

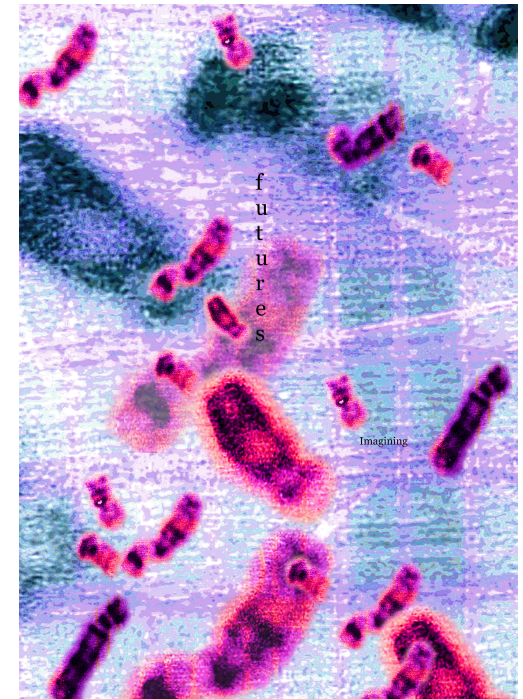


Unravelling Disability's DNA: Experience and Identity in a Genomic Age

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Background: Emergence of 'Generation Genome'



- New technologies and techniques are revolutionising the way prospective parents approach reproduction
- Whole genome/exome sequencing being integrated into NHS care
- No longer a question of what *can* be done with genomic technologies, but what *should be done*?

Background: Emergence of 'Generation Genome'



- What information is significant and what is not?
- Notion of 'disease severity' typically used to discern this boundary, however concept is not clearly defined
- Lack of definition universal guidance is striking given the relevance of this concept, e.g.:
 1. Abortion law
 2. Evaluation of potential screening programmes
 3. Return of incidental findings from sequencing

What is a severe disease?

- HFEA evaluates disease severity when granting licenses for PGD. Focuses on the following to determine severity:
 1. Penetrance of genetic variant
 2. Age of onset
 3. Symptoms and impact
 4. Treatment (what type/how invasive and effective)
 5. Quality of life
 6. Variability and range of possible symptoms
- Perspectives of people living with the conditions have generally been excluded.



Experiential Knowledge

Experiential knowledge refers to knowledge emerging from 'lived experience' of a phenomenon

Embodied Experiential Knowledge (derives from direct sensory experience)

Empathetic Experiential Knowledge (derives from close emotional ties between individuals)

(Abel and Browner, 1998)



Why ask affected families?

1) Various impacts for people living with genetic disabilities distinct from those from rest of UK population:

1. Change in public profile of disease
2. Expressivist objections
3. Reductions in research funding/social support
4. Reductions in peer-to-peer support

2) Experiential knowledge of affected families now relevant to the reproductive decisions of whole population



Imagining Futures Project

wellcome trust

Current Wellcome project on social and ethical aspects of genetic screening

Exploration of the views of **people with different types of genetic impairment** (n= 108) and **genomic sequencing volunteers** (n= 31) towards the use of whole genome/exome sequencing as a population screening tool (pre-conceptually, prenatally or in newborns)



Included Conditions



Cystic Fibrosis



Thalassaemia



Fragile X Syndrome



Haemophilia



Spinal Muscular Atrophy



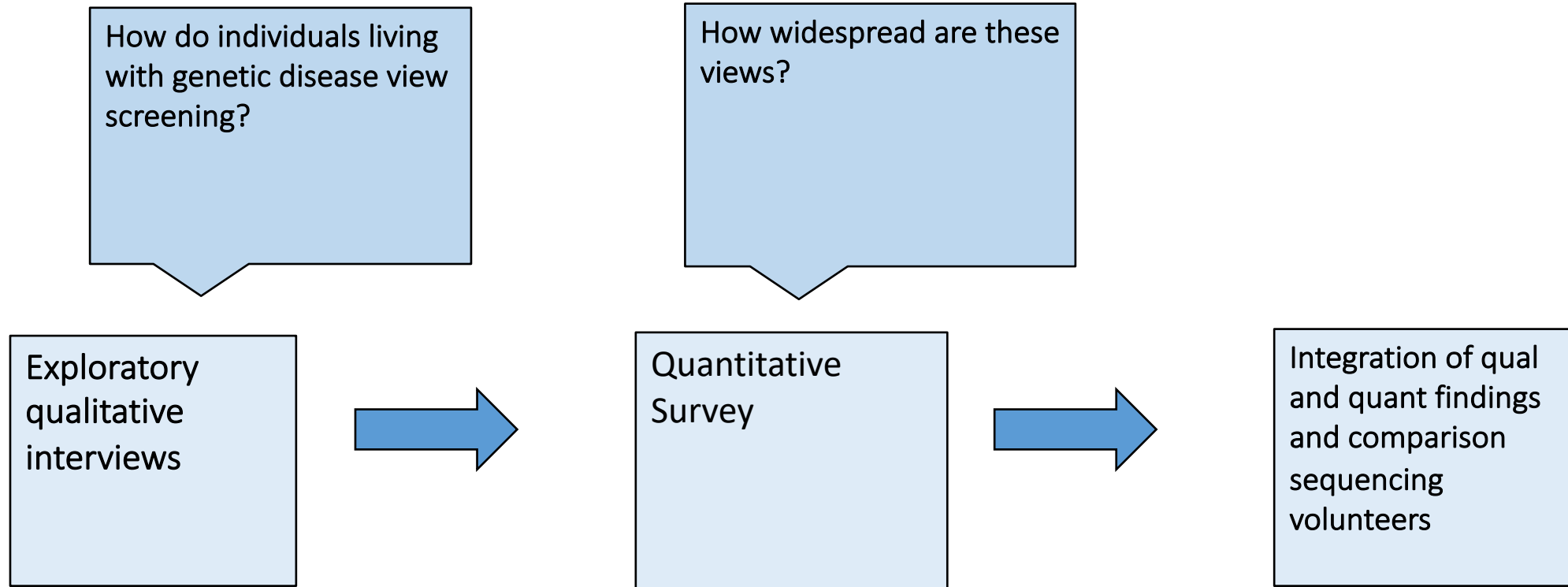
100,000 Genomes Project

What might future genetic screening programmes look like?

- **Pre-conception carrier screening:** blood test before a pregnancy is established.
- **Prenatal Carrier Screening:** pregnant women screened, as well as partner and foetus where indicated.
- **Newborn screening:** heel prick test of all newborns



Imagining Futures Study: Exploratory Sequential Mixed Methods Design



Affected Families (interviews n= 108, surveys responses n= 751)

	Haemophilia	Fragile X Syndrome	Thalassaemia	Cystic Fibrosis	SMA
Interviews					
Affected Adults	10	2	7	10	15
Family Members	12	15	8	8	21
Totals	22	17	15	18	36
Surveys					
Affected Adults	179	Launched July 2018	46	Launched Nov 5 th 2018	82
Family Members	149	Launched July 2018	141	Launched Nov 5 th 2018	255
Totals	414	In Progress	187	In Progress	337

Finding One: Genetic Condition As Transformative Experience for Parents

- Diagnosis of genetic condition most commonly an unanticipated event
- Mismatch between prognosis and lived experience of genetic condition within family
- Different reactions to child's diagnosis between sporadic cases and obligate carriers of haemophilia

Finding one: Genetic Condition as Transformative Experience

“When Finlay was diagnosed.....they were telling me my son had this horrible disease that I had only vaguely heard ofbut yeah I absolutely would have been one of those people saying ‘this isn’t right, I’m going to terminate’because I was ignorant really and knew no better”

[Jinny (38), Mother of Finlay, Haemophilia A(6)]

Finding one: Genetic Condition as Transformative Experience

“I think when I was first offered the testing, when I was pregnant, it took me a bit off-guardI’ve always been around haemophilia. My dad has it, two of my cousins have it...[..]... there was no way I would have had that test, because I wouldn’t have terminated for a condition....that we can manage....A kid with haemophilia would be really lucky to be born into a family like ours because we are so clued up you know?”

Emma, age 40, daughter of man with Haemophilia A, Theo born without the condition

Finding Two: Attitudes Vary Across Impairment Groups

“Oh God yeah I just hate it [reproductive genetic medicine]. I mean it’s genocide for the modern era, isn’t it? It’s portrayed as this sophisticated and progressive new thing, but in reality all they’re doing is bumping the babies off, aren’t they?”

[Rosie, age 20, Type II SMA]

Finding Two: Attitudes Vary Across Impairment Groups

“I think there is so much ignorance and that is why so many children continue to be born with unnecessary diseases like thalassaemia, it’s just not acceptable...[...].I think the problem is that parents are selfish and thinking only of their desire to have a child and not the child itself”

[Chiara, 44, thalassaemia]

Finding Three: The Meaning of The Experience and Identity Politics

- How far impairment incorporated into personal identity was significant to responses to genomic medicine and the way impairment experiences were interpreted.
- Dimensions of Experience: age of onset of condition, stability/deterioration, availability of and associated burden of medical treatment, illness and suffering, social conditions and stigma

Finding Three: The Meaning of Experience and Identity Politics

“I can understand why somebody might want to find out....but it’s just too close to home and too hypocritical for me to think it’s ok to abort someone with one condition when I’ve also got a condition...because that then opens the door to someone aborting me, or someone else with CF.”

[Seth, 32, Cystic Fibrosis]

Finding Three: The Meaning of the Experience, Disability, Impairment and Illness

“...some parts of it [life with SMA] don't bother me at all. And probably not the parts most people think. Most people think not being able to walk, or dress yourself....are the worst things about it..[...]...but they're...um....they're not actually. Especially for me when I've never done those things in the first place”

[Julia, 40, Type III SMA]

Finding Four: Lived Experience and Clinical Classifications of Genetic Conditions

“My sister [with type III SMA] hates having SMA. She....resents it every single day. But the funny thing is, everyone expects me to be the bitter one because I’m type II..[...]...[whereas] she can still walk. But.....I’ve never known any different, and she has”

[Rachel, age 24, type II SMA; Chloe age 26 type III SMA]

Finding Five: The Limits of Lived Experience

“The trouble is, how I thought about it [haemophilia] in my 20s and 30s, when I was having my own children, is very different to now. Back then it didn’t affect my joints too badly, you know....I was able to ignore it most of the time [unlike now].....Because yeah your haemophilia might be fine for the first 20/30 odd years of your life...but it doesn’t mean it will stay that way”

[Graham, 59, Haemophilia A]

Key Findings

1) The lived reality and social dimensions of genetic impairment is critical to reproductive attitudes amongst families and adults living with genetic conditions.

2 &3) The meaning assigned to that lived reality is greatly influenced by the nature of impairment experiences, but also the politics and psychology of personal identity, resulting in very different attitudes across and within impairment groups.

4) Classifications of disease severity solely through a medical lens do not 'map' onto the lived reality of genetic disease in a straightforward way and consequently are of limited use in predicting outcomes for diagnosed fetuses.

5) However, lived experience is also limited: subject to revision in light of changing circumstances/experiences

Concluding Thoughts: Making Use of Lived Experience?

How can/should the lived experience of disabled adults and families inform the goals and practice of clinical genetics?

Denmark- 'contact a family' programme

What are the challenges associated with using lived experience in this way?



Papers From This Study

Boardman F, Hale R, Young P. (2019). Preventing lives affected by haemophilia: A Mixed Methods Study of the Views of Haemophiliac Adults and Their Families Towards Genetic Screening, *Molecular Genetics and Genomic Medicine* [in press]

Boardman F, Hale R. 2018. Genetic Impairment and Selective Reproduction: The Views of Adults with Genetic Conditions Towards Genetic Screening, *Molecular Genetics and Genomic Medicine*, DOI: 10.1002/mgg3.463

Boardman F, Hale R. 2018. *"I didn't take it too seriously because I'd just never heard of it"*: Experiential Knowledge and Genetic Screening for Thalassaemia in the UK, *Journal of Genetic Counseling* (accepted, in press).

Boardman F, Hale R. 2018. Responsibility, Identity and Genomic Sequencing: A Comparison of Published Recommendations and Patient Perspectives on Accepting or Declining Incidental Findings, *Molecular Genetics and Genomic Medicine* [in press].

Boardman, F. 2017. Experience as Knowledge: Disability, Distillation and (Reprogenetic) Decision-Making, *Social Science & Medicine*, 191: 186-193.

Boardman, F., Young, P., Warren, O., Griffiths, F. 2017. The Role of Experiential Knowledge Within Attitudes Towards Genetic Screening: The Views of People With and Without Experience of Spinal Muscular Atrophy, *Health Expectations*, 21 (1): 201-211.

Boardman, F., Young, P., Griffiths, F. 2017. Impairment Experiences, Identity and Attitudes Towards Genetic Screening: The Views of People With Spinal Muscular Atrophy, *Journal of Genetic Counseling*, 27 (1): 69-84.

Boardman, F., Young, P., Griffiths, F. 2017. Population screening for Spinal Muscular Atrophy: a mixed methods study of the views of affected families, *American Journal of Medical Genetics Part A*, 173(2):421-434

Boardman, F. 2014. The expressivist objection to prenatal testing: the experiences of families living with genetic disease, *Social Science & Medicine*, 107, 18-25.

Boardman, F. 2014. Knowledge is Power? The Role of Experiential Knowledge in Genetically 'Risky' Reproductive Decisions, *Sociology of Health & Illness*, 36 (1): 137-150.

Dissemination and Impact

- **Policy**

Contributed published papers and comments as part of evidence review conducted by UK NSC national consultation on SMA screening.

Contributions to Nuffield Council on Bioethics reports (NIPT and genome editing)

Contributions to policy debates Equality and Human Rights Commission

- **Public**

Art installation [in]:valid, café scientifique, Radio 4 presentation, Funzing talk, articles in the Conversation and the Independent.

- **Professional**

Talks to genetic counsellors, embryologists, support group staff, pharmaceutical company (Biogen), publications in professional magazines e.g. Spine, SMA News Today

- **Academic**

Papers, academic conferences, incorporation of findings into teaching (substantive and methods)



Articles in the media

- **Boardman F, O'Neill H.** 2018. Should we edit the genomes of human embryos? A geneticist and social scientist discuss, <https://theconversation.com/should-we-edit-the-genomes-of-human-embryos-a-geneticist-and-social-scientist-discuss-100355> 24th September.
- Boardman, F. 2018. On Genome Editing, The Independent, <https://www.independent.co.uk/voices/genome-editing-human-genes-designer-babies-illness-traits-diseases-disabilities-nuffield-council-a8451491.html> 17th July.
- **Boardman, F. May 2018. BBC Radio Four, Four Thought**, invited talk and Q&A with live audience, Big Cosy Bookshop, Coventry. <https://www.bbc.co.uk/programmes/b0b0v5xb>
- **Boardman, F.** 2017. Why we should be worried about gene carrier screening, The Conversation, 2nd August, <https://theconversation.com/why-we-should-be-worried-about-gene-carrier-screening-81124>
- **Boardman, F.** 2016. Should we edit out genetic disease? The Conversation, 9th May, <https://theconversation.com/should-we-edit-out-genetic-disease-58654>
- **Boardman, F.** 2016. Identity, disability and the genome. Bionews, 11th April, issue 846, http://www.bionews.org.uk/page_638169.asp
- **Boardman, F.** 2013. Screening Dilemmas: Disease, Disability or Something In-Between? Bionews_25th November, issue 732, http://www.bionews.org.uk/page_365114.asp