GENETIC TESTING-HOW DOES IT AFFECT FAMILIES?

EXECUTIVE SUMMARY

The aims of this research were to find out:

⇒ how genetic testing for cancer affects family relationships

⇒ whether genetic testing changes the way in which individuals think about who is family

⇒ whether views are the same or different within a family

The individual stories we have gathered provide information about the long term effects of genetic testing, which will be helpful to those who are thinking about having a test.

KEY POINTS FROM THE RESEARCH

Respondents suggested that:
Choice in genetic testing is important but sometimes responsibility to others in the family is more so.

It is the responsible thing to know about your genetic status for your children's sake, for yourself and to help research.

Those that declined to have a genetic test acted irresponsibly but should have the right to choose.

Although knowledge is a good thing, having knowledge of a gene that gives an increased risk of cancer can be like "living under a big black cloud."

The research found that:
Respondents held a positive and optimistic view of science and medicine

Genetic testing can make you aware of unexpected connections to others that sometimes you would rather not know.

Although those in this study were identified by a genetics department as a "family" the ways in which people decided who was close to them sometimes had little to do with genetics or biology – Some family relationships were beyond question (children and parents) whilst other family-like relationships were chosen.

Genetic status did not appear to be important in family relationships. There was a lack of knowledge within the family about who was tested and who has the gene despite public meetings and "family" appointments. The gene was not talked about.

ABOUT THIS "FAMILY"

The people who contributed to this study come from one large extended family who were one of the first internationally to have the MLH1 gene identified. MLH1 is one gene known to cause Lynch Syndrome– previously called Hereditary non polyposis colon cancer (HNPCC). This makes their experience different to those undergoing testing today because the medical approach to genetic testing has developed since that discovery. Their contribution however is important in discovering how genetic testing can be experienced by families and the consequences of those experiences.

ABOUT LYNCH SYNDROME

Lynch Syndrome– previously called Hereditary non polyposis colon cancer (HNPCC) describes a predisposition to bowel and other cancers of the digestive and urinary systems. In women the reproductive system is also involved. Those who have the MLH1 gene have an increased risk of developing cancer at an earlier age than the general population.

Authors and Funders

Author-Lorraine Cowley

Supervisors-

Dr Janice McLaughlin

Dr Tracy Finch

Dr Emma Clavering

Professor Sir John Burn

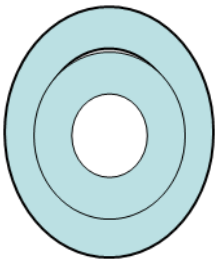
Funder– Cancer Research UK

CANCER RESEARCH UK

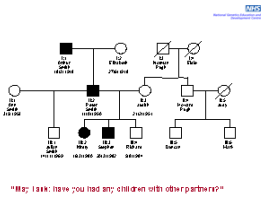
WHAT WE DID

A sample of 15 of the 50 people from this extended family who were offered a genetic test agreed to be interviewed. All of those participating had been tested for the known gene causing Lynch Syndrome. They discussed their family relationships using family photographs, a social map diagram and the genetic family tree. At a second interview we discussed their experiences of genetic testing with most of the respondents. The interviews were recorded and transcribed then analysed. In the analysis, common ideas were identified about genetics and family.

Family Photograph



Social Map



Genetic Family Tree

IS IT A CHOICE OR A RESPONSIBILITY?

Genetic testing was viewed as both a choice and a responsibility. For some the choice to have a gene test was felt to be an easy and obvious choice. It seemed most important to talk about choice in genetic testing when thinking about those who did not want to be tested. Those who did not want to be tested were thought of as being scared or the type of person who did not want to know. This was described as frustrating for some who experienced family members that didn't want a genetic test. Although it was understood to be an individual choice by all those in this study, there were occasions when that choice became a responsibility. This was most obvious where children were concerned.

"I suppose a lot think ignorance is bliss but I'm not that type. I like to know for myself but I like to know for my kids you know?"

CHILDREN– A REASON TO BE TESTED

Although over half of the people in this research felt that they had a choice in who was close to them, one relationship was never questioned, that of parents and children. When thinking about having a genetic test, all of those who had children thought that it was important to be tested for them. Some had parents who had declined a genetic test and although they would not have acted the same way because they had children, they accepted that their parents had a right to choose.

LOOK AFTER YOURSELF– ANOTHER REASON TO BE TESTED

Those who participated in this research shared a set of beliefs about genetic testing. Those beliefs were:

- That belonging to this family poses a health risk (for some)
- That health risk (an increased risk of developing cancer) can be identified through genetic testing
- Through genetic testing they can gain access to medical intervention (Screening)
- Screening can reduce the health risk (Cancer)
- Cancer can be avoided or the success of treatment can be improved through screening
- That life can be saved as a consequence

"If they didn't have the test and they didn't have children? It's up to them. You can't force people to have tests. It's got to be up to the individual I think."

Although those with MLH1 gene have a higher risk of developing cancer, getting cancer is not a certainty. Equally those that don't have the gene can get cancer by chance. People from a family with Lynch Syndrome today can get access to screening without knowing their genetic status if their risk is thought to be high. Some acknowledged that for those who declined testing, having knowledge of their genetic status if they were a carrier may be too worrying and could therefore be harmful to their health.

"And just being armed with the knowledge I suppose that if anything did happen you've given yourself the best chance of being able to fight it I suppose…"

"I owe a little bit back… you feel obliged. So when they approached me about the research… would I be interested in coming on board… ? I had no hesitation in saying yes, obviously to try and help things"
"It's technology and we should use it you know… "

RESEARCH AND MEDICAL ADVANCEMENT

The family identified in this research are unique in their position as the first to have the MLH1 gene identified. Their history with medical research spans generations and most participants told stories of "family" gatherings in surgeries for medical investigations before the discovery of MLH1. They told of their experiences of having their eyes examined, mouth swabs, handprints and finger prints taken. Some expressed gratitude to the GP who first recognised a pattern of cancer in their family, to genetics and to medical research. All expressed a strong belief in research and some expressed a responsibility to participate as a way of repaying what they felt was a debt. All of the participants in this research had a favourable view of research and medical advancement.

"I mean the family were going and saying well we think we've got cancer you know and doctors were saying to them all no you know. You can't have… and they were saying well yeah we do think we have"

BEFORE GENETIC TESTING

IMPLICATIONS FOR PRACTICE

Those working with families should not assume that people on a genetic family tree will be close

When asking family members to pass on genetic information it would be useful to work with their individual view of family, alongside the genetic family tree.

Those who have a long history with the genetics service and think genetic testing is a positive experience (regardless of the outcome) may feel more of a responsibility to help further research.

Geneticists need to be mindful of this and take care not to over burden those individuals with requests to participate in research.

Genetic testing (having a blood test to identify a known gene) and screening (having a colonoscopy to identify polyps) can be commonly mixed up and assumed to be the same thing.

Geneticists need to continue to make the distinction and make others aware that they do not have to have a genetic test to access screening.

Families offer advice to each other about whether to have a genetic test or not. Genetic counsellors aim to help someone to make up their own mind as an individual.

It would be helpful to find out from those having genetic counselling what their family thinks of them having a test to understand the pressures the individual is under.

Screening for those with a gene that causes an increased risk of cancer should be provided in the agreed timeframe as this causes anxiety when it is not.

Genetic counsellors should not talk with an individual about genetic tests for adult cancer causing genes in terms of benefit to their children as this puts pressure on the individual to be tested.

Acknowledgements:
Thank you to all those who participated in this research, for generously giving up their time and sharing their valuable experiences.

Further information can be found at: http://ghr.nlm.nih.gov/condition=lynch-syndrome