“Framing the trajectories of decision-making in predictive and prenatal genetics”

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Aiming to look at decisions in settings …

- Where the decision is largely personal and perhaps less likely to be driven by “medical need” or the professionals’ perspective, as in:

- (i) Predictive genetic testing: especially for adult-onset neurodegenerative disorders (**Huntington’s disease**)

- (ii) Prenatal genetics: approach to managing the pregnancy (including **diagnostic testing** or decisions about **terminating the pregnancy**)

Focus on the ‘Life-World’ rather than the ‘World of the Clinic’

- We have aimed to recruit participants as co-researchers, using three types of data to gain insight into what shapes their ‘decisions-in-context’.
- This gives the participants full control over our knowledge of their deliberations
- Our data consist of:
    - consultations
    - diaries/audio-logs
    - closing interviews

The plan for our presentation:

- Angus: the prenatal (fetal cardiology) clinic, including:
    - our appeal against a REC decision on the consent process
    - clinics, diaries, audio-logs and interviews
- Shane: the predictive testing clinic (most of our data)

Consent process

- As designed, our process was to provide information about the project at least a day before recruitment

- To record the first clinic on basis of a provisional, oral consent (when patient is often still uncertain about the clinical process, let alone the research)

- To discuss the project in more detail after the clinic and then EITHER delete the data OR document the consent (and recruit to full participation)

- Insistence by REC on initial signed consent was a real problem for recruitment (impacting colleagues as well as patients)

Approval process

- Project start date February 2018, recruitment to run until July'20
- Initial '**distorting**' REC approval July 2018
- Amendment lodged November 2018
- Unfavourable opinion April 2019
- Appeal lodged with HRA April 2019
- REC considered HRA opinion and our response to further conditions; gave approval July 2019 (HALF WAY POINT)
- Updated NHS R&D approvals over next couple of months
- Recruitment process working well in antenatal are: then COVID

Clinic Consultations

- Very solicitous …. focus on mother's comfort
- A lot of technical fetal echo data …..
    - that *(to generalise)* interests Dads more then Mums *(who ask whether the baby will survive)*
- Explicit expressions of empowerment and non-directiveness
    - although it is not clear that a statement of non-directiveness achieves the goal it asserts (i.e. is performative)

Prenatal case: Down's syndrome with cardiac anomaly, including an **Audio Log**.

Characters: M (mother of the fetus), D (father of the fetus) and MM (mother of M)

Clear understanding of the complexity of the fetal cardiac anomaly (septal defects, underdeveloped ventricle, need for multiple surgical procedures, perhaps a transplant)

Paediatric cardiologist warned them that the cardiac surgeons in another hospital might refer to the cardiac problem as life-threatening.

Multiple offers were made by different members of staff to say that a termination of the pregnancy would be available

Mum's diary - 1

- Concern that other people would think them cruel to put the baby through multiple surgeries

- Wish to keep secret the fact that they had been offered ToP … as people may feel they should have done this

- Close family members have been (implicitly) critical and given details of other babies who died after multiple operations for heart defects

- Mixed feelings about several friends who have announced they are pregnant: "Why do they have to get pregnant now??"

Mum's diary - 2

- Repeated expression of the baby dying after she had set up a strong bond/attachment. She "would not cope"

- "I feel like I will look at our child and just cry every day either through fear of losing him/her or feeling sorry for everything that child has been through because I chose to bring him/her into the world"

- Has been reluctant to talk about her feelings with professionals (but might talk with older child's health visitor)

- Cried when she saw happy scene of child with Down's syndrome greeting her dad at nursery; she still sometimes wished the baby didn't have Down's

Mum's diary - 3

- Wants even more than before to work at home (in financial role), so her training course really matters to her
- Anxious in case she doesn't pay enough attention to her 3 year-old daughter
- "Not knowing or dwelling on things is better for me. Researching every angle of something and then storing that in my mind for when it does/doesn't happen is not good for me …. Throughout the whole pregnancy I have been very well looked after. There isn't anything that I would change in the support that I had. *What I would change is my own thoughts and actions, but I'm working on that now*"

Audio Log of family conversation

- Raw hurt …. alongside mundane aspects of everyday life
- Discussion on how a couple can discuss the question of terminating a pregnancy. Undue influence by the man?
- Mum’s mother giving support, being non-directive
- Discussion about staff and whether they were trying to “influence” the couple’s decisions
- There was also a discussion about being recorded and participation in this ethics research (data not included here)

Raw Hurt

MM: Christ, that baby's got a lot to go through.

M: I was like fucking hell, could there be anything else?

……

D: A late stage, you're halfway through aren't you? So we know there's a baby in there. We've had three scans, we've seen the baby three times moving about. **I don't think that I could live with myself if we decided to terminate now**.

MM: I know it's so hard. ….. But then you've also got to think **is it fair on the baby**, what that baby's got to go through?

M: ….. I don't want any of this to be happening. But I'm so scared of giving birth and getting an attachment with that baby, and then it's your physical child has died. **That's what frightens me, I don't want to lose a child**.

The Life World: Other Stuff

M: … **We've got [Daughter] to think of as well, she's going to be going through all this as well**. Having her brother or sister that's in hospital all the time, and we've got to think of her …. It's just shit isn't it really, shit fucking situation. Now I do feel like I want to give up everything. Work, college, everything. **How the hell am I supposed to concentrate now on college and pass exams** and shit, and go and listen to their problems all day?

MM: When are you back, next week?

M: Monday. **What would you do, Mum?** How would you get yourself through this?

MM: I don't know.

Risk to the baby

M: I'm just scared that I'm not going to have this baby, or if I do have this baby it's not going to be for long. That's what I'm scared of.

MM: I know, but don't think like that. Don't think like that.

M: I know, but when you've got someone sat telling you it's a life threatening condition, it's like fucking hell, **that baby is clearly not going to live a long happy life then is it**?

MM: But it might love, he or she might. Another sandwich anyone?

Undue Influence?

D: **Yeah, I don't want to feel like I'm pushing my thoughts on you though**. I mean I think you probably have more of a say than I have because it's your body

M: **It's just as much your child as mine**. I'd never go over your head on anything. Because not only have I got to live, like I say, for the rest of my life thinking what if? it's also then I've got rid of a baby that you wanted. **So I'd never go, "He was just saying he doesn't want to talk me into anything"**, because it's my body and I'm going through it. … Yeah I've got to live with that I got rid of a baby, and what if? what if? all my life. … I'd never go over your head.

D: **You can easily turn that around though and say I told talked you into keeping the baby**, and then we've just had to go through a lot of.

MM: Yeah, it works both ways doesn't it?

Professional recommendation to terminate (?): 1

M: I just felt like I was getting okay with the Down's syndrome, and oh right there's a heart problem and it'll need surgery. That's okay.

D: But I think maybe two or three times today I think **they've told us that if we did want to terminate they'd support**. But they were mentioning it more this time than on the last scan.

M: Yeah, well **all three people we saw said it today**.

D: Yeah.

M: **All three of them, termination is still an option**.

D: And the one doctor said it's your right, it's your right, and I don't know, it just didn't sound right to me.

Professional recommendation to terminate (?): 2

MM: Was it sounding like they were saying they think you should or?

M: The one doctor said "**oh it's your right to terminate**", I think he said advisable. And what he meant was it's advisable that we're given the option.

D: Yes, he wasn't advising us to have no, he was saying he needs to.

M: He said **"I've got to give you the option to terminate**". That's what he was saying.

D: Yeah.

M: Because when he said that, oh you know de, de, de, it's advisable. **I thought, "so you're basically saying you think that we should then"**. That's what I was thinking.

Interview: attitude towards disability shaped by early experiences of disability

PT: Yeah, yeah and I hate to say it but yeah, I, you know, in work if they had, they used to have disabled groups in playing Boccia and things like that in the main hall and I would go, I wouldn't walk along that corridor, I'd go up and around, to avoid somebody coming over and asking me, engaging in conversation, and yeah, I, I would go out of my way to avoid, because I was nervous.

INT: Right, right, but then … were you nervous because the woman next door had actually come and, and pulled your hair and actually basically, then attacked you almost?

PT: I think so yeah, yeah, and I, I didn't know how to speak to people, you know ….. how to …. communicate …

Genetics and the Life World

- Patients and families have sophisticated discussions about the problems they face and the ways in which others (professionals and family members) attempt to influence them

- Decisions about reproduction
    - depend upon judgements of how worthwhile our lives are
    - depend upon (early) experiences of disability
    - these judgements have consequences within the family // society

- In some contexts, stigma is a key factor that shapes quality of life

Diaries

- Keeping a diary was spoken of as positively helpful by several participants
- Helped to maintain relationship with partner
- Off-loading in diary spared the partner
- Organised thoughts so that discussion with partner was more focused and helpful
- Could be a valuable research tool - but perhaps not so helpful in practice if shared with clinicians

Audio logs and recording conversations

- We had two family conversations recorded:
- One was the very rich one around the Down syndrome foetus with cardiac anomaly
- The other followed a HD predictive clinic appointment where the partner was not present. The conversation was a simple account of the clinic and did not generate particular insights
- **In future projects**, I would strongly encourage patients to keep both diaries and audio logs, and to capture informal family conversations if at all possible