

Persevering to take a predictive genetic test

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Theories of how patients make their decisions on genetic tests

- ▶ Stage theory of the process of making a decision (Taylor, 2005)
 - ▶ 'pre-contemplation non-engagement stage, contemplation or engagement, and maintenance or review stages'
 - ▶ During contemplation and engagement stage, patient comes to one of four positions
- ▶ Discursive patterns used in clinics (Sarangi, 2004)
 - ▶ 'Gaining knowledge as a Basis for Future Action'
 - ▶ 'Needing to know as a subjective necessity'
 - ▶ 'Downplaying What Can Be Known'

Genetic/genomic counselling in predictive neurodegenerative clinics

- ▶ 3 stages
- ▶ Stage 1
 - ▶ Accepting the reality of the disorder
- ▶ Stage 2
 - ▶ Deciding to go ahead with a predictive genetic test
- ▶ Stage 3
 - ▶ Readying oneself to accept the result

Modelling decision making using Habermas

- ▶ Discourse ethics focuses on finding a coordinated agreement among people with competing interests
- ▶ Focuses on how individuals can use reasons to persuade others to come round to their view
- ▶ Employments of practical reason
 - ▶ What should I do?

	Pragmatic	Ethical/existential	Moral
Sphere	Fixed preferences, focusing on processes How to I get what I want?	Clarifying self-understandings/ meaning of a good life “Who am I/Who do I want to be?”	Conflicts of interest disrupting social cohesion
Discourse	Recommendations or program of action	Advice about the conduct of life or how to realise a life project “Is this the right thing to do, all things considered?”	Justifying and applying norms

Methods

- ▶ Ethnographic observations of clinics
- ▶ Patients as co-researchers
 - ▶ Diaries
 - ▶ Audio recordings
- ▶ Post-clinic interviews

Predictive neurodegenerative genetic clinic data (Huntington's Disease and Motor Neuron Disease)

	Gender	Pre-clinic	Discussion clinic	Decision clinic	Results clinic	Diary etc	Interview
B PD 1	M		Yes			Yes	
CPD 1	F		Yes	Yes		Yes	Yes
CPD 2	M		Yes	Yes		Yes	Yes
CPD 3	F		Yes				
CPD 4	M		Yes	Yes		Yes	Yes
CPD 5	F		Yes	Yes		Yes	Yes
CPD 9	F					Yes	
CPD 10	M			Yes		Yes	Yes
CPD 12	M		Yes	Yes			
CPD 13	M		Yes				
CPD 14	M		Yes			Yes	Yes
CPD 19	F		Yes			Yes	Yes
SPD 3	F (MND)	Yes					
SPD 4	M	Yes					
SPD 6	F	Yes	Yes		Yes		
SPD 8	F	Yes		Yes			
Ex PD 1	F (MND)		Yes				
Ex PD 3	F		Yes				
Totals	18	4	13	7	1	9	7

Discussing the test

- ▶ Quick glance at a moral discussion
- ▶ Cases where the patient action plans or interests potentially conflicts with other people's interests
- ▶ Pragmatic discourse
 - ▶ Strong sense of personal preferences
- ▶ Ethical/existential discourses
 - ▶ Who am I and who do I want to be in the future?
 - ▶ What is the best course of action for me in this situation?
- ▶ Outlier cases
 - ▶ The role of challenge

Moral discourse

- ▶ Takes shape where personal interests come into conflict with interests of others
- ▶ Clinicians and counsellors manage moral conflicts by suggesting or supporting moral norms

Moral discourses

Patient grounding conversation in baby	Patient: Okay. That's fine and my last question is about if I get a positive result, when I could get tests for the baby?
Taking up discourse	Clinical geneticist: I think, yeah well for, for her ... Patient: Yeah.
Use of "we", focus on risk	Clinical geneticist: ... what, what we would ... we would say is the chance of her developing any problems as a child are very tiny. Patient: Mmm. Clinical geneticist: Are remote. Patient: Yes.
Articulating norm	Clinical geneticist: And so we, we would strongly suggest not testing but waiting until she is old enough to ask. Like you have. Patient: Okay, that's fine.
Finding consensual space	Clinical geneticist: We, we wouldn't want to test a child, just occasionally we test a young child if, if they are developing problems that could be from Huntington's.
Agreement	Patient: Okay.
Justification	Clinical geneticist: Then, then there's a reason to do it.
Agreement	Patient: Yeah, but that's fair, I just ... Clinical geneticist: But that's very unusual.
Voice of others	Patient: Yeah I just heard from my grandmother that they also did the test on some children but ... yeah. Clinical geneticist: We, we would, we would not really ...
Moral discourse	Patient: Yeah I, I thought that as well that she's going to be old enough to ask about it. Clinical geneticist: She has to decide for herself I think. Male partner: Yeah. Patient: Yeah.
Suggesting of model	Clinical geneticist: Is probably what you should have said. (C PD 10, clinic 2)

Primarily pragmatic discourse

- ▶ Quite certain of their preferences
- ▶ Focused on getting through clinics as a way of getting to their test result
- ▶ Resisted efforts to question their preferences

Primarily pragmatic discourse

Grounding ethical/existential discourse

Agreeing, focuses on time

Reflects back

Switches ground to pragmatic discourse

Reflects back

Offers summary

Returning to main topic

Continues pragmatic discourse

Apology

Complaint

Ethical/existential within pragmatic

Clinical genericist: Okay, but in terms of it being a blow, and then.

Patient: Yes, I'll need some time to, I'll obviously need some time to.

Clinical genericist: People do.

Patient: Because I'm looking at swapping jobs and stuff, and I'm trying to stay flexible at the moment. Because I know with this I will need some time to adjust either way, so that I'm planning for that either way, whether it be good or bad.

Clinical genericist: You'd rather carry on with the process now, and then sort out work, rather than sort out work.

Patient: Well I'm sort of juggling, I'm looking at some freelance stuff.

Clinical genericist: It can fit in either way.

Patient: Yeah, it's sussing out some freelance stuff. Because it's, one of the things, because I started this process back in May last year. Once I made the decision and finally made that phone call to [HD Specialist nurse] going right, let's just do this, kind of want to get on with it. And it's just been like oh, it's been like nearly a year, and it's just like "Oh god, really?"

Clinical genericist: Yeah, I'm sorry.

Patient: Because it takes a long time to, even though I'd kind of decided I wanted to do it, it took time to actually make that phone call to start the process. Waiting list, oh a couple of months. And then I was like "Oh okay", and then it was a lot longer than that. Yeah, I'm not under any illusions that I'm going to be trotting out and going it's all marvellous. It will take time to adjust to, and I'm prepared for that. (CPD 5)

Ethical self-reflection driving pragmatic discourse

- ▶ Describes reflection that arrives at a clear description of personal preferences
- ▶ Clarity enables description

Ethical self-reflection driving pragmatic discourse

Authenticity

Clinical geneticist: Yeah, yeah and does that [support from friends] make you feel that is your decision you're making though or is it again, it can be quite a difficult thing to?

Confirming self-clarification

Patient: Yeah, I, I don't feel as though I'm pressurised into it, I don't feel as though I have to you know, I, I have to do one thing to appease, yeah no, I, I'm always quite um, yeah I just like to deal with the facts, for me not knowing would wind me up more than knowing.

Encouraging

Clinical geneticist: Yes, yeah.

Repeating

Patient: I'd rather have the certainty of it, and then I could plan my life and move on, I don't want to always have this cloud lingering over me.

Encouraging

Clinical geneticist: Yes, yes.

Patient: It would just, I know some people quite like the bliss of not knowing, and would just rather be.

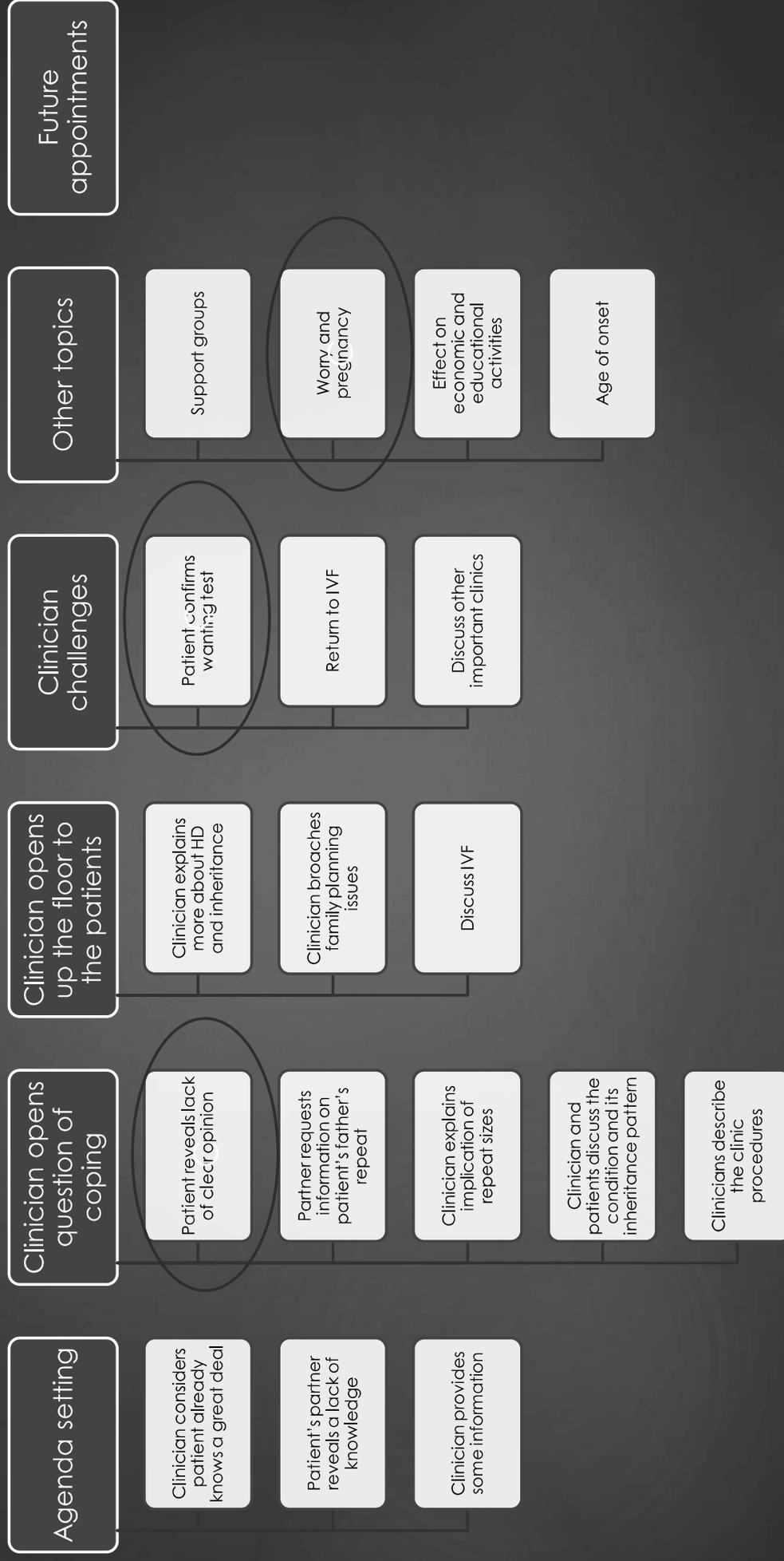
Clinical geneticist: Uh huh.

Patient: I, I like to have all my chips on the table, I like to just know what I've got in front of me, and you know. (B PD 1, clinic 1)

Difficult cases

- ▶ Cases that lie outside of the categories
- ▶ Raise questions about counselling

Topics covered in first clinic



Clinician opening question of coping

- Context 87 **Clinical geneticist:** Because obviously it's a change in risk.
- 88 **Patient:** Yes.
- Coping 89 **Clinical geneticist:** How have you kind of coped with that ...
- Recounting sequence 90 **Patient:** [Overlap] I don't know, I think ... I don't know, I think up until the point of
91 dad having a confirmation [unclear-03:40] my time to kind of think oh, it's to do with
92 me now a little bit, you know?
- Indicating agreement 93 **Clinical geneticist:** [Overlap] Yeah, yeah.
- Context 94 **Patient:** Because until it was confirmed for my dad either way it then only mattered
95 to me then, really. Because if dad ...
- Indicating agreement 96 **Clinical geneticist:** [Overlap] Yeah ...
- Starting of realisation 97 **Patient:** Um, wasn't having the, the, the genetic disorder, then I wouldn't have it
98 then. So I'm only starting to kind of think about it for me now then, really.
- 99 **Clinical geneticist:** Mm.
- Differences 100 **Patient:** Um, you know I kind of ... I know things will get sorted for dad but obviously
101 it's different, kind of, um, time of my life compared to him then, really, you know?
- 102 **Clinical geneticist:** Yeah.
- 103 **Patient:** So, um, but yeah. (C PD 1, clinic 1)

Opinion remains absent (from minute 46)

Opening floor Challenge	1125 1126	Clinical geneticist: Yeah. Are there ... So ... But you're kind of feeling at the moment that this is not really for you?
Just wanting to know	1127 1128 1129	Patient: That's generally my thing. Cos again, in a, I mean, a similar fashion to my dad I'd rather just know and then deal with it rather than the kind of, what if it may be there aspect of it, then.
Indicating understanding	1130	Clinical geneticist: [Overlap] Yeah.
Knowing planning	1131 1132 1133	Patient: You know? But it was more around knowing and then, as I said, in terms of us planning for anything that what our options were, which obviously you and me are much more [unclear-46:56] come in here. Cos it was either going to be ... in my head it was either going to be if I didn't have HD then we would just carry on as whatever.
If test is negative	1134 1135	
Indicating agreement	1136	Male partner: Yeah.
Interest in IVF	1137 1138 1139 1140	Patient: But if it did then, you know, other options then, really. As I didn't ... IVF ... I didn't ... I don't know much about IVF or anything like that so I'd have to look into what's involved with that, but if it's possible then, you know, we could certainly chose that as an option ...
Indicated agreement	1141 1142	Male partner: [Overlap] Yeah ... Patient: If we come to it then, really. So ... (C PD 1, clinic 1)

1263 **Patient:** [Overlap] Okay. Yeah. I'm just thinking in terms of ... cos I don't
1264 think ... I think we're obviously going to go the kind of marriage thing
1265 comes up first, cos obviously in that time I could be showing some early
1266 signs. If I then have a pregnancy on top of that and they're going to
1267 exacerbate then, potentially, you know ...

1268 **Clinical geneticist:** I haven't heard about any.

1269 **Patient:** No. It's not ...

1270 **Clinical geneticist:** [Overlap] it's not, it's not something I've looked into but,
1271 but HD ... I suppose because we also have seen lots of families ...

1272 **Patient:** Mm.

1273 **Clinical geneticist:** With HD, or coming for counselling, lots of different
1274 topics around HD get discussed, and that hasn't come up actually.

1275 **Patient:** [Overlap] Okay.

1276 **Clinical geneticist:** So I'm guessing there isn't particularly ...
[...] (C PD 1, clinic 1)

Final diary entry before results clinic

Emotional day. Worst yet. [describes attending HD clinic with her dad]. Dad (and later partner) hopeful I will be the same, however makes me angry that they (or anyone) cannot give me any guarantee on that, and I may not reach old age. Dad only had 3 months to process and likely only have 10 years to worry/be under medical scrutiny etc, but I am facing 40/50 years and worry about employment/mortgage etc and don't know how I would cope. The nurse picked up that I was due my results next week and asked about my engagement ring/family etc., and must have seen this worried me as she offered me to call her after my results. Talked with clinic/team about what support Dad can access – got upset (in private) about potentially caring for Dad as becomes more symptomatic/end of life whilst potentially having to go through symptoms and treatment etc.

Later, attended wedding fayre and was very excited/happy about planning for future. Later, netball match which was physically tough and we lost, so became angry/upset about this – probably more unreasonable than normal. Partner and I then had disagreement as felt I had been pushing him away, thinking negatively and wallowing in situation by not trying to make myself feel better. Agreed would access counselling/therapy regardless of outcome.

Problematic clinics

- ▶ Focus more on the realities
- ▶ No clear preference has been formed
- ▶ Less attention on the meaning of the test result either for pragmatic or for ethical/existential purposes

Typical	40 41	Clinical Geneticist: [Clinician begins with overview of process] 'Cause some people are very clear straight away...
	42	Patient: Yeah.
	43	Clinical Geneticist: and other people need a bit more time to think about it.
Thinking time	44 45	Patient: I've had six months, I've thought about it six months ago, so... I'm pretty sure that I want to go ahead.
Past referral	46 47	Clinical Geneticist: Yeah, okay, I mean you, you, I think your GP [tuts] sent to us first like a couple of years ago.
Wasn't ready	48	Patient: Yeah, yeah, and then, I don't know, I think back then I, I don't think I was ready...
Indicated understanding	49	Clinical Geneticist: No.
Personal change	50 51	Patient: ... and, and now I think, after I've had my little 'un, erm, I'm ready to go ahead now.
		[Discussion of previous referral leading to geneticist inquiring into the change of mind]
External motivators	61 62 63	Patient: Just the way that I've seen my dad recently, over the past six months he's gone downhill really, and it's just something that's not, it's playing on my mind, I just, I would rather find out now rather than later.
	64	Clinical Geneticist: Mm.
	65	Patient's mother: Okay.
	66	Patient: I'm pretty quite headstrong so... (C PD 3, clinic 1)

Family traumas Is ready	394 395	Patient: Erm, with everything, all of the family, we've dealt with a lot as a family anyway so I, I just think I'm ready now, I don't think...
Indicates understanding	396	Clinical Geneticist: Yeah.
Timing	397	Patient: Like I said I've put it off once before I really don't want to do it again.
Raising challenge	398 399	Clinical Geneticist: No, no, I mean don't, you know I think if, you, if you clearly want to go ahead then we'll do it.
Indicated understanding	400	Patient: Okay.
Reflecting statements	401 402 403	Clinical Geneticist: You know so don't worry about that and I, I suppose, but I wouldn't want you to be sort of saying, er, yes I want it now, if you didn't really want it now but just thought it would be difficult to get referred back or anything.
Concern	404	Patient: Mm.
Offering more time	405 406 407 408 409	Clinical Geneticist: 'Cause I think if, you know, if you wanted more time then once you're at this stage, once we've seen you here then you can phone up and, you know if you leave it, if you want, you know, another month or two or ten, you know, you can leave it like that that once you want to carry back on, you can just phone up and we'll slot you back in, you don't have to go back getting referred again.
Restating reasons	450 451 452 453 454	Patient: 'Cause I spoke, the person that actually I went to the hospital with my dad, it was about three or four months ago, it was [HD specialist 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 that bad 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.
Lightens challenge	455 456 457 458 459	Clinical Geneticist: [Laughs] okay, I mean I, yeah... I just wanted to make sure that you're, not make sure, but make sure that you... If you sit here saying you want it, we'll do it, it's just I wanted to make sure that you're not, erm, doing it because you feel that if you walk away again, erm, and want to leave it for a little bit that, that we'll make you go through a palaver to get back, you know, I don't want you to think that.

Challenge	639	Patient: ... 'cause I've dealt with everything before now, so I just think I just want to get it over and done with [laughing].
	640	Clinical Geneticist: That Oh no, I get the message that you want, you want to get it over and done with.
	641	Patient: Mm.
Challenge	642	Clinical Geneticist: That But that's a funny way to put it.
	643	Patient: I know.
Challenge	644	Clinical Geneticist: That, you know if we didn't do it 2019, then in 2020 you wouldn't get it done?
	645	Patient: No I probably, I would probably put it off then [laughs].
Challenge	646	Clinical Geneticist: That [Laughing] but that sounds like you've still got quite a lot of reservations about it?
	647	Patient: Yeah, I, obviously it's not a nice thing to go through, I know that but I just think if I don't get it done now, I, I
	648	probably, I don't know, I don't know, there's a lot of things that goes on, that's really hard to explain [laughs]. All I know is
	649	that I've put it, like I say I've just, I've put it off once, I would want to put it off again.
Pattern	650	Clinical Geneticist: That [Laughing] but when you put it off before you said you weren't ready for it?
	651	Patient: No.
	652	Clinical Geneticist: That So that was fine, that was sensible to put it off if you weren't ready for it?
	653	Patient: Mm.
Challenge	654	Clinical Geneticist: That And you're telling me that if you, it sounds like you've screwed yourself up to do it now.
	655	Patient: Yeah.
	656	Clinical Geneticist: That But, and you wouldn't be able to do that again like next year.
	657	Patient: Mm.
Seeking agreement	658	Clinical Geneticist: That And it just makes me, I mean, I'm not going to stop you having it done, you know, it's up to you.
	659	Patient: Mm.
Challenge	660	Clinical Geneticist: That But, but, I'm just worried that you're sitting there really in two minds and you're feeling pushed by
	661	circumstances, by the fact that you got to the clinic, this sort of thing.
	662	Patient: Mm.
	663	Clinical Geneticist: That And that, the fact that you've got to this clinic room today, like that's not a good reason for doing it
	664	today, you know, and we'll fit in with, if there was a better timing for you, we'll fit in with that.
	665	Patient: Yeah, I know that like, I'll have a couple more sessions I think just to, er, properly think about it.

Conclusions

- ▶ There is a discourse that emerges during the clinics
- ▶ This is concerned with the reasons for continuing to securing a predictive test result
- ▶ These tend to constellate around some combination of pragmatic and ethical/existential reasons
- ▶ But in some cases, patients have less well formed views on the offer of a test