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Evaluation of an educational conference for persons affected by hereditary frontotemporal degeneration and amyotrophic lateral sclerosis

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ABSTRACT

Objective: There are limited studies exploring the support and education needs of individuals at-risk for or diagnosed with hereditary frontotemporal degeneration (FTD) and/or amyotrophic lateral sclerosis (ALS). This study evaluated a novel conference for this population to assess conference efficacy, probe how participants assessed relevant resources, and identify outstanding needs of persons at-risk/diagnosed.

Methods: We implemented a post-conference electronic survey that probed participants' satisfaction, prior experience with resources, and unmet needs. Along with multiple-choice, free-text items were included to gather qualitative context.

Results: Survey completion rate was 31% (115/376 attendees who were emailed the survey). There was positive interest in pursuing genetic counseling among eligible responders: 61% indicated they planned to seek genetic counseling because of the conference, which was significantly more than those who were undecided (21%) or did not plan to seek genetic counseling (18%). Qualitative data demonstrated need for additional education, support, and research opportunities.

Conclusion: Conference reactions indicate this is a valued resource. Results indicated the importance of raising awareness about existing resources, and the need for further resource development, especially for at-risk communities.

Innovation: While most resources are developed for caregivers' needs, this unique program targets at-risk individuals and unites ALS and FTD communities.

1. Introduction

Frontotemporal degeneration (FTD) and amyotrophic lateral sclerosis (ALS) are two adult-onset neurodegenerative disorders that are now conceptualized as an FTD-ALS spectrum given overlapping genetic etiology [1,2]. FTD is characterized by the accumulation of pathology in the frontal and temporal lobes resulting in prominent behavioral, executive function, and language disturbances, and it is the second most prevalent cause of young-onset dementia after Alzheimer's disease, affecting 15–22/100,000 Americans [3–6]. ALS is characterized by loss of upper and lower motor neurons resulting in progressive paralysis and eventually death, with an average disease duration of 2–5 years and affecting an estimated 5/100,000 Americans [7,8]. There is substantial clinical overlap between ALS and FTD: up to 15% of patients with ALS also meet criteria for FTD, with up to 50% experiencing attenuated symptoms [9,10], and up to 30% of patients with FTD develop motor dysfunction [11,12]. There is also genetic overlap between ALS and FTD, most commonly the *C9orf72* pathogenic repeat expansion [2]. Approximately 10% of patients with ALS and 40% of patients with FTD have a family history of the disease [12], and about another 10% of individuals with apparently sporadic disease have a genetic etiology. Identification of monogenic causes of FTD and ALS has allowed for predictive genetic testing for asymptomatic family members [13]. For individuals at-risk or diagnosed, identifying a genetic cause has important implications for clinical management and planning. Moreover, while there is no cure, there are several ongoing clinical trials that target genetic forms of FTD and ALS [3,7,14]. Thus, genetic testing is being increasingly offered to and accepted by those with ALS and FTD, although practice differs among neurology clinics [15–17].

Previous studies of predictive genetic testing in FTD/ALS were mostly designed to determine whether predictive testing is safe from a mental

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health perspective [18–20], rather than understanding the broader psychosocial needs of this population. Though rapid advancements in gene-specific clinical trials may provide a source of hope, the science is progressing faster than the development of education and support resources. The historic lack of resources for persons with FTD/ALS and other young-onset dementias, as well as for their care partners, has been revealed through qualitative studies and community needs assessments [21–23]. In contrast to the growing body of work regarding care partner needs, there has been little investigation of the specific educational needs of people with or at-risk for genetic FTD or ALS [24]. As more families will be identified with genetic forms of FTD/ALS, it is imperative to further our understanding of the educational needs of persons diagnosed, persons at-risk, in addition to their care partners.

While the resources available to those with or at risk for FTD/ALS are limited, lack of education may lead to the under-utilization of the resources that do exist. For example, FTD and ALS are clinically, pathologically, and genetically overlapping diseases; thus advocacy efforts, research studies, support groups, and educational offerings that focus on either FTD or ALS can often benefit both populations. However, some families have experience with only one of these conditions and may not be aware of these overlapping risks. Educational resources, including genetic counseling and conferences, are a first step to inform families about the connection between FTD and ALS, so that they may access the full range of services available.

The Penn FTD Center held its first annual Penn Familial FTD/ALS Conference in February 2021 to provide education, support, and connection opportunities for those who are impacted, personally or professionally, by familial FTD and/or ALS. This conference was, to our knowledge, the first interactive meeting specifically dedicated to genetics issues in the FTD/ALS population. Given this novelty, we sought to determine attendees' reactions to this conference via a post-conference survey. The goal of this survey study was threefold: to evaluate whether our conference helped meet participants' needs, to determine how the community was levaraging existing resources, and to inform future FTD/ALS resource development by identifying unmet needs, especially for persons at-risk.

2. Methods

2.1. Participants and procedures

On Saturday, February 27, 2021, the Penn FTD Center hosted the inaugural Familial FTD/ALS Conference. This virtual event was available at no cost to attendees and was recorded (see Supplemental Table 1 for the recording). Topics were generated by genetic counselors, neurologists, nurses, and social workers to cover information typically requested in clinical encounters. Topics included the genetics of FTD/ALS, the state of research in FTD/ALS, and ways to find support and cope (see Supplemental Fig. 1). Two panel discussions highlighted individuals with a family history of FTD/ALS who shared their experiences with deciding 1.) whether to pursue predictive genetic testing and 2.) their life experiences after having learned their predictive genetic testing results. The event concluded with three interactive breakout sessions, and participants self-assigned to the "persons diagnosed", "persons at risk", or "supporters" session. The event was advertised by FTD and ALS advocacy and research networks, clinical care teams around the US, and the Penn FTD Center electronic mailing list. The target audience included individuals at-risk for or diagnosed with FTD or ALS, those that care for or support individuals diagnosed or at-risk for FTD or ALS, and people with a professional interest in FTD/ALS. Individuals were invited to share the registration information broadly.

We sent registrants and attendees an email invitation to complete an anonymous survey after three days, with reminders at eleven days and eighteen days. The survey was closed three days after the final reminder, on March 19th, 2021. Study data were collected and managed using Research Electronic Data Capture tools hosted at the University of Pennsylvania (UPenn) [25,26]. The study was classified as a quality improvement (QI) protocol by UPenn's Institutional Review Board (IRB); as such, no informed consent was required but implied informed consent was obtained when individuals voluntarily completed and submitted their survey responses.

2.2. Instrumentation

A brief survey was developed to assess participants' satisfaction with each session (each talk was rated on a 5-point Likert scale from "very poor" to "excellent"), as well as the conference overall (eight items about the conference quality rated on a 5-point Likert scale from "strongly disagree" to "strongly agree"). The survey also assessed participants' experience with FTD/ALS (e.g., which diagnoses are in the family), and relevant resources (e.g., which FTD/ALS resources have been accessed previously, if they were thinking about genetic counseling, if they wanted similar conferences in the future), via questions with predetermined responses from a drop-down list. Two free-text prompts were included for qualitative context: 1) "Please tell us any topics you would have liked to hear about during the [conference] that were not discussed or not discussed as much as you wanted," and 2) "Please tell us any other thoughts you have about the [conference]." Basic demographic information such as age, gender, and country of origin was collected. Participants self-reported their relationship to FTD/ALS selected from a single-choice drop-down list (i.e., "person at risk", "person diagnosed", "family member of a person diagnosed", "supporter for a person at risk or diagnosed", "health or research professional", or "other"). Given the paucity of resources targeted towards persons at-risk/diagnosed, it is important to know how this population accesses resources: we hypothesized that the conditions which have surfaced in the family (ALS or FTD vs. both) may influence which resources individuals have accessed in the past. Thus, participants reported their familial status, referring to whether there was either FTD or ALS in the family, both, or neither. The full survey is available in Supplemental Fig. 2.

2.3. Data analysis

2.3.1. Quantitative methods

We calculated descriptive statistics of participant characteristics and responses to survey items. We assessed responses from persons at-risk, persons diagnosed, supporters, lay persons, and health/research professionals. Because this conference was specifically targeted for the needs of people directly affected by FTD/ALS (at-risk and diagnosed), we compared responses from people at-risk/diagnosed vs. supporters. We also compared responses of members of families with one condition (either FTD or ALS) to families with both conditions. Chi-square analyses determined whether participant responses differed by participant group.

2.3.2. Qualitative methods

Free-text responses were optional survey items. All responses were subjected to content analysis by two coders (LD and KAQC). Nine categorical codes were generated and defined (Supplemental Table 2). Codes were either created a priori, based on probes for specific feedback (e.g., "conference style") or emerged after review of the data (e.g., "individual stories,"). Content was analyzed within the context of participants' self-reported relationship to FTD/ALS, such as whether the participant was at-risk/diagnosed or a supporter. Coders independently assigned codes to each response, and consensus was reached for any discrepancy. Further analysis led to reorganization of the nine codes into four overarching themes. Of note, more than one theme could be present in open-ended feedback.

3. Results

Table 1 summarizes the age, gender, and familial status of the survey respondents who attended the conference ($n = 115$). Conference attendance could not be accurately determined (see 4.3 Limitations), however of the 376 attendees who were emailed the survey, 115 (31%) responded. Survey respondents were mostly located in the United States ($n = 108$, 94%). Participants were asked to select all that applied from a drop-down list of

Table 1
Characteristics of Survey Respondents. Demographic characteristics are presented for survey responders including age, gender identity, self-categorization of relationship to FTD/ALS, and family FTD/ALS status.

	At Risk/Diagnosed	Family	Supporter	Health/Research Professional	Other
n	28	29	32	18	8
Gender (%)					
Male	5 (17.9)	5 (17.2)	7 (21.9)	2 (11.1)	1 (12.5)
Female	23 (82.1)	24 (82.8)	25 (78.1)	16 (88.9)	7 (87.5)
Non-binary	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
Other	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
Prefer not to say	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
Age (%)					
0–19	1 (3.6)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
20–29	1 (3.6)	2 (6.9)	0 (0.0)	5 (27.8)	2 (25.0)
30–39	8 (28.6)	2 (6.9)	0 (0.0)	4 (22.2)	0 (0.0)
40–49	8 (28.6)	5 (17.2)	3 (9.4)	0 (0.0)	2 (25.0)
50–59	3 (10.7)	4 (13.8)	5 (15.6)	2 (11.1)	2 (25.0)
60–69	4 (14.3)	8 (27.6)	12 (37.5)	1 (5.6)	1 (12.5)
70–79	3 (10.7)	7 (24.1)	10 (31.2)	6 (33.3)	1 (12.5)
80 +	0 (0.0)	1 (3.4)	2 (6.2)	0 (0.0)	0 (0.0)
Family Status (%)					
FTD only	9 (32.1)	17 (58.6)	25 (78.1)	–	–
ALS only	5 (17.9)	5 (17.2)	0 (0.0)	–	–
Both	14 (50.0)	7 (24.1)	7 (21.9)	–	–

Dashes indicate not applicable.

answers regarding, “What were you hoping to gain from this conference,” and the most common response was “research updates” (n = 79, 69%), followed by “a desire to learn more about FTD/ALS” (n = 69, 60%), “learning more about genetic testing and counseling options” (n = 58, 50%), “hearing about others' experiences with genetic testing and counseling” (n = 51, 44%), “learning about support resources” (n = 42, 37%), “learning about coping skills” (n = 24, 21%), and “connecting with others in the same situation” (n = 22, 19%) (Supplemental Fig. 3).

Sixty of the respondents provided open-ended feedback about the conference or topics of interest. The four overarching themes included attitudes towards the conference content and style, desire to know more about developments in research and treatment, improving access to education and resources, and sharing personal stories (Supplemental Fig. 4).

Analyses probed three questions: whether our conference helped meet the needs of the at-risk/diagnosed community, how the community was levaraging existing resources, and what gaps in education, research, and support needs remain for persons at-risk and diagnosed with FTD/ALS.

3.1. Did our conference meet participants' needs?

Nearly all participants felt they learned something new from the conference and that their questions were answered (Fig. 1). Persons at-risk and diagnosed were more likely to “strongly agree” that the conference met their expectations ($\chi^2(1, n = 60) = 4.5, p = 0.033$) and that the conference was helpful to them ($\chi^2(1, n = 60) = 5.1, p = 0.024$), compared with supporters. Next, we assessed whether presentations were clear and accessible

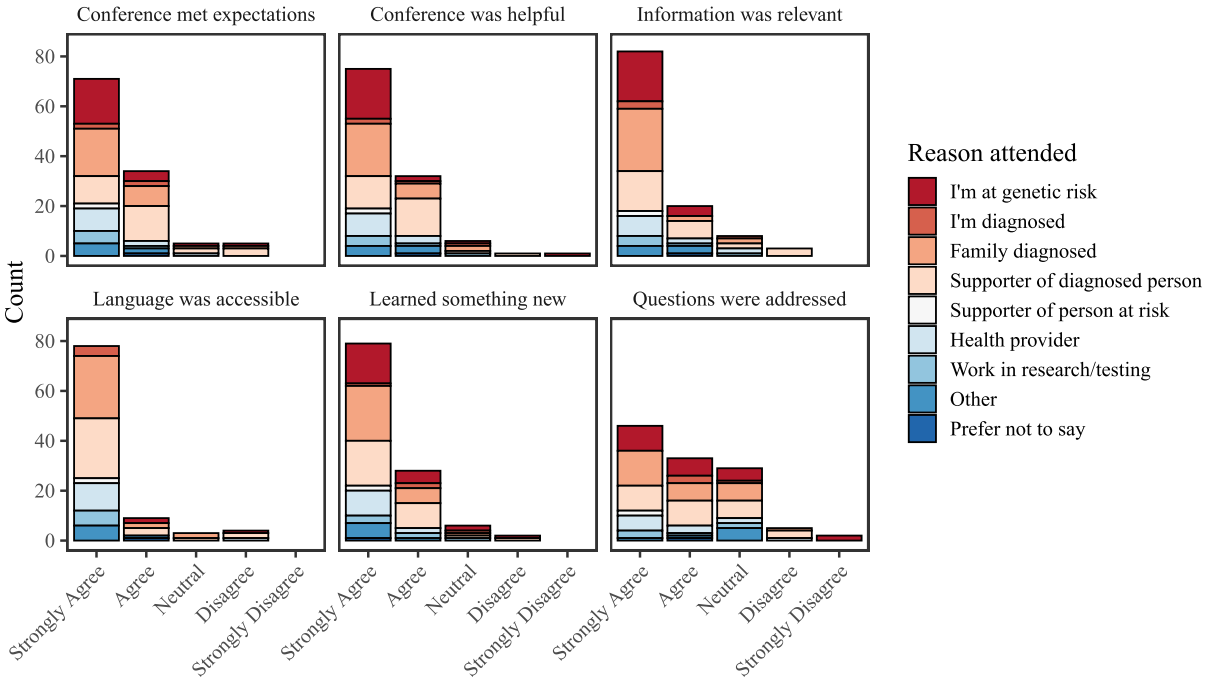

Fig. 1. Attitudes towards the Familial FTD/ALS Conference. Responses to six questions about reactions to the Familial FTD/ALS Conference are presented based on participant self-characterization of relationship to FTD/ALS.

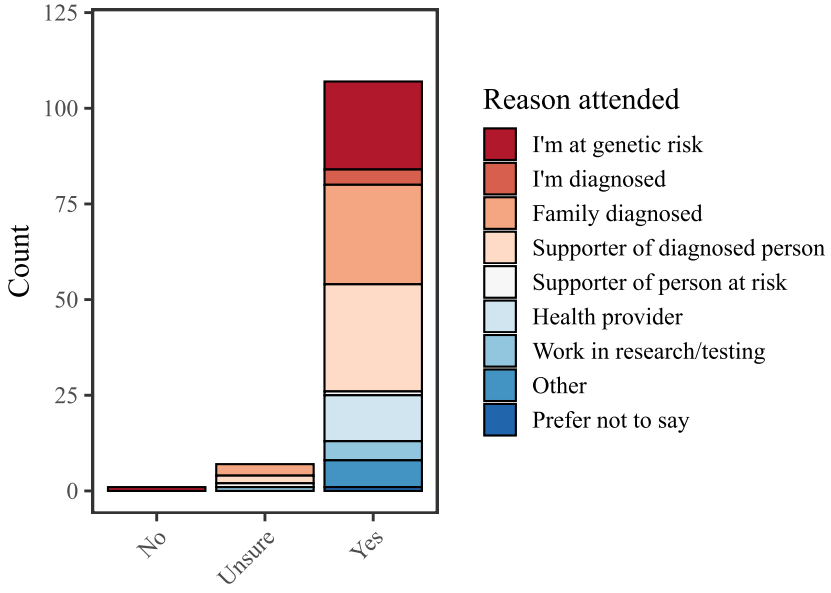

Fig. 2. Participant interest in a similar conference in the future. Participant responses to the question of whether they would have interest in a similar conference in the future are displayed, based on participant self-characterization of relationship to FTD/ALS.

to all: both lay persons and professionals agreed that speakers used understandable language, with no significant difference between these two groups ($p = 0.61$). Nearly all respondents ($n = 107$, 93%), regardless of their relationship to FTD/ALS, indicated they would be interested in attending a similar conference in the future (Fig. 2).

3.1.1. Attitudes towards conference content and style

Nearly half of the open-ended responses ($n = 29/60$, 48%) included positive feedback and expressions of gratitude for the conference and material covered. For example, comments included, "found the conference to be extremely valuable," and "there's a clear need for more of this in the community." Several people expressed relief and appreciation for being connected with the community, such as "it was comforting to know we are not alone in this battle." Many people praised the panel discussions included in the conference, noting that the volunteers who shared their experiences were "so clear and so real with their stories." While few commented that they would prefer an in-person conference, others felt the virtual format was useful: "thank you for making it virtual and accessible." However, four responses expressed dissatisfaction with an aspect of the conference. These included comments indicating a desire for more detailed information, frustration with technology, and difficulty with the technical language used during some presentations. We note that most respondents felt the language used in the conference presentations was accessible (Fig. 1).

Fifteen responses included suggestions to improve conference format. Given how well-received the panel-style presentations were, it is not surprising that there were requests for more shared stories. Reactions to the breakout sessions included comments about technology issues, the group dynamic, and time constraints. One participant noted that time zones pose a challenge.

3.2. How are participants leveraging existing resources?

A goal of the conference was to educate attendees about existing FTD/ALS resources so that they can maximize the use of the limited services at their disposal. Genetic counseling is critical resource for the target population. Respondents were asked, "Are you thinking about reaching out to a genetic counselor or encouraging someone to reach out to a genetic counselor because of the conference?" (Fig. 3). Of those who had not yet sought genetic counseling ($n = 57$), 35 (61%) answered "Yes," that they plan to seek genetic counseling; a one-sampled chi-square test confirmed that this was significantly larger than "No" ($n = 10$, 18%) and "Unsure" ($n = 12$, 21%) responses ($\chi^2(n = 57) = 20.3, p = 0.00050$).

While resources for either ALS or FTD are appropriate for all persons along the FTD/ALS spectrum, Fig. 4 indicates that people are more likely to seek out the resources specific to the conditions in their family, confirmed by chi-square analyses showing that persons with FTD only or ALS only in the family are more likely to have used just FTD or just ALS resources respectively, while persons with both conditions in the family were more likely to seek out both ALS and FTD resources ($\chi^2(6, n = 88) = 82, p = 1.1 \times 10^{-15}$).

3.3. What are the remaining needs of the community?

3.3.1. Research and treatment

Fourteen open-ended responses (23%) included a desire for more information about the latest research and treatment options. This encompassed requests for results from ongoing trials, and general research updates about the "progress on FTD research and treatments." Some respondents also wanted more information about how to participate in research, such as "it would be helpful to learn specific current or upcoming trials that at-risk or affected individuals can consider." Several individuals had questions about how to access and navigate brain donation options for postmortem research. There was also a desire for assistance in navigating the increasingly complex offerings: "touch on the clinical trials available and explain about how we know if it's a good one to explore. How do we know which one to take part in?" Some respondents noted the need for guidance on the "best therapeutic agents for persons with FTD and what ones not to use." Individuals wanting more research resources included supporters, family members of persons diagnosed, and community members, and it is important to note that 30% ($n = 3/10$) of the at-risk individuals who submitted free-form responses spontaneously mentioned the need for more research information.

3.3.2. Education and resources

Twelve open-ended responses (20%) included requests for additional education and materials, or discussed the need for improvement in this space. Examples included requests for "information on psychologists who are knowledgeable and experiences in FTD and can be helpful to the supportive person," and "a clearer path to resources." Although addressed extensively in the conference, some individuals requested more information about how to access genetic counseling and testing; navigating care can

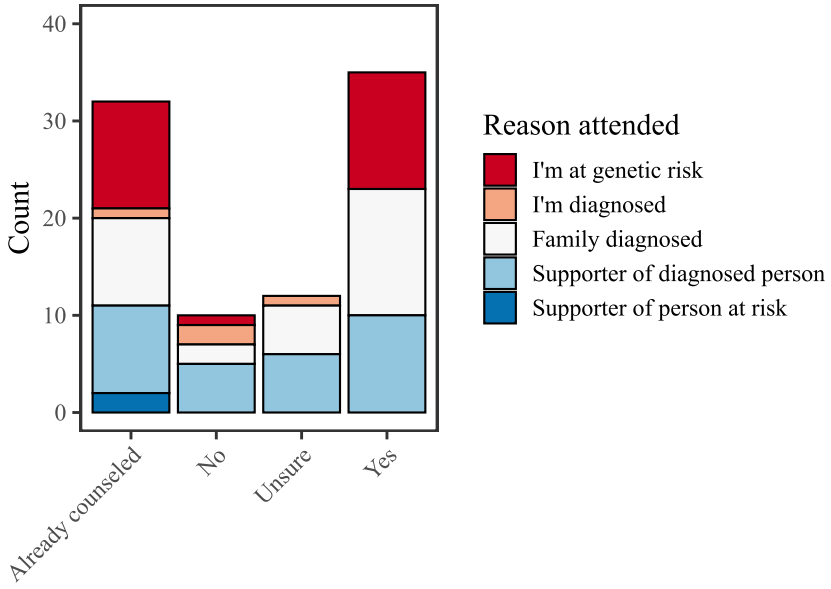

Fig. 3. Participants' self-reported intentions to seek genetic counseling. Participants responses to a probe assessing whether they would seek genetic counseling for themselves or others due to the Familial FTD/ALS Conference are presented based on participant self-characterization of relationship to FTD/ALS.

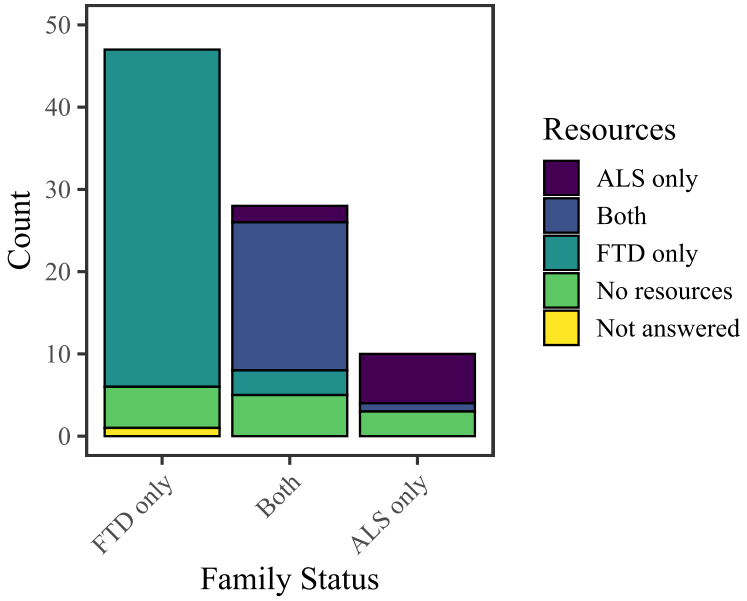

Fig. 4. Previously accessed FTD/ALS resources. Participant responses regarding FTD and ALS resources they have used previously are plotted based on which conditions are present in their family.

be extremely overwhelming, and these comments highlight the need for clear guidance about accessing resources.

Eleven responses (18%) included a request for a deep dive into specific topics. For example, some individuals wanted more information about their family's specific subtype of FTD, such as primary progressive aphasia (PPA). Others requested more information about family communication, reproductive options, legal considerations, and topics related to caring for a person diagnosed. Several responses noted that the information presented in the conference was most helpful for those early in their FTD/ALS journeys and requested information for those further along in the process. One participant stated, “It's almost like there should be two tracks for events like this – newbies and long-haulers.”

Seven responses (12%) included a desire to know what to expect from the diseases. These comments were all made by a supporter or family member of a person diagnosed. More information was requested about “Everyday life stages” and “Warning signs of disease progression as well as a timeline of progression.” This concern is common to all FTD/ALS community members, regardless of the genetic status of the patient. These sentiments underscore the uncertainty that comes with a diagnosis of ALS or FTD, and the need for continuing conversations between families and their neurologists throughout the diagnostic and disease process. See Supplemental Table 1 for resources that may help clinicians and/or community members navigate FTD/ALS spectrum disorders.

3.3.3. Sharing personal stories

Seven individuals (12%) shared personal stories via the open-ended prompts. This included detailed information about individuals affected in a family, stories of families' genetic testing experiences, expressions of concern related to disease in the family, and comments about specific hurdles

or supports families have identified in their own journeys. For example, a participant shared:

"I really enjoyed it. We have lost 6 to ALS/FTD and we have 2 living with it now. We have [many] living descendants from my grandparents, so it will strike again. I join these events, learn all I can and then update my extended family every couple of years. This was confirming of other information I already knew and provided some additional info I didn't know. I've been studying this for 14 years now. Thank you!"

These responses indicate a need for platforms for connection and expression.

4. Discussion and conclusion

4.1. Discussion

This study aimed to assess whether our novel conference met the educational and support needs of individuals diagnosed with or at risk for familial FTD and/or ALS, identify what resources the community is currently accessing, and determine remaining gaps in care. There is need, not only to develop resources for at-risk/diagnosed communities, but to evaluate those resources to ensure efficacy: survey responses indicated that the conference best served those directly affected by the disease (whether at genetic risk for or diagnosed with FTD/ALS). Individuals do not appear to be maximizing the full breadth of resources available, but indicated they were more likely to access genetic counseling because of the conference. Finally, qualitative data suggest there remain unmet needs in this community, such as psychosocial counseling, education about the conditions, and connection with research.

Perhaps the most valuable, but likely underutilized, resource in existence for members of families with hereditary FTD/ALS is genetic counseling. Studies across specialties consistently demonstrate the utility and efficacy of genetic counseling. Examples include increased empowerment, self-efficacy, knowledge of management/surveillance recommendations, as well as reduced anxiety and guilt for patients and family members [27–30]. This conference showcased how genetic counseling can be valuable to anyone impacted by familial FTD/ALS and can help with the decision of whether to pursue genetic testing; many individuals reported they were likely to seek genetic counseling after attending the conference. Given that FTD and ALS are conditions on an overlapping molecular and clinical spectrum, resources for ALS or FTD may serve both communities. Even so, our results indicated that individuals were more likely to seek out only those resources that are related to the condition that has presented in their family. This finding highlights the need for increased awareness about advocacy and education resources for the continuum of FTD/ALS, so that families can maximize their use of resources given the limited number available. Still, health care providers must tailor the resources they provide to the needs of the individuals, even as needs fluctuate over time. For example, when a person begins family planning, they may request information about alternative reproductive methods, or when a loved one's symptoms progress, the family may begin to think about brain donation.

Individuals at risk for FTD/ALS expressed substantial eagerness to be involved in research, including treatment trials. Many of the current interventional trials require knowledge of one's genetic status. Thus, as more people begin to navigate the decision-making process for predictive genetic testing, the influence of clinical trial availability on testing decision-making needs to be better understood and explored. Additionally, responses demonstrated that the growing number of available studies, while hopeful for advances in treatment, simultaneously creates another decision-making dilemma for families. It is crucial that neurologists and other team members initiate these conversations and are prepared to address questions about research and treatment, with options that are individually tailored.

Open-ended responses revealed a desire for more information about what to expect in the future. The uncertain nature of illness can surface in the form of ambiguous loss, a loss that is not concrete or as clearly definable as death [31]. Providers can assist with navigating this uncertainty. For example, neurologists can help individuals understand the typical symptoms of FTD and ALS to prepare and anticipate potential needs. Genetic counselors are trained to facilitate adjustment to uncertain information or circumstances, which commonly arises when considering genetic conditions and risks [32]. Key counseling strategies include clear and honest communication about uncertainty, adapting counseling to the needs of the person in that moment, and focusing on what is known despite the remaining uncertainties [32]. Managing uncertainty is an essential component to adaptation, and longer-term therapy may also be beneficial to help individuals learn to live and cope with the inherent uncertainty of their situations [33,34].

Comments about connecting with the panelists' stories, desire for longer break-out sessions, and people stating that the conference made them feel less alone in their journeys suggest that our conference and other resources should include personal story telling and interactive group activities, rather than solely academic lectures. We observed that many people used the open-ended questions as a platform to share their stories, and this was also true of the conference break-out sessions. Narrative storytelling can be both therapeutic and a tool for advocacy, which may lead to meaning making and regaining a sense of mastery or control, essential components of the coping process [35,36]. Narrative groups have been positively received in the Huntington's disease community [37] and may be a particularly effective therapeutic approach for familial FTD and ALS, as a genetic condition or test result can impact many family members simultaneously [38]. From our clinical experience and responses to this survey, it appears that building a sense of community and accessing relevant resources can be particularly challenging for asymptomatic at-risk individuals. There is a clear need for more support groups (especially virtual groups that can reach a broad audience including those in rural or underserved communities), as well as therapists knowledgeable in these conditions and additional forums for individuals to share their stories and experiences with FTD/ALS.

4.2. Innovation

Our study explored the utility of a novel conference dedicated to genetics issues in familial FTD and ALS. Despite the increasing availability of predictive genetic testing, there has been little previous exploration into the needs of individuals living at risk of developing FTD and ALS, or other at-risk communities. There may be shared factors among people living at risk for a variety of conditions, which should be taken into consideration with future resource development. For example, the UPenn neurogenetics team offers a quarterly, virtual, "positive predictive" support group for asymptomatic persons at-risk for a variety of adult-onset neurodegenerative conditions. We have found that while the symptoms of the diagnoses may differ, there is shared experience related to living at-risk. This is another reason why merging ALS and FTD at-risk communities is beneficial: while the conditions differ (though fundamentally connected), the at-risk experience is shared. Often the ALS and FTD communities operate in isolation, which is a missed opportunity for shared resources. ALS and FTD advocacy groups and professional societies should consider joint patient/family education initiatives such as our conference, as well as professional workshops and clinical trial planning initiatives. This study serves as a foundation for future joint resource development, as we have demonstrated perceived value of a conference targeted to the full FTD/ALS spectrum.

4.3. Study limitations

This study was limited by virtual methods for contacting conference attendees. For example, if several individuals from one household watched the conference together, only one email address was collected for the group. Surveys were sent to registered email addresses ($n = 326$) and email addresses used to log into the conference for those who were not registered ($n = 50$), and therefore some attendees were not sampled nor included in the survey response rate. Inter-coder reliability was not calculated for qualitative coding, as we used consensus to apply multiple codes to each response. Limited demographic data were collected as part of this survey. In the future, we will collect additional demographic

information, including race, ethnicity, socioeconomic status, and education level to evaluate potential disparities in access to resources or differing needs. We will consider implementing pre- and post- conference surveys to better capture how the conference impacts individuals' knowledge and attitudes regarding genetic counseling and testing, research, and other topics. The needs and desires of individuals who were motivated to attend this conference and complete the survey may differ from others in the general FTD/ALS community.

4.4. Conclusion

While the Penn Familial FTD/ALS Conference is an important addition to the resource arsenal of this community, there is a clear need for more resources spanning research and treatment, education, support, and more, especially for those at-risk. Genetic counselors are crucial to the development and dissemination of resources, as they work to assess the needs of their patients, and to fill any identified gaps such as through connection forums and educational resources. Our study demonstrated the diverse needs of those facing familial disease, and the demand for knowledge in a rapidly advancing field. Still, access to genetic counseling may be limited: neurologists and other care providers should refer individuals who are at-risk or diagnosed with FTD or ALS to genetic counseling. Given limitations of genetic counseling resources, alternative service delivery methods for genetic counseling should be developed and pursued.

Author contributions

All authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Author KAQC confirms that she had full access to all data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis. **Laynie Dratch**: conceptualization, methodology, investigation, data curation, writing – original draft preparation, and writing – review and editing. **Weiyi Mu**: conceptualization, methodology, writing – review and editing. **Elisabeth McCarty Wood**: conceptualization, methodology, writing – review and editing. **Brianna Morgan**: writing – review and editing. **Lauren Massimo**: writing – review and editing. **Cynthia Clyburn**: writing – review and editing. **Tanya Bardakjian**: writing – review and editing. **Murray Grossman**: conceptualization, methodology, supervision, project administration, writing – review and editing. **David J. Irwin**: supervision, writing – review and editing. **Katheryn A. Q. Cousins**: data curation, software, formal analysis, visualization, writing – original draft preparation, and writing – review and editing.

Human studies and informed consent

This study was determined to be a quality improvement initiative that does not meet the definition of human subjects research per the UPenn IRB. No formal consent process was needed; individuals provided implied consent with voluntarily submitted responses. We confirm all patient/personal identifiers have been removed or disguised so the patient/person (s) described are not identifiable and cannot be identified through the details of the story.

Data availability statement

The data that support the findings of this study are available within the article and from the corresponding author upon reasonable request.

Declaration of Competing Interest

Laynie Dratch receives consulting fees from Passage Bio and has received honoraria from the Muscular Dystrophy Association (MDA) and NSGC. Cynthia Clyburn has received honoraria from the MDA. Tanya Bardakjian is employed by Sarepta, a gene therapy company, and has financial relationships with Novartis, Invitae, Vigil Therapeutics, and Genome Medical. Murray Grossman participates in treatment trials sponsored by Passage Bio, Alector, and Prevail, and is a member of the Medical and Scientific Advisory Board of AFTD; he also receives funding from NIH and Department of Defense. Brianna Morgan receives funding support from the NIH and P.E.O. International. David J. Irwin is on the scientific advisory board of Denali Therapeutics. None of these sources of funding represent a conflict of interest. Katheryn A. Q. Cousins, Weiyi Mu, Elisabeth McCarty Wood, and Lauren Massimo declare no conflicts of interest.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.pecinn.2022.100108.

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