Received: 22 December 2021 | Revised: 1 May 2022 | Accepted: 7 May 2022

DOI: 10.1002/jgc4.1591

ORIGINAL ARTICLE

National Society of Genetic Counselors WILEY

Genetic counseling and diagnostic genetic testing for familial amyotrophic lateral sclerosis and/or frontotemporal dementia: A qualitative study of client experiences

Ashley Crook[1,2] | Chris Jacobs[1] | Toby Newton-John[1] | Alison McEwen[1]

[1]Graduate School of Health, University of Technology Sydney, Chippendale, New South Wales, Australia

[2]Faculty of Medicine, Health and Human Sciences, Centre for MND Research, Macquarie University, Sydney, New South Wales, Australia

Correspondence
Ashley Crook, Graduate School of Health, University of Technology Sydney, Chippendale, NSW, Australia.
Email: ashley.k.crook@student.uts.edu.au

Abstract

Genetic counseling and diagnostic genetic testing is part of the multidisciplinary care of people with amyotrophic lateral sclerosis (ALS, commonly called motor neurone disease, MND) and frontotemporal dementia (FTD). We explored client experiences of genetic counseling and diagnostic testing to inform the care of future families. Semi-structured interviews with individuals with ALS/MND/FTD or their relatives were conducted. The study was designed to include a wide variety of participants with varying disease status and abilities. Genetic counseling and diagnostic testing experiences were explored using interpretive description methodology. Bioecological theory was used as the framework for the reflexive thematic analysis. Eighteen individuals with ALS/MND/FTD or their relatives from 13 Australian families participated. Three themes were identified: sharing knowledge, (un)supportive care, and 'circumstance is everything'. Consistent with bioecological theory, one's genetic counseling experience was informed by individual circumstances, time, and proximal factors. These informed the level of information and support required in the genetic counseling process. Although some client circumstances cannot be changed, efforts could be made to enhance genetic counseling experiences by improving interactions between the client and their care team. Some clients may benefit from further discussions regarding the familial implications of genetic testing, and greater support with family communication. Clients' needs were derived from the data and will contribute to genetic counseling consensus guidelines.

KEYWORDS
amyotrophic lateral sclerosis, communication, frontotemporal dementia, genetic counseling, genetic testing, motor neurone disease

1 | INTRODUCTION

Amyotrophic lateral sclerosis (ALS, commonly termed motor neurone disease or MND in Australia) and frontotemporal dementia (FTD) are adult-onset, progressive, and neurodegenerative conditions that can be genetically linked. Diagnostic genetic testing, and the genetic counseling that accompanies it, is part of the multidisciplinary care of ALS/MND/FTD patients (Roggenbuck & Fong, 2020;

Turner et al., 2017). We explored client experiences of genetic counseling and diagnostic testing to inform the care of future families and the development of genetic counseling consensus guidelines.

Around 20% of ALS/MND/FTD patients will have an underlying pathogenic variant associated with familial disease identified on diagnostic testing (Blauwendraat et al., 2018; Shepheard et al., 2021). As a genetic etiology can be identified in both familial and apparently sporadic ALS/MND/FTD (Blauwendraat et al., 2018; Fostinelli et al., 2018; Shepheard et al., 2021), all individuals with ALS/MND/FTD should be offered genetic testing (and genetic counseling), irrespective of whether a suggestive family history is present (Roggenbuck & Fong, 2020; Turner et al., 2017). The genetic testing performed in this setting is an initial search for pathogenic variants to determine whether a molecular diagnosis can be made. It is commonly called diagnostic testing and may occur as part of the clinical diagnosis of ALS/MND/FTD in an individual, or it may occur later, when a clinical diagnosis has already been made.

Despite the above recommendations, diagnostic testing is not consistently offered, and health professionals may have differing views to patients regarding the value of testing (Klepek et al., 2019). Currently, life-prolonging therapeutic options are limited, and no therapies can prevent disease onset, but treatment trials that target individuals with specific underlying pathogenic variants in ALS/MND/FTD genes are emerging (Miller et al., 2020). Interest in genetic testing is expected to increase based on this availability (Benatar et al., 2006). In addition, if a pathogenic variant is identified, predictive and reproductive genetic testing may become available to biological relatives.

There are currently no consistent, evidence-based genetic counseling approaches to diagnostic testing of individuals with ALS/MND/FTD and their relatives, and few studies on this topic have been performed to date (Crook et al., 2021). As consumer input is key to ensuring that health care services best meet the needs of its clients (Santana et al., 2018), we sought to fill this research gap by exploring client experiences of diagnostic genetic testing and counseling in Australia and gaining suggestions to inform the care of future families. The Process-Person-Context-Time (PPCT) model of Bronfenbrenner's bioecological theory (Bronfenbrenner, 2001; Bronfenbrenner & Evans, 2000) guided our analysis.

What is known about this topic

There are inconsistent approaches to genetic counseling and diagnostic testing for individuals with ALS and/or FTD. Few studies have investigated the client experience of genetic counseling and diagnostic testing in ALS and FTD, which can inform future service delivery.

What this paper adds to the topic

Individual client and family circumstances at the time of genetic counseling and testing informed the level of information and support required and attitudes and responses toward genetic counseling and testing. Client experiences of genetic counseling and testing could be enhanced by improving their interactions with health professionals, support associations, and the wider family.

2 | METHODS

2.1 | Study design

The conduct, design, and reporting of this study follow the Consolidated Criteria for Reporting Qualitative Studies (COREQ) checklist (O'Brien et al., 2014; Tong et al., 2007) (Appendix S1). We adopted a pragmatic stance and used interpretive description to guide research design (Thorne, 2016).

The University of Technology Sydney (UTS) Medical Research Ethics Committee approved this study. Recruitment and data collection were conducted by the primary author, a female PhD student and genetic counselor with experience in qualitative research and working with ALS/MND/FTD families.

2.2 | Participants

We sought individuals to participate in a one-off, in-depth, semi-structured interview about their experience of diagnostic genetic testing and counseling for ALS/MND/FTD. Participants were eligible if they had previously been involved in some aspect of the genetic counseling discussion regarding diagnostic testing (regardless of whether they proceeded with testing). As patients may have cognitive or communication impairment due to disease progression, they may have unique needs or not be involved in the genetic counseling discussion at all. Therefore, clients/participants included patients, family members, and carers. Additionally, all participants were required to reside in Australia, be aged over 18, understand the purpose of the study, and provide informed consent in English.

2.3 | Procedures

Specific strategies were implemented to ensure a sensitive and safe research design given that the research topic may be upsetting (Liamputtong, 2007), and individuals living with ALS/MND/FTD may be a vulnerable population (highly dependent on medical care and/or with a cognitive or communication impairment) (National Health and Medical Research Council, 2018).

The recruitment strategy was designed to require participants to make the initial contact; then, consent after time and further information about the study had been provided so that their choice to participate was autonomous (Appendix S2). If no response was received after the initial recruitment contact or after the PICF was

emailed, participants were followed up by email once. By providing multiple interview and consent options, we expected that some participants would be better supported to share their experience jointly with someone else and/or to feel more confident communicating in their preferred way. A combination of interview types has previously been used in qualitative research for people with other medical conditions or when discussing a sensitive topic (Heath et al., 2018; Liamputtong, 2007; Morris, 2001).

Interviews were carried out between October 2020 and April 2021. An interview guide included closed questions about participant demographics, open-ended questions, and further probing questions to clarify or obtain further information about the research topic. Additional closed questions were included to ensure a format suitable for participants with communication and/or cognitive impairments (Appendix S3). The guide was pilot-tested before use. A distress protocol was developed (Appendix S4), informed by previously published distress protocols (Draucker et al., 2009), national guidelines (National Health and Medical Research Council, 2018), and research team consultation.

A professional transcription agency transcribed the audio-recorded interviews. The primary author verified and de-identified all the transcripts, replacing names with pseudonyms. The transcripts were not returned to participants for comment or correction.

Two additional strategies were used to ensure participants had the capacity to make an informed decision to participate. When providing consent, all participants indicated if they had an enduring medical power of attorney or guardian who made medical decisions for them. If they answered yes, additional consent from this person was required before proceeding with the study. Secondly, at the start of the interview, all participants were asked four yes/no questions about the study based on information provided in the PICF (e.g., Does this study involve a blood test? Will this study involve you answering some questions?) (Appendix S3). This strategy has been used in a recent study on stroke survivors (McGrath et al., 2019). The interview would not proceed if any answers were incorrect.

2.4 | Data analysis

Data collection and analysis occurred concurrently, per the interpretive description approach (Thorne, 2016). Data analysis was inductive, following the reflexive thematic analysis method (Braun & Clarke, 2006, 2019). The data were managed using NVivo 12. Two authors (AC and AMc) independently reviewed and coded all transcripts. All authors met regularly to discuss, reflect, and agree on emerging themes. Differences were resolved through discussion. Subgroup differences (e.g., individuals from familial vs sporadic, ALS/MND only vs ALS/MND/FTD families, individuals from the same or different families, and clinical status of the participant) were explored throughout the analysis.

2.4.1 | Bioecological theory

Once generated, all themes were mapped to the PPCT model of Bronfenbrenner's bioecological theory (Bronfenbrenner, 2001; Bronfenbrenner & Evans, 2000; Rosa & Tudge, 2013). Versions of Bronfenbrenner's theory have been widely used to study various social phenomena (Rosa & Tudge, 2013), including genetic counseling experiences (Hoskins & Werner-Lin, 2013; Tillerås et al., 2020). Bioecological theory provides a framework for understanding how the four PPCT elements simultaneously shape one's genetic counseling and testing experience (Bronfenbrenner & Evans, 2000; Rosa & Tudge, 2013). Proximal *processes* are at the center and include reciprocal interactions between the individual and other people, objects, and symbols (Bronfenbrenner & Evans, 2000). Individual *person* characteristics are next, followed by *context*. Context involves four inter-related, nested systems in one's environment: the microsystem (where proximal processes occur), mesosystem (interrelations among microsystems), exosystem (external systems that indirectly influence), and macrosystem (the norms and values of a particular culture) (Bronfenbrenner & Evans, 2000). Proximal processes are influenced by both person characteristics and the context in which they occur. Time includes the length and frequency of time during which the individual has been exposed to a particular process or context and includes time periods that are immediate or across the lifespan (Bronfenbrenner & Evans, 2000). As this was not a longitudinal study, the time component could not be thoroughly assessed. Still, time across the life stage undoubtedly informed experiences, and this is detailed in the results.

2.5 | Trustworthiness and validity

Several steps were taken to ensure trustworthiness of the study findings (Lincoln & Guba, 1985). The primary author's previous clinical and research experience and disciplinary orientation (in genetic counseling) are acknowledged as the research design instrument and demonstrate their credibility in this qualitative inquiry. The primary author wrote reflexive field notes electronically throughout the study to reflect on the data collection and analysis process and ensure rigor and transparency. A record of the research steps undertaken was also kept, and regular team meetings allowed for peer debriefing. To ensure confirmability, two investigators independently conducted the analysis, which is a form of analytic triangulation. Data triangulation was also demonstrated as participants included different clients from within and between families.

Using information power (i.e., the more relevant information a sample holds to answer the research question, the fewer participants are required), we took into account the aim of the study, sample specificity, the use of established theory, quality of dialogues, and analysis strategy to guide an adequate sample size (Malterud et al., 2016). We agreed that the complete data set, which includes accounts from 18 participants, was sufficient to address our study question.

TABLE 1 Participant and family characteristics

Participant characteristics	Number of participants (18)
Sex	
Female	13
Male	5
State	
New South Wales	7
Victoria	5
Queensland	4
Western Australia	2
Highest level of education reached	
Did not complete high school	2
High school certificate	4
Trade or associate diploma	3
Bachelor degree	3
Post-graduate	6
Children	
Does not have children	3
Has children	15
Children, age < 18[a]	8
Children, age > 18[a]	7
Adopted or stepchildren, age > 18[a]	2
Participant status	
Personal history of ALS/MND	3
Personal history of suspected FTD[b]	1
At-risk relative, predictive testing: result unknown[c]	6
At-risk relative, predictive testing: PV inherited	3
At-risk relative, predictive testing: PV not inherited	1
Relative of a person with ALS/MND/FTD, unknown if at risk	1
Spouse of a person with ALS/MND/FTD	3
Family characteristics	**Number of families (13)**
Additional Family history[d]	
ALS/MND only	7
FTD only	0
ALS/MND and FTD	5
No additional family history	1
Testing status	
Diagnostic testing proceeded, PV detected:	11
C9orf72	7
SOD1	2
TARDBP	1
Could not recall	1
Diagnostic testing proceeded, PV not identified	1
Diagnostic testing declined	1
Primary health professional who facilitated diagnostic testing	
Neurologist	7
Genetic counselor	2
Clinical geneticist	1
Social worker	1
Could not recall	2

Abbreviation: PV, pathogenic variant.

[a]Some participants fulfilled multiple categories.

[b]Participant was awaiting further investigation to confirm the diagnosis.

[c]Result either pending, or participant was untested.

[d]Family history is in addition to the person who underwent diagnostic testing. For spouses, the family history listed includes the spouse's relatives.

3 | RESULTS

3.1 | Participant characteristics

Thirty-three individuals expressed interest in the study. Eight did not respond, and seven were deemed ineligible (six had not experienced diagnostic testing discussions; one was outside Australia). The final sample consisted of 18 participants (mean age 39.44, range 19–58) from 13 different families (Table 1). Two conjoint interviews were conducted, resulting in 16 interview transcripts (nine Zoom video, six telephone, and one email interviews). The mean length of the 15 audio interviews was 59 min (range 23–105). Two interviews occurred over multiple days (one email exchange and one Zoom interview that had to be rescheduled). Experiences occurred as early as the early 1990s for two families, but the majority experienced diagnostic testing discussions more recently, between 2010 and 2020. The primary author had no previous clinical or research relationships with any study participants. There were no apparent differences in participant responses based on the demographic and clinical variables (e.g., location, clinical, and disease characteristics of the participant and family, whether they had children or the health professional type who facilitated testing).

Experiences of genetic counseling and diagnostic testing were informed by both interactions with others, and individual characteristics, including previous and current experiences, attitudes, and beliefs. Two key aspects of genetic counseling were identified, occurring either concurrently or independently: the genetic testing process and managing knowledge within the family. Three themes were formulated and mapped to Bronfenbrenner's PPCT model of bioecological theory (Bronfenbrenner & Evans, 2000): *sharing knowledge*, *(un)supportive care*, and 'circumstance is everything' (Figure 1). The themes are further summarized in Table 2 and the text below.

3.2 | Sharing knowledge

3.2.1 | Interactions with health professionals and support associations

Efforts by some health professionals to provide adequate information led clients to feel well informed throughout the genetic counseling process (Quote 1, Table 2). For others, information from health professionals about the availability of testing and counseling was lacking, even in the presence of a family history suggestive of familial disease. In terms of sharing the knowledge about genetic testing and associated implications within the family, some health professionals attempted to help (particularly when there were family-related barriers to communication). Still, information regarding genetic testing pathways was not always clear (Quote 2, Table 2). Other health professionals created barriers for relatives wishing to access more information:

> *I could not go ahead with the genetic testing here in [home state] without proof that it was in my family. I rang [Mum's neurologist]…He said "I'll only release [results] to a medical professional"…He wanted us all to come in…to do the counseling altogether…[but] everyone [in the family] has got opposing views on it all.*
>
> Alanna, mother: ALS/MND and FTD (gene positive).

Some clients became interested in testing after accessing online information from support associations. Some gaps in the information available online or through support associations led to difficulties for relatives wishing to access genetic counseling and predictive testing (Quote 3, Table 2).

Minimal pre- and post-test genetic counseling were provided to some. Consequently, some felt they did not make an informed decision regarding undergoing testing:

> *[The clinical team] didn't seem to be very interested [in discussing genetic testing]…I would have liked them to have helped with letting us know what things mean and the pros and cons.*
>
> Patricia, husband: ALS/MND (gene negative)

Pre-test genetic counseling discussions were deemed more relevant for some than others:

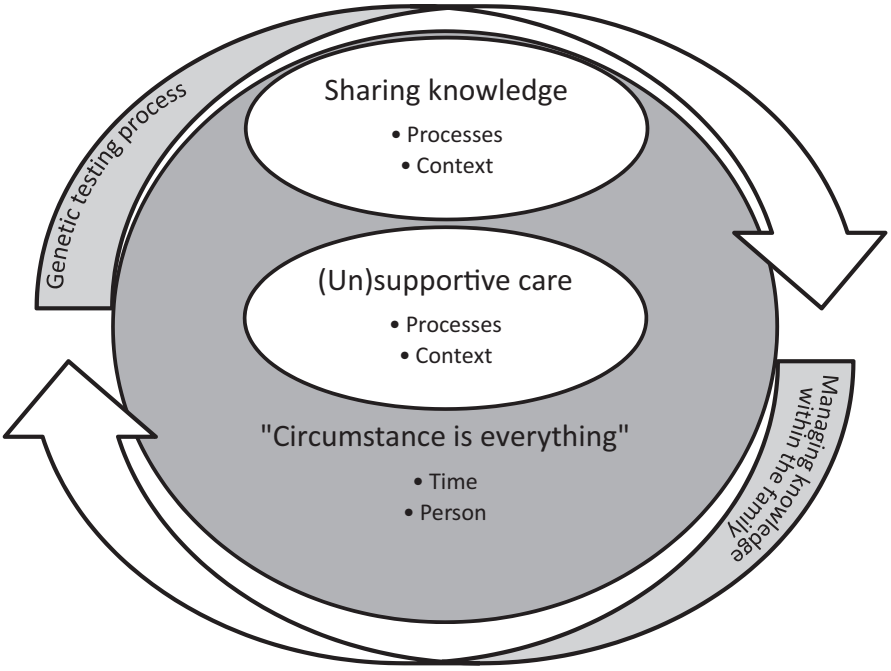

FIGURE 1 Visual outline of findings. Three themes were formulated and mapped to Bronfenbrenner's PPCT model of bioecological theory (Bronfenbrenner & Evans, 2000). The first two themes, *sharing knowledge* and *(un)supportive care*, encompass two key genetic counseling goals: Providing information and support. In both of these themes, interactions with others were at the forefront and could be either barriers or facilitators to a positive genetic counseling experience. These interactions occurred between the client and members of the family (e.g., carers and relatives) and those external to the family (e.g., health professionals, peers, and support associations). Underpinning these two themes was the third theme, 'circumstance is everything'. Circumstances were unique to each individual and influenced the level of information and support required and provided. Two key aspects of genetic counseling were identified, occurring either concurrently or independently: The genetic testing process (including decision-making and adjustment after results) and managing knowledge within the family (including family communication about testing and family history)

TABLE 2 Summary of themes

Theme, predominant PPCT categories and description	Subthemes and supporting quotes
Sharing knowledge • Proximal processes • ContextTo make an informed decision about whether to undergo genetic counseling and testing (and subsequently share knowledge within the family), clients needed to be aware of its availability and how to access further information. Clients relied upon information sourced from interacting with others, which served as either facilitators or barriers to informed decision-making. When barriers were present, some clients advocated for themselves to access further knowledge.	**Interactions with health professionals and support associations** 1. *[The genetic counselor was] really knowledgeable…She'd prepared this folder full of… specialists…and psychologists I can see. And information, hotlines and websites…[My General Practitioner/ GP] said he's never received such an informative letter…[The counselor had also drafted a letter to share with my family] to say "hi, I've been tested for this"…that was really useful.* Fabian, FTD suspected. Mother: FTD (untested). Fabian underwent diagnostic testing (gene positive) 2. *I had been contacted by [my father's] doctor when he made the decision to get genetic testing, to be told that it was taking place…[and that] I would have access to the results…They told me that it would be available but not what to do when it came to accessing it…It would be ideal…even if they say, "hey when you're interested, call this number or…contact this email address".* Estelle, father: ALS/MND (gene positive) 3. *The first thing I did when Ryan got his positive genetic test, called [my state MND association]…But there was very little information on…where to go and what to do…[They] gave me…an [interstate] organization who did genetic testing…[I wanted] local info.* Lara, brother Ryan: ALS/MND (gene positive) **Information sharing within the family** 4. *I raised testing. And [my neurologist] agreed readily…[Because of my mother's diagnosis] I was knowledgeable about MND…so I wasn't shocked by [my] diagnosis or in despair. I just wanted info.* Ryan, ALS/MND (gene positive) 5. *I knew my grandmother passed away from MND…But I never thought that it was genetic until Mum got it…My mother and my aunties never told me about…[my great aunt] dying of MND.* Alanna, mother: ALS/MND and FTD (gene positive)
(Un)supportive care • Proximal processes • ContextParticipants reflected on the level of support required and provided throughout the genetic counseling and testing process (support to facilitate adjustment to the information provided and share information within the family). Some interactions with others resulted in access to appropriate personal or professional support. Other interactions were barriers to supportive care.	**Support from the managing team** 6. *She explained everything so calmly and so caring…Our experience with the genetic testing itself…I couldn't fault.* Nina, husband: ALS/MND and FTD (gene positive) 7. *[My mother and I mentioned genetic testing to the neurologist. He]…told me to stop worrying…He didn't even want to discuss it…It wasn't until we actually sat down with [the genetic counselor, who]…reduced the anxiety around it…From [the neurologist]'s perspective, he probably deals with this all day, every day. And as soon as you start talking about those things, it probably raises a lot of anxiety. But we already had it.* Simone, mother: ALS/MND (untested) 8. *[Mum] said that she had the paperwork [regarding the availability of predictive testing] for us kids…But it was on her conditions. We weren't to go and get tested until Dad had passed away. She was to know the answers…It would have helped if the doctor…realized…how Mum would be…Maybe there should have been a family meeting that the doctor was present at.* Olivia, father: ALS/MND and FTD (gene positive) **Support from others** 9. *My husband hasn't handled it very well…we never discuss it because he gets very upset…[I am supported] by my GP and…a friend.* Alanna, mother: ALS/MND and FTD (gene positive) 10. *I…needed somebody to talk to…I spoke to friends…but they don't understand…[I saw] my GP…had a bit of a cry…But…she didn't have [more than]…a fleeting understanding of MND…I was the expert for her on the whole topic…I didn't know where else to go.* Lara, brother Ryan: ALS/MND (gene positive) 11. *My sister was a little bit cautious [about my proceeding with diagnostic testing]…My dad, however, was 110%. "Yes, do it…You get a yes or no answer, and we deal with it either way"… That helped me make my mind up that I'm definitely doing it.* Fabian, FTD suspected. Mother: FTD (untested). Fabian underwent diagnostic testing (gene positive)

(Continues)

TABLE 2 (Continued)

Theme, predominant PPCT categories and description	Subthemes and supporting quotes
'Circumstance is everything' • Time • Person[a]The unique lived experiences of participants informed how they approached genetic counseling and testing and/or managed knowledge within the family at that point in time. Some experiences were ongoing, others related to previous individual/family experiences. These unique circumstances informed information and support needs and related hopes, fears, and expectations toward genetic counseling, testing, results and managing knowledge within the family. By discussing their own experiences, participants reflected on whether genetic counseling (and diagnostic testing) should be a part of routine care.	**The timing of testing** 12. *It should have been done 11 years ago [when my husband first developed symptoms]…We didn't want it to be MND. But…it explains so much.* Nina, husband: ALS/MND and FTD (gene positive) 13. *[My brother] agreed to…have [genetic testing]. But he did not want the results. [They were] only to be given to me. [He] signed all the forms. "You can test…but not till (sic) I'm gone."* Genevieve, brother: ALS/MND (gene positive) 14. *[One thing that I would have liked] is the option of telehealth with a video aspect…Since the pandemic hit, they've all been phone appointments. A lot of the times…Kevin has been really upset and is…completely…disengaged. He's needed a moment, and…they've got no sensor that's happening.* Isabelle, husband Kevin: ALS/MND (gene positive) **Responses to genetic information within the family** 15. *[When my brother was diagnosed with MND], the diagnosis…had to be kept…from [the rest of the family]…I'm still bitter about that…It is now up to me to inform [them] with the instructions…of only telling them…when we have a good treatment or cure. Not happening. Basically, when they are essentially of reproductive age and maturity, we're going to be having that talk…If I'm not supposed to tell other family members…they miss the opportunity…to have that testing themselves.* Genevieve, brother: ALS/MND (gene positive) 16. *[My neurologist and I] came up with a cunning plan…to tell [my family] if you want to have the testing, that's fine…But it's probably not reasonable [until]…therapies emerge that are related to delaying the course…or preventing its onset.* Kath, ALS/MND (gene positive) **Attitudes toward counseling and diagnostic testing for others** 17. *People should have the right to know that it's available. They can then make the choice whether they want to go ahead with it or not.* Nina, husband: ALS/MND and FTD (gene positive) 18. *You've gotta (sic)…give [patients] the opportunity [to]…have this sample taken…for research purposes if that's what they choose to do, so that…their legacy is something positive…[So my answer is] yes, and early…It's not fair to give it late. It's not fair to not tell them that there may be a genetic component.* Genevieve, brother: ALS/MND (gene positive)

[a]Person in this setting may represent several client types: patient, family member and/or carer.

> *[Before having the testing,] there was no actual questioning or anything…I don't think I would have liked to be overloaded with information.*
>
> Kevin, ALS/MND (gene positive)

> *They were trying to minimize any delay in getting this result…But…if we had…gotten pregnant…[while] waiting for the results, we might be feeling very different about the…information we had before we got the results.*
>
> Isabelle, husband Kevin: ALS/MND (gene positive).

After testing, some felt they received inadequate follow-up, and questions remained unanswered:

> *[After the results were told to me], I made contact a couple times to say "can we talk about this?"… And I haven't heard [back]. So it's been a bit lost.*
>
> Kath, ALS/MND (gene positive)

3.2.2 | Information sharing within the family

Within the family, information was shared by informal family conversations and information other family members had received from outside agencies. In many cases, open knowledge about the family history of disease informed interest in testing (Quote 4, Table 2). In contrast, some participants were unaware of their family history, nor was it openly discussed. Genetic testing only became important once a closer relative was diagnosed (Quote 5, Table 2).

While some found it straightforward to speak to their family about their testing decisions and the results, others felt they needed to consider this further before sharing information with family members:

> *I haven't told [my daughter]. But there's a letter for her that my husband will give to her if she gets married or if… she's thinking of having children. And then she can make the decisions based on…information at that time.*
>
> Kath, ALS/MND (gene positive)

3.3 | (Un)supportive care

3.3.1 | Support from the managing team

Some felt well supported by the health professionals who facilitated genetic counseling and testing (Quote 6, Table 2). Others eventually found a health professional who addressed the client's concerns and anxieties about genetic risk (Quote 7, Table 2).

The option of further counseling or support to help facilitate adjustment to the diagnosis and result was not mentioned by all managing teams, and pre-existing thoughts regarding counseling meant it was not normalized:

> *No one that I'd ever known had openly…sought counseling…[I'd] never even considered it*.
>
> Kevin, ALS/MND (gene positive).

> *[After the diagnosis, Kevin wasn't] able to have a conversation about it because it was too distressing…That led me to asking, at one of the neurology appointments, what they normally offer in terms of ongoing or additional…support…For Kevin, me suggesting some of these things was less appealing to him because…it wasn't seen as what was normal…because it hadn't been offered by a neurologist*.
>
> Isabelle, husband Kevin: ALS/MND (gene positive)

Some felt more support from health professionals toward family communication would have been helpful (Quote 8, Table 2).

3.3.2 | Support from others

Aside from the health professional who arranged testing, participants sought support from family, friends, other health professionals, ALS/MND/FTD support associations, and connected online with others in similar circumstances. In some cases, distress about the familial risk prevented close family members from providing support (Quote 9, Table 2). Some at-risk relatives felt alone adjusting to the news that they were at genetic risk and found it difficult to access adequate professional or social support (Quote 10, Table 2). When sharing knowledge within the family, responses varied, and the level of support from family members sometimes informed whether clients proceeded with diagnostic testing (Quote 11, Table 2).

3.4 | 'Circumstance is everything'

3.4.1 | The timing of testing

At the same time as undergoing genetic counseling and testing, clients may have been under tremendous stress, with testing occurring alongside attempts to address and adjust to other disease-related symptoms.

> *Nothing about this disease is easy.*
>
> Kath, ALS/MND (gene positive).

The importance of prompt access to genetic counseling and testing was more relevant for some than others. For some, the results confirmed a diagnosis of ALS/MND/FTD, ending a long diagnostic journey (Quote 12, Table 2). Some patients had unique needs or wishes, often related to the progress of their disease (Quote 13, Table 2). Experiences where the counseling was adapted to the client's needs were looked upon favorably.

Some thought that outside factors such as the COVID-19 pandemic impacted the level of care provided at the time (Quote 14, Table 2).

3.4.2 | Responses to genetic information within the family

Responses within a family to a clinical or genetic diagnosis varied and, in many cases, had a substantial impact on family dynamics and communication. Some responses resulted in barriers to information sharing and supportive care for members of certain families (Quote 15, Table 2).

Some barriers to sharing knowledge and accessing support related to the disease experience in the family:

> *I'm not talking to my sister at the moment. [There is] family estrangement because of [MND] and what happened with Dad.*
>
> Olivia, father: ALS/MND and FTD (gene positive).

Others were unrelated:

> *[Regarding my son,] I'm separated from his mother… Our relationship isn't that great…He will need to know [but]…I've just got a bigger issue of when and how I tell his mother.*
>
> Fabian, FTD suspected. Mother: FTD (untested). Fabian underwent diagnostic testing (gene positive)

Sharing information within families was also hindered due to the beliefs of the client. One participant felt that even a discussion about the availability of predictive testing could lead family members to proceed and was cautious about initiating a conversation.

> *My cousin…[is] getting curious and concerned about his potentially having the gene…He hasn't asked me…And I haven't needed to tell him that [he is at risk] yet…I don't want to push people into knowing unless they want to know.*
>
> Estelle, father: ALS/MND (gene positive).

Others decided to frame this information a certain way with their relatives, given they did not perceive any immediate benefits of predictive testing for other family members (Quote 16, Table 2).

3.4.3 | Attitudes toward counseling and diagnostic testing for others

Informed by their own experiences, most participants were positive about routinely discussing the option of diagnostic testing (Quote 17, Table 2). Some suggested that patients should have the opportunity to contribute a DNA sample to research without knowing their results so relatives could access DNA in the future (Quote 18, Table 2). Others were more cautious, particularly concerning ALS/MND/FTD patients who were unable to give consent, where there were few clinical benefits to the person with ALS/MND/FTD:

> *If the person who had the disease is capable of making the choice,…[they] should be offered it…[But] if…their mental capacity is not there, then why can't they live not knowing…about it?*
>
> Alanna, mother: ALS/MND and FTD (gene positive).

4 | DISCUSSION

The primary aim of this study was to explore client experiences of genetic counseling and diagnostic genetic testing to inform the care of future families. Experiences demonstrated that the genetic counseling process included two distinct phases that occurred concurrently or independently: genetic testing and managing knowledge within the family. Consistent with bioecological theory (Bronfenbrenner & Evans, 2000), experiences were informed by interrelationships between clients and their environment. Proximal processes within one's microsystem (i.e., direct interactions with others) were an important part of participants' experiences. Some clients felt well informed and supported throughout the genetic counseling process through interactions with other family members, health professionals, support associations, peers, and online information. But, interactions were not always easy or accessible, and clients had to seek informative and supportive care. Individual client and family circumstances or characteristics at the time of genetic counseling and testing (i.e., the person and time aspects of bioecological theory) informed the level of information and support required and attitudes and responses toward genetic counseling and testing. Circumstances varied due to disease progression, lived experience of disease, family communication and dynamics, and external stressors such as the COVID-19 pandemic. While some circumstances remain unchangeable, the findings demonstrate that interactions between the client and their care team could be improved. These findings and further implications for research and practice are outlined.

Participants in this study included patients, their spouses, and relatives who had all experienced genetic counseling discussions related to diagnostic testing. The majority were counseled by a neurologist. One was counseled by a social worker, which is a routine part of care in that location. The outcomes of diagnostic testing concern both the patient and their relatives (Mendes et al., 2018) and as soon as a diagnosis of ALS/MND/FTD is made, at-risk relatives may already have feelings of uncertainty and fear concerning their possible familial risk (Crook et al., 2021; Howard et al., 2021). As the clinical and psychological implications of diagnostic testing are vastly different depending on whether you are the patient, their spouse, or their relative, we had anticipated that each subgroup may have unique needs in the diagnostic testing process. Instead, we found no apparent differences in the subgroup analysis, based on reviewing the clinical and demographic variables (e.g., clinical/risk status of the participant, ALS/MND/FTD experience, location, and the health professional type who facilitated testing). This may be due to the relatively small sample size included in the study. It is also possible that generalizations cannot be easily made as unique client/family circumstances, dynamics, and responses informed experiences and needs. This supports previous findings that the decision to proceed with genetic counseling and testing is informed by personal, familial, and practical factors (Crook et al., 2021).

The potential benefits of genetic testing and counseling for at-risk relatives rely on clients sharing testing and counseling outcomes within the family. The study results demonstrate that some participants found sharing knowledge in the family to be burdensome, irrelevant, or unnecessary. Some decided to protect others from the burden of knowing (Mendes et al., 2018), perhaps easier in ALS/MND/FTD than some other neurodegenerative diseases (e.g., Huntington's disease) as it is not always heritable. Similar to studies in familial cancer (Smit et al., 2021), family communication was informed by perceptions about risk management options, the degree of closeness in the family, and a sense of responsibility. Family communication was a longitudinal process (Mendes et al., 2018): some communicated with their family as part of the testing process, others after testing was complete, and others deferred communication entirely. Although the focus of pre-test counseling may be on the implications of testing for the client, the impact of testing on relatives and family communication should also be explored (Mendes et al., 2018). Given that genetic counseling support improves communication in some families (Forrest et al., 2008), follow-up genetic counseling regarding family communication may be required even after testing is complete. Family-centered interventions and support resources for both clients and health professionals to assist communication may also be beneficial. For clients, this may include information sheets or personalized family letters to support dissemination, narrative group sessions, or group consultations with the option of separate confidential discussions. Additional training regarding family systems theory, narrative practice, family therapy, and communication may be beneficial for health professionals (Mendes et al., 2018;

TABLE 3 Client needs as indicated from the research findings. Supporting quotes are available in Appendix S5

Support needs	Information needs

• Clear and sensitive communication that is adapted to the client's needs
• The opportunity to have questions and concerns addressed
• Time and space to consider options
• The option of multiple persons attending counseling and testing discussions
• At-risk relatives also supported to facilitate adjustment to the diagnosis and consider the option of predictive testing (if relevant)
• The option of additional appointments, counseling or support options and resources
• Follow-up after testing, if indicated
• An exploration regarding family communication and assistance with this (if necessary)
	• Timely awareness about the availability of counseling and testing
	• Practicalities on the genetic testing process:
	  • Pathways to access genetic counseling and testing
	  • What condition(s)/gene(s) are being tested
	  • Testing timeframes and limitations
	  • The likelihood of pathogenic variant detection
	  • Insurance implications and information regarding privacy and confidentiality
	  • Contact details of the testing team
	• Implications of proceeding or not with testing for the patient and their family, including research opportunities
	• A clear plan for follow-up and results disclosure
	• Locally relevant information, including resources to refer back to
	• Pathways to access further support/information (if wanted)

Roberts et al., 2020; Smit et al., 2021; Stopford et al., 2020; Young et al., 2019).

Consistent with previous studies (Fostinelli et al., 2018; Wagner et al., 2017), participants supported genetic counseling being offered to all individuals with ALS/MND/FTD. Due to disease progression, some suggested the discussion should be made as early as possible. Others raised concerns regarding testing of persons without the capacity to consent for themselves. As the potential benefits may not outweigh the risks for some patients and families, counseling and testing should be offered as an option rather than mandatory. Genetic counseling that addresses both the patient and their family's concerns should be accessible and is now recognized as a fundamental right of people living with ALS/MND (International Alliance of ALS/MND Associations, 2021). The results from this study highlight the value some (but not all) participants still placed on the importance of pre-test counseling to assist with decision-making. Similar to the Huntington's disease diagnostic testing approach (Craufurd et al., 2015), the findings highlight that there can be a variety of clinical situations where diagnostic testing can be indicated. Flexible approaches that adapt the pre-and post-test counseling to the client's needs, take into account individual circumstances, and work to ensure the best possible outcome for both the person with the disease and their family are key (Craufurd et al., 2015). Genetic counselors are well equipped to provide client- and family-centered genetic counseling regarding diagnostic testing, but clinical genetics health professionals are not always a part of multidisciplinary ALS/MND/FTD care, and timely access may be difficult (Salmon et al., 2021). Regardless of the health professional type who facilitates genetic counseling and diagnostic testing discussions, avenues to access more information and support outside of formal genetic counseling or testing discussions could be helpful as not all clients will require the same amount of information and support pre-and post-testing (Crook et al., 2021).

4.1 | Study strengths and limitations

Qualitative methods were a major strength of the study, providing contextual insights into client experience and needs. Still, limitations exist regarding the study methods and implications for research and practice. We sought to include a wide variety of participants with varying disease status and abilities and hoped that our research design would support this with multiple interview and consent options. Unfortunately, all experiences may not have been accounted for as we experienced difficulty recruiting the breadth of participants we had hoped to ascertain. In particular, it was difficult to access FTD-only families, including individuals with FTD, which may explain why there are limited studies in this area (Crook et al., 2021). We were also only able to recruit participants residing in four of six states in Australia. Recruitment bias may have also occurred by recruiting individuals who had to be engaged with support associations. Direct recruitment from patient management clinics (ideally, with consecutive sampling) may have been more representative of the participant population than relying on potential participants seeking research opportunities through support associations. While we endeavored to ensure trustworthiness, the study may have benefited from synthesized member checking (Birt et al., 2016). Despite these limitations, the findings provide valuable insight into an under-studied and emerging area of practice.

4.2 | Practice implications

Although one's clinical circumstances, family history, and dynamics cannot be changed, the findings demonstrate that clinical practice could be improved by improving client interactions with the health professionals and support associations facilitating genetic counseling discussions. A summary of the needs inferred by participants in this study is detailed in Table 3 (further information available in Appendix S5). Many of these needs are not just unique to genetic counseling and testing and have been previously reported as a requirement of those who are burdened by living with or caring for someone who is affected with ALS/MND/FTD (Gentry et al., 2020; Weisser et al., 2015).

The question remains on what is the minimum amount of discussion or exploration required to constitute an informed and supported decision to proceed (or not) with genetic counseling and diagnostic

testing. These aspects will be further considered in our subsequent study, where we plan to develop guidelines for genetic counseling and diagnostic testing in ALS/MND/FTD that are informed by the input of consumers and health professionals.

5 | CONCLUSIONS

Interest in genetic counseling and testing for ALS/MND/FTD is expected to increase as further management options become available for people with ALS/MND/FTD and their relatives. Consistent with bioecological theory, one's genetic counseling experience was informed by individual circumstances, time, and proximal factors. Although some client circumstances cannot be changed, efforts could be made to enhance client experiences of genetic counseling and testing by improving interactions with health professionals, support associations and the wider family. Some clients may benefit from further discussions regarding the familial implications of genetic testing and greater support with family communication. We expect the client needs ascertained from this study will contribute to the development of guidelines for genetic counseling in ALS/MND/FTD. By improving genetic counseling experiences, we hope that some of the burdens of the disease lived experience could be reduced.

AUTHOR CONTRIBUTIONS

Alison McEwen: Conceptualization; formal analysis; methodology; supervision; validation; writing – review and editing. **Ashley Crook:** Conceptualization; formal analysis; investigation; methodology; project administration; visualization; writing – original draft; writing – review and editing. **Chris Jacobs:** Conceptualization; methodology; supervision; validation; writing – review and editing. **Toby Newton-John:** Conceptualization; supervision; validation; writing – review and editing.

ACKNOWLEDGMENTS

The authors would like to thank all participants who generously provided their time and input. We would also like to thank Dr Emma Power, speech pathologist and academic at the University of Technology Sydney, who provided guidance on conducting research with individuals with cognitive and communication impairment. Open access publishing facilitated by University of Technology Sydney, as part of the Wiley - University of Technology Sydney agreement via the Council of Australian University Librarians.

COMPLIANCE WITH ETHICAL STANDARDS

CONFLICT OF INTEREST

Authors A Crook, C Jacobs, T Newton-John, and A McEwen declare that they have no conflict of interest.

HUMAN STUDIES AND INFORMED CONSENT

The University of Technology Sydney (UTS) Medical Research Ethics Committee approved this study. The research was undertaken in compliance with the Australian Code for the Responsible Conduct of Research and National Statement on Ethical Conduct in Human Research. Informed consent was obtained from all study participants. Many participant identifiers have been removed, so the person(s) described are not identifiable.

ANIMAL STUDIES

No non-human animal studies were carried out by the authors for this article.

DATA SHARING AND DATA ACCESSIBILITY

The data are not publicly available due to privacy or ethical restrictions. Further information is available in the supplementary material or by contacting the corresponding author.

ORCID

Ashley Crook https://orcid.org/0000-0001-5578-0460
Chris Jacobs https://orcid.org/0000-0002-9557-9080
Toby Newton-John https://orcid.org/0000-0003-4219-4985
Alison McEwen https://orcid.org/0000-0001-8705-1190

REFERENCES

Benatar, M., Polak, M., Kaplan, S., & Glass, J. (2006). Preventing familial amyotrophic lateral sclerosis: Is a clinical trial feasible? *Journal of the Neurological Sciences*, *251*(1–2), 3–9. https://doi.org/10.1016/j.jns.2006.05.069

Birt, L., Scott, S., Cavers, D., Campbell, C., & Walter, F. (2016). Member checking: A tool to enhance trustworthiness or merely a nod to validation? *Qualitative Health Research*, *26*(13), 1802–1811. https://doi.org/10.1177/1049732316654870

Blauwendraat, C., Wilke, C., Simón-Sánchez, J., Jansen, I. E., Reifschneider, A., Capell, A., Haass, C., Castillo-Lizardo, M., Biskup, S., Maetzler, W., Rizzu, P., Heutink, P., & Synofzik, M. (2018). The wide genetic landscape of clinical frontotemporal dementia: Systematic combined sequencing of 121 consecutive subjects. *Genetics in Medicine*, *20*(2), 240–249. https://doi.org/10.1038/gim.2017.102

Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology*, *3*(2), 77–101. https://doi.org/10.1191/1478088706qp063oa

Braun, V., & Clarke, V. (2019). Reflecting on reflexive thematic analysis. *Qualitative Research in Sport, Exercise and Health*, *11*(4), 589–597. https://doi.org/10.1080/2159676X.2019.1628806

Bronfenbrenner, U. (2001). Human development, bioecological theory of. In N. J. Smelser & P. B. Baltes (Eds.), *International encyclopedia of the social & behavioral sciences* (pp. 6963–6970). Pergamon.

Bronfenbrenner, U., & Evans, G. W. (2000). Developmental science in the 21st century: Emerging questions, theoretical models, research designs and empirical findings. *Social Development*, *9*(1), 115–125. https://doi.org/10.1111/1467-9507.00114

Craufurd, D., MacLeod, R., Frontali, M., Quarrell, O., Bijlsma, E. K., Davis, M., Hjermind, L. E., Lahiri, N., Mandich, P., Martinez, A., Tibben, A., Roos, R. A., & Testing of the European Huntington's Disease Network (EHDN). (2015). Diagnostic genetic testing for Huntington's disease. *Practical Neurology*, *15*(1), 80–84. https://doi.org/10.1136/practneurol-2013-000790

Crook, A., Jacobs, C., Newton-John, T., Richardson, E., & McEwen, A. (2021). Patient and relative experiences and decision-making about genetic testing and counseling for familial ALS and FTD: A systematic scoping review. *Alzheimer Disease and Associated Disorders*, *35*(4), 374–385. https://doi.org/10.1097/WAD.0000000000000458

Draucker, C. B., Martsolf, D. S., & Poole, C. (2009). Developing distress protocols for research on sensitive topics. *Archives of Psychiatric Nursing*, *23*(5), 343–350. https://doi.org/10.1016/j.apnu.2008.10.008

Forrest, L. E., Burke, J., Bacic, S., & Amor, D. J. (2008). Increased genetic counseling support improves communication of genetic information in families. *Genetics in Medicine*, *10*(3), 167–172. https://doi.org/10.1097/GIM.0b013e318164540b

Fostinelli, S., Ciani, M., Zanardini, R., Zanetti, O., Binetti, G., Ghidoni, R., & Benussi, L. (2018). The heritability of frontotemporal lobar degeneration: Validation of pedigree classification criteria in a northern Italy cohort. *Journal of Alzheimer's Disease*, *61*(2), 753–760. https://doi.org/10.3233/JAD-170661

Gentry, M. T., Lapid, M. I., Syrjanen, J., Calvert, K., Hughes, S., Brushaber, D., Kremers, W., Bove, J., Brannelly, P., Coppola, G., Dheel, C., Dickerson, B., Dickinson, S., Faber, K., Fields, J., Fong, J., Foroud, T., Forsberg, L., Gavrilov, R., & LEFFTDS Consortium. (2020). Quality of life and caregiver burden in familial frontotemporal lobar degeneration: Analyses of symptomatic and asymptomatic individuals within the LEFFTDS cohort. *Alzheimer's and Dementia*, *16*(8), 1115–1124. https://doi.org/10.1002/alz.12095

Heath, J., Williamson, H., Williams, L., & Harcourt, D. (2018). "It's just more personal": Using multiple methods of qualitative data collection to facilitate participation in research focusing on sensitive subjects. *Applied Nursing Research*, *43*, 30–35. https://doi.org/10.1016/j.apnr.2018.06.015

Hoskins, L. M., & Werner-Lin, A. (2013). A multi-case report of the pathways to and through genetic testing and cancer risk management for BRCA mutation-positive women aged 18-25. *Journal of Genetic Counseling*, *22*(1), 27–38. https://doi.org/10.1007/s10897-012-9521-y

Howard, J., Mazanderani, F., & Locock, L. (2021). Life 'on high alert': How do people with a family history of motor neurone disease make sense of genetic risk? Insights from an online forum. *Health, Risk & Society*, *1-17*, 179–195. https://doi.org/10.1080/13698575.2021.1946488

International Alliance of ALS/MND Associations, T. (2021) *Fundamental rights for people living with ALS/MND*. Retrieved from https://www.als-mnd.org/support-for-pals-cals/advocacy/pals-and-cals-rights/

Klepek, H., Nagaraja, H., Goutman, S. A., Quick, A., Kolb, S. J., & Roggenbuck, J. (2019). Lack of consensus in ALS genetic testing practices and divergent views between ALS clinicians and patients. *Amyotroph Lateral Scler Frontotemporal Degener*, *20*(3–4), 216–221. https://doi.org/10.1080/21678421.2019.1582670

Liamputtong, P. (2007). *Researching the vulnerable: A guide to sensitive research methods*. Sage.

Lincoln, Y. S., & Guba, E. G. (1985). *Naturalistic inquiry*. SAGE Publications.

Malterud, K., Siersma, V. D., & Guassora, A. D. (2016). Sample size in qualitative interview studies: Guided by information power. *Qualitative Health Research*, *26*(13), 1753–1760. https://doi.org/10.1177/1049732315617444

McGrath, M., Lever, S., McCluskey, A., & Power, E. (2019). Developing interventions to address sexuality after stroke: Findings from a four-panel modified Delphi study. *Journal of Rehabilitation Medicine*, *51*(5), 352–360. https://doi.org/10.2340/16501977-2548

Mendes, Á., Metcalfe, A., Paneque, M., Sousa, L., Clarke, A. J., & Sequeiros, J. (2018). Communication of information about genetic risks: Putting families at the center. *Family Process*, *57*(3), 836–846. https://doi.org/10.1111/famp.12306

Miller, T., Cudkowicz, M., Shaw, P. J., Andersen, P. M., Atassi, N., Bucelli, R. C., Genge, A., Glass, J., Ladha, S., Ludolph, A. L., Maragakis, N. J., McDermott, C. J., Pestronk, A., Ravits, J., Salachas, F., Trudell, R., Van Damme, P., Zinman, L., Bennett, C. F., … Ferguson, T. A. (2020). Phase 1–2 trial of antisense oligonucleotide Tofersen for SOD1 ALS. *The New England Journal of Medicine*, *383*(2), 109–119. https://doi.org/10.1056/NEJMoa2003715

Morris, S. M. (2001). Joint and individual interviewing in the context of cancer. *Qualitative Health Research*, *11*(4), 553–567. https://doi.org/10.1177/104973201129119208

National Health and Medical Research Council, A. (2018). *National Statement on Ethical Conduct in Human Research*. https://www.nhmrc.gov.au/about-us/publications/national-statement-ethical-conduct-human-research-2007-updated-2018

O'Brien, B. C., Harris, I. B., Beckman, T. J., Reed, D. A., & Cook, D. A. (2014). Standards for reporting qualitative research: A synthesis of recommendations. *Academic Medicine*, *89*(9), 1245–1251. https://doi.org/10.1097/ACM.0000000000000388

Roberts, J. S., Patterson, A. K., & Uhlmann, W. R. (2020). Genetic testing for neurodegenerative diseases: Ethical and health communication challenges. *Neurobiology of Disease*, *141*, 104871. https://doi.org/10.1016/j.nbd.2020.104871

Roggenbuck, J., & Fong, J. C. (2020). Genetic testing for amyotrophic lateral sclerosis and frontotemporal dementia: Impact on clinical management. *Clinics in Laboratory Medicine*, *40*(3), 271–287. https://doi.org/10.1016/j.cll.2020.05.002

Rosa, E. M., & Tudge, J. (2013). Urie Bronfenbrenner's theory of human development: Its evolution from ecology to bioecology. *Journal of Family Theory & Review*, *5*(4), 243–258. https://doi.org/10.1111/jftr.12022

Salmon, K., Anoja, N., Breiner, A., Chum, M., Dionne, A., Dupré, N., Fiander, A., Fok, D., Ghavanini, A., Gosselin, S., Izenberg, A., Johnston, W., Kalra, S., Matte, G., Melanson, M., O'Connell, C., Ritsma, B., Schellenberg, K., Shoesmith, C., … Genge, A. (2021). Genetic testing for amyotrophic lateral sclerosis in Canada – An assessment of current practices. *Amyotroph Lateral Scler Frontotemporal Degener*, *1–8*, 305–312. https://doi.org/10.1080/21678421.2021.1980890

Santana, M. J., Manalili, K., Jolley, R. J., Zelinsky, S., Quan, H., & Lu, M. (2018). How to practice person-centred care: A conceptual framework. *Health Expectations: An International Journal of Public Participation in Health Care and Health Policy*, *21*(2), 429–440. https://doi.org/10.1111/hex.12640

Shepheard, S. R., Parker, M. D., Cooper-Knock, J., Verber, N. S., Tuddenham, L., Heath, P., Beauchamp, N., Place, E., Sollars, E. S. A., Turner, M. R., Malaspina, A., Fratta, P., Hewamadduma, C., Jenkins, T. M., McDermott, C. J., Wang, D., Kirby, J., Shaw, P. J., & Morrison, K. E. (2021). Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. *Journal of Neurology, Neurosurgery, and Psychiatry*, *92*, 510–518. https://doi.org/10.1136/jnnp-2020-325014

Smit, A. K., Bartley, N., Best, M. C., Napier, C. E., Butow, P., Newson, A. J., Tucker, K., Ballinger, M. L., Thomas, D. M., Jacobs, C., Meiser, B., Goldstein, D., Savard, J., Juraskova, I., & PiGeOn Authorship Group. (2021). Family communication about genomic sequencing: A qualitative study with cancer patients and relatives. *Patient Education and Counseling*, *104*(5), 944–952. https://doi.org/10.1016/j.pec.2020.10.022

Stopford, C., Ferrer-Duch, M., Moldovan, R., & MacLeod, R. (2020). Improving follow up after predictive testing in Huntington's disease: Evaluating a genetic counselling narrative group session. *Journal of Community Genetics*, *11*(1), 47–58. https://doi.org/10.1007/s12687-019-00416-9

Thorne, S. E. (2016). *Interpretive description: Qualitative research for applied practice* (2nd ed.). Routledge.

Tillerås, K. H., Kjoelaas, S. H., Dramstad, E., Feragen, K. B., & Lippe, C. (2020). Psychological reactions to predictive genetic testing for huntington's disease: A qualitative study. *Journal of Genetic Counseling*, *29*(6), 1093–1105. https://doi.org/10.1002/jgc4.1245

Tong, A., Sainsbury, P., & Craig, J. (2007). Consolidated criteria for reporting qualitative research (COREQ): A 32-item checklist for interviews

and focus groups. *International Journal for Quality in Health Care*, *19*(6), 349–357. https://doi.org/10.1093/intqhc/mzm042

Turner, M. R., Al-Chalabi, A., Chio, A., Hardiman, O., Kiernan, M. C., Rohrer, J. D., Rowe, J., Seeley, W., & Talbot, K. (2017). Genetic screening in sporadic ALS and FTD. *Journal of Neurology, Neurosurgery, and Psychiatry*, *88*(12), 1042–1044. https://doi.org/10.1136/jnnp-2017-315995

Wagner, K. N., Nagaraja, H. N., Allain, D. C., Quick, A., Kolb, S. J., & Roggenbuck, J. (2017). Patients with amyotrophic lateral sclerosis have high interest in and limited access to genetic testing. *Journal of Genetic Counseling*, *26*(3), 604–611. https://doi.org/10.1007/s10897-016-0034-y

Weisser, F. B., Bristowe, K., & Jackson, D. (2015). Experiences of burden, needs, rewards and resilience in family caregivers of people living with motor Neurone disease/amyotrophic lateral sclerosis: A secondary thematic analysis of qualitative interviews. *Palliative Medicine*, *29*(8), 737–745. https://doi.org/10.1177/0269216315575851

Young, A. L., Butow, P. N., Tucker, K. M., Wakefield, C. E., Healey, E., & Williams, R. (2019). Challenges and strategies proposed by genetic health professionals to assist with family communication. *European Journal of Human Genetics*, *27*(11), 1630–1638. https://doi.org/10.1038/s41431-019-0447-9

SUPPORTING INFORMATION

Additional supporting information may be found in the online version of the article at the publisher's website.

How to cite this article: Crook, A., Jacobs, C., Newton-John, T., & McEwen, A. (2022). Genetic counseling and diagnostic genetic testing for familial amyotrophic lateral sclerosis and/or frontotemporal dementia: A qualitative study of client experiences. *Journal of Genetic Counseling*, *31*, 1206–1218. https://doi.org/10.1002/jgc4.1591