Haemochromatosis: Susceptibility and Risk

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The construction of risk estimates in a cancer genetics clinic … and their implications

Aims of Haemochromatosis study:

- To study the consequences of population screening for genetic disorders based on the case of haemochromatosis.
- To study how risk of haemochromatosis is being understood and managed by both medical science and its users.

Aims of Cancer Genetics Study:

o To study the processes involved in assembling cancer genetic risk

o To study the implications of such processes for professionals, patients, and NHS.

o Field of study - breast, ovarian and colorectal cancers

Cancer Genetics Research Data and Sites:

Referral Meetings (when professionals first consider a possible ‘case’) (N=20)

Review meetings (when professionals review data on genetic risk for individuals and categorise them as low, moderate or high risk) (N=16)

Interviews with users of the service (‘patients’/clients)(N=57)

Consultations between professionals and clients (N=26)

Also Laboratory Observations + Videoconference materials + 100 referral letters + GP interviews

Haemochromatosis: Research Data & Sites

In-depth Interviews with:

- Haematologists and clinical geneticist [8]
- Blood donor homozygotes [11]
- Affected patients (from Cardiff, Cambridge, Southampton, and members of Haemochromatosis Society) [22]
- Ongoing Data Collection:
- Blood donor homozygotes (Cardiff) [10]
- Affected patients (Cardiff and Cambridge) [14]
- Members of Haemochromatosis Society [22]

Interpretations and management of ‘risk’ a key focus for both studies

Risk assessments central to clinical genetics. Such assessments determine the flow of resources to patients.

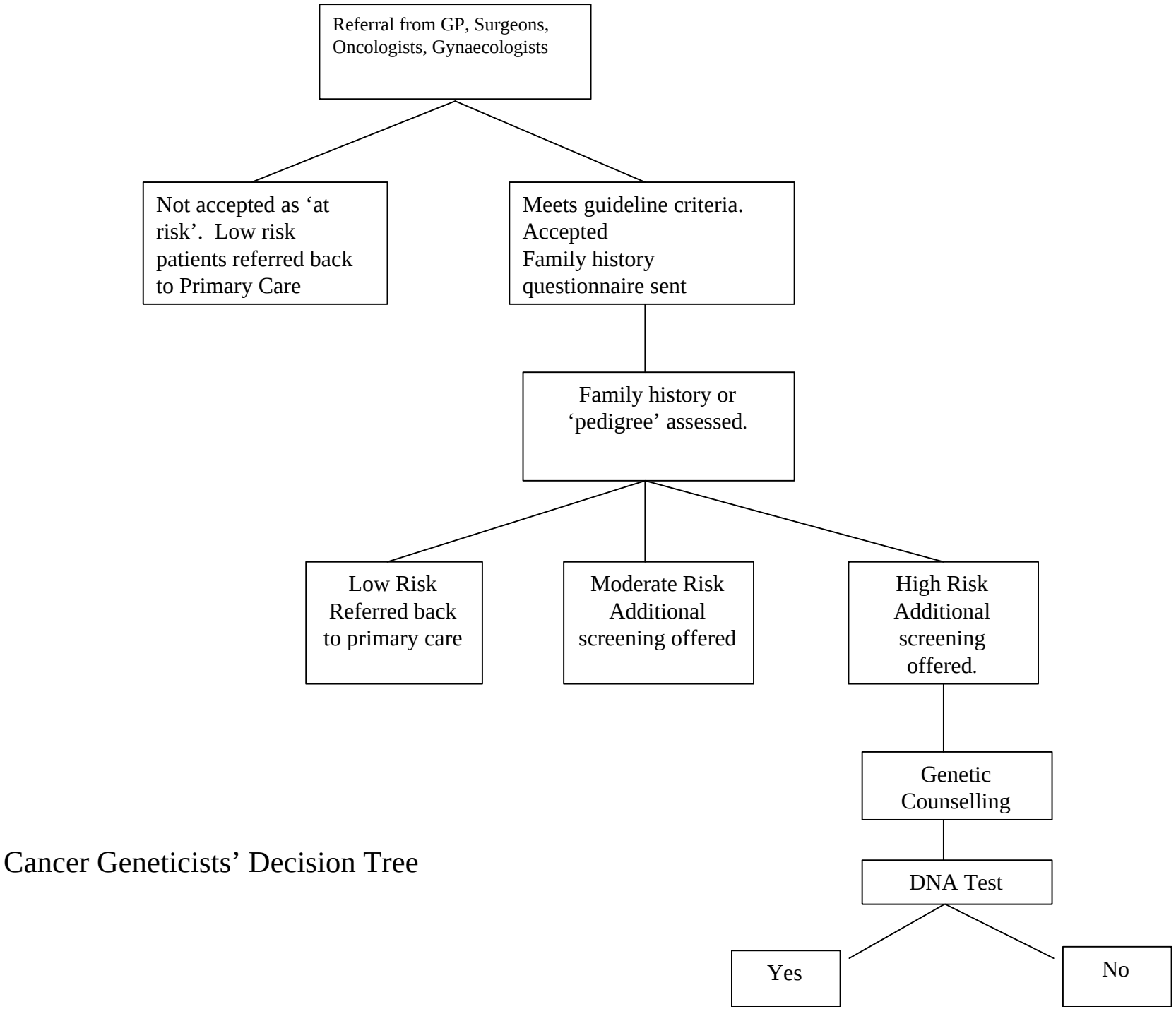
Yet risk assessments are seldom about certainty (more often involve degrees of uncertainty)

Assessments are malleable and variable - difficult to stabilise

C.f. White Paper (*Our Inheritance, Our Future: Realising the Potential of Genetics in the NHS*)

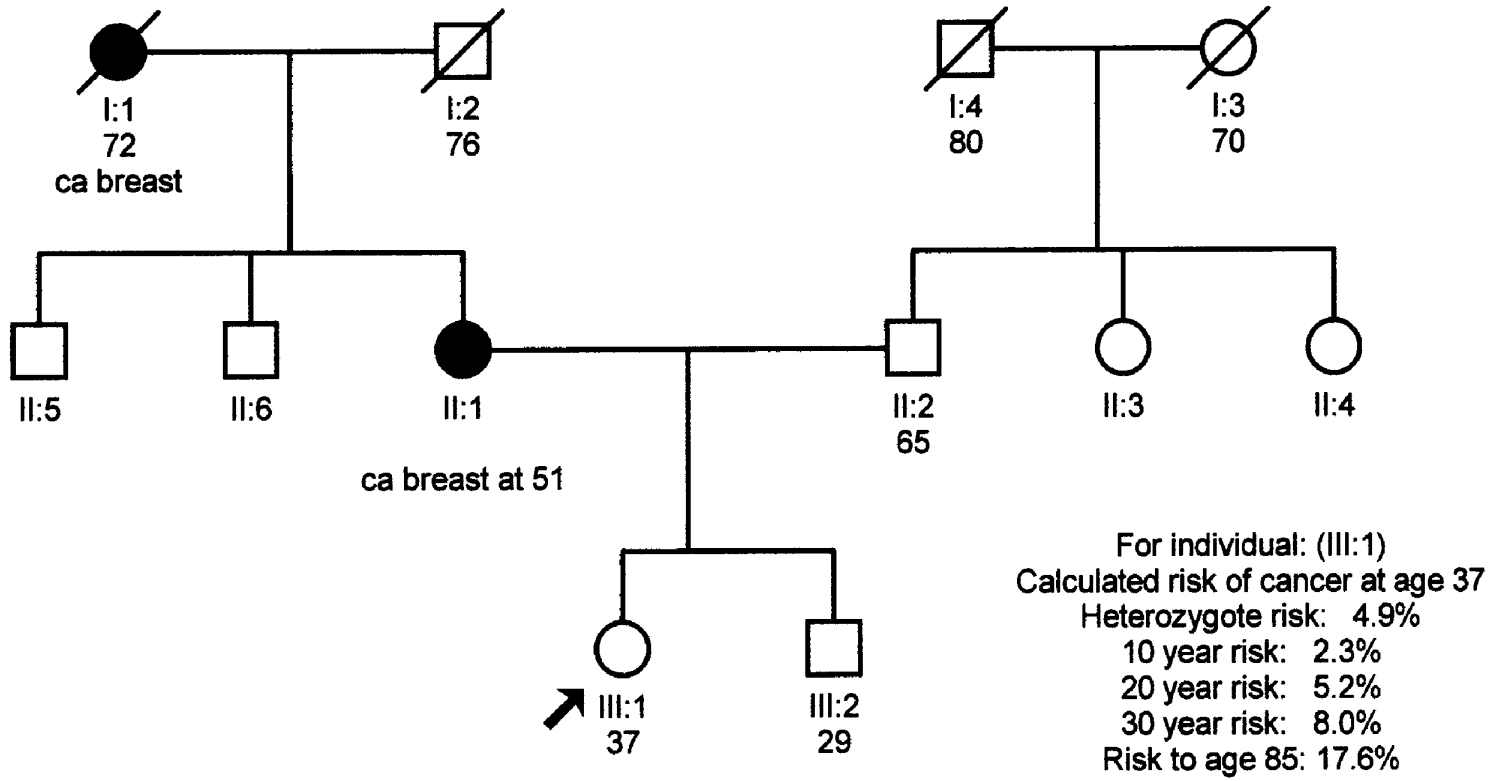
The Referral Criteria for Breast Cancer

- 1 first degree relative diagnosed at 40 years or less
- 2 first degree relatives at 60 years or less
  (on the same side of the family)
- 3 first or second degree relatives any age
  (on the same side of the family)
- 1 first degree male breast cancer
- A first degree relative with bilateral breast cancer



Cancer Geneticists' Decision Tree

A Pedigree Drawn (and the risk calculated) by CYRILLIC



So what do the clients make of such assessments?.....

Lay people also make risk assessments.

They tend to do so on the basis of family history - but use different styles of calculation

“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. [assessed by clinic as moderate]”

When lay and professional risk assessments coincide patients are more likely to be satisfied.

Even if the ‘news’ is not apparently very good...

FW: Mm, so how do you feel about that, being told that you are at moderate risk?

P16: That is quite acceptable I think. ... It just... it makes me... I am glad because for me to be told that I will be monitored at the age of 40 is great..... its nice to know that there is that support, you know you will be slotted in there and you will be looked after or whatever

Similarly a Haemochromatosis homozygot:

AB So you are being monitored from time to time?

JS I was told I was told I was.

AB Yes.

JS Whether I am or not I don't know but in my little mind I'm happy knowing that Big Brother is out there checking my blood for me when I'm not looking.

AB Right because you're a regular blood donor, you donate every now and again?

JS Yes so I think as long as I'm blood letting, that is the cure or the medication whatever you want to call it, I don't know the term, for the symptom is blood letting. Well I'm doing that every 4, 5 months anyway so I feel happy in myself that this blood letting process is going on. Somebody up there, this unseen person is monitoring may be and they told me they are so I'm happy to let them carry on.

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As with cancer genetics patients, haemochromatosis patients identified at risk often develop a false sense of security because they are being monitored.

[...] But I mean I, the last mammogram or the last but one I had I was called back. Well it didn't stop me panicking over that you know. I didn't really, I know they say don't panic but you do until you get the all clear sort of thing. So I think they [haematology clinic] must've just been very good at not, at you know allaying your fears about it because they haven't, they've definitely given me the impression that as long as I do this [routine clinical bleeding] I haven't really got anything to worry about. Now I don't know if that's a false impression but that's how you know I sort of see it. That as long as I do this you know it won't, and I'll ask things like that, may be I have been under a false sense of security.

However, being deemed to be at ‘population’ risk or low risk (or no risk) is not always welcome, and can generate a sense of dissatisfaction - this is specially so when professional and lay assessments of risk fail to coincide

FW: Does the fact that they have told you that you are [low risk], has it reassured....?

P22: No it hasn't no. I don't know. I suppose I had it in my head anyway that I think I am at higher risk, because it is just so close family. I

FW: And how do you feel about [being low risk]? Do you feel reassured?

P29: No. Not at all. Its just seems to be too much co-incidence that so many people in my family all on my mum's side and all blood related, you know. My Nan, her brother and her sister died. My mum's sister. 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.

A Haemochromatosis patient:

AB Do you feel your mood is sort of on a different plain when your ferrotins are higher than normal?

R Yes, yes.

AB Do you feel that there is some sort of correlation?

R Yes.

AB You feel more depressed or more irritable when there is saturation of iron

R Yes, yes

AB That's interesting and he's not able to sort of pick on that?

R No he just nods his head and says, looks at his computer and says "Ah well the results are doing this," and I say "Well look I'm feeling like this, this and this," "Well it might be that, it might not," but I don't get any information. So I suppose a lot of the information that I've got has been from the internet and my own GP...

Some implications ..

Risk status determines ‘resources’ made available to patients.

Patients tend to focus on the resources rather than the risk status and can gain a false sense of security from being ‘monitored’

Where there is disjunction between patient and professional assessment of risk there tends to be dissatisfaction. (Disjunction most common in ‘low risk’ assessments).

As a result, patient and professional views about ‘what is needed’ in the clinical service often diverge