Experience as Knowledge : Prenatal and Genetic Testing Decisions For Spinal Muscular Atrophy

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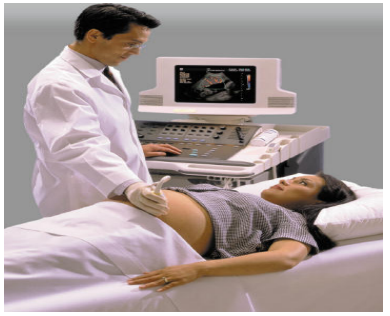
Context of Study



- Increasing number of antenatal screening tests and prenatal genetic tests available; women encouraged early on to consider the possibility of disability/disease
- Cystic Fibrosis heel prick test to identify ‘carriers’. Ideas about kinship being challenged as new genetic identities discovered.
- Human Genetics Commission (2011) ‘Informing Options, Increasing Choice’ , genetic screening on the horizon.
- Concerns amongst feminist writers about the effect these tests have on women- ‘the tentative pregnancy’ (Katz Rothman,1986) as well as issues of power and control
- However- potential for women to exercise greater reproductive autonomy? (Sharp and Earle, 2002; Brooks 2001)

Context of Study

- Disability Rights Supporters have also raised concerns. The ‘expressivist objection’ (Parens and Asch, 2000).
- Concerns about ‘overly medicalised’ portrayals of disability in antenatal screening consultations and discussions around selective termination Vs the actual experiences of families living with disability.
- Tensions between these two perspectives: ‘Feminism, Abortion and Disability: Irreconcilable Differences?’ Sharp and Earle (2002)

Experiential Knowledge

- Emergence of ‘experiential knowledge’ as an ‘authentic’ form of knowledge in the literature. It is derived both from direct sensory experiences of the world (embodied experiential knowledge) or through the experiences of others (empathetic experiential knowledge (Abel and Browner, 1998: 315)

- Growing body of research into the way in which knowledge accumulated from everyday life and embodied experiences can challenge medical understandings (Lippman, 1999; Etchegary et al., 2008) and also can be drawn on in medical decision making (Hallowell, 2006; Downing, 2005;Parsons and Atkinson, 1992).

Experiential Knowledge

- Disability rights supporters have drawn on experiential accounts of impairment and disability in families to challenge dominant constructions of disability.
- Experiential knowledge has political significance for feminist writers as well as disability rights supporters.

My Research

- How far does experience of disability and impairment, both embodied and empathetic, within families affect genetic testing decisions (including prenatal testing)?

- How do families negotiate the ‘irreconcilable differences’ suggested by Sharp and Earle which are brought to the fore by the existence of genetic technologies?

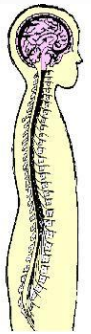
Spinal Muscular Atrophy

- After Cystic Fibrosis, SMA is the most common (potentially fatal) autosomal recessively inherited condition (i.e. single gene disorder requiring both parents to carry one copy of the gene to transmit SMA)

- It affects 1 in every 6,000 newborns in the North West European population (Dreesen et al., 1998)

- Three main types: SMA type I, SMA type II and SMA type III of varying severities. SMA type I is currently the most common genetically inherited condition causing infant mortality in the UK and America (SMA Foundation, 2009)

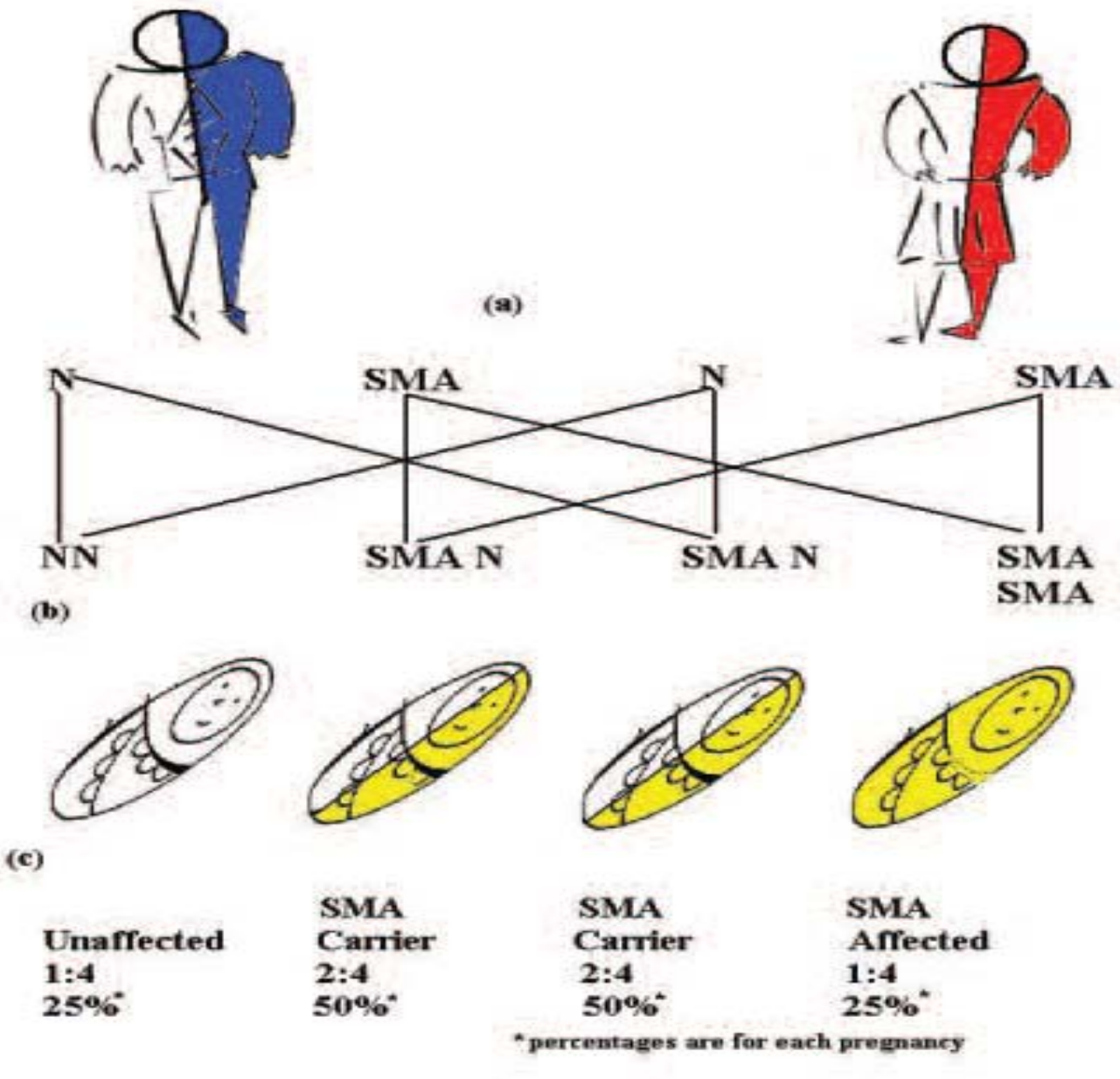
Spinal Muscular Atrophy



- SMA is characterised by a degeneration in the anterior horn cells in the spinal cord (responsible for relaying nerve ‘messages’ between brain and muscle). Results in generalised (and often severe) muscle weakness and premature death in infants.

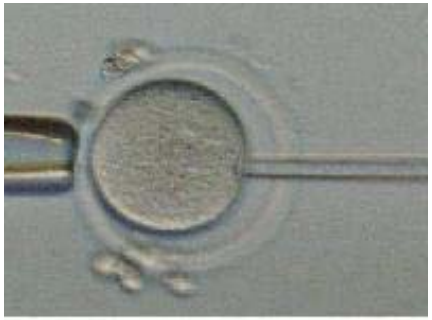
- Typically understood that those with milder forms of SMA may be ambulant until late teens and gradually experience increasing muscle weakness into adult life. Children diagnosed with Type II SMA are rarely able to walk at all and need help with most everyday activities, and Type I SMA, most frequently diagnosed in early infancy, typically causes death before the age of 2.

Inheritance of SMA For Two Carrier Parents



Genetic Testing Available for SMA

- Adult Carrier Status Testing (over 18s)
- Prenatal Genetic Testing (for families known to be at risk)
- Pre-Implantation Genetic Diagnosis (for families known to be at risk)
- SMA is not currently screened for in routine antenatal screening, although there have been calls for this form of testing to be introduced (Prior, 2008)



Methods

- Recruited 61 participants with SMA in their family. Primarily through the Jennifer Trust for SMA (JTSMA), a national charity which currently supports 2,000 families and individuals affected by SMA in the UK.
- Also recruited through disability parenting websites, disability organisations, personal websites set up by people with SMA and personal contacts
- 59 in-depth interviews were carried out (44 over the telephone, 10 via e mail and 5 face to face). Interviews lasted on average 1 hour and ten minutes each. E mail interviews were conducted over periods ranging from 3-8 months.
- 25 participants were diagnosed with SMA, 36 had SMA in their family.
- 79% of participants were female. Reproductive decision making highly gendered.
- Participants were at different points of reproductive decision making.
- All recordings transcribed verbatim and analysed using Nvivo7 using a modified grounded theory approach over a period of 9 months.
- All participants were sent copies of their completed transcripts for verification purposes. Participants asked for consent for quotations to be used in presentations.

The Uses of Experiential Knowledge in Reproductive Decision Making

- Participants presented their experiential knowledge in different ways to support/justify their reproductive decisions and present them as responsible.

- Reproductive decisions were always made in the context of familial and social relationships: within emotional bonds, shared experiences and obligations. Many participants experienced competing responsibilities arising from the context of these relationships.

Experience as Warning/Reassurance: *"I'm sitting in the prettiest spot in the whole argument"*

Experiential knowledge treated as a certainty in reproductive decision making and used as either reassurance/warning to justify particular decisions. E.g. Fraser who lost two children in early infancy to SMA:

….the ability to under go prenatal testing [after the deaths of 2 babies, and before going on to having child without SMA] was a God send for us, because no one would want that if they could avoid it and I think everybody would say the same who is affected by it. I don't see any dilemma at all with type I testing, the test is there and I think every parent should take advantage of that. I can't understand where they're coming from if they don't…I know some parents face a big dilemma around type II testing, but if type I is the biggest genetic killer of children under the age of 1 in the UK, which I'm told it is, then I'm sitting in the prettiest spot in the argument in a way because I don't see any dilemma with using the test. You know, and very few people could argue against my position…I know some people talk about type Is living past their first birthday, but that wasn't going to be the case for our children, so I think I'm sitting in the securest spot in the whole argument, I think.

(Fraser)

For other participants, their experiences of SMA in their family and in the families of others were reassuring:

I suppose actually in hindsight now, SMA's been a positive thing really. I think that a lot of sort of what I've done and what I've achieved has been hugely down to sort of having the personality to overcome the problems that have come along with SMA. Um I think when you sort of know your own experiences and those of all my friends with SMA and when you think about having children and whether they will be affected by SMA, the way I look at it is, 'well I've coped and I'm fine with everything' so, you know, it's not all bad. I mean there're always going to be a certain amount of people who feel it's irresponsible to bring in a child to the world knowing that they're going to be disabled or because you're disabled yourself, but they tend to be the people who have no experience of disability themselves, they don't know what it's like and it's just their perceptions… and that's just part of the harshness of life and I don't take a lot of notice of them.

(Rhona, Diagnosed with SMA Type III)

The Burden of Experiential Knowledge: *"I'm stuck between a rock and a hard place"*

For some participants, experiential knowledge could be burdensome as it made reproductive decision making more complex:

Yeah I think I'm in quite a difficult position with that really, with having children. Because my first thought was that I would never bother with any sort of testing, if we were going down that route, because I know SMA quite well really and I see what a wonderful person Megan is, and you know we often say that her condition has given her strength, mental strength that she may never have developed, so I know it on that level, but… I also know the other layers of it…um [pause] I suppose it sort of sounds cold to say it but, it's human nature to sort of think that if it could be preventable for somebody…Is it going to be really difficult or is it something that you can take on? Of course you're going to want the child that doesn't have to struggle and things, no one wants to see their child go through that. So I've got that, but on the other hand I've got 'what would that say to Megan?' It would be like me saying that she wasn't important or that her life wasn't worthwhile with her condition, that there was something wrong with her. So where you go with that….um….Yeah I'm stuck between a rock and a hard place [laughs].

(Claire, the sister of Megan who was diagnosed with SMA Type II in early childhood)

What would I be saying about my own life with SMA and everyone else's living with SMA if I got rid of a baby with SMA?

(Sarah, diagnosed with SMA Type II and whose sister is diagnosed with SMA Type III)

Experiential Knowledge, Ownership and Privilege: *"they can only see it from where they are, not where I am"*

Strategic privileging of certain forms of 'authentic' experiential knowledge over others could act as an emotional buffer against complex decisions within families, but also a means by which to invalidate those perspectives that would define the decisions as irresponsible:

I definitely feel that no one can ever know what SMA is really like until they live with it themselves, you know, they've got it. Because other people, outsiders, no matter how close they are to you, they could be in your family even, they can't put themselves in your position. And my brothers, they don't really know how much help I need, because while I was at home with them growing up I was quite capable of doing quite a bit for myself, it's only since I've moved out and lived on my own that now I need PAs [personal assistants] all the time…They [brothers] know I need help, but I don't think they know how much help I need. So it's really just between me and my PAs. Not even my friends...it's not something you...talk about...everything that's physically hard is usually done in the house, you know all the personal help and looking after the house and everything, no body sees that apart from the person who's helping me. And even then, how do they know how it feels to you? So yes it's so individual in that respect, you can't know unless you've lived that life.

(Gill, Diagnosed with SMA Type II and whose able-bodied brother, Luke, underwent carrier testing before having children)

Conclusions

- Experiential knowledge mediates reproductive decision making in complex ways.
- By drawing on the suffering of previous or existing family members and defining this experience as a ‘warning’ as to the certain suffering of future generations, participants were able to justify their decisions.
- Those who opted to avoid prenatal testing and selective termination frequently spoke of SMA as a condition that did not involve suffering, but a disability that could be ‘overcome’ and around which a happy and fulfilling life could be built.
- Both of these strategies could assist reproductive decision making as they offered a ‘way out’ of the complex dilemmas associated with the decisions.

Conclusions

- However, for some participants, experiential knowledge introduced new complexities. Experiential knowledge of SMA could invalidate certain ways of justifying reproductive decisions.

- Experiential knowledge could be trapping- to prevent another life with SMA was considered tantamount to rejecting their current child with SMA, or devaluing the lives of others with SMA (the ‘expressivist objection’; Asch, 2001).

- Medical, genetic and experiential knowledge were intertwined in the way in which participants presented their decisions.

Practice and Policy Implications

- Screening for SMA may become available in the UK in the future and Cystic Fibrosis testing (heel prick test for carriers) is already underway. More people going to have to make decisions about the use of prenatal testing with and without knowledge of the condition in their family.
- Disability rights supporters have argued that more knowledge about day to day experiences with given conditions should be made available to prospective parents undergoing antenatal screening and prenatal testing.
- Attitudes to disability in future generations can be influenced by the birth of an affected family member. However, experiential knowledge can also cause distress and constrain the choices of prospective parents. It may not always be a helpful resource and had the potential to disrupt relationships within families.
- The expressivist objection is an important consideration in families living with genetic disability and needs to be acknowledged. Experiential knowledge of the condition can exacerbate these concerns.

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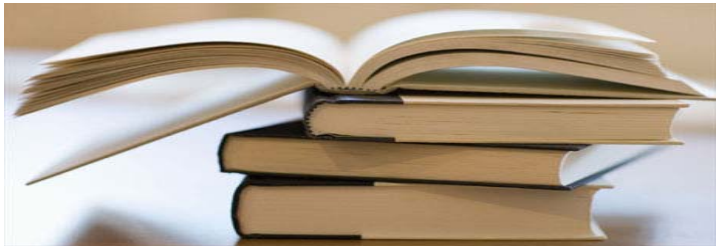

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