

Current Debates:
Do we own our genes?

Current Debates: Do we own our genes?

- Part One: Genes, genetics and genetic counselling
- Part Two: Impact of genetic testing
- Part Three: Public understanding of genetics

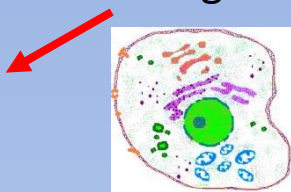


Medical Genetics

Liwsa Kim Protheroe-Davies
Principal Genetic Counsellor



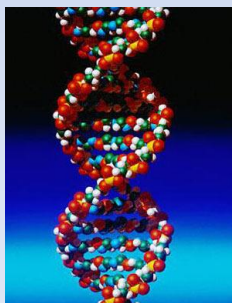
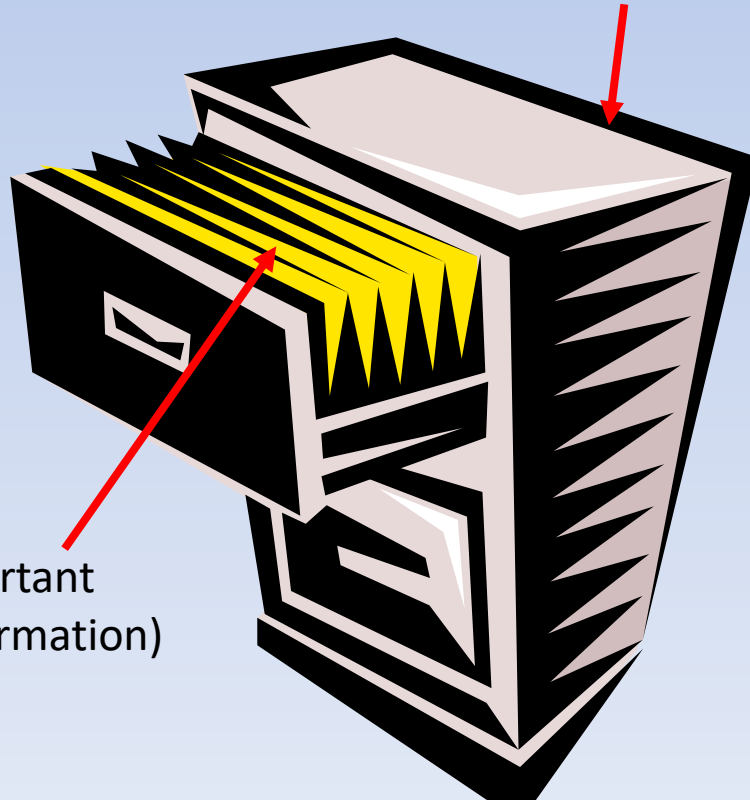
Building = Cell



Office = nucleus

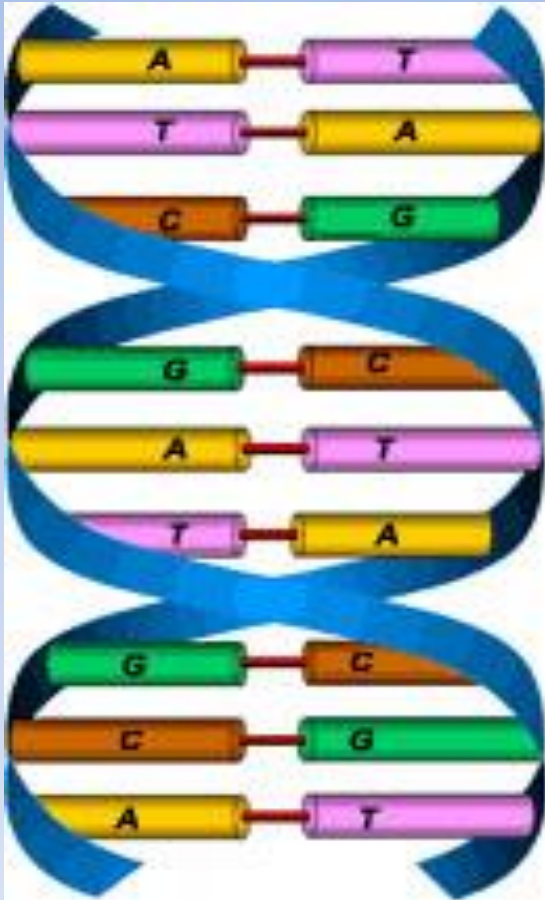


Filing cabinet = chromosome



File = DNA (important information)

What is DNA?



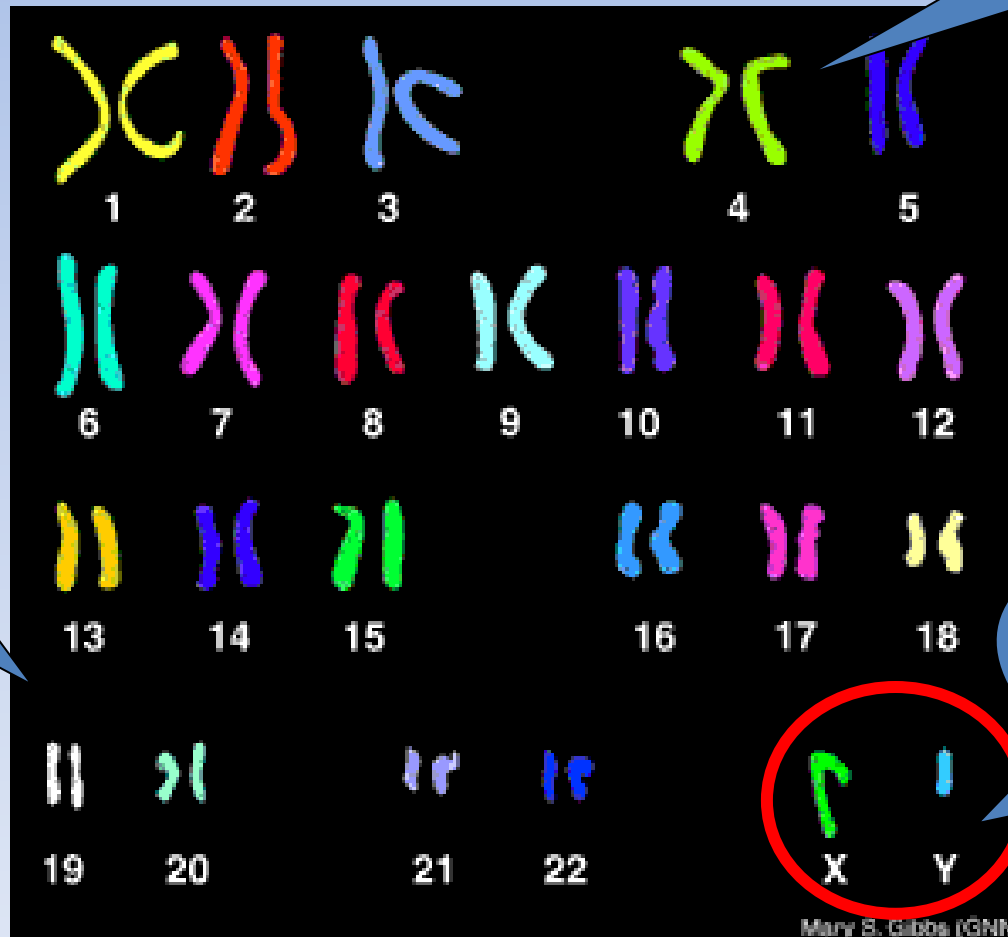
- Bases – T,A,G,C
attached together by a sugar backbone.
- Every 3 bases make an amino acid.
- Amino acids are the building blocks of proteins.

How do we see our genes?

Range in
Size
1 -22

23
Pairs

46
Chromosomes



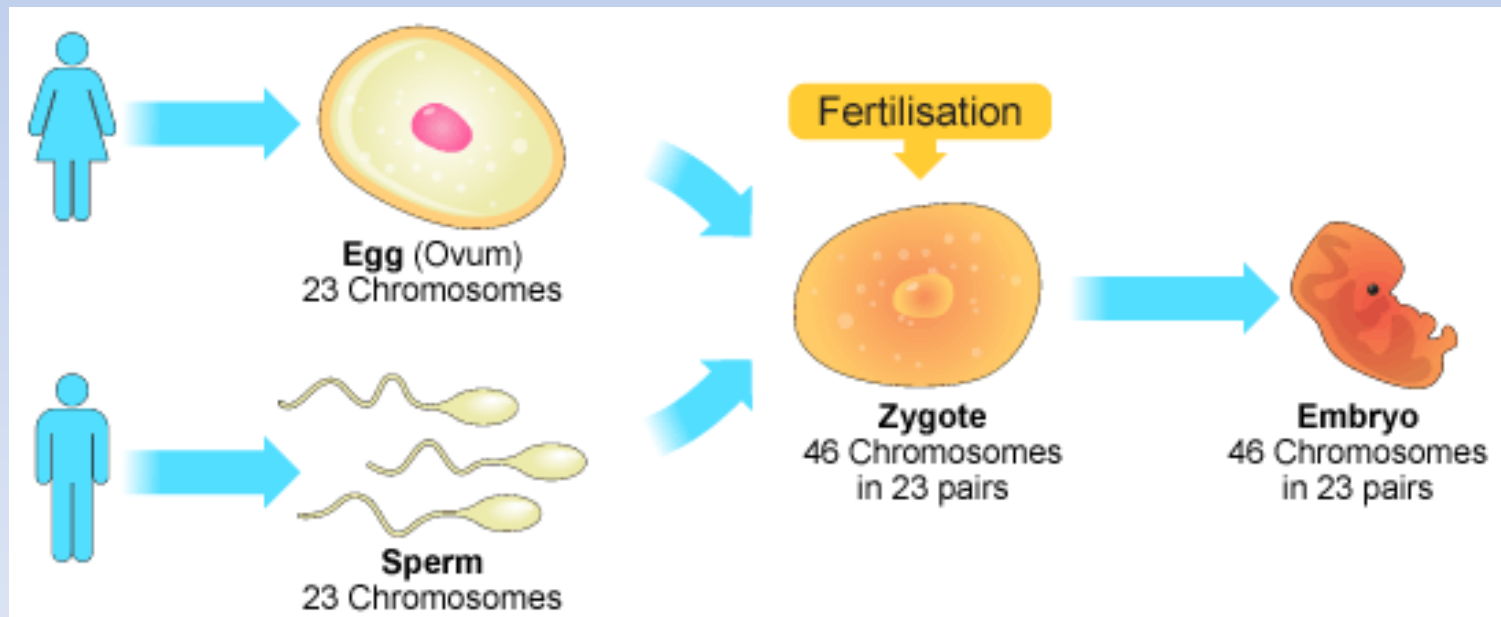
Sex
Chromosomes

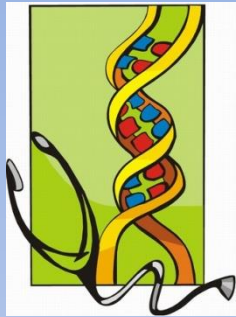
Inheritance

Chromosomes in Pairs

Only one of each pair passed on to children in egg/sperm.

Should have 2 copies of each gene – called alleles.

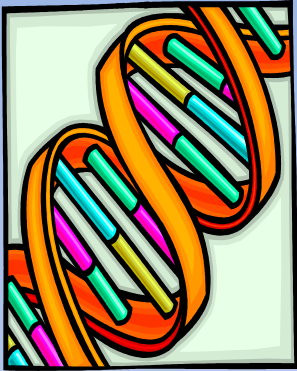




Clinical Genetics



- Spelling mistakes in the DNA code causes problems.
- Extent of the problem depends on the size, nature and location of the mistake.
- Most genetic syndromes are caused by deletions or duplications of the genetic material. i.e. too much or missing info.



Genetic detectives



- Requesting notes
- Cancer Registry
- Histology/Pathology
- Request of Information



Welsh Cancer Intelligence
& Surveillance Unit
Uned Arolygiaeth
a Gwybodaeth Cancr Cymru



Impact of Genetic Testing

The Cancer Perspective

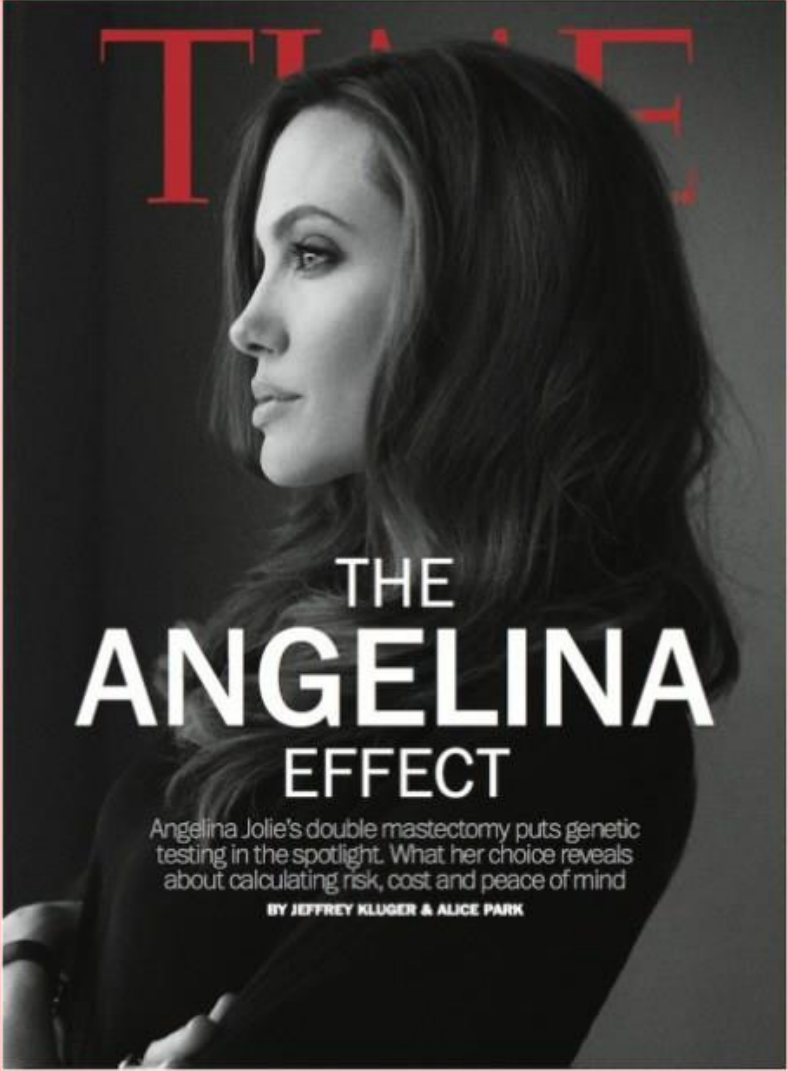
Angelina Jolie tested positive for BRCA gene.



FIND OUT IF YOU SHOULD BE TESTED:
BeAwareFoundation.org/ask-the-doctor

getty images

MAY 22, 2013



TIME

THE ANGELINA EFFECT

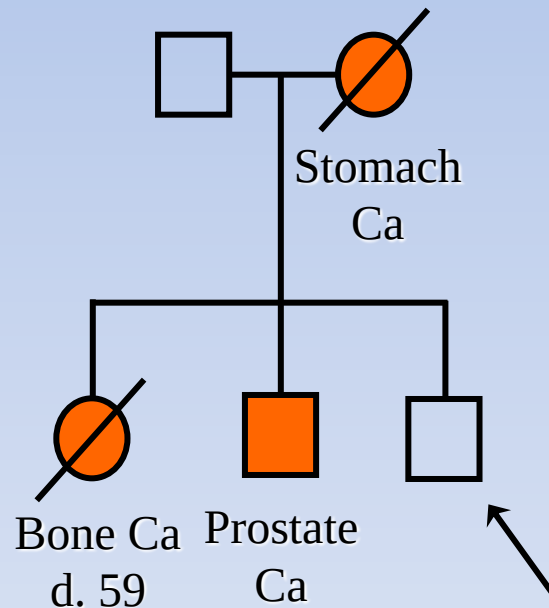
Angelina Jolie's double mastectomy puts genetic testing in the spotlight. What her choice reveals about calculating risk, cost and peace of mind

BY JEFFREY KLUGER & ALICE PARK

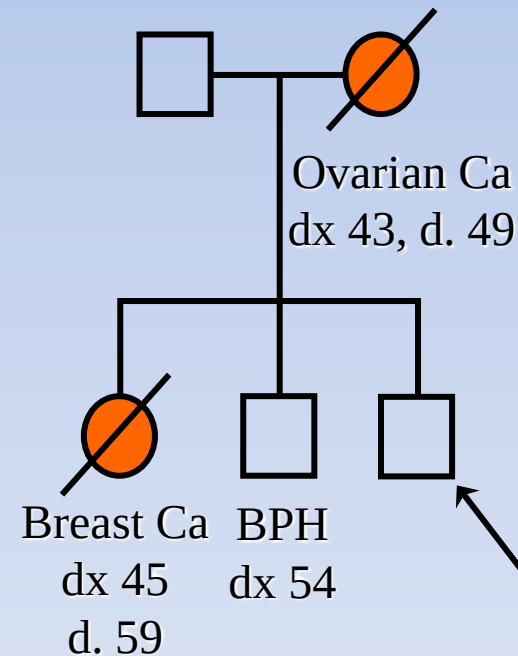
TIME.COM

Importance of Verifying Family History

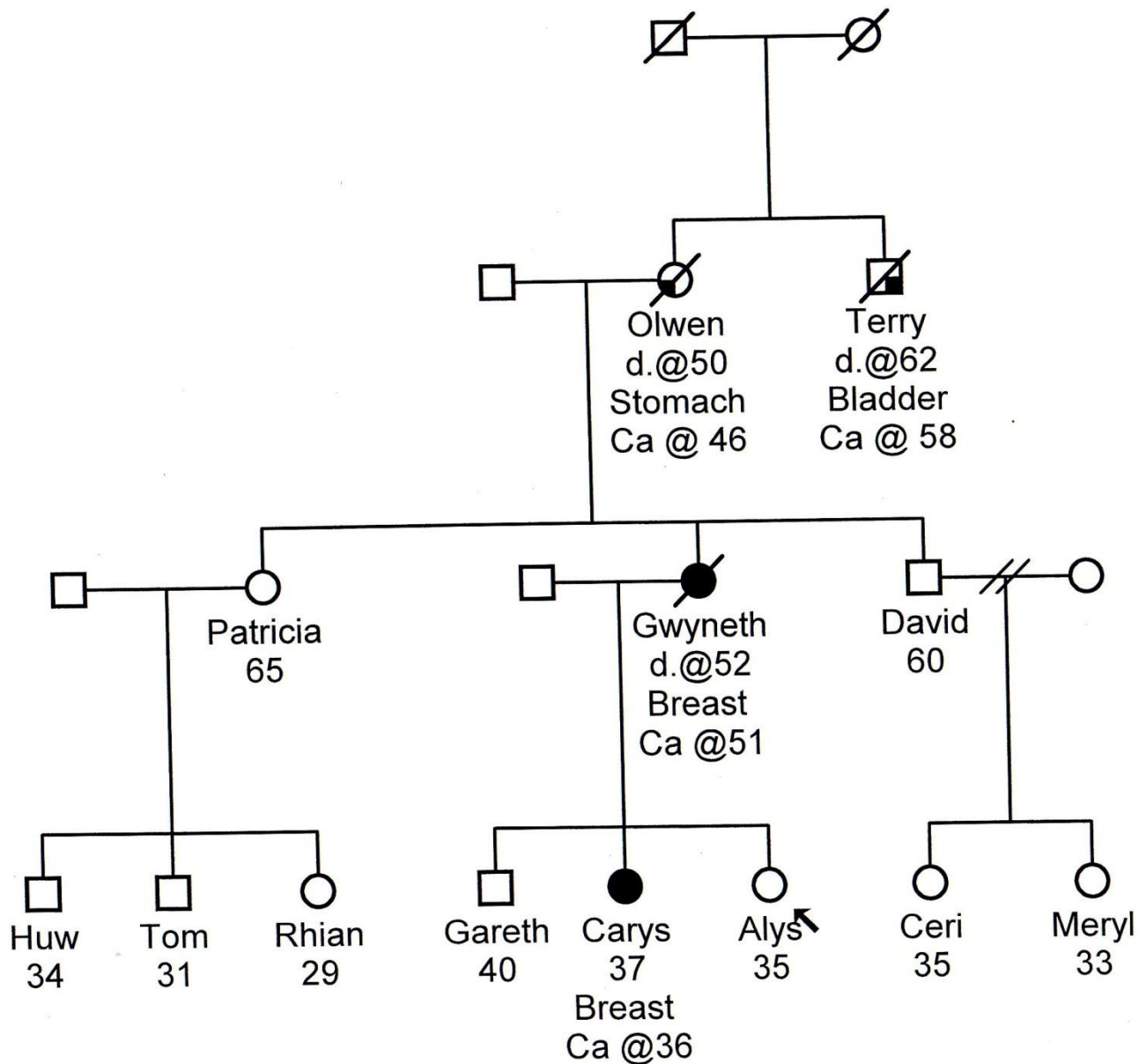
Verbally reported pedigree



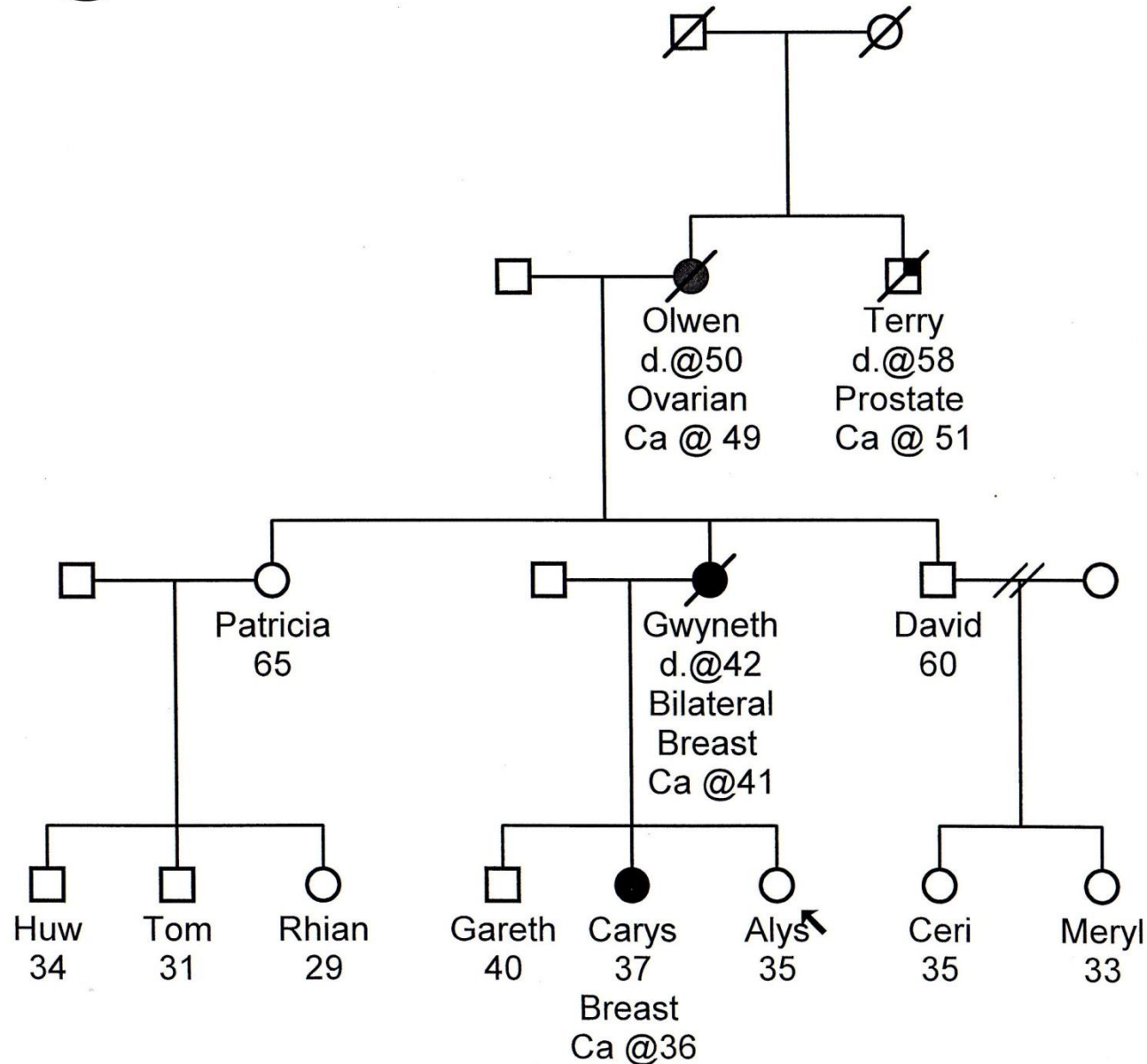
Revised pedigree based on pathology reports



Pedigree from FHQ



Pedigree after confirmations

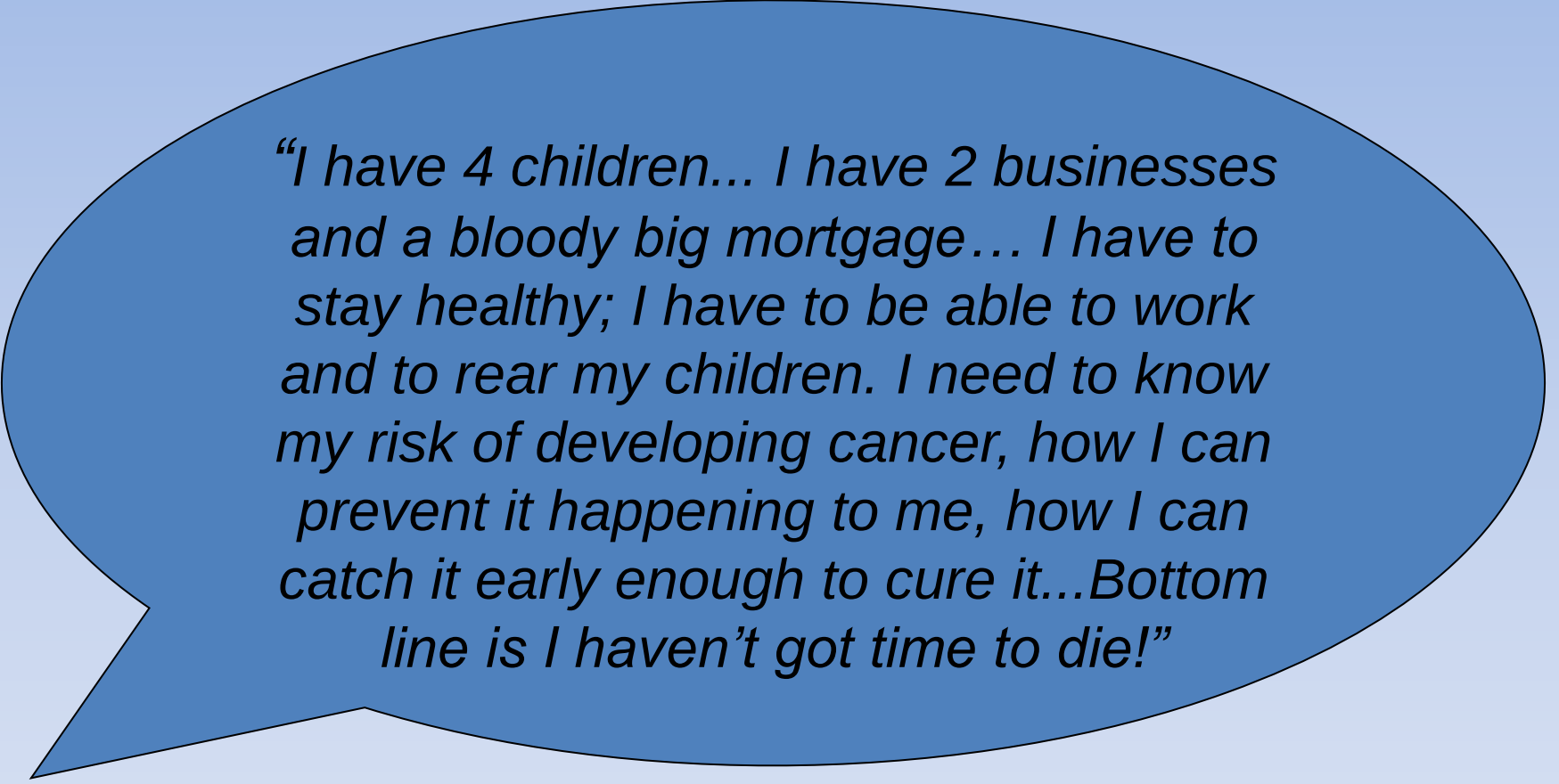


- <http://www.cancergeneticsstorybank.co.uk/withegenewevegots.htm>
- <http://www.cancergeneticsstorybank.co.uk/noteasythingtotellanybody.htm>

What do we really understand
about genetics?

Genetic risk, motivation and behaviour

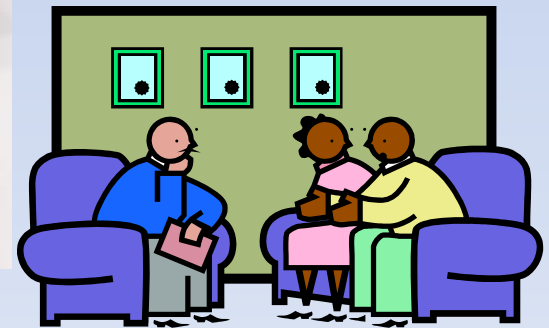
Genetic risk, motivation and behaviour



“I have 4 children... I have 2 businesses and a bloody big mortgage... I have to stay healthy; I have to be able to work and to rear my children. I need to know my risk of developing cancer, how I can prevent it happening to me, how I can catch it early enough to cure it...Bottom line is I haven't got time to die!”

Cancer genetics patient/participant in CARIAD trial

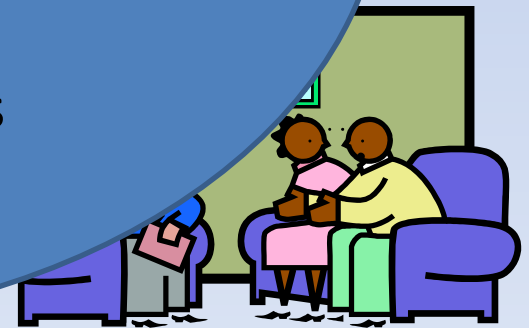
Genetic risk, motivation and behaviour



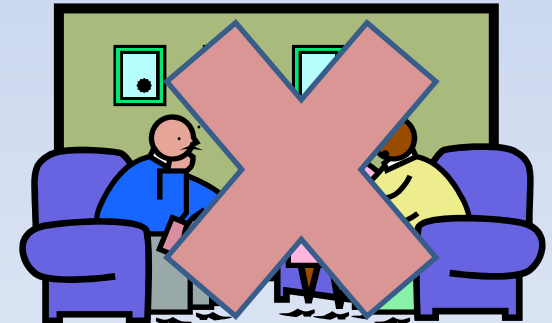
Genetic risk, motivation and behaviour



- Attitudes & beliefs
- Risk comprehension
- Emotional responses
- Delivery of genetic information
- Individual differences



The future (is here)



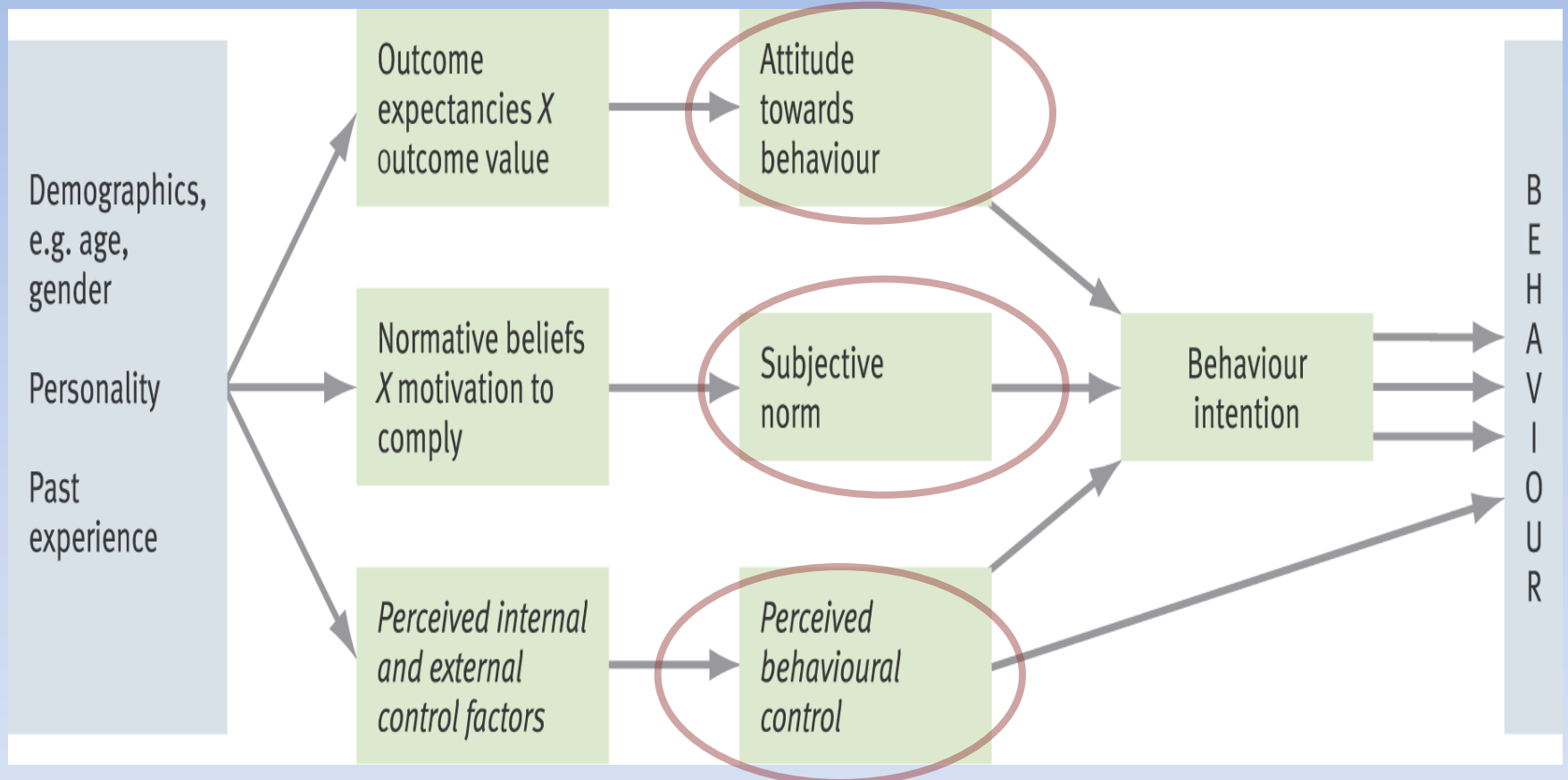
What we know #1

Attitudes influence health behaviours.

How well do attitudes predict interest in genetic testing?

- Greater interest in genetic testing linked to:
 - more positive attitudes towards testing
 - positive beliefs about early diagnosis and treatment
- BUT
 - Negative attitudes associated with concerns about confidentiality and discrimination

The Theory of Planned Behaviour



Attitudes towards genetics in the general population

- What attitudes do the general population hold towards genetics and genetic testing?
- How do these attitudes influence behavioural choices and interest in genetic testing?

Attitudes towards genetics in the general population

Hutchings, Fitzgerald & Phelps (2013)

- Understanding attitudes through experimental social psychology paradigm
- Reaction time computerised response task
 - Faster reaction indicative of stronger attitudes

Hutchings, Fitzgerald & Phelps (2013)

Figure 1: Example of attitudes towards target words

	Good	Bad	Exciting	Scary
Faulty Gene	7%	93%	9%	91%
Altered Gene	43%	57%	27%	73%
	Ethical	Unethical	Exciting	Scary
Genetic Diagnosis	93%	7%	34%	66%
Genetic Screening	75%	25%	34%	66%
Genetic Profiling	55%	46%	50%	50%

Implications for public health genomics

- What you say (or how you say it) matters!
- Certain words used by health professionals or in health literature may trigger automatic negative responses towards genetics/genomics
- Awareness of certain words associated with negative attitudes may give insight into individual's attitudes, beliefs and understanding of genetics/genomics

Future research.....

- What attitudes do non-genetic specialists hold towards genetics?
- How well do these health professionals understand genetic information?
- How well can they deliver this to patients?

Attitudes towards genetics in the general population

- What attitudes do people hold towards people with genetic conditions?
- How do these attitudes influence behavioural choices?
- Hypothetical dating scenarios manipulating severity of genetic condition in potential “mate”

Attitudes towards genetics in the general population

- Participants indicated less intention to invest in a relationship with potential mates with more serious genetic conditions $F(1, 41) = 4.28, p = .007$.
- Interaction between condition and positive-negative word valence for relationship investment.

E.g. participants who reported “genetic test” as being “bad” significantly less likely to consider investing in a relationship with an individual in the non-treatable genetic condition than those who saw “genetic test” as “good”.

Implications for public health genomics

- Implications of negative attitudes towards genetics on wider lifestyle choices.
- A new form of “genetic discrimination”?
- NB: Modern prejudice cannot be captured explicitly

Attitudes towards genetics in the general population

Intention to receive a hypothetical genetic test

	Mean	SD
Higher scores reflect more positive responses (scale of 1-7)		
Intention: How likely would you be to have a genetic test	5.59	1.61
SN: Most people who are important to me would approve of me having a genetic test	5.91	1.40
SN: Having a genetic test may have implications for family members	4.64	2.10
PBC: I could easily have a genetic test if I chose to do so	4.87	1.61
PBC: I would be concerned that other people could find out my genetic test result	3.47	2.13
AR: If I did not have a genetic test I would later wish I had	5.10	1.80

Can we (or should we?) change attitudes?

- Information about genetics needs to tap into these key factors
- BUT need to tap into implicit attitudes – asking people what they think may not be linked to behavioural choices...
- Potential for attitude change intervention

What we know #2

People's understanding of genetic risk
influences behaviour

Understanding impact of genetic risk information

- Objective/Clinical risk status
 - Subjective risk status
- ≠

*“I find it hard to understand that if so many people in my family have had breast cancer that they felt I was not at risk. I keep thinking ‘what if they got it wrong!?’
CARIAD trial participant*

Understanding impact of genetic risk information

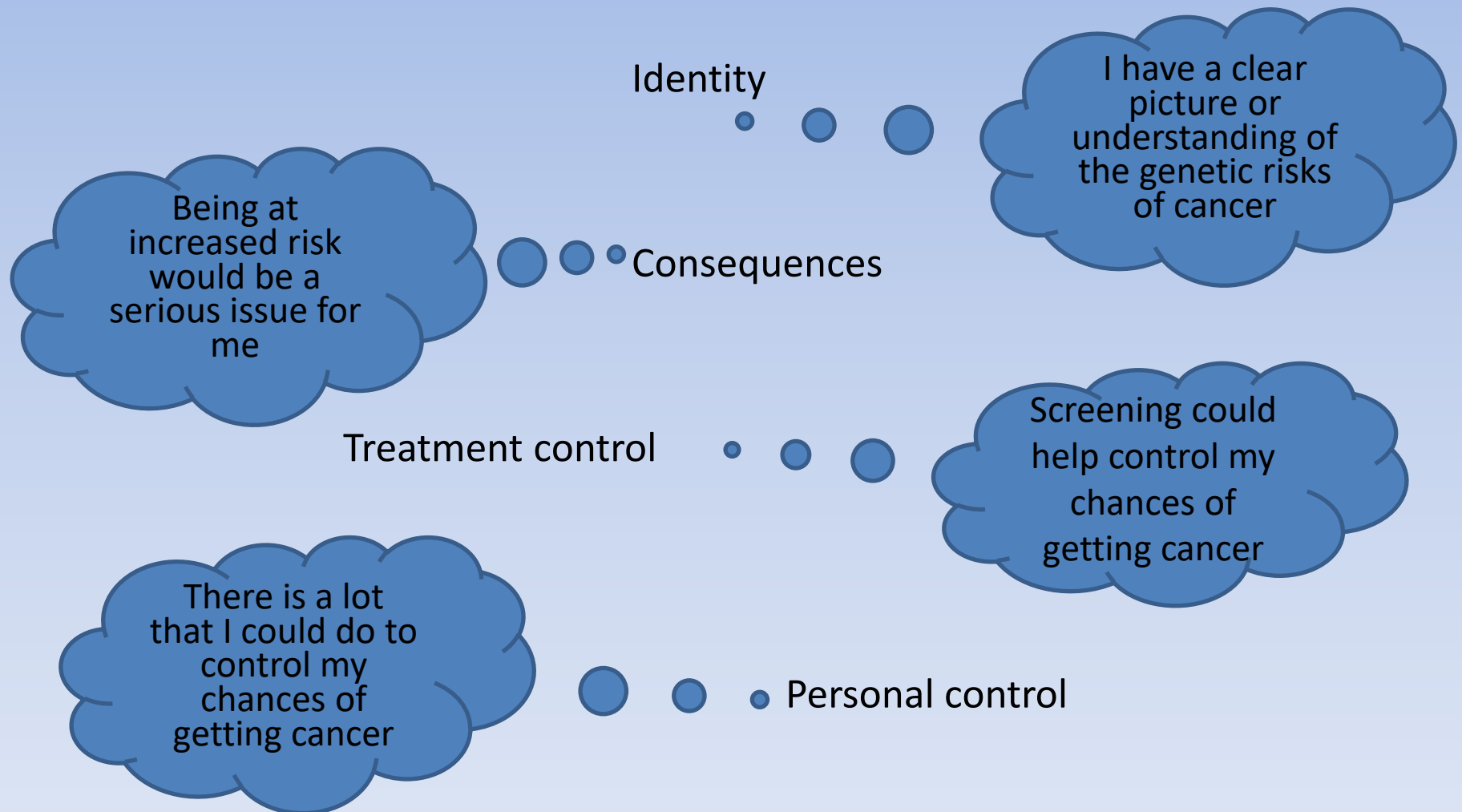
- Objective/Clinical risk status
 - Subjective risk status
- $\neq$
- Subjective risk better predicts subsequent health behaviours and distress
 - Need to explore the gap between the two in consultation/counselling to influence healthy behaviours

Self-regulation model (Leventhal et al., 1992)

- People are “common-sense scientists”
- Actively process information about genetic risk or a genetic condition
- Form a cognitive and emotional response based upon beliefs about:
 - Identity (label, signs & symptoms)
 - Causes (genetic, psychological, environmental)
 - Consequences (e.g., emotional, financial, physical)
 - Timeline (acute, chronic, cyclical)
 - Curability/controllability

Self-regulation model (Leventhal et al., 1992)

Representations of genetic risk based upon beliefs about:



Self Regulation Model and Genetic Risk

- SRM suggests poorer psychological outcomes in those who believe that:
 - being at genetic risk has serious emotional, physical or financial consequences
 - little they can do to control their chances of developing the illness
 - little belief in treatment, screening or surgery controlling their chances of developing the disease
 - do not understand their genetic risks

Self Regulation Model and Genetic Risk

- May be inaccurate!
- Illness representations acquired through:
 - Media
 - Personal experience
 - Personal experience with a genetic disease is a major determinant of its representation and subsequent decisions (Shiloh, 1996)
 - Family & Friends

Self Regulation Model of Genetic Risk

- **Influence preventive behaviour and coping responses:**
 - Seeking medical help (over or under-use)
 - Adherence to treatment (e.g., screening, medication)
 - Emotional reactions to risk/symptoms (e.g., avoidance, hypervigilance)
 - Engagement in self-care behaviour (e.g., BSE, healthy lifestyle)

Implications for public health genomics

- Accurate information about causal factors if not solely genetic
- Education about controlling/reducing symptoms/disease/genetic risk through health behaviours
- Education about over-reliance on medical treatment/medication in controlling risk

Future of psychological research in this area....

- Research needs to move away from the focus on patients with recognised family histories who receive education and counselling about these issues as part of their NHS-based treatment
- Need to consider the knowledge, understanding and attitudes of the general population in relation to genetic testing.
 - Role of decision-explorer support?
- Education/training of health professionals re: genetics?
- Attitude change interventions for health professionals?

Conclusions

- Knowledge of genetic risk can influence decision-making and adoption of healthy behaviours in a positive or negative way
- Depends upon:
 - Attitudes
 - Beliefs about causal factors, controllability and cure
 - Psychological distress
- Health professionals need to be aware of their influence over all of these processes and help people to make the right decisions for them

Conclusion:
Do we own our genes?