor. And regards looking like my mother, I look like my mother and my father, so I couldn't really say that to be honest. But obviously I think it is a hereditary thing. There is no way about it really.

Lay assessments of risk often differ also

P19: I mean I sort of, you know you read magazines and on the TV and they talk about 1 in 3 or 1 in 5 people in the population. And then you do a quick calculation in your head and there is my mum, my uncle and my auntie. And so that really puts me higher than 1 in 3. So I reckon I have got about 50:50 chance.

The role of 'genes' in disease causation is often understood to be mediated by environmental factors

P10: (Assessed as high-risk). My mother grew up in quite a large family. There was 9 well there were 10 of them, but one was killed. And the day that war broke out they were moved into a new council house in the village where they lived. And towering over this council house was this pylon... I mean we spent ages as kids swinging on the pylon. Now my auntie… insists that they have all got it because of this pylon. She never lived at the house and that is why she hasn't got it, and that is why she is never going to get it.

Which may be partly why being assessed by professionals as 'low-risk' can be disconcerting.

Interviewer: And how do you feel about [being assessed as 'low-risk']? Do you feel reassured?
P29: No. Not at all… so many people in my family all on my Mum's side and all blood related, you know. You know there are just too many. That puts things... you know reading a letter which says 'you are not in a higher risk' does not put my mind at rest at all. Not at all… I don't know, I was waiting to speak to you to see if there are any roads that I can go down to see whether I carry this gene.

Though many of those assessed as being at 'high-risk' are 'glad'

P41: You say what do I think about it [i.e. being assessed at high risk of inheriting a mutation]? Yeah I am glad.

A stance that demands explanation in itself …

Lay reactions to risk assessment

It was clear from our interviews with users of the CGSW that they too performed risk calculations. Interestingly, they often did this by focussing on facial and other forms of bodily resemblance with family members. Yet, however calculated, it was evident that where lay and professional risk assessments coincided users were more pleased with the CGSW. On the other hand where risk assessments diverged, users could be somewhat disgruntled. Divergence was most likely for the low-risk clients. It is not immediately clear why this is so. However, our data suggest that what clients focus on in a risk assessment are the resources that follow the risk screening, possible testing, advice and counselling rather than the risk itself. As a result, high-risk clients often expressed contentment at the chances of being 'looked after', 'kept an eye on' and 'checked out', whilst low-risk clients often expressed ideas of being rejected and excluded from surveillance procedures. For some, being low risk meant that they 'were not even patients'.

It seems evident that genetic risk assessment procedures for late onset disorders can significantly redraft a person's subjective health status. Such redrafting can have important consequences for the perception and demand of health service provision. It also has consequences for our concepts health and illness, the sick-role and the nature of the 'patient' in the modern world.

About the Project

This study was one of twenty projects funded by the ESRC in Phase 1 of its Innovative Health Technologies programme. Our research used mainly interview and observational methods to examine the ways in which risk estimates are constructed, communicated and understood. We contacted ninety-two consecutive patients who had been referred to the regional cancer genetics service and sent them an information leaflet about the study. Fifty-eight of these people agreed to take part in an in-depth, face to face interview, in which they were asked about their experiences of receiving a risk estimate and how the new genetic knowledge had affected their everyday lives. Most of these interviews took place in the patients' own homes, lasted an average of 45 minutes and were audio-recorded. Nineteen of these patients, most of whom had been categorised as 'high' or 'moderate' risk, had been invited to attend a clinical consultation with the CGS team, and we requested their permission to sit in on and observe these appointments. We also observed sixteen of the 'risk review' meetings and twenty-one of the 'referral' meetings that were held by the staff before the patients arrived. These two main methods of data collection were complemented by seven days of observation in the laboratories where screening and predictive testing took place, and a content analysis of 100 referral letters to the CGS and 12 telephone interviews with the general practitioners who had referred patients to the service.

For further information contact:
Professor Andrew Webster, IHT Programme Director
Department of Sociology, University of York, Heslington, York YO10 5DD
Tel: +44 1904 43 3064/4740 ◆ Fax: +44 1904 43 4702/3043 ◆ E-mail: iht@york.ac.uk
Web site: http://www.york.ac.uk/res/iht/