The Fragile X Community's Views on Screening

Recently, Dr Felicity Boardman and colleagues from Warwick Medical School conducted a study to investigate the implications of genetic screening programs. Very little was known about what families living with Fragile X Syndrome (FXS) and other genetic conditions think about screening, particularly carrier screening which is carried out before a pregnancy is established (pre-conception genetic screening), during pregnancy (pre-natal screening) and after birth (post-natal screening).

Thank you to all those who took part in this research. The initial summary results are presented below. We will keep you informed about progress following on from this study, as well as any more in-depth findings which become available.

This data will be used to create academic publications, and as evidence to submit to the next UK National Screening Committee review of Fragile X screening policy. We will keep members updated as academic papers and reports become available.

Content warning: this article discusses some challenging themes including pregnancy termination and discusses aspects of Fragile X using medicalised language.

Note: Quotes used throughout the article have had minor details amended in order to protect the participants' anonymity.

Who took part in the research?

We received an overwhelmingly positive response from members of the Fragile X Society, both to the survey, and the one-to-one interviews. In total, we received back 323 surveys and we undertook 17 in-depth interviews with 19 people living with different Fragile X associated experiences, which we used both to help us design, but also interpret the survey findings.

This report will give you a brief overview of the data.

Survey Participants

We had a range of people respond to the Fragile X Screening Survey (UK). Table 1 shows that the majority of responders were female (82%), aged 46 or over (73%), with a bit of a split between whether participants were religious (53%) or not (43%). The most common religious affiliation was Christianity.

In terms of relationship to Fragile X:

- 20 (6%) participants reported having full mutation FXS
- 30 (9%) reported being diagnosed with Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS) or Fragile X Associated Premature Ovarian Insufficiency (FXPOI)
- 273 (85%) took part either because they were a pre-mutation carrier and/or they had affected family members.

The vast majority of these responders were parents of a person affected by FXS (75%), with the largest group of these parents (86%) having (at least) one male child affected by FXS.

Interview Participants

The 19 in-depth interview participants were also majority female (15 participants, 79%). Ages ranged from 17-75, but the average age of participants was 48, reflecting a similar demographic to survey participants. The split between those diagnosed with a Fragile X related condition was also very similar to survey participants:

- 3 (16%) participants having a diagnosis themselves: two were diagnosed with FXS (1 male, 1 female) and one was diagnosed with FXTAS (female).
- The other 16 participants (84%) were relatives/pre-mutation carriers. Of these relatives/carriers,
    - 11 were parents of a child with FXS (69%), with 4 of these parents having more than one child with FXS (36% of total parents).
    - A further 4 avoided having another child with FXS through using pre-implantation genetic diagnosis (1), prenatal diagnosis (2) and the use of a donor egg (1).
    - The final 3 parents with one affected child chose not to have further children, or had already completed their family by the time their child was diagnosed with FXS.
    - The remainder of the relatives included in the study were the grandparents (2), cousins (2) and spouse (1) of people diagnosed with a Fragile X-associated condition.

Results of the Research

Living with Fragile X

Participants in both the survey and interviews were asked about their views on what the experience of living with Fragile X was like, including how they viewed quality of life and support from wider society. Overall, 77% of survey respondents thought that people with FXS can have a happy and fulfilling life (35% of whom strongly agreed), but nevertheless 56% thought that FXS causes people to suffer. The interview data illustrates how these views could sit together in the way that the condition was experienced:

“I mean I think my son [with Fragile X syndrome] enjoys life. He's a very happy child, but obviously at time he's very upset when he doesn't understanding something, or he's doing like aggressive behaviour or naughty behaviour and of course you have to tell him off. And he gets very upset by that, he takes it very, very personally. He has a lot of confusion and frustration which must be very hard for him. but I think he has… as a child I think he has a very good quality of life but when he becomes an adult I’m not sure how that will be”

-(mother of male child with Fragile X syndrome)

Key theme: Lack of support and recognition

One of the key points that came out strongly in both the interview and surveys was the lack of support from wider society for families and individuals living with Fragile X associated conditions. Only 26% of the 323 survey participants thought that families were well supported by society and 73% thought that most difficulties associated with Fragile X related conditions could be overcome, or at least minimised, with better social care and resource provision (31% strongly agreed).

"you have to fight for everything"

-(mother of two children with Fragile X syndrome)

Some of the interview participants also mentioned that the relative invisibility of Fragile X syndrome and related conditions could create barriers to support and understanding,

"I think the moment you see someone with a wheelchair or some sort of physical disability you're instantly more generous with your time, with your thought process, etc, but you just see an ordinary looking person on the street, for example, but they are like looking around, looking a bit dazed and confused, people wouldn't think to help or offer… you know, or worry about them. Do you know what I mean? it's that kind of thing. It's the physical versus the mental I guess and people respond to what they can see in front of them. If they can't immediately see it, they question if it's even there at all"

-(mother of an adult male with Fragile X syndrome)

"This Fragile X thing, I don't know how much that makes them stand out. I know he comes home from school [son with pre-mutation] and he says some of the kids at school think I'm weird, which makes me think, well maybe it's more obvious than we think it is. But I don't know if you were to look at him that you'd say that he's got something ….So people know what someone with Downs looks like, but we don't see anyone with Fragile X in the media. No one speaks about it that I'm aware of, so as far as… we're in this little world there's only us that have it and can you explain to you what it's like. So the result of that is that he doesn't get supported with it because no one can see it, and even if it's pointed out they don't know what it means"

-(mother of teenage son with pre-mutation)

"What upsets me is people just put it down to naughty behaviour, bad children, you know if he's having a meltdown in the supermarket and he's lashing out, being aggressive and people stare. I've stopped talking about Fragile X now, I just say it's autism as I think we've come some way with the understanding about autism. But with most people I won't even get that far, they'll just tut or stare and assume I am the worst parent in the world with the most awful child in the world, and quite frankly it has stopped me going out sometimes."

(mother of young male child with Fragile X syndrome)

"…..it's society that perhaps may need to change a little bit more, not our children. You know, if society does change sometimes a little bit differently our children wouldn't have quite so much problems, you know, we wouldn't as a family have had quite so much problems, but that's probably actually asking for too much really isn't it?"

(mother of two daughters with Fragile X syndrome)

Fragile X Community's Views on Screening

Pre-conception screening

Both survey and interview participants were asked their views on pre-conception genetic screening, a form of genetic screening whereby all members of the population would be screened to find out if they are carriers of the Fragile X pre-mutation, in order to inform future reproductive decisions.

Overall, 82% of survey respondents supported the idea of screening the population before reproduction occurs, with the most common reasons for supporting such a programme being that:

1. Increase awareness of Fragile X in society, enabling more informed reproductive decisions (87% agreed, 52% strongly agreed).
2. It would alert pre-mutation carriers of future health risks (87% agreed).

Despite these benefits of pre-conception genetic screening, however, some of the more ethically complex aspects of this type of screening were also seen as problems by survey participants.

For example, 60% of participants, despite supporting pre-conception screening as way to increase informed decision-making and awareness, nevertheless felt that people who do not have direct experience of FXS in their family- who are identified by screening- wouldn't be able to make a truly informed decision about having a child with FXS without having experienced it or lived with it themselves.

This view also came up in the interviews. Whilst pre-conception screening was widely supported, and seen by some participants as *'necessary'*, others raised the concern that people would be forced to make decisions with that information that they may be ill-equipped to tackle,

"What I would say is that nobody knows what it is like to bring up a child with Fragile X, Down Syndrome, you know, Tourette's Syndrome… unless you're doing it. I don't know how they're going to be able to make those decisions"

(mother of adult male with Fragile X syndrome)

25% (1 in 4) survey participants also thought that pre-conception genetic screening is a form of social engineering, which was also discussed frequently within the interviews,

"My concern is that we are going down the route of having the perfect child, you know, with all the genetic complications, so I think we are. If screening is passed, in a few years' time will we be saying I want child with blue eyes, blonde hair that looks like a model, or whatever… And I think that's not fair, I think we're interfering too much. And the world is nice because of the variety of things and the world is better because of its variety. And if we start off down that route I think we can create more problems."

(mother of teenage son with Fragile X syndrome)

Prenatal Screening

Prenatal screening would involve pregnant women finding out if they are premutation carriers during pregnancy, and potentially also having the foetus tested, if they are found to be a carrier. Prenatal screening emerged as the most controversial form of genetic screening amongst survey and interview participants because it is aligned with pregnancy termination.

In spite of reservations, however, 79% of survey participants responded that they would support the introduction for such a programme for pregnant women. The strongest reason for supporting prenatal screening was the idea that it would allow pregnant women to make informed decisions about whether or not they want to have a child affected by Fragile X associated conditions (81% agreement) and 43% thought termination of pregnancies is acceptable for conditions like FXS, for which no targeted treatments or 'cure' exists (24% disagreed).

Interestingly, the slight majority (51%) thought public attitudes to people living with FXS and their families will be affected if FXS becomes a routinely scanned for condition, and just under half (48%) thought that pregnant women who find out for the first time that they are a premutation carrier are more likely to terminate a pregnancy affected by FXS than someone coming from an already affected family.

The difference in perspective that living with Fragile X associated conditions gives families was also reflected on by the interview participants,

"If I was pregnant and I found out yeah umm I'm not sure I would have gone through with it [pregnancy]. Which sounds awful to say, and I'm glad I didn't know really as I would have said 'that's absolutely something I can't cope with', and I would have missed out on my [son]. Even saying that out loud gives me goose bumps….but, you don't know what you'll miss out on at that point, and you can easily convince yourself you're doing the right thing because when you read about all the problems they have, yes it sounds terrifying. You think they'll have a terrible life, you think YOU'LL have a terrible life, as a parent…and that hasn't been the case"

(Mother of young male child with Fragile X Syndrome)

For other participants, however, the ability to choose overrode the potential increase in Fragile X associated pregnancy terminations that could occur alongside a prenatal screening programme,

"*If you can give people the choice, then that should definitely be available. And people that preach about the sanctity of life and such things are either… I mean they're usually people that have no experience of exactly what it's like. I think anyone who knows the reality of what it's like would never judge a person for saying 'no thanks' and I absolutely would have done if I'd been given that choice back in the day. Although I wouldn't be without [son] now, my life could have gone an easier path and we would have all suffered less as a family…. and I…I just think we should let people choose that easier path if they aren't cut out for this one" (mother of adult male with Fragile X syndrome)*

"I just think there are very few people in this world who are genuinely able and willing to bring up a disabled child. I'm not saying it's not better for the nation as a whole to have people with Fragile X in it because [name] is a great life enhancer, there's no, you know… I think the affected individuals (people) in the end can add greatly to life's rich tapestry, but I don't think that bringing them up it is something that most people find easy can do and with a few people, you know, the child is at risk of harm. So choice is so important."

(Father of daughter with Fragile X syndrome)

Survey participants were somewhat split on the question of whether it would be a loss or a benefit to society to have less people with Fragile X associated conditions coming into the world. 31% thought it would be a loss to society to have fewer people with FXS coming into the world, 30% disagreed and the remainder of participants were unsure. Part of the ambivalence also came out in the interviews with some participants wanting to separate out the condition from the person,

"of course I would think it was a loss if my son was not here, it would be a loss to society, but his Fragile X syndrome you can get rid of thanks"

(mother of young male child with Fragile X syndrome)

Whereas other interview participants had clearer views in opposite ways:

"Surely it can only be a benefit to have less people coming into the world with this condition? Forget about society, it's a huge burden for that person individually, so I can't see any problem with getting rid of it"

(Uncle of 4 children with Fragile X children)

"….the emotional and human side of me says it would be a real loss because we actually become more human when we develop caring relationships and when we

learn to actually give and take. And we would lose something as a society if we didn't actually have that caring role within us. So I think it's probably a loss."

(mother of young male child with Fragile X syndrome)

Newborn screening

80% of survey participants were supportive of newborn screening for Fragile X associated conditions. Key reasons for this support were a belief that it would enable parents to make informed decisions about subsequent pregnancies (91% agreement), would lead to better health and social care for the family (85% agreement), would spare the family the difficulties and delays with obtaining a diagnosis later on (83% agreement). A further 77% of survey participants thought that newborn screening is also a good way to identify pre-mutation carriers in the general population, which could be used to inform their future health and reproductive decisions.

There was strong evidence from the interviews that delays getting a diagnosis were common, and the four parents who had multiple children with Fragile X syndrome had all had their second child before their first child was diagnosed. A further parent had gone on to have another child before the diagnosis of their first, but their second child was unaffected. Participants mentioned lack of awareness of Fragile X conditions amongst health care professionals as a key barrier to an earlier diagnosis. This was particularly the case for girls with FXS whose symptoms may be less pronounced than their male counterparts.

The key concerns about newborn screening that came out of the interviews were the effects it might have on early bonding between infant and parent (although only 12% of survey participants agreed this was a concern) and also that it might rob a family of a care-free time when the child is still symptom free (only 15% of survey participants agreed this was an issue).

"…finding out when you've just had the baby, you're in quite a vulnerable space anyway, it could really scare you up at a time that really should be focused on bonding. I don't know, but I could see how it may cause problems if you think you've got this perfect baby and they're saying 'well actually he has all these problems and he got it from you', it might taint the happiness and the whole experience. I'd rather leave that time alone."

(mother of young male child with Fragile X syndrome)

" To be honest, I'm so glad that I didn't have the diagnosis when he was first born because … he wasn't treated differently, you know, he was terrible in the supermarket as a baby but we had to go because there was no other way, you know, he had to sit there in the trolley and that's it. So yeah, we had a period of time when we were just a normal family, and to me that's priceless really."

(mother of young male child with Fragile X syndrome)

However, for other participants a late diagnosis could be a destructive experience for the family,

"It was actually the 'non-diagnosis' that caused all of the problems for us, because we [parents] started blaming each other for everything and how he was behaving, and we were accusing each other of not doing enough with him, of not trying hard enough, knowing earlier could have saved us from that really stressful time"

(mother of young male child with Fragile X syndrome)

Summary and conclusions

As technologies relating to genetic testing are developing and becoming ever more available, it is essential that the views of individuals and families living with genetic conditions are taken into account.

Attitudes to screening are complex. In this study, support was shown for all forms of screening (prenatal screening least popular), but participants simultaneously had reservations about screening programmes, particularly around how information about Fragile X conditions would be communicated to the public, the ignorance of the general public about Fragile X conditions, and the potential reduction in people being born with Fragile X associated conditions who would have a lot to contribute to society. Many participants resolved this conflict by distinguishing between what they thought should be available for the general public, and what they would personally do themselves if faced with the reproductive dilemmas that can be associated with screening programmes.

Of note, the present article focussed upon the views of parents, though it will be essential to listen to the voices and experiences of people with Fragile X Syndrome (full-mutation) to ensure their views are represented in the planning around screening, much like the increasing self-advocacy seen in the Down Syndrome community. Should screening be offered in the future, it will be essential to ensure that comprehensive information is available to those undergoing screening which includes the views of people with Fragile X Syndrome and their families, as opposed to purely medical information. Education and understanding of Fragile X is key, as is understanding of learning disability.

Note: Views expressed in the article should not be taken to represent the views of the Fragile X Society.