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Lindsay Prior
Using technology to transfer expertise.
Problems and potential in cancer genetics.
E·S·R·C
ECONOMIC
& SOCIAL

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Using technology to transfer expertise.

Problems and potential in cancer genetics.

Currently computer based support systems available that provide:

- Protocols for diagnosis
- Protocols for prescribing.
- Probabilistic systems for predicting risk or for prognosis. ←

From the standpoint of policy makers such systems :–

- Serve to standardize the quality of care;

- Help integrate the primary/secondary care interface.

- Serve to transfer expertise

From the standpoint of secondary care specialists –

- Improve rate of ‘appropriate referrals’ etc

- Enable expertise to be dispensed ‘at a distance’

What kinds of issues arise with cancer genetics?

Examine examples of issues that arise in discussion between genetic counsellors and clinical geneticists

And their implications for the primary/ secondary care interface

NC2: ... This is JS [male patient]. Born 1973. Em, he is 28. His father died at the age of 28 with ca stomach. His mum died @51 with breast cancer being diagnosed at 49. His grandfather died of an unknown primary, this is er., maternal, grandfather, and mothers' fathers' aunt – so that is great aunt? Great aunt? Yes. Died of breast cancer. He doesn't know how old she was, but her daughter has just been diagnosed with breast cancer. Other side, father's side of the family all alive and well. Er. Extremely distressed, extremely worried, has just done an elective where he has just seen a lot of cancer patients for the very first time. Went to see his locum GP who said, 'don't be absolutely ridiculous there is no link', and he would like us to em, follow him up really. I spoke to the GP, his real GP, and she is very happy for the referral to stand, and that if we actually get some evidence to either reassure him or to take him on board.

CG1: Did she actually say what she thought? Did she give an opinion?

NC2: The GP?

CG1: Yeah

NC2: No, she just said 'we've had a difficult locum'.

The implications ….?

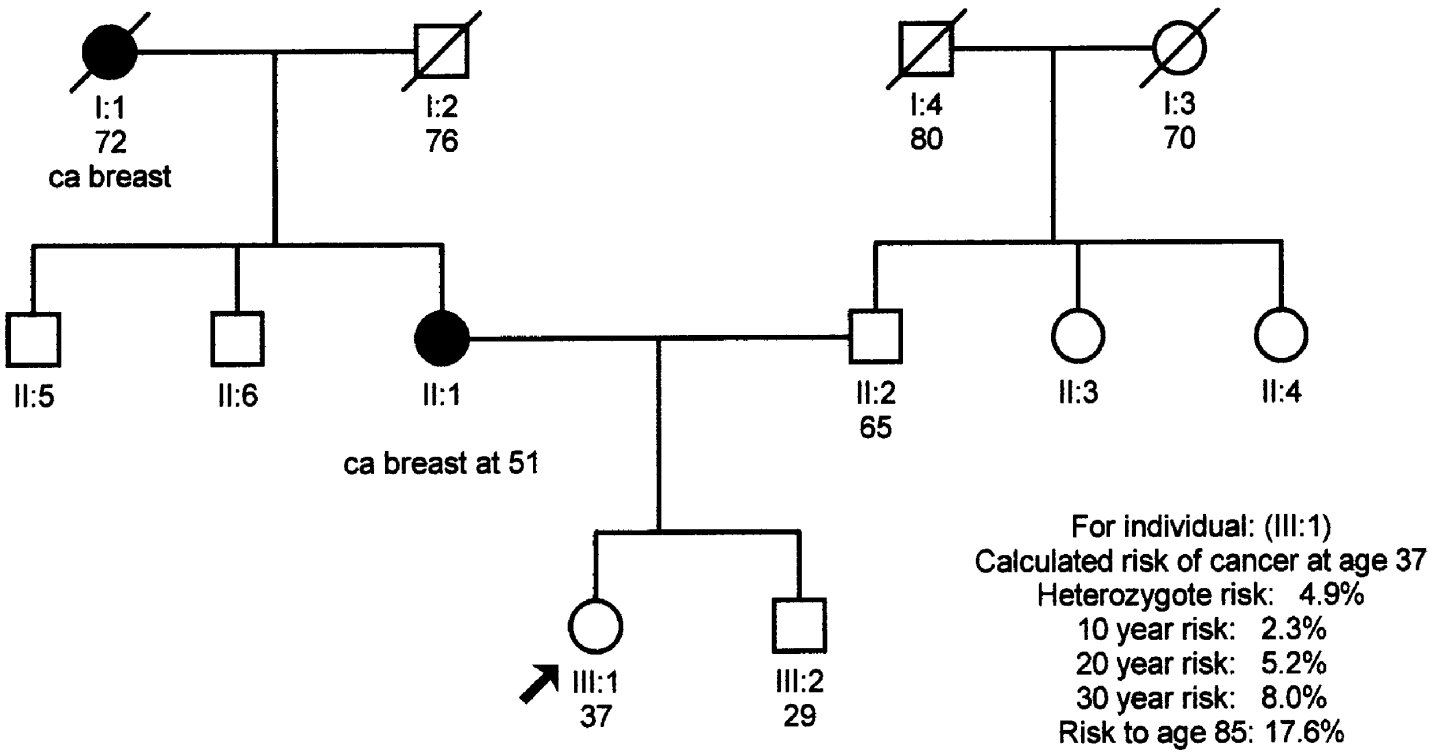
- Complicated issues of inheritance - that require investigation.
- Anxiety and confusion on behalf of the patient
- Need to assess a genetic risk with some accuracy.
- Conflicting messages from medical professionals.

Computerised decision support devices can deal with some (but only some) of these issues.

E.g's CYRILLIC, RAGS

Thus CYRILLIC can both draw a pedigree and calculate a risk for an individual

A Pedigree Drawn (and the risk calculated) by CYRILLIC



NC4: This is JH who is 32. 'This lady's 35 year old sister has just been diagnosed with breast cancer. She herself is 33 and is naturally concerned. There are other sufferers of the disease in the family. An aunt was also diagnosed in her early 30s...'. It's from her GP.

NC2: It's a really good GP

CG1: Yes, the thing is if you really start to tease [the pedigree] apart there are lots of black lines all over the place, they are all on different parts of the family. Her grandmaternal's niece. 40s

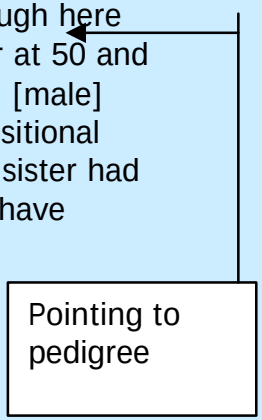
NC2: That's 3rd degree

CG1: Well that is 3rd degree, yeah. And then 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 [CYRILLIC] puts her into a high – oh! – 24.6%. Mm.

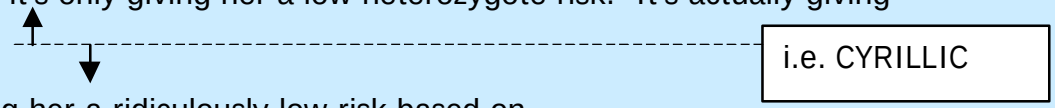
NC2: What did you think, because you had some good thoughts about this one.

CG1: This is one that I would put into a high risk group. Can you think why I have decided to put her into a high risk group?

NC1: This is a referral [flicking through notes] … It's a lady HW who is 50 now 'extremely fit 50 year old whose grandmother was diagnosed 45 with breast cancer and died at 48. A second cousin who died 36 of breast cancer. In addition a male second cousin who died in 70s of breast cancer.' That's a fairly impressive family tree. So here she is. So its her – its coming up through here isn't it, its her mother's, father's, sister's daughter W had breast cancer at 50 and another sister had breast cancer and also ca oesophagus at 36. And R [male] here also had breast cancer at 73 but he also was investigated for transitional cell carcinoma – er bladder carcinoma. And then we have her father's sister had breast cancer at 61 and her daughter had breast cancer at 30. So we have confirmed these two which is a bit…



CG1: Well I mean both sides are….
NC1: On this side, maternal grandmother had breast cancer at 46. But interestingly you've got this prostate here you see at 63.
CG1: Mm
NC1: And an endometrial
CG1: It's a tricky one isn't it
NC1: Mm. Its probably more dominant on this side
CG1: I suppose the reassuring thing is that her parents have survived to 82 and 80.
NC1: You see it's only giving her a low heterozygote risk. It's actually giving her a low risk



CG1: Its giving her a ridiculously low risk based on….
NC1: But I just think it looks very interesting. Basically we have got 1,2,3,4 anyway on one side of the family haven't we. 5.
CG1: Yes. I mean there is a part of me, my knee jerk reaction is that there is so much going on on 3 branches of this family that would almost all put her at moderate risk, and turn round and say right is there any reason for putting her at a high risk?
NC1: I would be inclined to… well to bring her in anyway.
CG1: Certainly it needs to be discussed whatever.
NC1: Yeah

Problems with the decision-support devices

- Different devices give different estimates of risk.
- In cancer, risk calculations are only as good as the epidemiology that they are based on.

E.g. CYRILLIC has no males in its sample and underestimates risk for women of Ashkenazi Jewish descent.

- The calculation is also only as good as the family history (checked through a 'backroom' system)
- Risk is 'dynamic' - e.g. favourable test outcomes for close relatives can change the picture dramatically.

The technical difficulties

1. Checking the family history

2 Interpreting the results

3 Understanding ‘risk’ - esp. the dynamic of risk

Better systems?

Better epidemiology ?

Better training?

But are we dealing merely with technological problems here?

NC3: ... received the referral. Mother was diagnosed at 51. And her grandmother also had breast cancer and died at 72. Well it says died at 71 here but on the questionnaire it says it was found at 72. I'm a bit unclear about when exactly the diagnosis was. She is very very anxious.

NC2: She's put her risk at 100%.

CG1: [gasps inaudible]. Hah. Well according to our criteria, even on the absolute criteria she doesn't meet them because of having one person at 72. We tend to be a bit more flexible by taking an average age of under 60, but she doesn't meet an average age of under 60 either.

NC2: I think on the referral it said 60.5 when we added them up.

CG1: Oh right! Now, has anybody actually spoken to her?

NC3: I have spoken to her briefly.

NC2: In order to find all of that, didn't you?

CG1: What I was thinking of is that this sounds like it is going to be somebody who if we turn around and say she is low risk – which is the category that we ought to be putting her into – is going to be pretty upset.

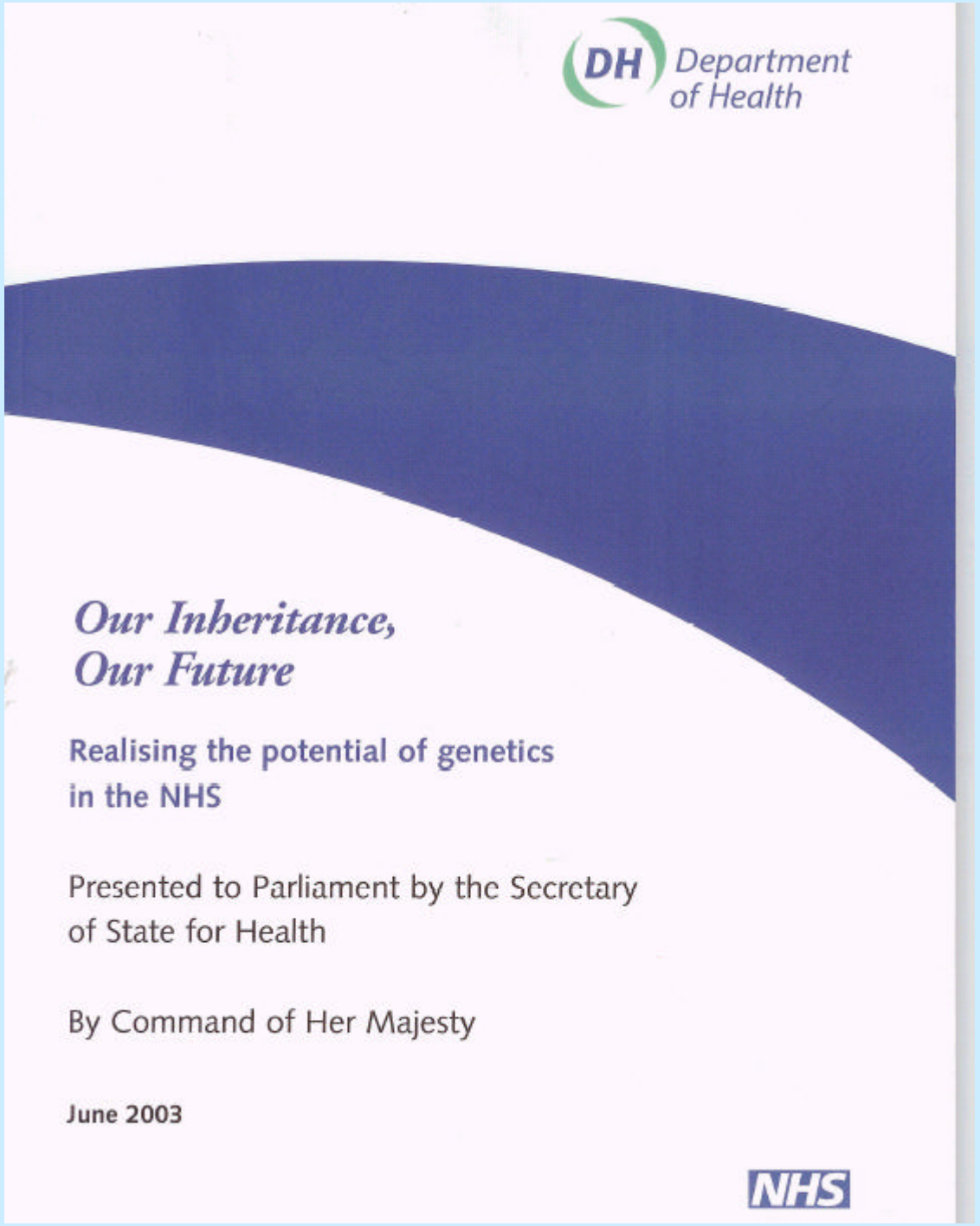
% Referrals Not Meeting Criteria. CGSW (Cardiff) 2001

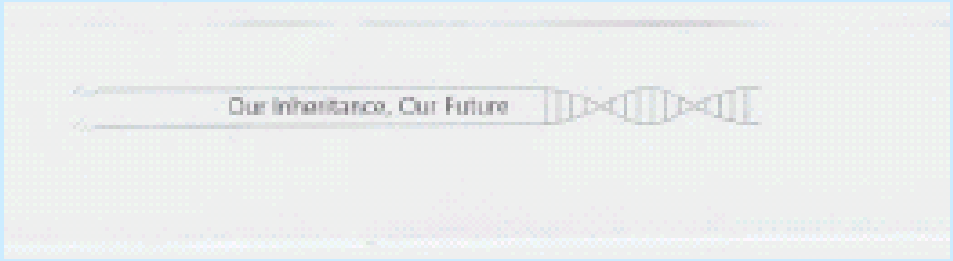
Type of Cancer	% Not Meeting Criteria	N
Breast	13%	157
Ovarian	10%	31
Colorectal	23%	31
Total	14%	219

The consultation as a ‘negotiated order’

Number of letters referencing detail other than family history

	Number of letters/100
Cancer history	3
History of symptoms (breast, abdomen)	16
Mental health problems	7
Other physical health problem (e.g. Asthma, arthritis, MS)	6
Fit and well	10
Smoker	4
Menopausal	4
Nice/ pleasant patient	5
Patient worried/anxious	27
Social circumstances	22
Prescribed medication	20
Investigations	14


DH Department of Health
Our Inheritance, Our Future
Realising the potential of genetics in the NHS
Presented to Parliament by the Secretary of State for Health
By Command of Her Majesty
June 2003
NHS



Roles for Primary Care in Genetics

- Managing patients' concerns and expectations
- Identifying genetic conditions
- Assessing Risk

← A problem for low risk and 'no' risk

- Managing Risk
- Screening
- Testing
- Providing and co-ordinating long-term care
- Gate-keeping to specialist care

Conclusions

Technology = ‘machines’ + humans + social relations

Neither humans nor social relations can be reduced to the hardware.

Ergo ... ‘Downloading’ expertise (in genetics or elsewhere) is not a simple problem of getting the hardware right

Most importantly the ‘machinery’ of risk assessment is unable to deal with the anxiety or even the mildest forms of suffering that both primary and secondary care doctors have to deal with.

‘Technology - machines, instruments, drug treatments - like blinkers on a horse, restrict and define and thus simplify the viewpoint’. For, ...

‘What is lacking in [modern] medicine is an adequate consideration of the place of the person**S**.’

Eric J Cassell, *The Nature of Suffering and the Goals of Medicine.* (2004)