

# Programme Transcript - Inside the Ethics Committee

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## INSIDE THE ETHICS COMMITTEE

### Programme 3 – Sharing Genetic Information

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**PRESENTER: JOAN BAKEWELL**

**PRODUCER: LORNA STEWART**

#### **Bakewell**

A genetic fault carried on a single gene can pass life-threatening disease from parent to child. What happens when you discover you carry a faulty gene? Should you tell relatives who might also carry it? Would they want to know? What if their lives, or those of their children, depended on it? And what role should doctors play in all this - does their duty of care stop with their patient, or include relatives too?

Welcome to Inside the Ethics Committee.

It's October 2012. Andrew has been feeling unwell for several weeks. Then one night the stomach cramps become severe and he starts vomiting. He and his wife Elizabeth rush to the hospital and doctors find that Andrew's bowel is obstructed. He has emergency surgery, and they discover he has cancer.

**Elizabeth**

Just... shock... and... thinking that's really unfair. The nurse specialist was very encouraging, she said – hopefully they'll remove the tumour, have some chemotherapy and you'll be on your way, six months' time you can put it behind you.

**Bakewell**

Andrew has an adeno-carcinoma, the most common kind of bowel cancer. He's just 33 years old. His nurse specialist.

**Nurse**

There was evidence that cancer cells had gone into the lymph nodes, so he went on to have chemotherapy with a combination of drugs.

**Bakewell**

The schedule is gruelling for Andrew and Elizabeth

**Elizabeth**

I was working, our little one was at nursery and I was pregnant with our second child. We were trying to juggle childcare with hospital appointments, oncologist appointments, chemotherapy appointments, trying to keep the little ones quiet so that he could rest and obviously all the washing and the ironing and the cooking and the washing up – just mad, mad busy.

**Bakewell**

Whilst he's having chemotherapy, Andrew is referred to a genetics consultant. Having cancer at such a young age is rare and his oncology team suspects a genetic cause. The genetics consultant begins by taking Andrew's family history of cancer – and it's a long one.

**Genetic Consultant**

I could see that his father, first degree relative, had had bowel polyps at an unusually young age. That a paternal aunt had been diagnosed with ovarian cancer at the age of 38 and had died from her diagnosis at the age of 40. That a paternal first cousin had been found on post-mortem to have a kidney cancer....

**Bakewell**

And the list goes on into the wider family...

### **Genetic Consultant**

... who was diagnosed with bowel cancer and died from it at the age of 34. So although we know cancer is becoming more and more common this number of cancers in so many people, who are closely related to one another, at these mainly young ages, does suggest the presence of a hereditary predisposition.

### **Elizabeth**

It all seemed so obvious what a prevalence of cancer there was and I definitely thought how could I have not said something or noticed.

### **Bakewell**

Because Andrew has so many cancer-affected relatives on his father's side, the consultant suspects a genetic condition known as Lynch Syndrome. If Andrew's father has the faulty gene then Andrew has a 50% chance of inheriting it. This would mean he has a high risk of getting cancer in his lifetime – up to eight out of 10 people with Lynch Syndrome will go on to develop bowel cancer

A blood test confirms that Andrew does indeed have the faulty gene. He also discovers it's been known to be in his family, on his father's side, for some years but Andrew was never told he was at risk.

### **Genetic Consultant**

Clearly some communication had been lost and it does appear that Andrew's father perhaps didn't know of the diagnosis and it was on the paternal side of the family.

### **Bakewell**

Had Andrew known of his Lynch Syndrome earlier he could have been screened regularly. Colonoscopies look for tiny growths in the bowel, known as polyps, which carry a cancer risk.

### **Genetic Consultant**

Colonoscopy allows the detection of the bowel polyps and the removal of the polyps before they can become cancerous, so it reduces cancer risk. But also it allows the early detection of cancers and a low grade cancer like that has got much greater survival. If we had known that may

have allowed the earlier detection of his bowel cancer before it had already spread to his lymph nodes.

**Bakewell**

Elizabeth and Andrew are determined that the same thing will never happen again in his family.

**Elizabeth**

We went back home and we drew out the entire family tree. I think Andrew made a phone call or wrote a letter – a personal letter – to all of them.

**Bakewell**

At Christmas in 2013, a full year after his bowel obstruction, Andrew begins to feel unwell again.

**Elizabeth**

I wasn't sure whether it was just because it was the winter and not having seen the sun but actually he just looked a bit pale and I did have an inkling around about that Christmas time that there was something wrong.

**Bakewell**

A CT scan reveals that Andrew's cancer has indeed returned.

**Elizabeth**

I think Andrew was at work when he got the results over the telephone and so he would have told me when he got home that night. And we knew from way back in the first oncologist appointment that the place that you didn't want to get metastases was in your peritoneum.

**Bakewell**

Andrew's nurse specialist.

**Nurse**

The peritoneum is the lining around your abdomen and depending on how he did with chemotherapy his life expectancy was sort of in months, rather than years, unfortunately.

**Elizabeth**

And so for us we knew that that was pretty much it.

**Bakewell**

Andrew died in November 2014, two years after his original cancer diagnosis and 18 months after his Lynch Syndrome test.

Let's turn now to our panel.

Jonathan Roberts, of King's College, London and the Wellcome Trust Sangar Institute, a genetic counsellor who is currently researching how families communicate about genetics and Deborah Bowman, Honorary Professor of Ethics and Law at St Georges University London.

Jonathan, how common is it for people not to tell their family that they have a genetic condition?

**Roberts**

Experiencing challenges in communication with families is relatively common but for there to be complete non-disclosure is relatively rare.

**Bakewell**

And how do you encourage them to communicate?

**Roberts**

You can do things such as emphasise the benefit to other family members, so as we heard in the story before family members may not be aware of quite how beneficial that knowledge would be. So in Lynch Syndrome early screening is incredibly effective, especially colonoscopies, and you're looking for small polyps to see if they are harmless or potentially early onset cancers. And a lot of family members, once they realise the importance of this, are more willing to communicate. Other ways involve just reducing the potential barriers, so some family members might just not know how to start that conversation. So you can do things like draft letters that families can then cascade out and that way they know they're giving accurate information.

**Bakewell**

Deborah Bowman, confidentiality – it's the cornerstone of medical data and what we all believe – why is that so important?

**Bowman**

The assumption is and it's probably a valid assumption, that trust flows

from it and therefore that actually it would be difficult for us to go to our doctors or to a genetics counsellor or to a nurse if we couldn't be absolutely certain that what we were about to share was to be confidential.

**Bakewell**

But under what circumstances would it be ethical to break that confidentiality?

**Bowman**

So there are three broad categories under which you can share information that's confidential. The first is when you've got consent, the second is when it's in someone's best interest you can't get consent and that's often around communicating with carers. And the third is this broad category of the public interest, so where there's a serious risk to somebody else. It's an entitlement to share confidential information it's not a duty to do so and that distinction's important.

**Bakewell**

Does it apply to genetic...

**Bowman**

I mean the principles of confidentiality apply across whatever specialty you're working in, so what you'd be thinking about is is this a circumstance where there is a justification – it's a justification rather than an obligation – to share confidential information. And the presumption is always, always, always that you avoid breaching confidentiality if you possibly can, even where you think you have to share information you would only ever do that after lots of efforts with the patient to try and help them in sharing the information.

**Bakewell**

Okay but that throws the onus of confirming this information on the patient always and the patient's carrying a burden already, isn't there a case for easing the burden – why should the patient carry the burden?

**Roberts**

I think that's a very good point. I think there's a balance to be struck between whose responsibility it is. Generally speaking it's better for family members to contact other family members, they'll have a better idea of how the communication works, how best to go about it and the clinicians can be

there in support. But there is definitely an argument to say that clinicians could do more to support family members in doing that.

**Bakewell**

Deborah, whose responsibility is this?

**Bowman**

So it's interesting, I mean the way we're hearing it it sounds like responsibility's this parcel that everyone's trying to get rid of and actually I think most clinical practice isn't like that, it's about the clinician working with the individual patient and all the relationships within which that patient is situated to think how can we best support communication here. And I do think it's collaborative in the vast majority of cases.

**Bakewell**

Jonathan, suppose you've been through all the procedures that Deborah's suggesting, would you ever go against the wishes of a patient?

**Roberts**

There are rare cases where you might go against the wishes of the patient but you would have to understand that there's clear benefits to another patient for which you have duty of care and those decisions are normally made within a team.

**Bakewell**

So thank you panel for the moment because we turn now to an entirely different situation that concerns Lucy.

Lucy's story begins six years ago. It's 2010 and she's 28. Lucy's father has been unwell with depression for some time, then he telephones her with devastating news. For legal reasons, Lucy's words are spoken by an actor.

**Lucy**

My father actually rung me to tell me that he'd killed a family member. I initially didn't know if he'd gone mad or if this had actually happened.

**Bakewell**

Lucy's father asks her to call the police and tell them what he's done.

**Lucy**

I remember just sitting on the floor crying my eyes out, not really knowing what to do. Literally it felt like my world had turned upside down. The floor didn't even feel straight anymore. You just have nothing – no reality anymore.

### **Bakewell**

Lucy's father is remanded in custody, then taken to prison to await trial. Lucy and her sister plead for him to be taken to the hospital wing of the prison. He's been depressed for some time and they are concerned about his mental health.

### **Lucy**

He was ill for quite a long time before that, he was taking antidepressants, he wasn't really working, he hadn't worked properly for quite a while. He slept quite a lot. He considered attempting suicide a couple of times in the year leading up to this.

### **Bakewell**

Eventually he is assessed by two psychiatrists who diagnose him with depressive illness and suspect he has had a psychotic episode. He pleads guilty to the charge of manslaughter on the grounds of diminished responsibility.

### **Lucy**

I was relieved. For the first time I knew he was in a place where he was safe and where he couldn't harm anybody else as well.

### **Bakewell**

Lucy and her sister visit their father where he is being held under the Mental Health Act at a high-security forensic psychiatric unit.

### **Lucy**

It's quite an intimidating place to visit and on top of everything else intimidating given what he'd done.

### **Bakewell**

Then, after three years Lucy's father's social worker gets in touch, out of the blue, to say that his case has been taken to a tribunal and his time in the psychiatric unit will soon come to an end. He is to be released back into the community



## **Lucy**

We were alarmed to hear that he was being released into the community and fearful. We had absolutely no understanding of why that decision had been made. He's still unwell, he's not rehabilitated and in a way I feel like he wasn't being protected as well.

## **Bakewell**

Lucy calls her father's support team to explain her concerns and they visit her at home. They explain that her father is being released on health grounds because he has received a new diagnosis - Huntington's disease.

## **Lucy**

I remember being told that he didn't want us to know and I remember saying – don't worry, I won't say anything. At least we know what's wrong and he's going to get better. At that point I had absolutely no understanding of Huntington's disease.

## **Bakewell**

When the team leaves, Lucy decides to find out more about Huntington's.

## **Lucy**

I went on the internet, did some research and discovered what Huntington's disease was. And everything completely fell apart again, I completely freaked out, I just – I just couldn't believe it. It made sense and I then understood that he was unwell with this and had been for some time but I just didn't know how I was going to survive having to cope with another thing like this.

## **Bakewell**

Huntington's disease is a degenerative, fatal brain condition. The chief executive of the Huntington's Disease Association explains.

## **Chief Executive – Huntington's Disease Association**

The most common symptoms of Huntington's disease tend to be things like people having difficulty swallowing, difficulty speaking, their mobility will become affected and so they have difficulty walking. But also the cognitive and mental health problems develop and so people become very irritable, they might become aggressive, depression is a very common symptom.

**Bakewell**

Tragically for Lucy, it's also a condition which is passed genetically from one generation to the next.

**Chief Executive – Huntington's Disease Association**

If a parent has Huntington's disease you have a 50% chance of inheriting the gene that causes the illness. If you inherit the gene that causes the illness you will go on and develop symptoms. The prognosis is usually 10-15 years from onset of symptoms, some people will live to about 40-45 years old depending when they start the symptoms.

**Bakewell**

Lucy realises with horror that she herself is at risk of having inherited the condition.

**Lucy**

I can't even remember how I got through this period of time. I was just utterly distraught and I spent so much time crying my eyes out.

**Bakewell**

She's unsure whether to have the test to find out for certain whether she carries the gene or not. She refers herself to her local genetics counselling service.

**Lucy**

I remember sitting in the waiting room, looking around with all sorts of genetic information on the wall. I had to fight with myself not to absolutely lose the plot. And the genetic counsellor called me in and I just burst into tears and explained everything that had happened.

**Bakewell**

Lucy's genetic counsellor.

**Genetic Counsellor**

She had lost a close family member, she also had been told that her father had had Huntington's disease, she also knew that she was at 50% risk. That is a lot of bad news to deal with at any one time and for anybody coming to see us you would want to give them time to truly reflect on the impact of having such a test and not make that decision too quickly.

## **Lucy**

Continually I was debating with myself – do I test, do I not test, do I want to know, do I not want to know – what does it mean for other family members. And still at this point grieving for the loss of such an important person in my life, it was really difficult.

## **Genetic Counsellor**

An individual may think – well if I have the test and it's negative I can forget all of this. And that's tantalising there the whole time but of course the flipside is the cost of the results being opposite to that has profound effects for individuals.

## **Lucy**

Basically if you get a positive result that's it for the rest of your life and living with that daily just seemed like a massive burden and something that would be a huge struggle. Whereas if you don't test you've got the chance that you don't have the gene.

## **Bakewell**

Lucy decides not to have the test – not an uncommon decision at all. The Chief Executive from the Huntington's Disease Association again.

## **Chief Executive – Huntington's Disease Association**

Less than 20% of people who are at risk to Huntington's disease choose to go on and have the genetic test. And that's for all sorts of reasons but mainly because once you actually find out that you carry the gene you can't take that knowledge away and although it does tell you that you're going to develop the illness it can't tell you when that's going to happen or what symptoms you're going to have.

## **Bakewell**

Lucy begins to worry about developing symptoms: she notices things most of us take for granted and they make her anxious.

## **Lucy**

Everything you do, if you lose a car key or you forget something, anything, if you trip over on the street, everything you attribute to – oh well that's it, I'm about to die. I was just terrified that I was going to suddenly get Huntington's disease.

## **Bakewell**

The genetic team offers Lucy a neurological assessment. It shows that she is symptom free and she is relieved. But her risk of carrying the gene continues to play on her mind. Two years later, Lucy returns to the genetic counsellor.

## **Genetic Counsellor**

Lucy returned saying that she had decided that the time was right for her to be tested for the Huntington's gene. She felt she couldn't cope any longer with the not knowing and that as difficult as a positive Huntington's result was she felt that was going to be better to deal with.

## **Lucy**

Being put in this situation without any choice I was just trying to take back some control of my own life and that's basically why I made the decision in the end to do the test.

## **Bakewell**

Let's return to our panel now. With me still are Deborah Bowman and Jonathan Roberts.

And before we talk to you Jonathan, Deborah I've got a question for you. Now Lucy's father was convicted of manslaughter with diminished responsibility but it also seems that the doctor thought he had the capacity to make his own decision about medical information – isn't that contradictory?

## **Bowman**

I can see why it might appear so but capacity's decision specific, so the diminished responsibility defence would pertain specifically to his mental state in relation to that crime at the time when he was thought to be psychotic. We don't know when the decision to test was taken, he'd been an inpatient, he presumably had received treatment and may well have had capacity to make some decisions but not others, that's no unusual.

## **Bakewell**

Now Jonathan Lucy's been suddenly plunged into this dreadful situation about the Huntington's disease, it's a death sentence and there's no treatment now that's very different from Andrew's situation, isn't it, with the Lynch Syndrome. So how is the genetic counsellor's role different for one

and the other?

### **Roberts**

Huntington's disease is one of the most challenging cases that genetic counsellors face and I think one of the reasons for that is that there's not always a clear benefit to whether you should test or not to test outside of whether it's the right decision for that patient. So with Lynch Syndrome there's a clear benefit in terms of early screening. With Huntington's disease that clear benefit isn't there. So the most important thing is that a patient makes the right decision for them, that they're in control, that they've made an autonomous decision.

### **Bakewell**

Well we'll learn about that now because joining us is Matthew Ellison, who's the founder of the Huntington's Disease Youth Organisation.

Now Matthew you yourself are a Huntington's disease gene carrier, what was the background against which you went for the testing?

### **Ellison**

My father had Huntington's and I'd known that he had Huntington's since I was seven and it's always been in my head when I've been growing up that I'm at risk. I didn't really know that was 50% but I knew there was a possibility that this could happen to me. And it was when I got a bit older, I tested when I was 19, so when I was about 17, 18 I started doing more research about testing and my risk and thinking about that.

### **Bakewell**

We've heard that less than 20% of people go ahead and have the test when they know they're at risk. What convinced you that testing was a good idea?

### **Ellison**

Most people who don't want to get tested they kind of want that hope, they kind of want that normal life, kind of think – well it's a possibility, it's a risk but so many things are a risk that we don't really know about so we'll just leave it and see how life goes. But I think there's also that small percentage, like me, who do want to know that information and think well okay I can actually plan my life around this so I know that I might not be able to do things when I'm 35, 40, so let's try and do things now.

**Bakewell**

Deborah Bowman, is knowledge always a good thing?

**Bowman**

It's impossible to say definitively one way or the other. Some people have a greater tolerance for uncertainty than others. For some it's hope, for others it's intolerable anxiety. I think the other really confusing thing is it might be both for the same person. You might both want to know and not know. And that's why autonomy sounds so simple and it's so complex because actually we're all very complex and we're always shifting in what we think we might want in a hypothetical situation feels very different when it's an actual situation. And what we want when we're 20 might not be what we want when we're 40 and so on and so forth.

**Bakewell**

Jonathan?

**Roberts**

We're talking about whether knowledge is always a good thing. I think control is often a good thing and for some people control is choosing not to know, for some people control is choosing to know. And I think people are most at risk of an adverse reaction to a test result when they haven't felt in control.

**Bakewell**

I get the feeling Matthew that you feel in control.

**Ellison**

I do, yeah, it gave me control. I used it as motivation to do something with my life and to kick on almost, it was an inspiration for me, yeah.

**Bakewell**

So let's get back to the story of Lucy who has been tested for the Huntington's disease gene and returns to the clinic to get her result.

**Lucy**

I went to the clinic and was taken into the room by the genetic counsellor, which I remember vividly, and she sat me down, opened up the paperwork and said I'm really sorry to tell you that you have the gene, the Huntington's

disease gene.

### **Genetic Counsellor**

It is very hard to give somebody a result like this when you're doing genetic counselling. I counsel patients for all sorts of different reasons and very often there's a happy outcome but with Huntington's you have this lengthy disabling deteriorating condition where there's no treatment.

### **Lucy**

I can remember walking the short journey home feeling completely unsteady and shocked. I got myself home and just got into bed and spent the rest of the day crying my eyes out.

### **Bakewell**

And it's not only herself that she's concerned about. Lucy has a three year old son. He was just four months old when Lucy discovered her father had Huntington's disease. Planning ahead for her son was partly why she had the test.

### **Lucy**

Sometimes I just lie at night watching him sleeping wondering what the future holds and how this little person is going to cope. Finding out that I am gene positive and that he's 50/50 risk is going to destroy his life. If I'd known in the pregnancy I would have terminated to not put my son through this situation.

### **Bakewell**

There are a number of choices open to people with Huntington's disease who want to have children.

### **Chief Executive – Huntington's Disease Association**

People who carry the Huntington's gene can have children naturally and their children will have a 50% risk of inheriting the gene. The other option is something called pre-implantation genetic diagnosis and what that means is basically an IVF procedure, so a sperm and an egg are put together, an embryo is formed and that embryo is tested to see whether or not it carries the gene and then only embryos that don't carry the gene are implanted back into the womb.

### **Bakewell**

It's also possible to test for Huntington's disease during pregnancy after a natural conception whereby fluid around the baby is tested for the genetic fault via a process called amniocentesis.

But finding the genetic fault in an embryo automatically reveals that at least one parent must have it too. And most at risk adults choose not to find out if they have the gene.

For these people, who don't want anyone to know whether they have Huntington's, there is another option if they want to be certain that their offspring does not inherit it – it's called exclusion testing.

### **Chief Executive – Huntington's Disease Association**

What exclusion testing does is not actually test the specific gene but areas around the gene and it tracks back through generations, so it can tell whether a pregnancy is at a high risk of having the Huntington's gene or a lower risk, but it's not a 100% accurate, so there is a possibility that pregnancy could be terminated and actually the child isn't carrying the gene.

### **Bakewell**

But for Lucy and her son it's too late for any of these options.

After she becomes aware she has Huntington's, she decides to take legal action against her father's medical team. Lucy contacts a lawyer.

### **Lawyer**

Lucy has instructed us to bring a claim of negligence against the various hospital trusts that were treating her father who knew about his potential diagnosis and who chose not to disclose that diagnosis to her.

### **Lucy**

Not only was he in the active stages of the disease he'd killed the family member and is therefore not somebody who should be making any decisions for us as a family. The right thing would have been to disregard his wishes and to think about us as a family.

### **Lawyer**

There are competing rights here. There are the rights of the father to have his medical information kept confidential and there are the rights of the



family member, in this case Lucy, to learn of that information so as to take action to avoid very serious and potentially life threatening harm. For me, personally, it is by no means obvious that the right of an individual to have his medical information kept confidential should in any way trump the right of a relative to have information that will enable them to avoid very serious harm.

### **Bakewell**

For Lucy, having the information about her father's Huntington's disease would have allowed her to make an informed choice about her pregnancy and the possibility of passing on the faulty gene to her son.

### **Lucy**

To grow up knowing you're a 50/50 risk with a parent who is going to become unwell and there's no treatment, there's no cure and also you know that your grandfather has committed manslaughter whilst ill with this disease, it's a terrifying place to be and that is never ever going to be okay and there's nothing that can be done about that now because nobody bothered to open their mouths.

### **Bakewell**

Lucy's son is now three years old. Obviously he doesn't yet know his mother and grandfather carry the faulty gene.

### **Lucy**

After everything I've been through I can't ever imagine at the moment a time when I can sit him down. There's never going to be the right time for me. I know at some point in the future it is something that will have to be – it will have to be discussed.

### **Bakewell**

Telling a child about the risk of Huntington's disease and getting a test to determine whether they've got it are two very different things. Testing in childhood is not recommended. The Chief Executive of the Huntington's Disease Association.

### **Chief Executive – Huntington's Disease Association**

If you allow parents to make a decision for their children then you're taking away the choice of that child when they become an adult of making that decision for themselves.

**Bakewell**

Children are also not tested for Lynch syndrome.

In our first story, Elizabeth feels that her husband having died the most pressing issue now is screening her children as early as possible for signs of cancer. She knows all too well the impact of not having that screening.

**Elizabeth**

Not a day goes by that I don't wish things different. There's so many different times at which it could have happened, that somebody could have put two and two together and the outcome might have been different for Andrew. I believe he'd still be here, I believe that he would have gone for screening because he was that sort of person. And my children would have their daddy.

**Bakewell**

But it's not normal procedure to screen for adult onset conditions in childhood. The genetic consultant.

**Genetic Consultant**

In childhood the child isn't at risk, there are no health implications for children from this and so we would prefer to wait until the child is of an age when they can have a discussion with us and understand the implications of the testing, understand what the condition means and can make an informed decision.

**Bakewell**

For these reasons, genetic testing for the faulty gene is only recommended for adults. For anyone who tests positive, their own screening for early signs of disease usually starts 10 years before the age at which the youngest of their relatives developed cancer. But there's an age limit with this recommendation – the rule is that screening should start no younger than 25.

Elizabeth thinks her children should be screened much sooner than that.

**Elizabeth**

In Andrew's case one of his relatives was diagnosed at 16 with bowel cancer. And so for our children I think that the screening should start

before they're 18.

### **Bakewell**

Although Elizabeth feels that screening for cancer should start before it's recommended at the age of 25, she's not ready to have them tested for the gene. They're just three and five years old.

### **Elizabeth**

Would I want to know that both of them have got Lynch Syndrome now? I'd want to know if they both didn't but I certainly wouldn't want to know that one of them did. Sometimes you wish you could see into the future, don't you, but actually I think it's just as well we can't.

### **Bakewell**

For Elizabeth and her children, that future is tragically one without Andrew.

Now let's return to our panel: Deborah Bowman, Jonathan Roberts and Matthew Ellison.

Jonathan, we just heard that Elizabeth, the mother in the case, wants her children screening for early signs of cancer but not testing for the faulty gene. Would you ever do that – screen without knowing they're at risk?

### **Roberts**

For some families who are at increased risk of bowel cancer their family history alone means that increased screening would be recommended. The advantage of having genetic testing is it means you can avoid doing colonoscopies in cases where you don't need to. So colonoscopies do have a small risk and obviously they're particularly uncomfortable and they're not a procedure you want to be doing a lot of if you don't have a very good reason for doing it.

### **Bakewell**

Should screening start much earlier for Andrew and Elizabeth's two children because one family member developed cancer at 16?

### **Roberts**

The point at which screening starts is a question that needs to be managed between the clinical genetics team and the oncologists and surgeons because with all of these things it's balancing different types of risk.

**Bakewell**

Now Deborah, children are not usually tested for the gene faults that cause adult onset conditions like Lynch Syndrome and Huntington's until they're 18 – why is that?

**Bowman**

One relates to capacity and choice, although of course actually you can be quite a lot younger than 18 and have capacity and make reasonable choices. I think there are other reasons. I mean screening is not a neutral act, there are risks and benefits and actually the younger somebody is the more likely you are to think this is a decision that could or should wait.

**Bakewell**

Okay, well let's move on to telling children about a family condition. What is the age when you believe that a child can take on complicated information? Jonathan?

**Roberts**

I think it depends a little bit on how the family talk about a condition and what their story is. For some families this is something that is known about from a young age and children kind of grow up having little bits of information and more and more as they get older. For other family members it can feel like a bolt from the blue and I think one of the challenges is when there's a piece of information that's new and the parents aren't quite sure how to communicate that to the kids and then they feel as if they has to be a sort of one event.

**Bakewell**

Matthew, at the Huntington's Disease Youth Organisation you work with a lot of teenagers, the young people who are at risk, now do any of them wish they didn't know about having Huntington's in the family?

**Ellison**

Generally we don't get that, we don't get people contacting us saying I wish I didn't know that Huntington's was in the family, we get contacts saying I've just found out and I'm very scared or things like that. What we do at HDO is offer appropriate information to learn about Huntington's disease via our website because we get so many young people contacting us saying oh I've been on Google or whatever and typed in Huntington's

disease and just got the scariest information and fatal, no cure, it's terrifying for them. So we try to educate children and young people in an appropriate way for them, that's not going to be scary for them but it is going to educate them on what they're facing.

### **Bakewell**

Okay well so far we've been talking about testing and telling, what about preventing the gene being passed on in the first place, and avoiding it? Jonathan, what are the options for someone who's at risk of carrying a faulty gene and doesn't want to pass it on to their children?

### **Roberts**

So there are two main options. The first is testing a pregnancy using amniocentesis, if the pregnancy is found to have the HD gene then that pregnancy can be ended. The second is something called pre-implantation genetic diagnosis, this is a form of IVF but before any embryos are used they're tested and embryos that have the HD gene are discarded. There's a third option, that's a rarer option, if somebody really doesn't want to know their status but wants to test their pregnancy they can have something called exclusion testing. So this is done by using genetic markers from grandparents and you can find out if the pregnancy has inherited the same genetic material as the grandparent who had HD. It doesn't provide a clear result as amniocentesis or PGD, it can only tell you whether your pregnancy is at an increased risk or not. So the uncertainty goes up but the advantage is if you really don't want to know your status a parent can have exclusion testing.

### **Bakewell**

Matthew.

### **Ellison**

I got married two years ago and our plan was always to have children but not at risk. So we've looked at the options that Jonathan just explained and we've gone with PGD – which is the pre-implantation genetic diagnosis. So we're currently going through that at the moment. You're not guaranteed to be successful, far from it, it is a difficult process to go through as well for the woman, lots of injections, sometimes people say oh you don't need to pass on the risk and actually these options aren't simple, they're not easy for most people.

**Roberts**

I think that's a really important point when discussing the options with families. Each option has its own risks, its own ups, its own downs, same positives and negatives, and it's about couples choosing which of those they want to take and often there's no right answer, it's a very, very personal decision.

**Bakewell**

Now another thing we've heard about in this case is about Lucy is that she's bringing a legal case against her father's medical team. Now Deborah, why is her case contentious?

**Bowman**

The way in which the law is usually framed is that the duty of care relationship is about the doctor and his or her patient. So it is unusual in that it's seeking to extend that duty and then say that the way in which that duty was discharged or not was negligent.

**Bakewell**

We've heard Jonathan say that although the obligation is to keep the patient's information private there would be a situation in which a counsellor could breach it. So if this comes into law and Lucy is successful what will that do to the situation?

**Bowman**

Okay, so I think one of the reasons that there haven't been duty to warn cases, that's the term for these sorts of cases, has been a concern that if one were to impose on clinicians an obligation that extended beyond the patient in front of you that would distort the relationship with the patient in front of you, that they may not be honest, that they may not come and see you in the first instance, that they might give you partial information, that they wouldn't be able to be certain that you were looking after them, rather than looking after this wider population. And there are implications for how doctors and patients relate to each other of these sorts of judgements.

**Bakewell**

Now Jonathan if the law were to change how would it affect you?

**Roberts**

I think the distinction between there being a duty to inform and it being

justified to inform is a really important one. So I think in clinical genetics, currently working from the principle that there are certain cases where it's justified to breach confidentiality, the change from there then to being a duty is a massive shift and I think there's potential for that to change that level of trust.

**Bakewell**

Well wait a minute, we're dealing with a fatal illness here, we're dealing with an illness that has no treatment, that is going to be fatal. So there surely is a moral obligation to save life?

**Roberts**

I think the justification for breach of confidentiality is absolutely there and you could argue that moral obligation to give – to save lives provides the justification for breaching confidentiality and sharing data. And there's perhaps an argument that clinicians should feel more confident about doing that and...

**Bakewell**

It's a strong case isn't it?

**Roberts**

It's a strong case and there's an argument to be made that clinicians perhaps come down too hard at the minute on balancing confidentiality against sharing information. But I think there's a big difference between saying clinicians should be more aware of moral cases where they can breach confidentiality to saying they have a legal duty to do it, I think there's a very different perspective.

**Bakewell**

And it's very interesting, isn't it Deborah, the whole field of genetics, this field is only going to get more complicated and moral obligations, legal obligations, are going to multiply.

**Bowman**

I think there's something very interesting about genetics that it really challenges that idea of the dyadic two people relationship on which most medical ethics is predicated.

**Bakewell**

It's clearly going to be a very complicated future that we face.

Thank you to my panel: Deborah Bowman, Matthew Ellison and Jonathan Roberts.

But now let's find out what happened to Lucy.

She is bringing a negligence claim against the hospital trusts that treated her father.

She feels that her father was not able to make decisions for himself because of his illness, and that he should not have been allowed to make decisions for the family after he had killed a family member.

At a preliminary hearing, the trusts made an application for the case to be struck out, saying that it should not be heard. Lucy's lawyer.

### **Lawyer**

The basis of their application was that there is no duty of care owed by a doctor to anyone other than their patient. At that hearing they persuaded the judge that that was right and that there was no fair, just or reasonable reason why such a duty should be found in Lucy's case. Following that decision Lucy made an application to the Court of Appeal that she should have permission to appeal and that permission was granted. Unfortunately the hearing is not listed until spring 2017.

### **Bakewell**

When the appeal is heard it stands a chance of being a landmark legal ruling in the sharing of medical information.

### **Lawyer**

If Lucy were to win this case it would be a very significant change in the law in relation to duty of care owed by doctors to their patients. The way that I conceptualise it in my own head is that what this is in fact doing is redefining the definition of patient, so that yes doctors only owe duties of care to their patients but that in the context of genetic medicine the patient is defined not only as the person who provided the genetic sample but also as all those people who are affected by the information contained within that genetic sample.



**Bakewell**

For Lucy, the court case is just one of a whole list of things she worries about.

**Lucy**

It really goes in phases. If everything's going wrong in life a bit you think oh god I'm going to get ill soon and it all gets on top of you. And then if the sun's shining and you've had a nice holiday, I have days where I think – they might develop a cure and I will live until I'm 90 and see my grandchildren. But I don't allow actually myself to hold on to that, I just think it's every day as it comes at the moment, deal with one thing at a time. I don't look to the future, I don't really make plans, it's something that is out of my hands unfortunately.

ENDS