Screening for SMA: What are the Issues?

Imagining Futures: the Social and Ethical Implications of Genetic Screening

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Who Am I?

- Researcher based at Warwick Medical School with a background in health and social science
- Completed a PhD study in 2010 on how families living with SMA approach reproductive decision making and use genetic technologies
- In 2012 was awarded 3 years of funding (from ESRC) to explore the social and ethical implications of genetic screening
Overview of the Talk

- To describe the current research project and how you might get involved
- To highlight some of the key findings so far
- To outline how this research will be used in policy reviews
- To take questions relating to the research
Issues around prenatal testing and screening are extremely sensitive

A diverse range of perspectives exist—none are ‘right’ or ‘wrong’

In this presentation you will see some examples of these perspectives—these are fictionalised to protect identities.

Please be aware that people in this room may not feel the same way on these issues as you—be sensitive when raising questions/points!
Background Work—PhD Thesis 2006–2010

- One on one interviews with families living with SMA
- Asked about: experiences of life with SMA, uses of genetic testing, views about termination for SMA
- 61 participants with different Types of SMA in family
# Participants 2006–2010 Research

<table>
<thead>
<tr>
<th>Participants</th>
<th>Numbers</th>
<th>Female</th>
<th>Male</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnosed with SMA</td>
<td>25</td>
<td>21</td>
<td>4</td>
</tr>
<tr>
<td>Sibling of person with SMA, without SMA themselves</td>
<td>7</td>
<td>4</td>
<td>3</td>
</tr>
<tr>
<td>Parent of person diagnosed with SMA</td>
<td>24</td>
<td>21</td>
<td>3</td>
</tr>
<tr>
<td>Grandparent of person diagnosed with SMA</td>
<td>1</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Son or daughter of person diagnosed with SMA</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Partner or spouse of person diagnosed with SMA</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td><strong>Totals</strong></td>
<td><strong>61</strong></td>
<td><strong>49</strong></td>
<td><strong>12</strong></td>
</tr>
</tbody>
</table>
# Diagnoses in Families

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Number of Participants with that diagnosis in family</th>
<th>Female</th>
<th>Male</th>
</tr>
</thead>
<tbody>
<tr>
<td>Type I SMA</td>
<td>12</td>
<td>11</td>
<td>1</td>
</tr>
<tr>
<td>Type II SMA</td>
<td>32</td>
<td>25</td>
<td>7</td>
</tr>
<tr>
<td>Type III SMA</td>
<td>11</td>
<td>10</td>
<td>1</td>
</tr>
<tr>
<td>SMARD</td>
<td>3</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>SBMA</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Autosomal Dominant SMA</td>
<td>1</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td><strong>Totals</strong></td>
<td><strong>61</strong></td>
<td><strong>49</strong></td>
<td><strong>12</strong></td>
</tr>
</tbody>
</table>
Findings

- Diverse range of viewpoints on reproductive decision-making. Some very for, others very against, genetic testing for SMA

- Experiences of life with SMA were key to whether people felt they would want to prevent future lives affected by SMA
Examples of Findings

“Seeing how my parents dealt with my SMA I just know I could handle whatever disability came along in my own offspring. I feel committed to not having testing for any conditions, be it Down’s, or whatever, because of the family I’ve come from. We got on fine”

“Having genetic testing and selective termination was a no-brainer for us– we knew we couldn’t go through that again and I don’t see any dilemma with terminating for SMA, none whatsoever. SMA is a heart-breaking experience, even for those who’ve got the milder Types– you’d still not CHOOSE to have it, would you? And testing gives us a choice about this.”
Current Project: Imagining Futures

- Screening for genetic conditions has come onto the public agenda in recent years
- Screening different to testing. Screening refers to performing genetic tests on population, not families already living with SMA
- Some single gene disorders, like Cystic Fibrosis, are already screened for (heel prick test)
- Given the diversity of views on testing within families living with SMA, became interested in attitudes towards population screening
How might SMA be screened for?

1) Pre–Conception Genetic Screening

Individuals/couples screened for their SMA carrier status BEFORE conceiving a pregnancy. However, a couple could not be told in advance what Type of SMA they are carriers for.
How might SMA be screened for?

2) Prenatal Screening

All pregnant women screened for SMA carrier status. If she is a carrier, the father is screened. If both parents are carriers, a diagnostic test on foetus would be offered, however it could not diagnose the Type. Termination could be offered. Could identify some adults with adult-onset SMA!
How might SMA be screened for?

3) Newborn Screening

Blood taken from newborn and genetically tested to see if baby has SMA. This test will not be able to accurately diagnose the Type of SMA. It would also identify some carriers of SMA—parents would be informed of baby’s carrier status.
In May 2013, the UK National Screening Committee (advisory board to government) undertook national consultation on SMA Screening.

In December 2013, it was decided that screening was not recommended at this time.

Specific issues highlighted:

1. Not enough information about the total number of people affected in UK (and Type)
2. The test cannot determine whether children are likely to be severely or very mildly affected by the condition.
3. As the test is not reliable it may not help people make decisions about whether to have children.
4. It is not clear whether people would want to have the test or how families living with SMA feel about it.
Imagining Futures Project

Phase 1
In-depth interviews (approx. 30)

Phase 2
Nationwide survey (approx. 2,000 people)

Expert Panel made up of families living with SMA consulted throughout
36 interviews undertaken (from original sample 2006–2010)

Broad range of viewpoints:
“I think it’s important to give parents screening so they know and can decide if they want to go through with having a baby with SMA because not every parent is cut out for it….some parents just couldn’t deal with what comes along with having a child with SMA and then of course that’s of detriment to the child so I think the option to opt-out should be there”
“I don’t think screening is a route we should go down because I think that’s playing God. To me, it’s the same as saying that your life’s not as worth while if you have SMA and certainly there are many things in life that make people unhappy—mental health problems, domestic violence— but they can’t screen for those sorts of things! And really who’s to say that a baby with SMA shouldn’t have a shot at having their life? Who should have the power to say that?”
Viewpoint 3

“I absolutely think screening should be offered and offered before the kids are born with it. I was furious when I found out that a test in pregnancy exists for this condition and I wasn’t offered it before [son] was born. If they can do it, they shouldn’t withhold that from parents – for money reasons or whatever. No one should be forced to bring a child into the world who will suffer – and all because the parents didn’t know about it beforehand. By not doing the screening, they’re robbing parents of the chance to stop SMA happening to their child, and who wouldn’t want to stop it happening to their child?”
“I just don’t think a screening programme is feasible if it can’t tell you the Type of SMA, because I think if it’s Type I and the prognosis is very poor, then a termination might be the right option, however the waters get murkier when it comes to Types II and III, because they can have a great quality of life with the right support. So screening for me, it’s like giving people information— but just not enough that they can base a decision on— and it’s such a big decision. I just think if you can’t give accurate and balanced information then I think in some ways you’re better off staying ignorant and getting on with what life throws at you.”
Phase II

- SMA Screening Survey (UK)
- Designed to measure how widespread some of the views expressed in the interviews are
- Due to be out in September
- Online and paper versions to be made available
- To be completed by anyone with SMA in their family living in the UK
- Findings of the research to be submitted to the next policy review of the UK National Screening Committee (likely to be 2016–7)
Feel free to ask questions about the research.

Please be sensitive to others in the room who may have a different perspective to you.

If this talk has raised issues for you that you would like to discuss with a support worker, help is available. Please ask.
Where can I find out more about the research?

www.warwick.ac.uk/imagining_futures

https://www.facebook.com/imaginingfuture

https://twitter.com/ImaginingFuture