Exploring the experiences of living at risk of familial frontotemporal dementia

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Background:

Frontotemporal dementia (FTD) is the second most common form of dementia in those under 65 years of age. There is a genetic cause found in around 30% of cases and these follow an autosomal dominant pattern of inheritance meaning that the offspring of a mutation carrier has a 50% chance of carrying the mutation themselves. Those 'at risk' of FTD have the option of undergoing predictive testing to learn their genetic status however the majority choose not to find out their status.

There is a considerable amount of uncertainty attached to being at-risk of familial FTD including an unpredictable age at symptom onset and phenotypic heterogeneity. Therefore literature from other familial hereditary diseases cannot necessarily be extrapolated. The symptoms associated with FTD and related diseases such as MND, as well as the high penetrance of the most common FTD-causing genes may lead to psychological distress in those living at-risk. However, there is limited research into the effects of living at-risk, largely focusing on those who choose to undergo predictive testing, with no studies to date involving participants who choose not to know their genetic status.

The aim of this study was to explore the lived experience of those at-risk using a qualitative approach in order to identify target themes for future psychological intervention.

Methods:

Sixteen qualitative interviews were analysed using an inductive approach to thematic analysis. Participants were asked about their experience finding out about their risk, how they felt throughout different parts of their journey, their experience of genetic testing if this was performed, as well as the support they had or feel they would have benefited from.

Results:

Five themes were generated covering multiple facets of the at-risk experience. Emotionally, participants reported experiencing low mood and anxiety, underpinned by isolation, helplessness and uncertainty. The theme 'End of life' explores participant's views on assisted dying and seeing FTD as a terminal diagnosis. Talking to people who understand (including both peers and professionals) was commonly referenced as an integral source of support, with participants often wanting more peer support. However, a lack of understanding in both the general public and non-FTD specialist healthcare professionals was a barrier to accessing support for many people.

Conclusions:

Living at risk of familial FTD is a psychologically challenging and complex experience, independent of the challenges presented by predictive testing. For some individuals, more specific and target support may be required. Support facilitated by both peer 'experts by experience' and FTD-specialists may be particularly useful to those in need. Future research should explore tailored support strategies to assist people in coping with these challenges.

"I could convince myself that I saw early signs of it in myself. So when [I] forgot a word or, you know, left the door open, you would attribute it to that. …And that that actually got me very down at the time."

"There's this paralysis that sets in, I think, when you get the, it's not so much the diagnosis it's the fear of the diagnosis. And suddenly, like your life…hits this fork where there's this kind of terrible stark future."

Theme 1: Anxiety

- Fear was most commonly described in relation to risk, being a gene carrier, the FTD diagnosis and death.
- Participants worried about their risk, potentially having passed the gene to their children, their sibling and also their genetic result
- The onset of symptoms was also a source of worry, participants noticed that when they experienced normal lapses in memory and occasional tripping, they worried that they were experiencing early signs of symptom onset, however this tended to be momentary and mild.
- There is a large amount of uncertainty associated with being at-risk as FTD is unpredictable in terms of heterogeneity of symptoms, age of symptom onset and disease duration. This unpredictability meant that people did not know what to expect and struggled to plan and prepare for the future. The effect of uncertainty was significant for some people who decided that they would rather know their genetic status than live with the uncertainty of being at-risk, this tended to bring relief in these cases regardless of a negative or positive result. Others coped with this uncertainty by planning as though they were a gene carrier, planning things like future care, finances, insurance and making funeral arrangements.

"it's all…unknown. It's the uncertainty that a) in two years time, I could potentially be facing early onset dementia. Or I know at sixty three, I'm going to get diagnosed with motor neurone disease."

"it is just a kind of, it's a terminal diagnosis 8 years before you'll die, kind of thing. And that's pretty horrendous, I think, to live with"

Theme 4: End of life

- Participants talked about viewing a positive genetic result as a 'terminal diagnosis' or 'life-threatening news' and many reported living with the assumption that their life would end around the a similar age as their affected parent.
- A small number of participants reported that they would like to have the option of assisted dying when they were to develop symptoms, however understood that this would be an unlikely possibility due to the need for capacity to consent.

"I made up my mind that I would go, if I did get motor neurone disease that I would be going to Dignitas. So that's a kind of, a comfort to know that, you know, there is a way out…well, the difficulty is it's all about timing, isn't it? Because if I got FTD, I probably wouldn't do that because, you know, you have to be of kind of sound mind to be able to make these decisions."

"Just having someone there to talk to really. That's the best support, is someone, someone who understands it"

"actually discovering that this nice lady next to you was saying the same terrible, terrible things made you realise that you were part of some bigger process, that it was a kind of shared thing and…you weren't sort of alone in this whole process. That you weren't, that, you know, other people had got through it and survived, as it were"

"I did point out to [GP], I said, do you know about this disease? And [GP] said, no. I said, do you think the counsellor would? And [GP] said, I can't say yes or no, but possibly not. And I said, I don't want to have to explain what it is, to deal with the issue."

Theme 2: Talking to people who understand

- A general lack of support was reported. However, those who received support found support from 'someone who understands' most useful. This included peer support, support from family members and partners and also support from specialist academics and healthcare professionals.
- This source of support was helpful in normalising and validating their experience, as well as giving people a safe space to be frank about the difficulties they faced.
- A lack of understanding of FTD and genetic risk was a major barrier to support as the idea of needing to educate people prior to receiving support prevented them from pursuing usual support pathways such as speaking to a GP, counsellor or psychologist.
- Some of those who utilised local psychological services found it of limited use as counsellors lacked understanding of the specific problems they faced.
- Cognitive behavioural therapy (CBT) was of some use to those who pursued it, however it was reported that participants and their counsellors noted that some aspects of the model were not appropriate for their circumstance.
- Participants also commonly expressed a desire for more peer support.

"I do think some sort of a specialised one to one therapy with somebody that has a background knowledge, or is willing to have and kind of learn a background knowledge on it would be helpful"

"Meeting other people definitely in the same position, I think it would definitely help because you wouldn't feel so alone or so unique and like completely unique. And to discuss your experiences with those people. I think it's ultimately really good for both of you then, because you can really support each other "

"there was a a period … where I would would have said I went through a period of depression and I did go to doctors for that and you know, had had some medication to to help to help with that…but it was a fairly sort of, you know, miserable time. And it tended to occupy a lot of my, a lot of my kind of, you know, thoughts."

"I spent a number of days just sitting there crying"

Theme 3: Low mood

- General symptoms of lowered mood following disclosure of their risk including sadness, frequent tearfulness and in some cases depression, resulting in psychological and/or pharmaceutical intervention.
- A feeling of helplessness as there is nothing that they can do to prevent onset of the disease. Individuals also did not plan for later in life due to this pessimistic view of the future.
- Isolation was also reported due to the uniqueness of their experience and the lack of understanding outside those who were in a similar position. Participants reported being left to cope alone with their problems, however acknowledging this shared unique experience with others in a similar position helped in alleviating the feeling of isolation.

"it's just what I've got to face and I can't do anything about it"

"I felt like it was something that I had to do on my own and no one else can understand"

"it's almost like you're left out at sea."

"I didn't want to have to sit there and explain to someone the ins and outs of everything."

"I think there's things that I appreciate now that I might have had the same life and I don't think I would have appreciated them the same way"

Theme 5: Time is precious

- Doing things sooner rather than later and living life now was commonly mentioned as participants wanted to make most of their life while they are healthy.
- Some participants also talked about viewing their risk or genetic status as a positive as it encouraged them to live the life that they want to live, take opportunities, have a good work-life balance and appreciate the life that they have had.

"it does make you live for today, because tomorrow is not guaranteed."

"the life which I lead now is …pretty much my dream life in many ways….if you knew the date you were going to die, you'd want to squeeze in and squeeze as much juice out of life as possible."

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