Contingency and authenticity in deliberations on predictive genetic tests for Huntington’s Disease

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“Framing the trajectories of decision-making in the context of predictive and prenatal genetic and genomic tests”.

- Ethnographic research project
- Multi-site
- Decision making processes where patients are offered a predictive or diagnostic test
- Genetic tests, pre-natal tests
- Asking patients to keep a record on their thinking
The offer of a genetic test

**Test to find out if you'll get Huntington's disease**

If you have a history of Huntington's disease in your family (especially if a parent or grandparent had it), you can have a test to see if you'll also get it.

It's up to you to decide if you want to have the test. Lots of people at risk of Huntington's disease decide they'd rather not know until any symptoms appear.

If you do want to know, ask your GP for a referral to a genetic counsellor. You'll have several appointments with the counsellor. It's only done once all the benefits and risks have been explained.

The test involves checking a sample of your blood for the genetic fault that causes the condition. It can take a few weeks to get the result.

The Huntington's Disease Association has more information about genetic testing for Huntington's disease.

As a form of knowledge

- Relevance for the person
- Offer without specific expectation
- Complex and potentially dangerous knowledge
- Potentially transformative of the sense of self

https://www.nhs.uk/conditions/huntingtons-disease/diagnosis/
Deliberating a genetic test

“Genetic counseling is a service intended to help patients and their families gain sufficient information to help them make decisions about testing and technology, manage their genetic conditions, and cope with post-counseling/testing realities. As such, it is neither exclusively education nor is it exclusively psychosocial counseling” (Veach, Bartels and LeRoy, 2007, 725)

The structure of the clinic incorporates a delay. After referral to the clinic, a patient sees a counselor pre-clinic then a clinical geneticist. The clinician does not take a sample for testing at this stage. Rather, there is a delay before the second session. This delay was incorporated based on concerns surrounding the negative psychological effects that a positive test result may have on patients.

Is this delay still justified?
Conceptualising the patient journey

Double contingency provides the basic frame of interaction between self and other in social theory (Luhmann, 1995, Habermas, 1993)

Recent theorising has revealed the limitations of double contingency, and the importance of triple contingency (Strydom, 2001)

Given that the vast majority of patients do not seek predictive testing for HD, managing uncertainty in self-other relations is of paramount importance

The idea of contingency draws attention to the stabilisation and subsequent destabilisation of relations at the heart of the decision to take a genetic test

Contingency also raises questions of authenticity for the person involved
Conceptualising the patient journey

Begins when the patient first learns of the condition and of the possibility of a test

Places the patient in a tension laden situation of double contingency

Once the patient is old enough (usually at least 18, sometimes sooner) they face an offer of a test.

Their double contingent situation is resolved by established patterns of interaction and behaviour or,

These patterns are not be enough for everyone. Some look for more certainty to narrow the contingency of their relations.

Project focuses on the period in which patients have engaged with genetic counseling
Authenticity

• The value of authenticity involves the basic sense of resolution between the spontaneous subject and their social world

• The inauthentic person is a person who is pushed by "desires, by external circumstances, by false opinions or judgements, by the whims of fate" (Heller, 1999: 226). She is a 'passive slave' of her passions.

• The authentic person by contrast, is one who is 'pulled' by their own self-understanding
  • Authenticity means to remain true to the leap, to one's choice of oneself. **Authenticity is to remain true to oneself.** Authenticity has become the single most sublime virtue of modernity, for authentic people are people who remain true to their existential choice, who are pulled and not pushed, who are personalities (1999: 227, emphasis in original).

• Patients deliberations on the offer of a genetic test is a deliberation on their authenticity as actors in (contingent) self other relations
Patient’s experience of taking the test

- Uptake of predictive testing for HD among all at risk of HD is 17.4% (Baig et al., 2016)
- The main reasons for testing is to reduce uncertainty (70.7%) followed by to plan for the future (57.7%) (Baig et al., 2016)
- Reasons not to test include the absence of a medical intervention, a positive result only says the patient will get the condition if they live long enough, not when and so unhelpful when planning (Smith et al., 2013)
- Having a result has no or a small positive effect for people who do not have the gene change (Crozier, Robertson and Dale, 2015)
- People carrying the gene change appear more likely to suffer distress, but the distress noted may be an early symptom of the condition (Crozier, Robertson and Dale, 2015)
The decision to take a test

• Counselors prompt patients to reflect on the effects of different possible outcomes (Sarangi et al., 2004)

• Decision is set within social and family relationships (Atkinson et al., 2013, Smith et al., 2013)

• Patients adopt two approaches to deciding to participate in genetic testing
  • Decision evolves over time
  • Decision ‘happens so quickly that it is felt as a “fait accompli”’ (Manuel and Brunger, 2014: 1048)
Questioning the delay

• If only a small portion of those eligible are coming forward for testing, is the delay incorporated into the clinic structure justified?
• Are those coming in also the people who have thought about this the most?
• Does the delay serve any purpose? Moreover, given that genetic counseling is neither funded nor equipped to prompt a full examination of personal motives, is a limited check and delay warranted?
• Theoretically:
  • If the delay initiates reflection and discourse, this space allows new forms of rationality to permeate into existing states of affairs.
  • This involves a transition between discursive stages (Habermas, 1984, 1987)
    • Existing understandings come into question
    • Through reflection and discourse, more aspects of the situation come to the fore causing new understandings to emerge
    • These new understandings are integration in some way into everyday life
If a discourse takes place:

• More and more aspects relevant to the situation are identified, and examined
• The basis of the relationship between self and other is examined
• The patient thinks about how to take responsibility for themselves in all their determinations (extending now to biological determinations) and how to consciously continue their life history (authenticity)
Unquestioned authentic self

• C PD 4 was a middle aged with no biological children, whose dad received a positive result from a genetic test at around 50 years of age, and began to deteriorate in his early 60s.

• C PD 4 sought a referral to genetics upon becoming around the age of his dad when he was tested, to plan his retirement and to manage himself if he started to display symptoms

• C PD 4 thought things through and made his decision before the clinic:
  • generally I think I’m somebody who just shov-shovel it on the box and, you know, I’ve done my reflecting on that. [...] say’s his wife will say] I’m looking at every little thing, you know what I mean, and why have you don’t that like that, when you could have done it that way and saved five seconds in the kitchen or... Just little things like that I, I reflect on stuff, [...] It’s all, you know, it sort of builds, builds and builds, but a little bit but nothing overly [...] 

• C PD 4’s diary included a single one line entry on the need to tell his mum of the test.

• However, C PD 4 did want to find his authentic voice through testing:
  • my whole reason for getting tested was to make sure I didn’t find myself in a position where I didn’t realise I’d started displaying, you know, [...] I didn’t want to be in work and then suddenly go, “oh obviously I’m not well” and ignoring the facts and carrying on and carrying on, so I just wanted to have a plan. I just wanted to be able to say okay, if I’ve got it, I’ve got it and this is my plan, erm, if I haven’t got it, then great, I’ll carry on not really having a plan and just see where life takes me. (C PD 4, interview)
  • Again maybe some concerns, midsummer last year, about whether some of the cognitive early symptoms maybe was starting to affect me, but that was maybe more to do with how my job, and the pressure of my job, and the problem solving rationale analysis that I have to do in my job I felt was being affected. But whether that’s just work related stress, and not the disease. So there’s a few things. (C PD 4, Clinic 1)

• This patient had already thought through why he wanted the test, and sorted out his thinking before his first appointment. He had no need to enter into any further reflection

• But his thinking was already characterised by the desire to present an authentic self
Confirming the authentic choice

• C PD 14 was a young man in his late 20s whose sister was in the advanced stages of HD and whose early life had been severely disrupted by his dad’s HD

• C PD 14 had closed off discussion of HD until his wife began to ask about it. Slowly, he revealed his concerns to her. Through these conversations, C PD 14 began to think about his future with his wife and the possibility of having children, thoughts that led to a determination to have a predictive genetic test:
  • yes slowly I come round to it and erm probably the reason I did the test was more for assurance for ... for her, because I thought you know, if I have got it, it’s going to impact her a lot now she’s made that decision to ... to marry me and go through that process. [...] And the other side of it, I’ve seen people who’ve just gone ahead and had kids without knowing but I ... I just ... couldn’t. I don’t think I could live with that myself. I know there’s ... that’s ... that’s me personally and I’ve always said that if I do have it I want it to ... to die out with me and so that it (C PD 14, interview)
Confirming the authentic choice

- C PD 14 was then in a hurry to get the test and impatient with the clinic arrangements. In fact, he was in such a hurry he looked into going private for his test but decided against this when he realised the private provider also used the same protocol.

- But having thought he had thought about all the implications of the test, he was surprised by the reflections the clinic prompted in him:
  - it probably does need to take that bit ... bit longer. I’m not sure obviously as long as it ... it did but [...] it needs to take that bit longer, to make sure that person is ... it’s not just on a whim that they’re doing it, they’ve had months to think about it (C PD 14, interview)

- C PD 14 kept a diary using the notes app in his phone. On one entry he reflects:
  - I feel the appointment was useful as it stoped [sic] me in my tracks and allow [sic] me to rethink all elements of my decision over the days following the appointment. I felt I was trying steamrolling to results day and this allow me to stop and rethink things through. (C PD 14 Diary)

- In his diary he records using google and how he:
  - Looked for all good results on the HDA forum and tryed [sic] to envisage this, stumbled across a bad result from someone on the same situation as me and then decided to stop (C PD 14 Diary)

- Although he acknowledges that others would think more in the time between the two appointments, he felt it useful to reaffirm his decision:
  - I know that a lot of people feel very worried and that and they feel that ... they think of every opportunity ... er every eventuality but I felt like ... I probably have thought about that previously. And I think after that first session, erm within a couple of days afterwards I ... my mind was made up again. [...] I was a bit on the fence and yeah, it did stop me ... [...] but I’d go and then I ... then I actually thought about it a bit deeper and come back to the same side of it so ... so it was a case of yeah, just ... just go through a process then. [...] After I’d decided again that yes, I’ve come to the same answer, this is exactly what I want.

- Like C PD 4, C PD 14 had already concluded his thinking, and his decision was to resolve his authenticity
Revealing the inauthentic choice

• C PD 3 was a 28 year old woman with 2 small children, at the centre of a family with numerous ailments. She was heavily involved in looking after her dad who was becoming violent with HD, her mum had an electrolarynx, she had a daughter with mild cerebral palsy and her partner had a heart condition.

• C PD 3 had attended the genetic counseling service in the past, but had not had a test

• In her counseling session, she tries to convince the clinician she will be able to incorporate this new knowledge into her world:
  • I’ll just deal with it like I normally deal with everything... Yeah... [sniffs] But it, it’s different when you see it like that, isn’t it, completely different when you see it like that. But I think as well ‘cause there was reason, ‘cause I’m 28 now and they say the symptoms start kicking in when you’re 30. (C PD 3 – clinic 1)

• She observes how the condition manifested differently in her dad and uncle, but explains how it was circumstances that propelled her to genetics:
  • it was [HD nurse] that was the one referred me again ‘cause I just had an urge I’ve got to get it done and it, it was just the way that like, I see my dad go downhill like that but then I, I look at it from my uncle’s view as well and it’s just completely different. Erm, yeah, I think I need to do it for myself as well.
Revealing the inauthentic choice

• Picking up on this, the clinician points out how her decision to have the test has been driven by circumstance:
  Consultant: And you’re telling me that if you, it sounds like you’ve screwed yourself up to do it now.
  Patient: Yeah.
  Consultant: But, and you wouldn’t be able to do that again like next year.
  Patient: Mm.
  Consultant: And it just makes me, I mean, I’m not going to stop you having it done, you know, it’s up to you.
  Patient: Mm.
  Consultant: But, but, I’m just worried that you’re sitting there really in two minds and you’re feeling pushed by circumstances, by the fact that you got to the clinic, this sort of thing.
  Patient: Mm.
  Consultant: And that, the fact that you’ve got to this clinic room today, like that’s not a good reason for doing it today, you know, and we’ll fit in with, if there was a better timing for you, we’ll fit in with that. (CPD 3 – clinic 1)

• This patient agreed to participate in the project, but then stopped communicating with the researcher, so we have no further details of her thinking

• But the patient reveals an inauthentic choice as she describes coming to the clinic based on her fears about her dad’s disease progression, and the referral made for her by the HD nurse.

• This inauthenticity is picked up on by the clinician reflecting how the patient has “screwed” herself up for the test and was there because she was “pushed by circumstances”
Conclusion

• The authentic self in these deliberations, is authentic in the basic form of double contingency. Patients are primarily presenting themselves as authentic to those nearest to them, and to themselves. The question is to what extent their spontaneous actions are based on an “I” originating in their existential self, and to what extent is it originating in their biological self.

• Patients had already conducted much of their deliberations prior to their appointment. To the extent that there are any reflections, these focus on the their willingness to absorb the knowledge offered by the test. For some, there is nothing to reflect on, for others the chance to reflect provides an opportunity to confirm the decision. Others in our sample described becoming more at peace with the decision.

• Some patients, not discussed here, had had less time to think about their risks before coming to the clinic. These describe identifying additional topics for reflection in the interim period between clinics.

• The use of two clinics to discuss this decision does not appear to prompt additional reflection or discourse in many people. However, there are circumstances in which it does prompt reflection and provides an opportunity to identify situations where patients are seeking test results for inauthentic reasons, and so are at risk of being less able to cope with the information.
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