

# Journal Pre-proof

Genetic testing and family entanglements

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PII: S0277-9536(22)00163-0

DOI: <https://doi.org/10.1016/j.socscimed.2022.114857>

Reference: SSM 114857

To appear in: *Social Science & Medicine*

Received Date: 6 August 2021

Revised Date: 18 February 2022

Accepted Date: 22 February 2022



Please cite this article as: Dimond, R., Doheny, S., Ballard, L., Clarke, A., Genetic testing and family entanglements, *Social Science & Medicine* (2022), doi: <https://doi.org/10.1016/j.socscimed.2022.114857>.

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## Genetic testing and family entanglements

### Abstract

The development of the 'new genetics' in the early 1990's opened up a new space which required some patients and families to understand and navigate genetic testing. The social science literature that has grown alongside the 'new genetics', now spanning more than thirty years, has continued to explore and question assumptions about attitudes and responses towards genetic technologies. In this article we highlight how individual experience of genetic disease and personal responses towards genetic technologies can only be understood by considering their context. We focus on the rich literature on family within sociology, science and technology studies, anthropology, and family studies, to explore the myriad ways in which family is implicated in the patient experience of genetic testing. We explore these connections by drawing on a set of interviews held with individuals who have undergone a predictive test for a genetic condition, including Huntington's Disease and breast cancer. Five themes were developed: family disclosure, family gatekeeping, going for testing, individual and collective communication practices, and receiving a negative test result. To conclude, we highlight how these connections might be considered through the lens of entanglement, explaining the complex mechanisms through which family and genetics are intimately entwined.

### Key words:

Genetic testing; entanglement; family; communication; 'new genetics'; social science; Huntington's Disease; United Kingdom

Word count: 9038

### Introduction:

The aim of this article is to explore the entanglement between individual and family. It draws on interviews with individuals who were at risk of inheriting diseases such as Huntington's Disease HD or breast cancer, and who were offered the opportunity to take a predictive test which would reveal their diagnostic status. Within the clinic, family plays an important role in the diagnostic process, particularly for example in the case of rare genetic disease or for tracking patient development. Identifying a 'family norm' (McLaughlin and Clavering 2012) involves working out what might or might not be classified as a 'symptom'. The bodies of family members can also play a role in informing a diagnosis, they are measured explicitly and observed implicitly and, in some cases, relatives can receive their own diagnosis through this process (Dimond 2014). Family trees known as pedigree charts are often used to map biological relationships and track the pattern of inheritance. Mapping pedigrees centralises the individual within the context of social relationships, a process which continues to be employed alongside the development of genetic testing capacity. Family narratives are important in shaping this mapping, with relatives often being co-opted into the production of a particular and specific diagnostic journey for that patient, for example, by being asked to provide information about family history. Such genetic connections complicate the meanings of patienthood. The blurred boundaries between the personal and collective raise questions about ownership of genetic information, creating an ethical dilemma for policy and practice (Dheensa et al. 2016). These dilemmas are particularly acute in the context of pre-symptomatic testing for late onset conditions. A large body of literature, spreading across several decades, has focused on clinical and personal implications of genetic disease, recognising the need to support health professionals in encouraging communication (Mendes et al. 2016) and psychological adjustment (Leite et al., 2017). We agree with Koerner et al. (2010 p187) in their recognition that family are "participants rather than mere bystanders in the counselling process" with the implication that "it is not possible to talk to a person about genetics without talking about family". Understanding family communication is not just important for health professionals in

supporting disclosure to relatives 'at risk' (Young et al. 2019) but the role of family is central for understanding the personal experience of genetic disease and genetic testing.

### Family beyond the genetics clinic

The way in which an individual is entangled with others in the context of genetic disease has been understood through the framework of 'risky relations' (Featherstone et al. 2006). It is through these connections that diagnostic and risk information is communicated, confirming multiple routes for transmission, translation and assimilation of biomedical information. The role of family becomes crucially important for understanding the experience of genetic disease; it is an essential influence beyond the reach of the genetics clinic. The communication of information within the family has been explored in terms of barriers and facilitators, risk and responsibility, adaptation, and broader family communication strategies. When communicating to family members about genetic risk, Gaff and Bylund (2010) found that individuals often planned their strategy, thinking about who to tell, when and how, with reactions anticipated, where communication is a process rather than a single moment. Facilitators of communication include having close relationships with family members, feeling a sense of duty to others in the family, and being supported by practitioners in telling others (de Pinho Rodrigues 2020). Barriers include lack of understanding of the importance of communicating risk information to family members, and feelings of guilt, anxiety, grief and blame (Young et al., 2019). Genetic technology has reshaped the meanings of responsibility, including the expectation of making informed decisions based on new knowledge of genetic risk. The concept of 'genetic responsibility' has been used to consider the role of genetic information in the formation of social relationships and how it is entwined with expectations and obligations towards others (Arribas-Ayllon, Sarangi, and Clarke 2008; Weiner 2011). This includes for example, the normative expectation that a person would, and should, seek out genetic knowledge if it is available, and that

health information will be communicated to others deemed 'at risk', and is representative of a broader shift towards responsabilization within medicine (Leefmann et al. 2017).

#### Family studies - Doing and displaying family, and 'genetic thinking'

Whereas literature exploring the social aspects of genetics has developed alongside genetic technology, we acknowledge the influence of a longer history of a body of knowledge developed through 'family studies'. These prioritise the social over the biological, recognising diversity in family forms, where doing and displaying become heuristic devices to explore the nature of the bonds between individuals. Morgan (2011) highlighted how family was not just about 'being' a family. Family, he thought, was formed through sets of activities within a shared system of meaning. Building on Morgan's work, Finch (2007 p67) explored the process of 'displaying' family, pointing to the "social nature of family practices, where the meaning of one's actions has to be both conveyed to and understood by relevant others if those actions are to be effective as constituting 'family' practices." The concept of display was important because it was the process through which individuals and groups gave meaning to their actions - doing 'family things' enabled these relationships to be defined as 'family relationships'.

Nordqvist (2017) has built on the work of family theorists to develop ideas between the everyday life of the family and the role of 'genetic thinking'. She explored how ideas about genes and genetics were made meaningful in the context of families of donor conceived children, where the donor might or might not be a presence in their lives. For Nordqvist, 'genetic thinking' renders genetic relationships meaningful through processes of everyday family life, including identifying family resemblances between the child and family members, and discussion of inheriting 'family traits'. Nordqvist concludes that there is a close relationship between genetic talk and social practice, where genes 'do not speak for themselves'. Importantly, she recognises how it is through these

practices that genetic connections come into being, as she explains “genetic connections are *rendered* meaningful and so *become* meaningful” [emphasis in original, p 878]. In our work we show how biological relations also need to be rethought in terms of how those relationships are experienced – through displays of connection, responsibility, surveillance and emotion. Whereas Nordqvist says we need to think more about sociological implications “in which genetic thinking is rendered meaningful in contemporary family life”, we do the opposite in this paper. Here, we suggest that we need to think about the ways in which family practice is rendered meaningful in a context of the genetics clinic, and a society, where genetic relationships are often prioritised. In this article we consider these entanglements through the themes of family disclosure, family gatekeeping, going for testing, individual and collective communication and receiving a negative test result.

## Methods

This article draws on a set of interviews which formed one part of a larger ethnographic project, including observation of genetic counselling sessions, to explore how individuals made decisions about predictive testing as they went through the testing process. Predictive testing occurs when an individual has a genetic test which can diagnose a late onset condition in the absence of physical symptoms. Unless the result emerged as an incidental finding of a genetic test performed for another reason, such testing would only be performed when an individual is already known to be at high risk of a specific condition through a knowledge of their family history and the inheritance pattern of the condition. The nature of the testing and disease expression is beyond the scope of this article, but please see Clarke (2020) for further information. Participants were recruited through four regional genetics centres in England and Wales. Potential participants received a project information sheet with their appointment letter and were approached by a member of the research team after their clinical appointment to discuss the research project and address questions. Interviews took



place either in the person's home or online. Most of the interviews were conducted by Author 2, who is a social scientist, with some interviews conducted by Author 3 who is a psychologist.

Thirty people were recruited at the predictive genetics clinic to take part in the broader ethnographic project, of which seventeen agreed to be interviewed. The majority of these were engaged with clinical genetics because they were at risk of having Huntington's disease, while others were at risk of breast cancer or other more rare conditions. Most of the interviews took place six to eight weeks after their clinic appointment when they had received their test result for a disease that they were at risk of developing but without experiencing symptoms. The project did not change the participant's route through clinical genetics, or require any additional testing or appointments, although a genetic counsellor was available to them at their appointment to talk through any questions they had about the research. Ethical approval was gained through Wales Research Ethics Committee 1.

Transcripts were analysed using thematic analysis (Braun and Clarke 2021). The team began by reading the transcripts and meeting to discuss what aspects each found most interesting. The authorship team was made up of two social scientists, a psychologist and a clinical geneticist, which meant we all had diverse personal knowledge, from literature or experience, which could complement the analytical process (Becker, 1998). It was during this discussion that 'family' was discussed as an analytic category, and it was agreed that analysing the data in relation to this category could produce significant insights and form a valuable contribution to the literature. Author 1 began by reading each transcript several times. Using a basic word processor, all the references to 'family', 'family members' or situations which involved others were highlighted, and thoughts about the extract and any potential links to literature were noted. This process enabled the researchers to 'think' with the data, moving from the discrete data to more abstract ways of thinking, and a dynamic process described as 'reduction, simplification and complication' (Coffey and Atkinson, 1996). The overall process of thinking about each extract and applying codes to encapsulate their

meaning, resulted in a list of 15 codes, including communication general, communication barriers, diagnosis, risk, secrets, lies and protection. The authors then met again to discuss the codes and to ensure that all members shared the same interpretations of the data, including strong contributions from those who had conducted the interviews. The team discussed how the codes related to broader themes to help extend understanding of the role of 'family'. The result of these discussions was the delineation of the five themes explored in this article: family disclosure, family gatekeeping, going for testing, individual and collective communication practices, and receiving a negative test result.

## Results

### Family disclosure

The focus in this first section is how others in the family act as gatekeepers and communicators of information. Many of our participants described how it was through relatives that they initially found out about the disease and the implications for their own risk. Thus, how respondents might begin their patient journey, by finding out about the disease and their own genetic risk, was dependent on others.

Some participants found out about the disease following the diagnosis of a parent. Disclosure was often made by the non-affected parent, and often in the context of broader communication difficulties within the family. One participant (PD202) describes how he was told about his father's status as a teenager, which was communicated to him as a 'secret' by his mother in the context of his parents' frequent arguing, "they used to argue a lot and eventually she actually told me that, he's got it, and she said, 'don't tell him I told you this, but, he's got Huntington's". Another participant (PD308) was a child when she was told by her mother about the diagnosis of her estranged father, with her account describing family tensions about the appropriateness of

disclosing to children. These issues have long been debated within clinical genetics and academic literature (Clarke 1998) and this continues. The participant explains how her mother's sister disagreed with her mother for telling her at that age: "My aunty still says to this day 'I would never have told you when you was that young', but then would you tell me? There is no right time for that". The estrangement of her father was significant throughout her account, including when talking about testing within the family. She explained that, as an adult, she is the only one of her siblings to retain a relationship with her father and is the only one to have been tested.

These accounts are important for highlighting the gate-keeping work of family members, who are often the ones making decisions about whether, when and how to communicate risk information. Many of the participants were informed about the disease as an adult. This was the case for one person (PD209) who reported that she was told about the disease by her cousin, to encourage her father to be tested:

They, cos we didn't really know about Huntington's until dad had the fall and my, my cousin just text me and said "Get your dad tested for Huntington's, cos we think mum's got it, but she won't have the test, but get your dad to have it".

There are clearly different routes and purposes to communication. At times, discussion between family members resulted in the 'correction' of previous understandings of the disease and its pattern of inheritance. This was the case for one participant (PD201) who described how she was told vital information by a cousin's daughter, which contradicted her own understanding of disease transmissibility:

I was asking about how her dad was and she said that he was going for these different tests for his balance and things like that then really, so it was only from asking from her that she

mentioned about this Huntington's ... and I then told her in response then you know, "oh, no, it's not fifty, fifty, it only comes from the male side of the family" because that's what my dad had told me and she said "well that's not what they've told us" which is what led me and my dad to then obviously look into it ourselves then. So completely by, if it hadn't been for my cousin having the diagnosis now, I would never have had any inclination to wanna be tested.

In this case, the person had known about HD being in the family but had been told by her father that it was transmitted through the male line with the implication that she and her father would not be at risk. Knowledge of the genetic risk for the condition relied not just on her cousin being diagnosed but having that conversation with her about genetic risk. Thus, the cousin acted as an interpreter to genetic knowledge, by correcting 'lay' understanding, an explanation that had been communicated through the family and accepted as fact. Participant accounts suggested a cascade of information following a diagnosis. This was the case for one person (PD401), who described how her sister had shared information about the identification of BRCA with family members, identifying aunts, cousins and a brother as part of, and responding to, the information flow:

My sister phoned me up to say that she'd been in touch with [female cousin], and my auntie and everybody, they'd sort of rung round everybody and ... so that was when they told me that I needed to get it sorted really. ... I think my auntie and her daughter, my brother had already arranged for the testing and things.

Several participants highlighted a sense of responsibility to communicate risk information to family members. One participant (PD404) for example, experienced health problems following pregnancy, and was tested for MODY (Maturity Onset Diabetes of the Young). The diagnosis of a family member with MODY was significant in the context of a family history where elder relatives had possibly been

wrongly diagnosed with the more common kind of diabetes. Following her positive diagnosis of MODY, she explained her feelings of responsibility: “I felt like I should pass that information on, so that they’ve got the option and they’ve got the kind of knowledge and understanding of what the situation might be.” Another participant (PD402) described how he was asked to communicate test results by his brother, who was more distant from family members. This participant’s brother tested positive for the MAX gene (which links to an increased risk of developing tumours), and asked him to communicate the result to others because he was not in touch with them: “[brother] asked me then to go round everybody, rather than send it [photocopy of letter] to them, he wanted me to go round to every other family locally to tell them ... he doesn’t talk to any, he very rarely talks to me.” Of course, genetic factors do not always exert this sort of effect or operate in this way, but here the tie that binds people through genetic risk appears to overcome family divisions, with the better-connected brother being used as a resource, to act on his sense of responsibility by proxy. We can see how this is the case in communicating risk information to others, but we have heard similar accounts, where siblings or cousins might accompany each other for the genetic test, even when relationships are strained.

### Family Gatekeeping

Part of the important work of the clinic is to counsel patients in identifying the reasons why someone wishes to be tested, to think about the right time to be tested, and to anticipate and plan for their different futures depending on the result. We know from the literature that testing is pursued for many reasons, including to enable practical arrangements and prepare others for the physical burden of future decline if positive. Sometimes individuals want to be tested so that genetic information can inform the decisions of others, particularly younger generations. In this project the reasons people gave for testing varied, with some acknowledging the needs of others. This was the

case for one participant (PD210) who explained her reasoning in terms of her own family planning, but also referred to the expectations of her father and grandmother:

I just wanted to know like if I can have more babies in the future or ... because I wouldn't want to pass the disease to them. So I think that is what would [have] made me do it and also my, my, my dad's mum she wanted me to do the test and my dad as well, just because they wanted to know.

Although her justification for wanting to be tested was rooted in family planning, it was clear that there were other pressures from family members. These might not be the ultimate reason why someone is tested, but it can help to explain the experience of people who are in the position of deciding whether or not to be tested. Likewise, people choose not to be tested for various reasons, sometimes choosing not to be tested *yet*, or deciding they do not wish to be tested at all. The medical futility of a late-onset test in the context of a disease with limited treatment options was noted by some participants. However, several highlighted a disjoint in the family, where a tension was created because of different opinions about testing, but where access to a test was dependent on the agreement of others. We use the term family gatekeeping here to acknowledge the key role that one member might play in access to knowledge and its dissemination. In contrast, 'genetic gatekeeper' is sometimes used to refer to a professional gatekeeping role. In the UK for example, genetic clinicians and counsellors support patients through an extensive assessment period before being tested, and before receiving the results. Whereas individuals in the family might be keenly aware of their role and responsibilities as communicators of information, on occasion the role of gatekeeper is allocated to them. Several participants mentioned the difficulties of a younger member requesting testing, but where a positive result would reveal the diagnosis of their parent, who was choosing not to be tested. This is a particularly problematic outcome for families, and for the clinic. If the parent had not requested this information, then a clinic would often aim to defer the testing of the younger adult individual (at 1 in 4 risk) until a frank family discussion had occurred and

either the parent (at 1 in 2 risk) decided to seek testing first or at least had accepted that their child would be tested and that their own status might be revealed by this. If no agreement is forthcoming, however, the intervening parent cannot indefinitely block the testing of their own offspring. Several accounts of participants highlighted this important gatekeeping function and, in all cases, the younger person wanted to be tested for future family planning. One person (PD201) had requested a test but was told by the clinic that it would be ethically problematic because her father had not been tested:

We did even ask the question, is there any way that he can choose not to know, for me to be tested, but because of all the complications in terms of me knowing then, erm, then we had to go through dad being tested first then really and my dad didn't want to.

This woman's father did not want to be tested. However, following his positioning as family gatekeeper, he agreed to be tested in order to enable his daughter to access her genetic knowledge, which she acknowledged, "if I hadn't been in the equation then probably my dad wouldn't have probably wanted to have found out". Another participant (PD219) was acutely aware of this gatekeeping role. She had attended some counselling sessions with her mother and father, explaining that the clinician had suggested that her father attend, because "he saw it as abnormal that I was the one being tested, not my dad, and probably wanted to check that the relationship was okay".

Thus the participant recognised the significance of 'doing' family in the clinic in order for her to be able to obtain testing. In another example, when asked "what made you think about doing the testing?", the participant (PD209) replied by talking about her daughter who wanted to be tested for reproductive purposes, but where "her husband's not keen on having another baby if we've got Huntington's in the family". Here she explains how her daughter had requested testing, but this was denied because she had not been tested yet:

So when she went to the doctor, the doctor said “We can’t really do it unless your mum does it” ... so in the end, I, I sort of, I just sort of said “Okay then, I’ll, I’ll go for it and um and do it”, because obviously it’s affecting [daughter’s] life.

As others before her, this account highlights how she was placed in the position of family gatekeeper, and ultimately agreed to be tested, so that her daughter would be able to access that information to make decisions about extending her family. Once again, this highlights how personal decisions, in this case the choice to be tested, are entangled with the needs and desires of other family members. The shared nature of genetic knowledge, suggesting that family is one unit, can create tensions when we recognise how the family is made up of individuals with different circumstances and different needs.

#### Going for testing

In this section we consider how going for the test was identified by participants as a significant moment, where decisions were made about who to tell and who not to tell, and what to communicate. Communicating about the test itself is an aspect of family gatekeeping which has received less attention in academic literature, and context is important to note. One recent publication about the 100,000 Genomes Project found that individuals were more likely to disclose to family members that their genome had been tested when it was undertaken in the case of rare disease compared to cancer (Ballard et al., 2020). Weiner (2011) found that discussions about testing for familial hypercholesterolaemia (FH) were not dominated by a narrative of kinship, and that individuals did not often express an obligation to communicate potential risk information within the wider family. It is possible that the significance of going for a test lies in its potential to reveal a positive diagnosis, with subsequent implications for the disease status of family members. However, we identify here that going for the test is a significant moment in itself, with participants making



decisions about who they would and would not share the information with. We acknowledge that there are different reasons for secrecy, and of course, different time spans in which information might be deemed particularly sensitive. Although the purpose of this article is not to focus on the psychological or the ethical, future research might find value in thinking through differences in individual motivations towards disclosure, secrecy and responsibility.

Several participants were explicit about the withholding of this information, distinguishing their own role in going for testing, with broader knowledge about the disease in the family. For example, as one person (PD210) expressed referring to family members, “They knew about it [disease] but they didn't know that I was doing the test.” Likewise another (PD308) explained that she had been open about her desire to be tested, yet did not want to tell them when she was actually going for the test. Thus ‘going for a test’ is marked out as a specific moment for privacy, which is rather striking when made in the context of an otherwise generally open style of communication. Participants revealed considerable emotional work in thinking about the implications of communicating this kind of information. Going for the test was repeatedly identified as a moment involving thought and consideration about who would be told and who would not be told. For example, one person (PD219) explained that she had only told her parents that she was going for testing. She had a large family, and in her account she applied a dual process of identifying family members who were at risk of inheriting HD, and those whom she would not tell. She highlights one cousin, “Me and [cousin] don't have a good relationship, so I wouldn't tell her, because I'd worry about her just being kind of malicious, as she has been in the past towards me.” But in relation to telling another cousin, she highlights several distinct issues, including the timing of disclosure and that the cousin might be surprised that she had not been informed about the test before. However, she also reveals more significant aspects beyond disclosure:

Um, I think yeah, with [cousin], I swear it feels like a sense of like I'm betraying how their family are dealing with it, so her and her mum and dad have just decided like "ignore it, it's not a thing", so then if I come in and say "hey, I had the test ... and it's good news ..." and then it would just kind of disrupt what they've got going on, and maybe she would then sort of question what she should be doing. And I'd feel like I was being disruptive.

This account reveals the complexity of disclosing information. It particularly highlights the symbolic implications of the information she is disclosing and a deep ethics of care. The feminist ethics of care is based on an interdependent, relational ontology, where individuality is understood as formed in and through relations with others (Wouters et al., 2016). Tronto (1993: 128) for example identified that to attend to the other involves setting aside one's own will "in order to recognise and to be attentive to others", helping to explain how relationships and the moral obligation an individual can feel towards another becomes the site of care. In this account, going for the test was identified as having the potential to be translated as 'disruptive', but not for her own life and life options. The participant expressed concern about how her going for a test might be interpreted as giving directions about how others should behave. She describes the potential impact for her cousin's family, and for her cousin, whom she identifies as "emotionally vulnerable" and to whom she acts as a 'role model'. On the point of being disruptive, she explains further, displaying a sensitivity towards her cousin's different life approach, "if that's what's working for them, and helping them get through life, I don't want to mess it up for them", and then later, "I feel like if I say look, I've done this, then she'll start questioning what her attitude is, and then she speaks to her parents a lot, so she'll take it back to them. Um, and it just has this knock-on effect."

This is a complex account revealing considerable thought around communication about testing, about when to share information, recognising vulnerability in a younger cousin, and reflecting on and identifying differences between her own approach and that of her cousin's family. In this

context, such emotional work is invested because of the potentially disruptive effect on others of going for a test. Another participant (PD214) was aware of the persuasive power of communication, particularly for his sister who had been ill as a child, “if we started saying to my sister ‘Oh it could be that’ then we’re kind of forcing to have the test .... and my mum was always ... er with my sister ... you know, didn’t mention it because she didn’t want my sister to feel forced to have the test.” Thus many participants highlighted an ethics of care towards family members, recognising alternative life approaches, and expressing a reluctance to challenge or influence decisions and the perceived vulnerability of others.

#### Receiving a negative test result

Participants in this project expressed personal delight at receiving a negative result. One participant (PD204) for example, reported, “I felt good, I felt it was good news, erm, it was good news for me and good news for the people around me that love me so, erm, it was all good, it was all good.” However, going for the test, and receiving the results, were mixed with other powerful emotions, where the materiality of the test linked to the past and future of family life. The test was made meaningful in the context of witnessing his father’s deterioration:

I don’t want to say it didn’t really affect me, erm, it was obviously hard seeing my dad deteriorate at the start ‘cause going from nothing wrong with him to starting to display symptoms and the reality of seeing, you know, it’s not just a test.

We have described how participants in this sample went to great lengths to demonstrate their ethics of care towards other family members, including thinking about the timing of conversations about the test and disclosing the result. Participants highlighted how they invested in the emotional care of others in the family; for the most part this is the invisible work of being and doing family. This care is

also demonstrated when receiving a negative test, revealing mixed emotions, and an ambivalence between personal joy and concern for others. Being found to be negative for the disease was tempered with anxiety about how others might feel. Whereas parents were identified as people who might feel responsibility for passing the condition on to their offspring, in the context of a negative test, siblings or cousins were often identified as vulnerable, as peers going through similar experiences but with potentially different outcomes.

Participants were acutely aware of the potential for different fortunes between family members. One participant (PD214) identified this mix of emotion between caring for himself in managing the testing process and fear for his future, while caring for others in the family who have already been diagnosed. While welcoming his own negative result, he acknowledged that this was not the end of his experience, “I’ve not got it but then there’s obviously the bigger picture of erm the guilt with my sister, seeing my sister unwell and all that type of stuff that ...”. There is some recent literature on the social implications of different decisions towards testing (see for example Keogh et al., 2017) and the implications for practice (Geelen et al., 2011). Most of the participants in our sample received a negative test result, and this context is important to acknowledge when exploring the demonstration of care towards others. Indeed, many participants expressed the difficulties of going through testing alongside other family members, particularly because of the potential, or actual, misalignment and the differences in outcome. One participant (PD302) was tested for the breast cancer gene in her family and received her results a week after her sister received hers. While the participant was negative, her sister and cousin were positive, and she described how she felt after receiving her own results:

...just like sad for my sister really, and my cousin, and like when I messaged my friends and said oh, by the way, I got my result, and like my dad, and everyone was like, oh, I'm really happy for you, and I was like, I don't feel that happy at the moment, I just feel guilty.

This account reveals many mixed emotions, around the expectations of personal happiness amidst an acute awareness of the feeling of others. She describes what happened when she told her sister her result, but only after leaving the room when her sister came home from work:

She was like “are you all right?” Well no, she thought I ... she thought I'd obviously got it, and I was like “I don't have it”. She was like, “what did you run away for?” I was like “I feel sad for you”. She was like “oh, get a grip”. So like yeah, yeah, she was all right, yeah.

The tension between individual results compared with the fortunes for others in the family was highlighted by another participant, who had received a negative result, while her sister had not yet been tested. The account of another participant (PD205) also emphasises how the personal is entangled with the familial:

[...] even though it's a big relief you don't have, it's not that kind of jumping up and down jubilation thing because I couldn't really do that cos [sister]'s there and she, that's not the case for her so that changes that radically. .... and HD isn't out of my life, I may be having to look after her with it .... So it's not game over, it's a massive weight lifted obviously but it doesn't mean that you can completely ignore it for the rest of your life cos I still have other responsibilities. Mum would've been over the moon [laughter].

This account identifies multiple layers in the experience of receiving a negative result and making it meaningful. The participant describes the physical setting as a momentary barrier to personal expression. Her sister accompanied her for the test result, which meant that she felt she needed to monitor her own reactions. She also describes how her negative result did not mean it was ‘game over’, a sentiment reflected in the ‘bigger picture’ comment described previously. A positive test

result will obviously have implications for the imagined future of an individual and their family, but discussions about a negative test result can also raise emotive responses about the past, particularly in acknowledging the experiences of older relatives. Whereas the first participant described in this section contextualised the result in terms of his experience of watching his father decline, for one participant (PD205), it was her mother who was identified as prominent within this imaginary. In this case a negative result was made meaningful through recognition of the pleasure that their mother would have felt had she lived to be told this daughter's result.

In our sample, negative reactions to receiving a negative test result were reported, but primarily only through stories of other family members. For example, one person (PD202) described the reactions of an uncle who was positive for HD and who had refused to speak to his own brother, who had tested negative. However, there were spaces of ambiguity when participants had not yet received their own test results. For example, one participant (PD401), described going for a breast cancer gene test. She explained her emotions when her sister had received her test first, while she was still waiting for hers:

The day she had the test results, she phoned me, so I was over the moon for her. But I was like you know, still thinking I might have it. And she said yeah, "you'll be fine you know", she's trying to kind of reassure me and stuff. But yeah, the thought of her going through it would have been horrendous. But um we would have been there for each other, either way, whichever.

Here the participant highlights how she would respond to her sister's diagnosis, where thinking about her sister going through that process would be 'horrendous'. The words of the participant suggest important work is being performed here, that to be genetically responsible is to share the burden that another family member is carrying. Responsibility is demonstrated in the openness of

the statement, and the collectivising and sharing of the positive result. In their accounts, no participants prioritised their own happiness over their personal negative test above the potential pain of others.

A negative test was often accompanied by strong, and mixed emotions, as participants negotiated a strategy for disclosure, while recognising how their own negative test did not mean an end to their entanglement with the disease. It was also identified as a catalyst, not just for the communication of the test result, but for realising the investment of others in that result. Expressing this as previously being 'blinkered' highlights the extraordinary expectations of the boundary between the individual and collective in the context of genetic disease.

Although a negative test result can signal the end of a personal diagnostic journey, we have seen how it does not mean 'game over'. Other members of the family are noted as either still going through the testing process or as having received a positive diagnosis. But a negative test result can prove significant in other ways. For many participants, the negative test enabled the communication of the results. Several participants acknowledged a reluctance to enter discussions about the disease and the test before the results, and might not have discussed it at all if it was positive. This was the case for one person (PD202), who explained that he only discussed it with his father after receiving his negative test:

I knew that, erm, if I ever had, if I ever was positive for it I could never tell him cos I knew that he'd poss, he'd blame himself forever, so, erm, so that was a nice thing to tell him as well. I told him, yeah and, er, he was happy and he gave me a hug which is rare for our family, we don't hug very often, so that was good, he was well chuffed.

This participant explained the implications of these barriers to communication, particularly in terms of isolation. The father was identified as the only legitimate source of experiential knowledge, yet he was also the one who could not be approached because of the symbolic importance of the questions, “I guess not having met anybody else in this situation aside from my dad and I haven’t asked him, of course I couldn’t ask him because, cos, er, he could never know.” Alongside specific disclosure of test results, a negative test result was identified as a significant moment for enabling emotions to be revealed. For one person (PD214), the negative result marked the moment where he stopped focusing on himself, which he describes as being ‘blinkerer’, enabling him to be part of the emotional journey for the other members of his family:

So my thoughts and feelings about my family where I was a bit blinkered, because what would happen, I would always be ... I wouldn’t get as upset over it because I would always think well it could happen to me. So now that that’s gone, I feel myself getting a little more erm emotionally involved and upset about my family going through processes because I feel like I’ve kind of escaped it so that’s kind of where I am.

We can see how this blinkering could include being unaware of the emotional responses of family members towards his own risk, going for the test and receiving his results. Receiving the negative result and communicating it to the family enabled the emotions of others to become visible. The disclosure of his negative test result came with an unintended realisation:

I didn’t realise actually how much it had been impacting them. Because they were saying ‘Yeah we’ve been thinking about it.’ ... I’ve not seen my grandmother cry and she’s 82 years old. And she cried when I told her which was really nice. So that was ... that was ... you can tell how much it’s impacted them beforehand, to see that. [...] yeah, a relief, an outpouring.



While we have explored how the implications for others impact the experience of our participants, this account is important, because it returns attention to the individual. Describing a focus on oneself as being ‘blinkered’ suggests an assumption and an expectation that it is not appropriate to think about oneself in the context of a genetic disease, and where such transgressions require work in negotiating the spaces where this is possible and acceptable. It also offers an insight into the psychological impact of testing on individuals, and how this has the potential to become a barrier to the communication of genetic health information.

#### Conclusion: Family entanglements

This article contributes to a greater understanding of the implications of genetic testing by focusing on the role of family members. Individuals and families experience barriers and facilitators in communicating about genetic risk, and recent high profile legal cases have underlined the tensions these pose for health professionals (see for example Dove et al. 2019). A genetic disease has particular implications for family in terms of genetic risk. Here we underline how family are implicated in decisions about communication and disclosure. Mobilisations around a genetic test involve the emotional work and care of individuals in negotiating their communication practices with family members. We have shown how participants identify genetic information as both valuable and risky, with a desire to protect, manage emotions, identify vulnerability, and pay respect. This article contributes to current knowledge, and contrasts with much of the previous research, because we acknowledge that this work happens throughout the ‘patient’ journey, including before testing and when receiving a negative result.

Here we highlight the relevance of the concept of ‘entanglement’, which has been described as “to be entangled is not simply to be intertwined with another, as in the joining of separate entities, but to lack an independent, self-contained existence” (Barad 2007: ix quoted in Helosvuori 2020). We

draw on this concept to acknowledge the significance of local (familial) knowledge and experience in making genetic information and personal experience meaningful. The concept has previously been used to explore complex connections between patient participation, citizenship and professional expertise, such as in the medical consultation (Nunes et al., 2014), in tracking patient activism through changing relationships between public authorities, private firms and non-profit organisations (Rabeharisoa 2006) and understanding the role of institutional trust for those participating in clinical trials (Mwale 2020). McDougall et al. (2016) used ‘collaborative entanglement’ as a way to explore the tensions within intraprofessional care for patients with complex disease. Latimer and Lopez Gomez (2019) in their study of care work identified ‘intimate entanglements’ to explore the politics of intimacy and forms of belonging, and entanglements between the mundane and extraordinary. Helosvuiri (2020) in her study of IVF and childlessness talked about the lasting legacy of ‘lingering technological entanglements’. Entanglement has been used to show how systems (in our case genetic testing) are embedded in their social context. The role of family has been explicitly explored through entanglement, including blood cord banking (Beltrame, 2019) and the ‘relational conundrums’ of uterus transplantation (Guntram, 2021). The concept of local biologies was used by Gibbon (2017) to rethink the role of family knowledge and experience as a set of practices which shape experience and actions towards cancer testing in Brazil. She highlights a ‘recursivity’ (Franklin 2013) between communication in the clinic and patient experiences and understanding of genetic risk, where “biologies produced within these social contexts intersect, sometimes clash, but also loop back to shape a range of narrativised risk discourses and practices” (Gibbon 2017 p184). This looping effect is significant for understanding the experience of the participants in our study, where family forms the context which is shaped by, and in turn shapes, the decisions and experiences around genetic testing. The individual who might be invited for genetic testing cannot therefore be separated from their family and the family practices through which knowledge and experience are communicated and translated.

The strength of this project is its capacity to engage with patients during their initial contact with clinical genetics. This enabled data to be collected from patients going through the decision-making process as well as being interviewed after their decision process and their test result. There are however limitations, most notably that this project only represents the accounts of those who are engaged with clinical services and are therefore possibly more open to communication about and reflection on their experiences. Individuals who decline testing and do not engage with clinical services are an underserved population, and we acknowledge that this group is significantly underrepresented in research (Keough et al. 2017). In addition, the project focuses on the UK whose health care system supports genetic testing without charge and employs particular practices around genetic counselling, which means that the accounts of testing and decision making can only be understood within this specific context.

In this article we considered how the connections between the individual at risk of an inherited condition and the broader family are made visible and are negotiated in the context of genetic testing. Our aim is to bring back the family into a contemporary context which is heavily laden with the privileging and iconic imaging of genetics and genetic information, and where the diagnostic landscape is being transformed through technological developments and social expectations. Significant changes include the expansion of genetic testing capabilities such as whole genome sequencing, the rise of personalised and precision medicine, and increasing availability of direct-to-consumer testing, which are linked in part to a rhetoric of individual autonomy and consumer choice (Kelly et al., 2018). There are clinical implications, where diagnostic technologies no longer require blood samples from the patient's family members to inform a diagnosis, and therefore lead to less direct contact between professionals and family members. In this context, it becomes even more important to acknowledge and understand family entanglements which continue to shape communication and experience in this contested terrain between the individual and collective.

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#### Bullet points

We explored the early stages of decision making about predictive genetic testing.

The concept of entanglement was used to explore the role of family in genetics.

The family act as gatekeepers and communicators of genetic information.

Barriers to family communication include the desire to protect and respect others.

Family entanglements continue to shape communication and experience.