"IT TAKES AWAY WHAT MADE A PERSON THAT PERSON": RECONSTRUCTING IDENTITY IN RESPONSE TO GENETIC RISK OF BEHAVIORAL VARIANT FRONTOTEMPORAL DEMENTIA

by
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Abstract

There is limited research considering the personal experiences of individuals living with or at risk for developing behavioral variant frontotemporal dementia (bvFTD). FTD is known to have a significant genetic contribution. With advances in understanding the genetic component of FTD, rates of predictive and symptomatic genetic testing will increase. Though FTD is the second most prevalent cause of early-onset dementia after Alzheimer disease (AD), there is substantially more literature related to AD experiences. In dementia research broadly, care partners rather than persons diagnosed tend to be the informants. The data presented in this paper are derived from an exploratory project called Voices of Individuals: Challenges and Experiences of Behavioral Variant Frontotemporal Dementia (VOICE Of bvFTD) that qualitatively explores the specific experience of living with bvFTD diagnosis and risk from the perspective of the persons diagnosed and at-risk. Semi-structured telephone interviews were conducted to qualitatively explore how bvFTD may influence an individual's sense of identity, individuals' experiences of loss, how individuals cope and adapt to the challenges they face, and other factors that might be perceived to influence these processes. Participants with a diagnosis of bvFTD (n=6) and with genetic testing results conferring high risk of developing bvFTD (n=14) were recruited through two academic medical centers, the National Institutes of Health, ClinicalTrials.gov, and through FTD support resources. Interviews were transcribed and underwent thematic analysis. This paper considers how identity was shaped by the threat of developing FTD among the at-risk subset of VOICE Of bvFTD participants (n=14). Participants varied in their conceptualizations of their status, how they integrated FTD risk information into their current identity, and how they

anticipated changes to their identity in the future. FTD risk raised fundamental issues related to what constitutes the essence of a person, challenged people to wrestle with Cartesian dualism, and exposed the roles of time, social relationships, and social roles in the understanding of the nature of self. Understanding how at-risk individuals reconstruct their identities in response to their FTD risk status can inform clinical care, resource development, and future research.

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Introduction

Frontotemporal Dementia

Frontotemporal dementia (FTD) is a neurodegenerative disease characterized by progressive atrophy in the frontal and temporal regions of the brain that causes progressive changes in behavior, personality, language and cognition (Deleon & Miller, 2018). FTD is the second most prevalent cause of early-onset dementia after Alzheimer disease (AD) (Piguet et al., 2011). There is a significant genetic contribution to FTD (Rohrer et al., 2015; Warren et al., 2013; Onyike & Diehl-Schmid, 2013). Though there are multiple FTD phenotypes, this research focuses on the type that is both the most common and the most likely to be inherited: behavioral variant frontotemporal dementia (bvFTD) (Deleon & Miller, 2018; Seelaar et al., 2008).

There is a growing understanding of the link between FTD and amyotrophic lateral sclerosis (ALS). A pathogenic repeat expansion of *C9ORF72* is the most common mutation associated with familial FTD and FTD/ALS; it explains much of the co-occurrence of FTD and ALS in the same individual or within a family (Fong, Karydas, & Goldman, 2012). There are several other genes that impart dual risk for FTD and ALS including *TARDBP* and *VCP* (Synofzik et al., 2014; Guerreiro et al., 2015), and other genes that are associated with risk for just one condition, such as *GRN* which is only associated with FTD risk (Nguyen et al., 2018). Though FTD and ALS experiences are often intertwined, the Voices of Individuals: Challenges and Experiences of Behavioral Variant Frontotemporal Dementia (VOICE of bvFTD) project and this manuscript primarily focus on FTD experiences.

Individuals with bvFTD experience a progressive loss of interpersonal and executive skills. Persons diagnosed may exhibit disinhibition and personality change through inappropriate comments, hyperorality, lack of personal hygiene, excessive gambling, apathy, repetitive or compulsive behaviors, blunted affect, lack of empathy, and mental rigidity (Piguet et al., 2011; Warren et al. 2013; Deleon & Miller, 2018). Often, the beginning stages of disease resemble personality disorder-like changes, with later progression causing speech/language and cognitive loss. Many of the early symptoms involve breaking social norms, which presents challenges for care partners, loved ones that provide the primary emotional and practical support for the affected individuals (Massimo, Evans, & Benner, 2013a), in addition to the patients themselves. At the time of bvFTD onset, individuals are often in a relationship with dependent children and have a career upon which the family may be financially dependent (Shnall et al., 2013; Merrilees & Ketelle, 2010), causing unique social and financial challenges.

Greater understanding of the genetics of FTD has allowed for predictive genetic testing for unaffected family members of affected persons (Benatar et al., 2016). The Huntington disease (HD) protocol is currently the recommended guideline for predictive testing for FTD and other neurodegenerative diseases (Sorbi et al., 2012). As Crook et al. (2017) note, few studies to date have focused on issues related to predictive genetic testing for FTD or ALS, and to the best of our knowledge our study is the first to focus specifically on individuals at risk for both ALS and FTD due to genes that cause both diseases such as *C9ORF72*. The majority of the previous studies that focused on predictive testing for ALS and FTD were quantitative in nature and were most often designed to determine whether predictive testing is safe for this population (Molinuevo et

al., 2005; Steinbart et al., 2001; Paulsen et al., 2013), rather than understanding the broader psychological and social impact. As predictive genetic testing and diagnostic testing are increasingly utilized, many individuals have become aware of their likely future disease progression prior to the onset of the most severe or challenging symptoms. The group of individuals with knowledge of their genetic status is an emerging population that will continue to grow as better understanding of the genetics and continued improvement of diagnostic criteria improve the ability to identify individuals with bvFTD.

Dementia Worry, Personhood, and Patienthood

Dementia is a commonly feared diagnosis for many reasons. Dementia worry is a phenomenon that has been documented in healthy individuals with and without family history of dementia, as well as in those with early stages of AD (Kessler et al., 2012; Clemerson, Walsh, & Isaac, 2014). A key element of dementia worry is the threat to personhood and identity: dementia threatens the physical self, the identity as a human being, as well as the perception of a shared reality with others (Kessler et al., 2012). In the context of dementia, "personhood" describes a person's sense of self as an autonomous and whole entity who has purpose, self-worth, can experience peace and joy, can think, feel, make consistent choices, express individuality, and participate in meaningful social interactions (Touhy, 2004). Personhood is shaped both by a person's internal sense of self and by social factors such as how a person is viewed and treated by others (Boykin & Schoenhofer, 2001). The social context may include family, friendships, employment, and even research and clinical care contexts; social factors can be particularly threatening to personhood in the context of dementia (Kitwood, 1997).

One type of threat to personhood is "patienthood," which refers to a person's diagnosis and status as a patient becoming defining features of identity and narrative (Sabat et al., 2011). Centrality of patienthood to identity facilitates marginalization and negative stereotyping by creating a focus on difference and otherness (Sabat et al., 2011). Patienthood may present a threat to personhood by undermining one's sense of wholeness, self-worth, or other elements of personhood previously described. Diseases such as dementia that result in loss of independence, loss of skills, and need for frequent medical visits may be especially prone to cause a sense of patienthood.

Identity and the Self

The concepts of identity and self are difficult to define and related research has been based on a number of different models (Caddell & Clare, 2010). Various terms such as self, selfhood, identity, and personhood attempt to capture the essence of the individual (Tolhurst et al., 2014). In developmental and personality psychology, identity is often defined in terms of cognitive self-structure, who people believe themselves to be (Topolewska-Siedzik & Cieciuch, 2019). The majority of identity development research has focused on adolescence, as that is thought to be the most dynamic period in identity formation, although identity continues to develop across the lifespan (Topolewska-Siedzik & Cieciuch, 2019; Crocetti, 2018; Kroger et al., 2010). In a quantitative study of 3,216 healthy individuals aged 18-65 years, Topolewska-Siedzik and Cieciuch (2019) found that identity structure becomes more cohesive and well-functioning with age, and that role definition peaks in middle and late adulthood, the time at which bvFTD most often strikes and disrupts established roles. With genetic testing becoming more commonly pursued among diagnosed individuals, early to middle adulthood is often

when relatives of diagnosed persons first learn of their risk status and when they are first presented with the opportunity to pursue genetic testing. Thus, bvFTD presents a threat to established identity, and bvFTD risk often first appears as a threat in a formative period of identity construction.

Identity is formed by the individual's perception of the self as a continuous being, through past and present attributes, and through interactions with the social world that foster social identities (Tolhurst et al., 2014; Sabat et al., 2011). Dementia threatens identity by challenging all core pieces of what is thought to make a person a person. Little is known about how individuals at risk for developing dementia perceive, react, and adjust to these threats.

Several qualitative studies summarized by Spreadbury and Kipps (2017) have shown not only that early-onset dementia poses a threat to identity, but also that the impending loss of self may be sufficient to elicit grief responses. Individuals with early stages of young-onset dementia and their unaffected relatives express uncertainty surrounding loss of personhood; people fear the end of the self rather than physical death and are unable to imagine who they "will be" or how they will "think" if they develop dementia (Roach et al., 2008). This relates to a specific type of loss known as ambiguous loss in which a person is physically present yet psychologically absent (Boss,1999). Individuals with or at risk for developing bvFTD may feel a sense of loss regarding their identity and previously held ideas of the future. The very nature of FTD raises questions that reflect broader philosophical and psychological questions about the relationship between the mind, the body, and identity. Kitwood and others who have theorized about personhood as described above work within the Cartesian personhood framework that

separates the mind from the body, known as Cartesian dualism; in the context of dementia, the Cartesian perspective suggests that the person with dementia is mindless (Dewing, 2008). Since one aspect of personhood is the perception of the self as a continuous being, loss of the mind, and therefore cognition and memory, is a direct threat to personhood.

Exploration of these topics is crucial for the development of supportive interventions (Rentz, Krikorian & Keys, 2005). Individuals at risk for bvFTD may have experiences with redefining identity that differ in meaningful ways from those at risk for other more commonly studied neurological diseases such as Alzheimer disease due to the characteristic FTD presentation of changes in personality and social functioning, impairments that go beyond cognitive decline. While there has been some research that quantitatively assesses psychological outcomes after predictive genetic testing for neurodegenerative conditions in order to establish the safety of conducting such testing (Molinuevo et al., 2005; Steinbart et al., 2001; Paulsen et al., 2013), these studies have failed to consider more nuanced effects of learning risk status, and very few studies have considered those at risk for developing FTD. Thus, consideration of how identity reconstruction occurs after learning genetic risk for bvFTD is warranted.

Rationale for Current Study

FTD is incompletely understood from a clinical, genetics, psychological, and social standpoint. There has been little learned about the psychological impact of awareness of genetic risk for FTD. Improvements in clinical criteria and increased availability of genetic testing are resulting in more diagnoses being made at earlier ages, and more family members will be recognized as being at-risk. Individuals will

increasingly learn of their high risk for developing FTD prior to disease onset or decline in awareness with predictive genetic testing, and therefore attention is needed to address potential psychological and social implications of living at risk for bvFTD. Importantly, this is an emerging population for which clinical trials are becoming available. As more is learned about FTD and the growing promise of possible future drug therapy trials for at-risk individuals, rates of predictive genetic testing are likely to increase (Crook et al., 2017). Since more individuals will be identified as being at risk for FTD and at earlier ages, it is important to further our understanding of individuals' experiences and needs.

There has been a recent movement in research towards understanding the 'lived experience' of people with young-onset dementia, although efforts thus far have focused specifically on the diagnostic experience (Spreadbury & Kipps, 2017; O'Malley et al., 2019) and have not considered the experience of at-risk persons who have learned their genetic status. There has not yet been an in-depth investigation of how people at risk for developing bvFTD experience and adapt to their changing identity and threat to personhood, or how they might experience loss. Thus, the VOICE Of bvFTD project aimed to gather a deeper understanding of experience to better inform clinical practice, resource development, and future studies.

At its core, genetic risk for FTD raises fundamental questions for participants about their sense of self and identity. It brings out theoretical issues related to what constitutes the essence of a person, challenges people to wrestle with Cartesian mind-body dualism, and exposes the roles of time, social relationships, and social roles in our understanding of personhood. For individuals at known genetic risk of developing FTD, the potential for altered identity is twofold: change in identity due to the nature of the

FTD disease process and change in identity as a person living at risk. Thus, at-risk individuals must grapple with how identity may change with disease onset in the future, as well as how identity has and continues to change as a person living with knowledge of their FTD risk. In this paper we explore how FTD and the related disorder ALS present threats to identity, discuss the questions this raises for people at risk such as the uncertainty of when symptoms will develop and associated emotional valence of dread, and consider how centrally individuals incorporate their risk status into identity.

Methods

Participants

The participants of this study are a subset of the cohort of participants recruited for the VOICE Of bvFTD project. For the VOICE Of bvFTD project, individuals with a diagnosis of bvFTD or with genetic testing results conferring high risk of developing bvFTD were recruited through two academic medical centers, the National Institutes of Health, ClinicalTrials.gov, and through FTD support resources. There were two sets of inclusion criteria for VOICE Of bvFTD: one for diagnosed individuals and one for at-risk individuals. Eligible diagnosed adults were those who had a possible or probable diagnosis of bvFTD as per diagnosis by a neurologist, psychiatrist, or group consensus in a specialized dementia center if recruited through a clinical or clinical research site, or by self-report if through another site. Such individuals must have had symptom onset not more recently than two months prior to the interview date. Eligible at-risk adults were those who had genetic testing that identified a pathogenic variant in a gene known to confer high risk of developing FTD or a known familial mutation associated with FTD, confirmed by the individual's dementia or genetics provider or researcher, or by self-report if recruited through non-clinical or research sites. Such individuals must have had the genetic testing result disclosed not more recently than two months prior to the interview date. All research participants had to be 18 years of age or older and had to speak fluent English. This paper includes data from the at-risk participant cohort from the VOICE Of bvFTD project.

Procedure

Individuals from the two academic hospital recruitment sites who met inclusion criteria were identified from institutional databases by genetic counselors and researchers who assisted with recruitment for the VOICE Of bvFTD project at each site. Recruiters at the academic hospitals had clinical or research relationships with the individuals they were recruiting. An investigator at the NIH with extensive experience in neurodegenerative disease research identified individuals that met inclusion criteria from existing NIH dementia research cohorts. Potential participants were contacted through their existing clinical or research relationships via mail, e-mail, or telephone based on available contact information and preference of the recruiter. Any participant recruited through an academic hospital signed a HIPAA authorization form prior to the consent process. Individuals at the Penn FTD Center Caregiver Conference were provided with study flyers and contacted one of the authors (LD) if interested. Posts were made to ClinicalTrials.gov and a private *C9ORF72* Facebook group.

All study participants were provided with a written document that outlined the information covered in the verbal consent conversation prior to the consent phone call. Participants were encouraged to share the consent document with family members or support persons as they decided whether they would like to participate. Participants were also provided with a demographics form that they were asked to fill out prior to the consent phone call. Every participant participated in a consent phone call with LD. During this phone call, LD reviewed the consent information and confirmed eligibility by confirming inclusion criteria via collecting demographics information, as well as performing a consent comprehension assessment (Appendix I). After verbal consent was

given for the study, participants were scheduled to have a telephone-based interview with LD. For participants recruited from academic sites, basic medical information was collected from recruiters at the academic institutions after receipt of a signed HIPAA authorization form. For participants recruited from other sites this information was self-reported as part of the demographics survey.

To provide additional descriptive information on participants, just prior to the interview, each completed a brief nondiagnostic cognitive assessment via the Telephone Cognitive Screen (T-CogS) and insight probe after confirming that they still wanted to participate in the study. Each participant completed an approximately 40-90-minute interview that was audio-recorded and later transcribed. Phone interviews were conducted by LD between June and August of 2019. The interview was conducted using a semi-structured interview guide. If diagnosed participants requested that their care partner be with them during the interview then the care partner was allowed to join without responding to the interview questions. Three diagnosed participants had care partners present during their interviews for question clarification if needed. Several diagnosed participants completed the interview alone but had care partners who facilitated the research connection and were accessible if necessary during the interview. Immediately after the interview, the interviewer (LD) recorded notes on her reactions to the interview, major themes that arose, questions that did or did not resonate with the participant, and suggestions for future interviews.

Approximately 129 individuals were contacted about the VOICE Of bvFTD project. Thirty-seven individuals were reached during recruitment and expressed interest in participating in the study. They were sent study information packets and an invitation

to schedule consent phone calls. One individual declined participation at this point. Five individuals were identified as ineligible: two individuals were unable to consent due to their inability to pass the comprehension assessment and therefore did not meet inclusion criteria, and three individuals did not meet other inclusion criteria. Four individuals were lost to follow up. Once twenty interviews were completed, recruitment was paused. The remaining seven individuals are on a recruitment waitlist. Therefore, not including the individuals on the waitlist, the current response rate is approximately 15.5% (20/129). Of the twenty total participants, eleven were recruited from the University of Pennsylvania, seven from the NIH, one from a conference, and one from a Facebook group. Table 1 depicts recruitment by site and participant category. Twenty interviews and questionnaires were collected; fourteen were from participants with genetic risk for developing FTD, and six were from participants with an FTD diagnosis.

Table 1. VOICE Of bvFTD recruitment distribution by site and participant category.

Recruitment Site	Attempted Recruitment	Recruitment Responses	At-Risk Participants	Diagnosed Participants	Total Participants
Johns Hopkins	14	2	0	0	0
University of Pennsylvania	33	16	5	6	11
NIH	17	10	7	0	7
Conference	~30	2	1	0	1
ClinicalTrials.gov	unknown	5	0	0	0
Facebook group	~35	2	1	0	1
Total	~129	37	14	6	20

Instrumentation

Telephone Cognitive Screen (T-CogS): The T-CogS is a brief, valid, telephone-adapted version of the Mini-Mental State Exam (MMSE) (Kennedy et al., 2014). It takes up to ten minutes to complete (Newkirk et al., 2004). The T-CogS was modified slightly to reflect the updated telephone technology in question-wording and permitted responses. The T-CogS can be found in Appendix II. T-CogS scores were used to characterize the population and to allow for a measure to assess insight described below.

Insight Probe: Two questions were asked that prompted the participant to consider his or her performance on the T-CogS. While this was not a comprehensive assessment of insight, it provided a glimpse into the individual's awareness of his or her

mental capabilities. This probe took about 2 minutes per participant. The insight probe can be found in Appendix II. The purpose of this probe was to get a limited but basic understanding of to what extent insight was preserved, especially among diagnosed participants. This provided context for interpretation of the diagnosed participants interviews. If diagnosed individuals were to perform poorly on the T-CogS or insight probe, this would provide a nuance to consider when interpreting their interview responses.

Demographic Survey: Basic demographic information was collected from all participants. The survey also included several questions about experience with FTD and ALS such as whether there are other affected individuals in the family. See Appendix II.

Medical Information: Limited medical information was collected about participants. Information collected included diagnosis type (bvFTD or asymptomatic), genetic status (whether they have had genetic testing or not, positive or negative if had genetic testing) and which gene harbors the mutation if positive.

An interview guide was developed to reflect the specific aims of the study. The guide consisted mostly of open-ended questions with follow-up prompts to elicit specific information from the participants. Two versions of the guide were created such that the wording reflected the status of the participant as diagnosed or at-risk; both guides are available in Appendix III. The interview guide focused on experiences with FTD, but given the overlap between FTD and ALS, some experiences with ALS were explored. The major sections of the interview guide centered upon the FTD journey, personhood and identity, coping and management, future orientation, mental health, and overall experience.

Analysis

Interviews were audio-recorded. All interview recordings were transcribed by a third-party transcription company. Transcripts and observation notes were uploaded into MAXQDA, qualitative software that facilitated the coding and analysis process. Interview transcripts were coded and analyzed solely by LD using thematic analysis to identify common patterns and themes among the interviews. Interviews were analyzed in order to describe the ways that participants perceive their identity, discuss experiences of loss, and the coping strategies used to overcome the challenges posed by the disease. Several a priori codes were created based on topics from the interview guide, such as "coping mechanism," "reasons for testing" and "identity." The codebook was expanded with codes that emerged from the data, such as "genetics identity," "mortality," and "control." The preliminary codebook was applied to several transcripts from participants who were classified as at-risk and diagnosed. As transcripts were coded, the codebook was revised to include emerging concepts and previously coded transcripts were re-coded to incorporate any new codes. Eventually, code saturation was reached with no additional codes or concepts identified in subsequent transcripts (Hennink et al., 2017) and therefore the final codebook was confirmed. Though the codebook was developed and applied by a single coder (LD), the coder consulted with JO and LE, who have extensive experience in qualitative research, throughout the process, sharing coded segments of transcripts and discussing interpretations.

Qualitative analysis was performed using thematic analysis, which involves considering patterns and themes across the data. We investigated in what context the codes arose in the data, and how concepts may be interacting. Themes were analyzed

within the context of participants' experiences living with or at risk for FTD. Coded data within each theme were reviewed to select representative quotations from participants. Data collected from the demographic questionnaire and T-CogS/Insight probe measure was used to characterize the population. Data were analyzed across all interviews but with consideration of ways in which those who have developed symptoms may have differing experiences and viewpoints than those who have not developed symptoms.

During the coding and analysis process for the VOICE Of bvFTD project it became clear that while there were some similarities in experience between at-risk and diagnosed participants, there were also many nuanced differences that made it difficult to consider at-risk and diagnosed experiences together. Consequently, we decided to solely consider the at-risk participant experience in this manuscript; the diagnosed experience will be important to explore in a future paper. Thus, this manuscript only considers the at-risk subset of the VOICE Of bvFTD project participants (n=14) in the analysis. Interviews uncovered many themes, not all of which are described herein. This paper will focus on how at-risk participants conceptualized their identity and how those characterizations had been informed by their risk of developing FTD. Specifically, this paper considers the dual threat to identity for at-risk participants: the impact of the disease itself and the uncertain status of being at risk. In order to explore these concepts, several codes were examined including identity, genetics identity, sick vs. healthy, ALS vs. FTD, and status acknowledgement. Interactions between these codes and codes for fears, imagined future, and finding optimism/hope/strengths were considered. Representative quotations were identified after fully exploring each theme. Demographic information was used to consider whether individuals with shared characteristics, such as

time since genetic testing and age, seemed to share elements of their experiences. Broadly, experiences did not seem to differ based on these characteristics, but relevant observations are noted when applicable.

Results

This manuscript considers only the data from at-risk participants (n=14) in the VOICE Of bvFTD project cohort. At-risk participants had genetic testing between several months and seven years prior to study participation, with a mean of 2.07 (SD= 1.87) years since testing at time of participation. The study sample was 100% Caucasian (n=14), comprised of more females (n=10) than males (n=4), and well-educated with all at-risk participants having a college (n=5) or graduate (n=9) degree. Participant ages ranged between 31-63 years. All participants scored above 23 on the T-CogS, corresponding with the cognitively normal range on the MMSE. All participants had good insight into their performance on the T-CogS with most (n=10) estimating their exact score and the remaining (n=4) estimating only one point off from their actual score. For a summary of pertinent study sample characteristics, see Table 2.

Table 2. Demographics and summary of characteristics of at-risk study sample.

Characteristic	At-Risk Participants (N=14)	
Range of age at recruitment (years)	31 - 63	
Gender (N, %)	Female (10, 71.4%) Male (4, 28.6%)	
Race (N, %)	White/Caucasian (14, 100%)	
Gene Involved (Gene – N)	*C9ORF72* - 11 *GRN* - 2 *TARDBP* - 1	
Education (Highest Achieved – N)	College graduate – 5 Graduate degree – 9	
Range of age at time of testing (years)	30 - 56	
Approximate years since testing Average (Range)	2.07 (less than 1 – 7)	
T-CogS Score[1] (Score – N)	26 25 24	N=7 participants N= 6 participants N=1 participant
Self-Perceived Symptoms[2] (Confidence – N)	0 0-1 1	N=12 participants N=1 participant N=1 participant
Doctor-Perceived Symptoms[2] (Confidence – N)	0	N=14 participants

1. Telephone Cognitive Screen (T-CogS): Higher scores convey better performance. Range 0-26. Scores of 24-26 indicate normal cognition, and 17-23 indicate mild cognitive impairment.
2. Symptom perception number line: Participants were asked to endorse how confident they are and how confident they think their doctors are regarding their symptom status. 0 indicates confidence that the participant does not have symptoms, 2 indicates complete uncertainty about whether they have symptoms, and 4 indicates confidence that they do have symptoms.

To protect the identities of the individuals who participated in this study, participants were assigned pseudonyms. These pseudonyms and characteristics of the individual participants can be found in Table 3.

Table 3. Individual at-risk participant characteristics.

"Name"	Status	Age	Sex	Marital Status	Children (Yes or No)	Career Status	Time Since GT (Yrs)	Gene
Ella	At-risk	59	F	Married	Yes	Retired	3	*C9ORF72*
Anna	At-risk	63	F	Single	No	Retired	7	*C9ORF72*
Mary	At-risk	54	F	Married	Yes	Full-Time	3	*C9ORF72*
Leigh	At-risk	46	F	Married	Yes	Full-Time	3	*C9ORF72*
Drew	At-risk	47	M	Married	Yes	Full-Time	4	*GRN*
Mia	At-risk	44	F	Married	Yes	Full-Time	0	*C9ORF72*
Britt	At-risk	31	F	Married	No	Full-Time	1	*C9ORF72*
Nancy	At-risk	32	F	Single	No	Full-Time	0	*GRN*
Frank	At-risk	36	M	Married	Yes	Full-Time	0	*C9ORF72*
Eva	At-risk	49	F	Married	Yes	Part-Time	1	*C9ORF72*
Steph	At-risk	39	F	Married	Yes	Full-Time	1	*TARDBP*
Ally	At-risk	46	F	Married	Yes	Full-Time	2	*C9ORF72*
Luke	At-risk	44	M	Married	Yes	Full-Time	3	*C9ORF72*
Jeff	At-risk	31	M	Married	No	Full-Time	1	*C9ORF72*

Legend: F=female, M=male, Yrs=years, GT=genetic testing

Like any individual who has had predictive genetic testing for FTD, all participants had at least one family member (parent or sibling) with FTD, ALS, or both diseases. Thus, almost all participants had cared or were actively caring for someone with these conditions and therefore were affected by FTD in ways that had little objectively to do with their risk status. Participants' level of involvement in the care of their affected family members varied greatly. For example, some individuals were the primary care partners or legal guardians for parents or siblings and therefore had daily contact with their family members with FTD, whereas others lived far from diagnosed relatives and saw them infrequently with little to no involvement in their care. One participant, Eva (female, age 49), only had family history of ALS, not FTD, and several others had more

contact with family members who had ALS than those with FTD or FTD/ALS. All participants who had relatives diagnosed with FTD described a period of confusion when the affected loved one became symptomatic with FTD. This often began as observed differences in behavior and personality, as is typical of the disease course. Learning their genetic status changed the way they were affected by FTD, but how they integrated this information into their identity was undoubtedly shaped by their experience with the diseases in their loved ones.

Participants' motivations for having predictive genetic testing varied. Many participants had testing so that they could provide information to their children, and some had testing so that they could pursue alternative reproductive technologies such as in vitro fertilization (IVF) with preimplantation genetic diagnosis (PGD) to avoid passing down the genetic risk factor. Several participants chose to have testing so that they could participate in research studies. Other people just felt that they had to know their status to move forward with their lives and to be able to plan appropriately once they learned about the genetic risk in the family.

Conceptualization of FTD as a Threat to Identity

At-risk participants universally perceived the FTD disease process as a threat to identity. Frank (male, age 36) called *C9ORF72*-related FTD/ALS his "*worst nightmare of a disease*" that "*forces you to become an infant in an adult body*." Mia (female, age 44) said that FTD "*takes the essence of who you are away-- your personality."* Through witnessing family members' experiences with FTD, participants described the disease as *"soul destroying"* (Ella, female, age 59), and as having the ability to "*completely strip a*

person piece by piece by piece" (Britt, female, age 31). The common theme among these sentiments is the notion of FTD as a disease that reduces or strips away the core of identity. This sense of reduction and the image of becoming an "infant" demonstrates how the person with FTD is deprived of the personhood they acquired through a lifetime. Participants described how FTD causes people to retrograde to an earlier or a bare state as a reverse aging of sorts. Most participants feared losing their independence, again harkening back to the nature of FTD as retrograding. While they used varying language, all at-risk participants conceptualized FTD as a fundamentally reducing disease.

Participants grappled with Cartesian dualism, the philosophical approach that the mind is separate from the physical body. Many participants at risk for both FTD and ALS noted that they have engaged with the dilemma explicitly, as Frank (male, age 36) asked, *"The theoretical question of would you rather be able bodied but lose your mind or keep your mind intact but lose your ability to use your body."* Participants weighed which scenario seemed better or at least less bad. Several participants described how developing FTD would destroy who they are by destroying the mind:

> *Your brain is who you are. Once your brain is destructing at that point, you are there in body and soul; you're not there in your mind…as my body is shutting down, there'll be a progression into somebody I am not now. I will not be the same person I am now…So losing that part for me is like losing me.* (Mia, female, age 44)

Mia and other participants recognized that who they will be is not who they are now due to the disease. Mia's quote refers to an anticipated form of ambiguous loss of the self: the self is physically but not mentally present. This sentiment was also captured by Ella (female, age 59) when she said *"I want to die the same person that I am,"* after she

discussed how FTD would change who she was versus ALS in which the mind remains intact. Because of this feared loss of self, some participants acted to assert or preserve their identity. For example, Mia (female, age 44) decided to create videos for her children to document her life story before she was "gone." Many individuals experienced a threat to the permanency of their identity.

Another consequence of this loss of self is the loss of the self as a social being. Jeff (male, age 31) said:

> *You lose your personality, your sense of self, your sense of self to others. You are not who you were, you do not act in ways that benefit the way others think of you. I think that is scary, just we all are social people and we want to be respected and admired and thought of well, and FTD robs you of that.*

Jeff's description of the impact of FTD echoed the notion of losing pre-disease identity and introduced a social component to the loss. Social roles and relationships are a major way that people define themselves; by altering behavior and personality FTD degrades social status and capabilities.

In addition to the ways that FTD threatens social relationships, participants expressed fears regarding how FTD threatens their future ability to fulfill their social roles as parents and spouses. Frank (male, age 36) discussed his worry about how *C9ORF72*-related disease would strip him of abilities and roles that were a source of pride:

> *It has endangered some of the things that I take pride in as a person… it would force me to rely on other people for everything. It would force me to ask for help. It would eliminate my ability to care for my family and protect my family and maintain the house we live in and the cars we drive and make sure everyone's*

> *safe, like all those things that I take pride in as a husband and a father…and I'd be powerless to stop it. That's been the worst thing to think about.*

Frank's identity as a father and husband was threatened by FTD and ALS. Similarly, Jeff (male, age 31) said he did not want his future children to think of him as an incapable father if he changes due to FTD/ALS. Other participants worried about becoming a burden or embarrassment for their family members. For example, Luke (male, age 44) talked about his fear of a *"slow awkward un-dignifying death that bankrupts my family and drags them through a bunch of crap."* Through use of the term "un-dignifying" this quote also demonstrated the social component to identity perception, as how others perceived his demise mattered to Luke.

"How Will I Know?": Enduring Uncertainty & Dread

While genetic testing can resolve uncertainty related to whether or not an individual has a genetic risk factor for developing FTD, a result conferring high risk simultaneously engenders further uncertainty related to disease development. There is currently no way to predict if or when symptoms will develop, which symptoms will develop, or how severe or long a disease course will be for any one individual. Given that participants perceived FTD as threatening to identity, it is not surprising that all participants at risk for developing FTD described ways in which this uncertainty and dread of developing disease had become part of their lives.

When contemplating how they may change due to FTD, participants used strong language that indicated a sense of dread and anticipated negative downturn. Multiple participants used language such as feeling "*doomed."* Nancy (female, age 32) said,

"What's wrong with me? I'm cursed. I have this gene," and questioned whether she would ever consider herself *"normal"* again. Frank (male, age 36) summed up the impact of this dread: "*the fear of what was to come completely derailed my life."* Frank described how this fear caused crippling anxiety about developing the disease before treatment becomes available, and this resulted in his inability to form and maintain relationships or engage in daily activities without worrying about his risk. The risk status established an uncertain and ominous future that had present impact on participants' lives and self-perceptions.

Eva (female, age 49) described the uncertainty of her risk and her conceptualization of FTD as a monster: *"There's something with glowing red eyes under my bed and I know it's a monster. And it's not going to help me to just put a bedspread over it. There's still going to be a monster under my bed."* Eva expressed her desire to know as much as she could learn about the "monster" that represented FTD; she needed certainty as she was faced with profound uncertainty and a disease so dreadful that it was deemed monstrous. Many other participants similarly characterized the uncertainty their mutation status rendered as unsettling. Due to this uncertainty, participants were reminded of their mortality. Like many other participants, Drew (male, age 47) described a shift in his sense of self in which mortality became more salient, feeling his "*time horizon being foreshortened*." Jeff (male, age 31) described how he viewed both his *C9ORF72* status and FTD/ALS: "*Being gene positive it's basically a death sentence, and the execution style is the worst possible thing known to man…they should put serial killers through that not nice people like us.*" Knowledge of genetic risk for FTD forced people to confront not just their mortality, but also their sense of self as the *"execution*

style" is especially cruel in threatening the core of identity itself. Some participants experienced anticipatory grief regarding the potential loss of their expected future. Steph (female, age 39) said *"you kind of mourn what hasn't happened yet…it took me, oh, a good six months to like get used to…the fact that like I'm not going to live to be as old as I expected to live."* Thus, the uncertainty related to disease development also resulted in grief responses in addition to confronting mortality and the potential loss of the self.

Most participants knew firsthand what the course of FTD could entail via experiences with other family members. Because of this lived experience, participants had some idea of the nature of their likely fate, and this increased the sense of dread and doom. Luke (male, age 44) said, *"It's awful watching your parent or any family member go through that and it's just a little icing on the cake to be like, 'Oh, is that my fate too?'"* Similarly, Drew (male, age 47) described how his experience caring for his father and meeting other families at caregiver conferences eerily foreshadowed what may happen to him if he develops FTD:

> *[At the Caregiver Conference is] when my mutational status hits home and makes-- when I am most fearful…because it's presented in front of me. And because what I saw my father go through is typical and textbook…and it absolutely matches everybody else's…experience on what's going on with their loved ones...I just feel at that point that I'm doomed. That's what's going to happen to me and it's just a matter of time and I just have to live through it.*

For Drew, witnessing the cycle of FTD confirmed the reality of the disease course and his trajectory towards the same fate. At-risk individuals have extensive experience with what FTD and/or ALS can do to a person; this understanding tends to exacerbate dread and makes the threat real, salient, and seemingly inescapable.

Many at-risk participants recognized that it is unlikely that they will be the first to know that they are symptomatic if they develop FTD, as a characteristic component to FTD is lack of awareness or insight into one's own behavior. This fear of being symptomatic without knowing it contributed to participants' dread of wondering if or when they will develop symptoms. Ella (female, age 59) was consumed and stifled by self-doubt related to her risk of developing symptoms. She constantly worried that she would develop symptoms without awareness and inadvertently hurt herself or others; she stated that hurting someone else is what she feared the most: *"I have no confidence in any decision I make... I'm afraid of doing anything… I don't want to be responsible for a huge mistake."* She believed that the inability to trust her own judgement and brain with resulting self-doubt was the worst part of her risk.

As FTD causes people to lose self-awareness, participants devised other ways that they would detect the onset of their disease. Some participants confided in friends, family, or coworkers about their risk and asked them to inform them if their behavior changed. Many also engaged in self-surveillance for symptoms, although this often heightened anxiety. When participants forgot something or acted out of character it reminded them of their risk and raised worries that they could be symptomatic. For example, Nancy (female, age 32) said:

> *"I just have to constant[ly] remind myself not to look-- if I forget a word that this isn't a sign of symptoms, to not be paranoid of that…I remember after I found out I was positive I did lose my keys... I started bawling my eyes out…and that was the first time I had a fear. I was like 'Oh my goodness, the gene.'"*

All who noted similar worries rationalized that everybody forgets things from time to time. However, this surveillance for symptoms heightened many participants' worries about their risk, making the genetic status more salient.

Although some participants described surveillance for symptoms as related to their anxiety about becoming symptomatic, others found comfort in surveillance such that performance on cognitive testing and other evaluations from physicians was interpreted as proof that they were doing okay. For example, one participant described how constantly being checked at neurology appointments and research visits kept her from worrying. She said, *"I'm like 'Nope. Don't have it. When I have it, they will tell me,' so I don't feel like I have that constant concern that I have something wrong with me"* (Steph, female, age 39). This reliance on surveillance for reassurance was most often utilized by those who felt they were "just genetic carriers" or who rejected FTD risk as part of identity; these attitudes are described in more detail in the following section. Thus, for some people FTD was brought to the forefront of their identity through surveillance, but for others surveillance served as a tool to delay the centralization of FTD through denouncing its current impact.

Multiple participants contrasted FTD and ALS onset, emphasizing the nuances of FTD that make it harder to detect in the self: lack of self-awareness and its cognitive rather than physical nature. These nuances add a complicated layer of uncertainty and therefore fear. In addition to having a father with dementia that was assumed to be FTD after a *C9ORF72* pathogenic expansion was identified in the family, Luke (male, age 44)

had many relatives with ALS. Luke noted the difference between anticipating cognitive FTD and physical ALS symptoms:

> *What was even worse was the concept of the FTD piece because…there are so many natural things that could occur to make you question it anyway as opposed to for something physical like ALS, it's like, 'I'm pretty sure I'm okay. I got up and didn't drop anything today or trip and fall,' or whatever right, it's so much less clear about this, so that's a little more intimidating than the other.*

Luke recognized that the onset of FTD would not only be much harder for him to detect, but that there could also be more opportunities for false alarms. However, wondering how and when ALS onset may occur posed its own set of problems for some participants. For Frank (male, age 36), the physical nature of ALS led to intense surveillance for symptoms. After experiencing some tremors that worsened with anxiety, Frank convinced himself he was developing ALS until he eventually had a normal electromyogram (EMG). He described his intense ALS self-surveillance prior to having the EMG:

> *Everything I did in daily life became an ALS test. It sucked all the joy out of my workouts…[j]ust fearing that something would feel weaker. Every time I walked up the steps it was ALS. Every time I pull[ed] a door handle open, 'Does it feel any different?' Every single thing I did became a test. I was constantly saying tongue twisters to see if I could still enunciate my words clearly.*

While Frank said that he still engaged in surveillance after the normal EMG, he was better able to recognize when physical symptoms were due to anxiety. Frank's experience illustrated an all-consuming response to the uncertainty of disease onset, and the benefit of evaluation by a physician to at least temporarily quell anxiety.

Centrality of FTD Risk Status to Identity

While an FTD risk designation raised fundamental questions about what constitutes self and how a sense of identity is cultivated over a life course, participants simultaneously engaged with the threat of FTD diagnosis in their process of identity creation. Participants varied in how they dealt with the risk and therefore there were varying consequences for how and to what extent their risk was incorporated into their identity. The general stances participants took are best described as part of a continuum from FTD risk as central to identity to FTD risk as not central to identity.

FTD Risk as Central to Identity: FTD risk became central to identity for many participants, although there was variability in terms of the positive versus negative valence of these discussions. While FTD risk was very explicitly a part of identify for some, for others, this was discussed in a more subtle way. One participant described how his genetic status became a positive part of how he views himself:

> *I think that I am the first of a massive population of people who will soon…understand their genetic status for a variety of things, and I see myself as the pioneer, as Neil Armstrong navigating this world knowing, and so I feel like I've got a responsibility to tell others, to help others navigate this, to possibly facilitate others through this journey.* (Jeff, male, age 31)

By labeling himself as a "pioneer," Jeff assigned himself an identity centered around his genetic status. Though no other participants used the label of pioneer, a few highlighted their unique status as part of a small group of individuals who are aware of their risk for developing symptoms of FTD prior to any possible neurological decline. These

participants identified as part of a distinct group of individuals, and one described this new identity as being a "*rare bird"* (Eva, female, age 49). While this rarity was seen to confer special status and purpose in some ways, it also conferred isolation to some. Ella (female, age 59) said, "*It makes me feel so separately from everyone else, and I don't like being in a bubble by myself."* For Ella, FTD risk defined her as different and this was isolating. In his quote above, Jeff also conveyed a *responsibility* or *obligation* to act to benefit the FTD community so to speak, such as through advocacy, research, or connecting with others. Several other participants expressed similar feelings that spurred their research and advocacy involvement. Britt (female, age 31) described how she actively decided to make her genetic status part of her life through awareness of and involvement in research efforts. Drew (male, age 47) reiterated the value of participating in research as an asymptomatic person and referred to himself as "*a really good pincushion."* Thus, these at-risk participants built a shared identity as a new and perhaps pioneering group of individuals with awareness of their risk status without neurological impairment. They felt they could contribute uniquely to research, advocacy, and other efforts aimed at finding treatment.

A few at-risk participants assessed their risk in a way that allowed them to conclude that they were better people because of the experience of knowing. For example, Jeff (male, age 31) stated *"I think that I am the same person, probably a better person than I was."* He tried to look at life in a more investigative, mindful way and said:

> *Now I've got this new perspective on life, and I've gained this nugget of wisdom…You kind of take a snapshot. Am I doing it all right...I think that unless you have something in your life or someone or some teaching or some training that can snap you out of it you will never realize that life could be better.*

This approach demonstrated the ability of an at-risk identity to fundamentally and positively change the way a person viewed him or herself. Jeff (male, age 31) said that because of his awareness of his status he was able to "*love deeper and harder,*" to "*cherish relationships more,"* to feel wiser, and to take on the idea that he was "*truly blessed*."

Some participants referred to themselves as patients despite being asymptomatic. This suggests that the genetic status alone was important enough to impart patienthood. Nancy (female, age 32) described an experience in a support group that heightened her awareness of her risk and ultimately led to her self-characterization as a patient. When a man asked if it was wrong that he could not wait until his wife's battle with FTD ended many people said no, and the participant recalled feeling similarly when her own mother was sick. However, the participant also described being upset thinking about others wishing she would die:

> *As somebody with the gene, you sit there and think…'Great. Is my spouse going to say that about me? Are my children going to say that about me?'…You look at it as the caregiver and the family member of the person who has the disease, but now you also look at it as the person with the disease, because that can be you now…So when I hear people make that comment, it hurts me in a way that-- they don't know what it's like to live with the gene. But you can't be mad, because you thought the same thing at one time.*

The interviewer followed Nancy's comment by stating "but your perspective has changed," to which Nancy responded in agreement with *"because you're now technically a patient."* As Nancy navigated her roles as both a caregiver and a patient, FTD risk

became central to her identity. The risk induced a sense of patienthood, despite her asymptomatic status and the lack of clear role definition as she was "technically" but not obviously a patient, operating in an uncertain in-between phase. Interestingly, most at-risk individuals who called themselves patients or expressed similar sentiments still distinguished their current state from their anticipated future state: they noted that the risk status is not the same as having the actual disease. Participants' incorporation of patienthood into their identities may suggest that these individuals were convinced they would develop disease.

For some participants, the emotional valence of the risk status changed over time. Britt (female, age 31) transitioned from being embarrassed about others knowing her status to serving as an FTD/ALS advocate. Early on, her friends made a GoFundMe post to try to raise funds for the participant and her husband and she was *"mortified"* after it was posted, to the extent that she avoided signing onto social media or going home to her small town for fear of a *"pity party"* (Britt, female, age 31). After that post, she had to explain her status to many people. She later chose to get a visible tattoo that symbolized her risk; she saw dual purpose to her tattoo, as it served as a means of raising awareness and as part of her self-defined coping mechanism. Britt noted that she and her husband want to start a foundation using resources from the GoFundMe page to help other couples afford IVF with PGD, as they found it to be an expensive and challenging process to navigate. This signifies a change in Britt's relationship to society from someone to be pitied to someone who is an advocate. This transformation from humiliation to advocacy occurred within about a year after her results were disclosed.

FTD Risk as Less Central to Identity: Incorporation of FTD risk into sense of self was not universal among at-risk participants. Unlike those for whom FTD risk was central to statements about who they were, some participants identified as *"just a genetic carrier"* (Mary, female, age 54) and were readily able to identify other aspects of self that were quite separate from their risk status. Many of these individuals had the perspective of *"it is what it is."* These participants drew on the rationalization that all that had changed due to genetic testing was their awareness of their status. For example, one participant said, *"I was positive from the moment I was conceived. So I'm not any different than I was for all those years before I found out*" (Anna, female, age 63). Another participant, Luke (male, age 44), described how his attitude towards his status evolved over time to reach a similar sentiment as Anna:

> *I mean it was a certain level of anxiety but not like all the time…Because when you think about it as it applies to you, it's sort of like this hasn't happened yet but in fact it has. And then from there it starts to set in, not right away but over time that same mentality of well this was-- I've always had this, this isn't new and what you sort of have it in that frame of mind, all you really have is insight into what your predisposition[ed] for.*

This approach minimized the impact of risk disclosure by prioritizing the notion that the risk status itself was unchanged. Part of how Luke arrived at this stance was through making downward social comparisons. He described how comparing himself to diagnosed individuals participating in research allowed him to conclude that "*It's not nearly the big deal that you thought it might have been when you first started out."* By identifying others who were worse off per se, he felt more positively about his own situation and the risk status became less worrisome to him over time. It is possible that

many people at risk, like Luke, adjust their stance towards this "just a genetic carrier" sentiment over time as they continue to live without symptoms. This in fact was an explicit goal for one participant, who felt she needed to find ways to make FTD less central to her life. Nancy (female, age 32) described how her FTD risk became a major part of her cognition: *"It is always going to be on the back of my mind, at least for a while until I learn how to deal with it and not let it take over…I can't let it take over my life, but right now it's only a couple of months old."* She noted that she tried to diminish the centrality of FTD risk to her sense of identity, and compared this process to getting a new car:

> *You get a new car and that new car is…fresh on your mind. You're going to be talking about it maybe for the first couple months. It's the same thing. Once anything in your life changes, you're going to talk about it. And then as time passed, that car is not important anymore. It's just a car.*

Thus, after several months with FTD risk central to her identity, Nancy decided that she needed to actively prevent the risk from defining her and her thoughts. This entailed recognition of FTD as central to identity, as well as a desire and concentrated effort to redefine her self-identity in relation to FTD.

Participants who identified as "just genetic carriers" were people who knew their status for at least one year, and like Luke some were able to describe previous attitudes towards the status that were more anxious in nature. However, this shift towards a "just a genetic carrier" attitude is not universal as some of the individuals who expressed more negative and fearful sentiments indicative of a central role of FTD risk in identity described above knew their status for three or more years.

Thus, for some at-risk participants the genetic status was acknowledged but had not become a core element of identity by the time they were interviewed. However, some participants who took this "just a genetic carrier stance" and claimed minimal impact on their lives still simultaneously described new roles that were shaped by their risk, mostly as research participants and advocates. This stance may reflect participants' attempts to cope with risk via cognitively focusing on ways in which they are no different than they were prior to testing.

Other participants completely rejected FTD risk as part of their identity. One participant considered herself *"a fighter against what [she] consider[s] to be like medical injustices,"* such as cancer, but felt that she had *"other fish to fry"* besides FTD, referring to ailments of other family members. With regards to FTD she said, *"It's important that I fight it but I don't consider myself a sick person."* Since she was not symptomatic and did not feel that her risk status was affecting her, she chose to focus on others who were sick in her life, such as her husband. The way she viewed her status was, "*If somebody had told me I had high cholesterol I'd have the same response: 'Well, that's a pain in the ass. Let's take care of it.' I refer to it as a boil on my butt, if you don't mind my saying. It's really just-- it's a nonissue*" (Eva, female, age 49). Even though FTD was threatening to affect her, she still saw this threat as lesser than the current diagnoses of family members. FTD was clearly not integrated into her identity as it took a backseat to other problems of loved ones in her life. Other participants who rejected FTD as part of identity similarly engaged in this purposeful process of not letting FTD become central to identity. For example, Drew (male, age 47) actively chose to not alter his lifestyle or behavior after learning his risk. He said by living his life as he would have normally lived it, "*[it was]*

like saying 'fuck you' to the mutation and to the disease." However, such complete rejection of FTD risk as part of identity represented the approach of a minority of the at-risk participants. Thus, the centrality of FTD risk in the identities of participants varied, and many participants expressed multiple perspectives simultaneously.

Discussion

The purpose of the VOICE Of bvFTD project was to broadly explore how people think about their FTD diagnosis or risk, the emotional and behavioral effects of the diagnosis or risk, and the related challenges and experiences. In this manuscript, we explored how at-risk participants perceived the disease process of FTD as threatening to identity and how they incorporated their risk into their current identity. Additionally, we considered the questions raised by their risk and associated emotional reactions.

The limited research considering predictive testing in FTD or ALS has been focused on proving that the practice is safe and therefore feasible. Several studies have noted that there are no significant psychological implications to learning genetic risk for inherited neurodegenerative diseases by examining psychiatric admissions, suicide attempts, or lasting severe anxiety or depression (Molinuevo et al., 2005; Steinbart et al., 2001; Paulsen et al., 2013). However, this does not preclude the existence of difficult experiences not able to be captured by the outcome measures used and at the time points assessed. Additionally, this focus is too narrow to fully understand the experience of predictive testing and its impact. Of note, there is substantially less research related to the impact of FTD predictive testing compared to Alzheimer disease (AD) and other neurodegenerative diseases such as Huntington disease (HD), for which predictive testing has been available for almost two decades (Paulsen et al., 2013; HDSA, 2016). FTD results in altered personality and behavior without outwardly appearing motor signs like those seen in HD or initial memory impairment that is indicative of AD or other dementias. Thus, FTD presents unique challenges and reactions to predictive testing may differ. Furthermore, the measurement endpoints for previous studies of FTD and ALS

predictive genetic testing were assessed no later than three years post-disclosure which limits understanding of longer-term effects (Crook et al., 2017). Though our results fit with those of previous studies that suggest predictive testing can generally be performed without causing extreme or dangerous psychiatric reactions, our results also suggest that how people handle risk information is quite complex, variable from person to person, and far from static. Studies of quality of life and other measures designed to assess impact after testing need to consider methods to capture these nuances, such as by employing mixed-methods designs to consider broader effects and longitudinal designs to capture how people incorporate risk over time. Although it is important to establish lack of clinically significant psychological impact to justify the safety of predictive testing, to not elaborate on the challenges that do occur is a disservice to patients. It would be beneficial for providers to understand and discuss with patients the potential consequences of testing in pre-test, disclosure, and longitudinal post-test appointments, and therefore research needs to consider predictive testing experiences qualitatively in addition to reported quantitative measures.

Furthermore, future research should expand the quantitative outcome measures utilized beyond just assessments of severe psychological concerns. As identity reconstruction may be conceptualized as part of psychological adaptation, our data suggest that future research using a measure of psychological adaptation is warranted, such as the Psychological Adaptation Scale (PAS). The PAS is a quantitative assessment of adaptation to a chronic condition or disease risk (Biesecker et al., 2013). There are also quantitative measures of illness identity such as the Illness Identity Questionnaire (Van Bulck et al., 2019). Future research may consider the relationship between these variables

to assess how various identity restructuring processes might correlate with overall psychological adaptation and functioning to further understanding between these processes and outcomes for at-risk individuals. Since our data demonstrate that people may evolve in their attitudes towards their risk over time, research should also consider the longer-term evolution of identity reconstruction in response to risk to create interventions for those who struggle most with their risk status.

Studies that have assessed individuals' reasons for or against pursuing predictive genetic testing in FTD and ALS have found that some people pursue genetic testing to reduce ambiguity or uncertainty (Fanos et al., 2011; Steinbart et al., 2001; Riedijk et al., 2009), which was also the case for several of our participants. However, while predictive testing resolves some uncertainty related to the genetic status itself, it simultaneously engenders uncertainty about the future when it reveals a positive result conferring high risk of disease development. One major concern for participants in this study was the potential for development of FTD without self-awareness of disease. This and other fears about the future shrouded some, but not all, participants in unfathomable uncertainty and dread. In her work on uncertainty in illness, Mishel (1988) noted that it is not uncertainty itself that is inherently dreadful, but rather the implications of the uncertainty as shaped by the person's appraisal of the uncertainty as a danger or an opportunity. Due to the perceptions of the FTD disease process as stripping or reducing and the current lack of available treatment, it is unsurprising that many people appraised the uncertainty as a danger and therefore as dreadful. However, some viewed their risk as an opportunity to better themselves or help others. The uncertainty of genetic risk makes psychological adaptation challenging (Biesecker and Erby, 2008). Future research should explore what

causes some individuals to appraise the uncertainty of genetic risk as opportunity rather than danger, determine whether this approach is consistent with more positive psychological adaptation, and suggest counseling or other interventions that encourage this mindset if it is found to be beneficial.

At-risk participants were forced to consider what they view as the essence of a person, as this is threatened by their conceptualizations of the FTD disease process. Risk for FTD raised interesting theoretical questions grounded in Cartesian dualism that led participants to grapple with the relationship between mind, body, and identity. The ultimate questions underlying participants' attempts to make sense of their risk were the questions "who will I be?", and "who won't I be?" if I develop FTD. FTD risk also caused participants to confront related existential problems as it increased the salience of mortality. Although not described in detail in this manuscript, the majority of the participants grappled with the duality of risk for two neurodegenerative diseases, FTD and ALS. Future research should further explore how individuals think about and cope with the dual risk.

Most participants described ways they believed or feared their sense of self and identity would change in the future. Some participants described loss of psychological self despite physical presence as something they anticipated based on experience with family members. The physical presence but psychological absence of loved ones with dementia has been well-characterized as ambiguous loss (Boss, 2011). Ambiguous loss has traditionally been characterized as a relational phenomenon, such that the loss of a loved one is unclear due to physical presence with psychological absence as in dementia, or psychological presence with physical absence as in missing persons in war or natural

disasters (Boss, 1999). However, a novel type of ambiguous loss was identified by participants in this study: anticipated ambiguous loss of the self. Participants imagined the ways in which FTD would strip them of their core being and feared what identity would remain. They feared and grieved the impact this would have on themselves as well as on their families, friends, and others. This phenomenon of ambiguous loss of the self should be investigated further in other neurologic disorders for which predictive genetic testing is available. This should also be further investigated in people with early stages of dementia.

The centrality of FTD risk to identity varied greatly among participants, as did the emotional valence of the impact of the risk status on identity. Perhaps in our study altered identity is most readily recognized through participants' redefinition of existing roles and creation of new roles. Participants considered what their risk meant for their identities as parents, spouses, working individuals, and how it created new identities as research subjects, advocates, and patients. The process of refining identity due to FTD risk may be part of the psychological adaptation process. The cognitive theory of adaptation provides a framework to describe how people adapt through search for meaning, attempt to regain mastery or control, and bolster self-esteem in response to threat (Taylor, 1983). Certainly, receiving information about a genetic risk status for FTD or FTD and ALS can be threatening. As part of the process of adaptation, our participants had to continually assess what FTD meant to their identity, and how they would use that understanding to seek control and increase self-esteem. One strategy people utilized was making downward social comparisons to find positive aspects of their identities (Taylor, 1983).

The study sample uniquely included individuals who had known their status for several years, which created the opportunity to hear from people in various stages of adaptation and therefore various phases of incorporating FTD into identity. Our results suggest that the impact of risk status on identity can fluctuate over time. Some individuals intentionally took action to alter the centrality of risk to their identity, whereas others evolved in their perceptions due to certain experiences or with more time to process. This identity reconstruction, part of the cycle of psychological adaptation, is an iterative process that differs for each person, although there may be some commonalities. In this study, participants who tended to take the "just a genetic carrier" stance were those who had known their status for at least one year, perhaps suggesting that this is an attitude that people may adopt over time as they adjust to their risk. However, there were some individuals who had known their status for several years who described immense anxiety and dread. Thus, how individuals reconstruct their identity in response to risk is nuanced, and is likely influenced by more factors than just time since learning results. There has been little work to date that considers how people adjust to this information over time or what this means to individuals' sense of self. Better understanding of these processes may provide guidance about topics that need to be explored at clinical appointments with genetic counselors, neurologists, and other providers.

Additionally, understanding these processes over time could shape interventions designed to bolster psychological adaptation through identifying common ways that people successful and unsuccessfully incorporate FTD into their identity. A few interventions have been developed to support self and identity in individuals with

dementia and were summarized by Caddell and Clare (2011). However, Caddell and Clare (2011) and Oyebode and Parveen (2019) assert that more research is needed in this area. Furthermore, none of these interventions were designed for presymptomatic individuals at risk of developing dementia. Perhaps some elements from existing interventions may be translated into interventions for those at risk of developing dementia, such as exploring and enhancing role-identities as in Cohen-Mansfield et al. (2006) or creating ways to physically represent one's life history as in Massimi et al. (2008). However, many of the interventions described in Caddell and Clare (2011) focus on identity preservation through memory enhancement, which is not the core problem for presymptomatic individuals. Thus, research is needed to develop psychological interventions for individuals concerned with future loss of self with dementia, not just for those who are already symptomatic.

Participants conceptualized their status in a variety of ways through constructing illness identity, the degree to which a chronic health condition is integrated into a person's identity (Van Bulck et al., 2019). Some participants described illness identity stances that align with the existing constructs of enrichment, engulfment, acceptance, and rejection outlined by Van Bulck et al. (2019). Enrichment refers to feeling like a better person or having other positive impact from illness, engulfment refers to illness consuming identity, acceptance is when illness is incorporated as a piece of a larger identity, and rejection is dismissing the illness (Van Bulck et al., 2019). However, being at genetic risk for a condition without yet developing symptoms complicates this model of illness identity. Individuals who identified as "just genetic carriers" cannot be neatly categorized by the existing framework; they did not fully reject FTD as part of their

identity, but rather there was a temporal and symptom-based element to the incorporation of FTD into their identity. Another nuance not captured by the model arose from some at-risk participants who called themselves patients. "Patienthood" describes when being a patient is a defining feature of identity (Sabat et al.,2011). The definition of patienthood seems to resemble that of engulfment, however, most participants that expressed this were not otherwise exhibiting engulfment attitudes. Since there does not currently exist a commonly known role for being asymptomatic but at-risk, individuals can only define themselves as "patients" or "care providers." Thus, at-risk individuals may be trying to expand their own definition of "patient," as this is a concrete role with which they can identify. Though patienthood has usually been considered a phenomenon that may increase marginalization and negative stereotyping of persons diagnosed, our data suggest that how this perception of patienthood influences at-risk individuals may be worth exploration. Additionally, some participants could be characterized as aligning with different illness identity constructs simultaneously or evolving in their stance over time. For example, some participants described both feeling like they were better people for being able to contribute to the research (enrichment) and feelings of doom, self-doubt, and fear (engulfment). Thus, while the illness identity model provides a useful framework for thinking about the data, it does not seem capable of capturing the complexities of the participants' reconstruction of identity.

Limitations

The findings from this study are not intended to be representative of all individuals with known genetic risk for bvFTD. Our participants were well-educated, which often is associated with being of better financial standing. They may also have

been more savvy in identifying research, advocacy, and support opportunities and had the means to access these opportunities. Due to this, our participants may have had better access to counseling and other resources that helped them process what their status means to them. All participants in this study were Caucasian, which prevents consideration of how people from diverse backgrounds may experience this process differently. Many of the at-risk participants had the same gene implicated in their risk, *C9ORF72.* Pathogenic repeat expansions in this gene confer risk for both FTD and ALS, and therefore these participants may have differing experiences compared to those whose genetic risk is only for FTD. Participants were recruited from several sites, which introduces variation in the clinical care they received from genetic counselors and neurologists. How risk information is initially delivered may influence how people understand, perceive, incorporate and adjust to that risk (Fagerlin et al., 2011). Additionally, how the patient-provider relationship is perceived may influence these processes (Roter, 2006). However, including individuals who are cared for by different providers allowed us to capture greater variety of reactions to clinical encounters. With increasing uptake of predictive genetic testing as well as increased awareness and better diagnostic clarity for FTD, more individuals are receiving risk information and at younger ages. The reactions of younger individuals to risk may differ from the experiences represented by this sample and deserve consideration in the future.

This study was only able to capture experiences of people willing to participate in the research study, and therefore our data are unlikely to represent the full range of experiences of those at risk of developing FTD. We did not capture the experiences of at-risk persons who chose not to go through the testing process, as our inclusion criteria

required at-risk individuals to have undergone genetic testing with results conferring high risk of developing FTD. The experiences of individuals who do not know their status but know they are at risk based on family members' diagnoses or testing results may differ from those included in this study sample. It should be acknowledged that the interpretation of the participants' experiences is limited by the questions that were asked of them and is dependent upon the reflexivity of the primary researcher. However, the interviews were thorough and each participant was invited to discuss any uncaptured elements of their FTD experiences that felt important to share with the researcher. The data allowed for rich, thick description and a nuanced understanding of participants' experiences.

Practice Implications

The experiences described by participants in this study have implications for the practice of clinical providers such as genetic counselors, geneticists, and neurologists. It is evident from this study that individuals experience a wide variety of emotional and behavioral responses to learning their genetic risk status for bvFTD. Clinical providers should be aware of the diverse ways that people may respond to their risk and normalize these experiences. These differing reactions and experiences shape how risk is incorporated into identity. Integration of FTD risk into identity evolves over time, bringing new challenges and threats for the individual who also evolves in his or her coping and management strategies. Providers should help at-risk individuals anticipate that how they conceptualize their risk may change subtly or drastically over time.

Since individuals are continually processing what their risk means to them and facing new challenges, clinicians should continually assess and reassess individuals' needs for support. Providers should also help individuals identify their strengths in facing such challenges and help them consider how they may apply these resources or strategies to combat future challenges. Referral to therapy should be made when individuals who are at risk for FTD may benefit from further exploration of what this information means to them as part of the psychological adaptation process. Genetic counselors and other providers should remain prepared to start these discussions and facilitate patients' processing of the impact of identity restructuring. Most participants disclosed that discussing their experiences in detail as part of the interview was helpful, and many noted that they lacked opportunities to do so otherwise. Providers should invite these conversations during clinical appointments and make referrals to further counseling when

appropriate. This sentiment, as well as the finding that some individuals may have benefitted psychologically from medical surveillance, suggest the need for routine follow-up visits for presymptomatic individuals that include neurology assessments and genetic counseling. Currently, there is not a standard protocol regarding how often, when, and what follow-up should be offered to presymptomatic individuals after the initial genetic testing results disclosure. Many clinics follow the HD protocol (MacLeod et al., 2013; HDSA, 2016) which recommends follow-up shortly after the initial results disclosure, however there is no standard recommendation for longitudinal follow up and often genetic counseling is not included in follow-up clinical appointments. In the continued absence of a standard protocol, clinics should set institution-based goals for presymptomatic neurology and genetic counseling follow-up based on available resources.

Additionally, our findings showcase the importance of social roles in participants' characterization of identity and the unique ways that FTD threatens such roles. Given this, genetic counselors and other providers should strive to include family members and other supportive individuals in counseling sessions. This family-systems based therapeutic approach has been described in Rolland's work on the Family System Genetic Illness (FSGI) model, which considers the psychological and social challenges of genetic conditions for at-risk and diagnosed individuals as well as for their families, and how these changes differ based on disease typology and over the course of disease (Rolland and Williams, 2005). This approach may allow providers to better understand an at-risk person's situation and needs, and may facilitate problem-solving for families. When appropriate, genetic counselors should consider referring families to longer term

counseling services grounded in this approach. Future research should assess the impact of this approach to determine the extent of its benefits for patients and families.

Our results suggest that even individuals who seem to be coping well with their risk status in the present may have mildly impactful to nearly debilitating concerns about the future. Providers should explore how patients are dealing with uncertainty about the future. Interestingly, genetic counselors may need to pay special attention to individuals who adopt an "enriched" stance towards their risk status by declaring themselves a better person through the experience. People may adopt this attitude through positive coping mechanisms; however, it is also important to note that this may be reflective of defense mechanisms against feelings of shame (Kessler et al., 1984). Genetic counselors should be aware of this nuance, attend to comments that suggest an enriched stance with the co-presence of shame, and be prepared to explore feelings of shame with their clients as appropriate.

Uncertainty can be appraised positively, allowing room for hope, or may be appraised negatively, resulting in dread and fear depending on whether it is seen as a danger or an opportunity (Mishel, 1988). Genetic counselors and other providers should assess how at-risk individuals are thinking about the different elements of their lives that seem uncertain. They can then help at-risk clients by guiding them toward more effective coping strategies, helping them to redefine their goals, and helping them identify controllable aspects of life (Blesson and Cohen, 2019). Providers can also help their patients identify steps they can take to reach their goals and to gain more control (Blesson and Cohen, 2019).

With increasing hope in the neurogenetics field for gene-based and other therapies being developed in clinical trials, providers need to be aware of how this hope influences peoples' conceptualizations of disease and willingness to participate in research. This population is desperate for a cure, and our results suggest that some people not only are driven to participate in the research effort but feel a sense of responsibility and even obligation to do so. Thus, genetic counselors, neurologists, and other providers need to ensure adequate counseling about the benefits and limitations of research protocols. These conversations should begin in pre-test genetic counseling, as hope for the ability to participate in research evidently is a motivating factor for some individuals to pursue genetic testing. Providers must delineate the differences between research and treatment, as well as ensure that people are given the information and support needed to make informed decisions. The state of the science and formidable nature of the disease render this population especially vulnerable to coercion.

Further research should extend this preliminary understanding of how persons at genetic risk for bvFTD incorporate the illness into their identity and experience the disease. Larger-scale, longitudinal, and quantitative studies of the affective and behavioral impact of risk information may allow for more specific guidance regarding what types of interventions at what times may be appropriate to support this population. Future studies should also attempt to capture the experiences of a more diverse group of individuals in terms of race, ethnicity, and education. With predictive testing occurring more often and at younger ages, exploration of experiences of younger individuals will also be important. Studies that consider identity and experience of individuals who are at familial risk prior to having genetic testing could be useful for comparison to experience

and identity following confirmed positive status. As hope in clinical trials continues to rise, research should be conducted to explore how this hope may be both beneficial and detrimental in individuals' processing of their status.

Additionally, all individuals in this study were eager to contribute to the research efforts and most incorporated this as a positive part of their FTD identity. Research participation was part of the core motivation for pursuing genetic testing for several study participants, which aligns with reasons for testing outlined in other studies of ALS and FTD genetic testing (Steinbart et al., 2001; Crook et al., 2017). Many participants felt they benefitted from participation in this study; they disclosed that this study allowed them to discuss things that they otherwise would not have the chance to discuss, and that helping others through research made them feel good. Such experiences are not unique to participants in this study. Lakeman et al. (2013) argued that participating in qualitative research that allows for telling personal stories can itself be therapeutic, as it can help participants to make sense of their experience (Lakeman et al., 2013). Research participation may also provide at-risk persons with a sense of control, purpose and a clearly defined role in a time when much is uncertain. However, research will not be something that all at-risk individuals choose to pursue and not all will personally benefit. Regardless, clinicians should identify opportunities for at-risk individuals to contribute to research efforts and help at-risk individuals navigate the risks and benefits of participation when appropriate. All individuals should be informed of the existing opportunities as our data suggest that this can be a powerful source of hope and provide a sense of control or purpose for some individuals.

Appendices

Appendix I: Consent Documents

Verbal Consent Script

University of Pennsylvania
ADULT ORAL CONSENT SCRIPT

Study Title: VOICE Of bvFTD [Voices Of Individuals: Challenges and Experiences Of bvFTD]
Principal Investigator: Murray Grossman, MDCM, EdD
PI Version Date: Version 1, Date 5 June 2019

Hello, is this [name of participant]? My name is Laynie Dratch. I am a graduate student from Johns Hopkins. I am working on a research project for my master's thesis. You gave (provider *or* researcher *or* me) from (site name) permission for us to contact you about our research. (The provider *or* researcher *or* I) also sent you a packet of forms. I would like to tell you a bit more about our research project because you may be able to participate. Are you interested in hearing about my study and what your participation would involve? Do you have time to talk to me now? Are you in a place that feels safe to talk to me about this project?

[If not interested]: *Okay, thank you for your time. Have a great day!*
[If not in a safe place or now is not a good time]: Okay, when is a good time for me to call you back when you will have time to talk in a safe and private place?
[If yes]: Great! I'm happy you are interested in hearing more.

Key Information Summary
We are asking you to volunteer for a research study about life with or at risk for behavioral variant frontotemporal dementia. I will call it bvFTD for short. We want to learn how bvFTD impacts your day to day life, how you think about yourself, and what challenges you face. This is an interview study. We are asking you if you want to be in this study because you may be eligible to join as a person who is living with or at risk for bvFTD. You do not have to join this study. It is your choice whether you want to join or not. There is no penalty for not joining. In the next few minutes, we will talk about the details of the study, what we would ask you to do if you join, and the potential risks and benefits of being in the study. You can ask as many questions as you need to help make your decision.

What is the purpose of this research?
The study is run by Johns Hopkins University, the National Institutes of Health, and the University of Pennsylvania. The goal of our study is to learn more about experiences of living with, or at risk for developing bvFTD. We want to understand how bvFTD affects your day to day life, how you think about yourself, and how you overcome challenges. The study will involve interviews to help us learn as much as possible about living with bvFTD. We hope that this will guide future research, resource development, and clinical practice. We are planning on interviewing about 30 people who either have bvFTD, or have had genetic testing that showed they are at risk of developing bvFTD.

Why are you being asked to participate?
We are asking you to join our study because [you or your provider or your researcher] thought that you might qualify for the study. You might be able to join the study for one of two reasons. You might be able to join if you are a person with bvFTD and your symptoms started at least two months ago. Or, you might be able to join if you are a person who has had genetic testing results that showed high risk of developing bvFTD and you learned about the results at least two months ago.

What would you be asked to do if you join the study?
If you want to take part in the study, you will have at least two phone calls from us. This is one of those phone calls. During this phone call, which will take about 15 to 20 minutes, we will review the study in detail together. We will ask you some basic questions about yourself and your understanding of the study as part of the consent process, and to make sure that you are eligible for the study. We cannot include every interested person, so we want to make sure that each person we include is a good candidate for the project.

If you are a good match for the project based on the conversation we have today, then we will schedule another phone call. During that next call, we will ask you a few more questions about your thinking, and we will also complete the interview. The questions we ask you to assess your thinking are not going to be used for any diagnostic purpose. The interview questions will be about living with or at risk for bvFTD. I will be the person that you interview with if you participate. The interview will last about 30 to 60 minutes. When I call you for the interview, I will make sure that you are in a location that feels comfortable and private before starting the interview, just like I did today. We recommend that you avoid public spaces during your phone interview. The best place to do the interview is somewhere private and quiet. I will be in private room during the interview, like I am now during this phone call. We will audio record the interview. We will give you a $20 gift card to thank you for your time if you participate in this interview study. This gift card will be sent to you after your interview.

It is your decision whether or not you want to join our study. You do not have to join if it sounds like something you do not want to do. You may also choose to join now, but stop before the study is finished. There will be no penalty if you decide not to join, and there will be no penalty if you stop the study early. It will not impact any of your medical care. If you participate in the interview but want to stop early, you may still receive the $20 gift card.

What questions do you have so far?

What are the potential risks of the study?
There are no physical or medical procedures included in this study. This means that there is no risk of physical harm. You might become sad, anxious, or frustrated when answering some of the questions about your life with bvFTD. If there are questions that you do not want to answer, you do not have to answer them. You can also stop the interview completely at any time. If you become very upset during the interview, we will

suggest that you contact your provider. Or, if you are upset and do not want to talk to your provider, we can help you find someone else to talk to. If talking about your experiences makes you think of questions about bvFTD, we will refer you to someone who may be able to answer your questions. The other potential risk of the study is related to privacy and confidentiality. This is risk related to other people learning about your diagnosis. The research team does a lot to reduce these risks. We will talk about this more in a minute. You can also reduce the risk of people learning about your diagnosis by doing the interview in a private place.

What are the potential benefits of the study?
We do not expect you to personally benefit from the study. You might appreciate having the chance to talk about your experiences with bvFTD. You will not receive any treatments as part of this study. Even though you might not benefit directly from this study, you will help us learn more about the experiences of life with bvFTD. We may be able to use what we learn from you and other participants to improve resources and care for people with bvFTD in the future.

What will happen with the information I share?
There is a risk that someone outside the study will see your information. We will do our best to keep your information safe by using a participant ID number, removing the names of people and places from your interview data, and storing all information in secure ways – either locked in filing cabinets, or in a password-protected file on an encrypted computer. Let's talk about this in a bit more detail and explain what that means.

If you join the study, we will give you a participant ID number. This number will be used to label all of your study information. This way, the only information that connects your name to your study information is a password-protected file that includes the link between your name and your participant number. Only I will have access to this protected file. After your interview, we will destroy the link that connects your participant ID number to your name and contact information.

The interview will be audio-recorded and later transcribed. This means that a professional company will turn the audio into written text. The company will remove the names of any people and places that we say in the interview. This way, we can talk about people and places during the interview without having to worry about their information being included in our research. Once we have checked that the written version of the interview matches the audio recording of the interview, we will destroy the audio recording. This means that only the written version will be left, and this written version will not include any names of people or places. This written version is what we will use for data analysis, and for any papers that we write. If other researchers want access to our data, they also will only have access to this written version that does not have your name or the name of any person or place we talked about.

After we finish your interview process and send you your $20 gift card, we will destroy your name and other personal information including your contact information. At the end of the interview, we will ask if you would like to know about the results of the study. If

you would like us to send you an overview of the results, we will ask for your permission to keep your contact information in a separate, protected document.

Who do I contact with questions or concerns?
You may contact the VOICE of bvFTD team with any questions or problems with this research. Contact the University of Pennsylvania IRB Office if you have questions about your rights as a participant of this study, or if you feel you have not been treated fairly. The contact information is on the written version of the consent form that we are reviewing. This was sent to you in the packet you received.

Please reach out to one of these resources if you have questions or concerns:

Researchers' Contact Information:

Laynie Dratch, BA
Student Investigator / Associate Investigator
JHU/NHGRI Genetic Counseling Training Program
Baltimore, MD
Phone: 301-827-5029
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Murray Grossman, MDCM, EdD
Principal Investigator
The Penn FTD Center
Philadelphia, PA
Phone: 215-349-5863

University of Pennsylvania
Philadelphia, PA
Office of Regulatory Affairs Phone: 215-898-2614
IRB Phone: 215-573-2540

Do you have any questions? [Probe to assess the participant's understanding.]

Are you interested in joining the study?

[if no]: Okay. Thank you for your time. Have a great day!
[if yes]: Great! Like I mentioned before, we cannot take every person who is interested in the study. I would like to ask you a few questions to make sure you are a good fit for the study. Some questions will check to make sure that you understand the study, and some questions will ask for basic information about yourself and your family from the demographics form that you received earlier. Is it okay for me to ask you these questions now?

[If no]: Okay, when is a good time for me to call you back? *[If yes]: Thank you.*

[Complete Informed Consent Comprehension Assessment]

[Ask for Demographic Information]

[If eligible for interview]: Thank you for answering all of these questions. Based on what you have told me, you are a good match for the study. Let's go ahead and schedule the interview now.

Some people think it helps to think about their experiences with FTD before the interview. If you want to write or think about your experiences with FTD, you can do that between now and the interview. For example, you could think about what it felt like to get the [diagnosis or test result]. Do you have any questions for me? I look forward to our phone interview on (date /time scheduled). Thank you!

[If not eligible for interview]: Thank you for answering all of these questions. Unfortunately, we are not able to accept everyone into the study. Based on our conversation, you are not the best match for this study, and we cannot include you in the study at this time. The rest of the research team and I really appreciate the time that you have spent engaging with us. We hope that we might be able to offer more studies in the future. We want to thank you for your interest in our work and for your time. Do you have any questions for me?

Consent Information Sheet

University of Pennsylvania
CONSENT INFORMATION

Study Title: VOICE Of bvFTD [Voices Of Individuals: Challenges and Experiences Of bvFTD]
Principal Investigator: Murray Grossman, MDCM, EdD **Student Investigator:** Laynie Dratch
PI Version Date: Version 1, Date 5 June 2019

Key Information Summary

We are asking you to volunteer for a research study about life with or at risk for behavioral variant frontotemporal dementia (bvFTD). We want to learn how bvFTD impacts your day to day life, how you think about yourself, and what challenges you face. This is an interview study. We are asking you if you want to be in this study because you may be eligible to join as a person who is living with or at risk for bvFTD. You do not have to join this study. It is your choice whether you want to join or not. There is no penalty for not joining. This form includes details of the study, what we would ask you to do if you join, and the potential risks and benefits of being in the study. You can ask as many questions as you need to help make your decision.

What is the purpose of this research?

The study is run by Johns Hopkins University, the National Institutes of Health, and the University of Pennsylvania. The goal of our study is to learn more about experiences of living with, or at risk for developing bvFTD. We want to understand how bvFTD affects your day to day life, how you think about yourself, and how you overcome challenges. The study will involve interviews to help us learn as much as possible about living with bvFTD. We hope that this will guide future research, resource development, and clinical practice. We are planning on interviewing about 30 people who either have bvFTD, or have had genetic testing that showed they are at risk of developing bvFTD.

Why are you being asked to participate?

We are asking you to join our study because you might qualify for the study. You might be able to join the study for one of two reasons. You might be able to join if you are a person with bvFTD and your symptoms started at least two months ago. Or, you might be able to join if you are a person who has had genetic testing results that showed high risk of developing bvFTD and you learned about the results at least two months ago.

What would you be asked to do if you join the study?

If you want to take part in the study, you will have at least two phone calls from us. During the first phone call, which will take about 15 to 20 minutes, we will review the study in detail together. We will ask you some basic questions about yourself and your understanding of the study as part of the consent process, and to make sure that you are eligible for the study. We cannot include every interested person, so we want to make sure that each person we include is a good candidate for the project. If you are a good match for the project based on that conversation, then we will schedule another phone call. During that next call, we will ask you a few more questions about your thinking, and we will also complete the interview. The questions we ask you to assess your thinking are

not going to be used for any diagnostic purpose. The interview questions will be about living with or at risk for bvFTD. Laynie Dratch will be the person that you interview with if you participate. The interview will last about 30 to 60 minutes. When Laynie calls you for the interview, she will make sure that you are in a location that feels comfortable and private before starting the interview. We recommend that you avoid public spaces during your phone interview. The best place to do the interview is somewhere private and quiet. Laynie will be in private room during the interview. We will audio record the interview. We will give you a $20 gift card to thank you for your time if you participate in this interview study. This gift card will be sent to you after your interview.

It is your decision whether or not you want to join our study. You do not have to join if it sounds like something you do not want to do. You may also choose to join now, but stop before the study is finished. There will be no penalty if you decide not to join, and there will be no penalty if you stop the study early. It will not impact any of your medical care. If you participate in the interview but want to stop early, you may still receive the $20 gift card.

What are the potential risks of the study?
There are no physical or medical procedures included in this study. This means that there is no risk of physical harm. You might become sad, anxious, or frustrated when answering some of the questions about your life with bvFTD. If there are questions that you do not want to answer, you do not have to answer them. You can also stop the interview completely at any time. If you become very upset during the interview, we will suggest that you contact your provider. Or, if you are upset and do not want to talk to your provider, we can help you find someone else to talk to. If talking about your experiences makes you think of questions about bvFTD, we will refer you to someone who may be able to answer your questions. The other potential risk of the study is related to privacy and confidentiality. This is risk related to other people learning about your diagnosis. The research team does a lot to reduce these risks. We will talk about this more in a minute. You can also reduce the risk of people learning about your diagnosis by doing the interview in a private place.

What are the potential benefits of the study?
We do not expect you to personally benefit from the study. You might appreciate having the chance to talk about your experiences with bvFTD. You will not receive any treatments as part of this study. Even though you might not benefit directly from this study, you will help us learn more about the experiences of life with bvFTD. We may be able to use what we learn from you and other participants to improve resources and care for people with bvFTD in the future.

What will happen with the information I share?
There is a risk that someone outside the study will see your information. We will do our best to keep your information safe by using a participant ID number, removing the names of people and places from your interview data, and storing all information in secure ways – either locked in filing cabinets, or in a password-protected file on an encrypted computer.

If you join the study, we will give you a participant ID number. This number will be used to label all of your study information. This way, the only information that connects your name to your study information is a password-protected file that includes the link between your name and your participant number. Only Laynie will have access to this protected file. After your interview, we will destroy the link that connects your participant ID number to your name and contact information.

The interview will be audio-recorded and later transcribed. This means that a professional company will turn the audio into written text. The company will remove the names of any people and places that we say in the interview. This way, we can talk about people and places during the interview without having to worry about their information being included in our research. Once we have checked that the written version of the interview matches the audio recording of the interview, we will destroy the audio recording. This means that only the written version will be left, and this written version will not include any names of people or places. This written version is what we will use for data analysis, and for any papers that we write. If other researchers want access to our data, they also will only have access to this written version that does not have your name or the name of any person or place we talked about.

After we finish your interview process and send you your $20 gift card, we will destroy your name and other personal information including your contact information. At the end of the interview, we will ask if you would like to know about the results of the study. If you would like us to send you an overview of the results, we will ask for your permission to keep your contact information in a separate, protected document.

Who do I contact with questions or concerns?
You may contact the VOICE of bvFTD team with any questions or problems with this research. Contact the University of Pennsylvania IRB Office if you have questions about your rights as a participant of this study, or if you feel you have not been treated fairly.

Please reach out to one of these resources if you have questions or concerns:

Laynie Dratch, BA
Student Investigator / Associate Investigator
JHU/NHGRI Genetic Counseling Training Program
Baltimore, MD
Phone: 301-827-5029
Email: Laynie.dratch@nih.gov

Murray Grossman, MDCM, EdD
Principal Investigator
The Penn FTD Center
Philadelphia, PA
Phone: 215-349-5863

Lori Erby, PhD, ScM, CGC
Primary Associate Investigator
Medical Genomics / Metabolic Genetics Branch
NHGRI | Bethesda, MD
Phone: 301-443-2635
Email: lori.erby@nih.gov

University of Pennsylvania
Philadelphia, PA
Office of Regulatory Affairs Phone: 215-898-2614
IRB Phone: 215-573-2540

*Note: The consent forms and information sheets included in this document are those from the University of Pennsylvania. Parallel versions of these consent forms with Johns Hopkins contact information were also used.

Consent Comprehension Assessment

Informed Consent Comprehension Assessment

Participant type: Individual with bvFTD Individual at risk for bvFTD

Now I am going to ask you some questions on the consent form we just reviewed together. It's important that all participants understand the study.

1. Explain to me what this study is about. Can you tell me why this study is being done?

Sufficient Questionable Insufficient

2. What will you be asked to do if you enroll in the study?

Sufficient Questionable Insufficient

3. Are there risks to you of being in the study? What are the risks?

Sufficient Questionable Insufficient

4. Is there a chance you will benefit from being in the study? How might you benefit?

Sufficient Questionable Insufficient

5. Will this study impact your clinical care?

Sufficient Questionable Insufficient

6. What do you think about being part of a study that is designed to help others?

Sufficient Questionable Insufficient

7. Who's decision is it whether you enroll in the study? Can you say no? Would any bad things happen if you said no? How would you let the investigator know if you wanted to stop?

Sufficient Questionable Insufficient

Determination: **Able to Consent** **Not Able to Consent**

Interviewer will repeat sections of consent form for any assessment question responses deemed questionable or insufficient; targeted education may be used to improve potential subjects' understanding. A final determination that an individual is able to consent requires that the individual is found sufficient on all necessary items.

Appendix II: Assessments

Demographics Questionnaire

Demographics Survey

1. What is your current age? _____

2. What is your gender? Please circle your response.

Male Female Other _____

3. What is your race? Please circle your response.

1. White/Caucasian
2. Black/African American
3. American Indian/Alaska Native
4. Asian/Pacific Islander
5. Other _____

4. What is your ethnicity? Please circle your response.

1. Hispanic/Latino
2. Non-Hispanic/Latino

5. What is your marital status? Please circle your response.

1. Married
2. Divorced
3. Widowed
4. Single
5. Other _____

6. How many biological children do you have? _____

7. Do you have any other dependents (ex: adopted children)? Yes (how many?) ____ No

8. What is the highest level of education that you have completed? Please circle your response.

1. Less than high school
2. High school graduate
3. Some college
4. College graduate
5. Graduate degree

9. What is your most recent past or current occupation? _____

10. How would you describe your current employment status? Please circle your response.

1. Employed, Full-Time
2. Employed, Part-Time
3. Unemployed
4. Retired
5. Other _____

11. At what age were you formally diagnosed with FTD? _____ OR circle: Not Applicable

12. At what age were you first diagnosed with dementia? _____ OR circle: Not Applicable

13. At what age did your symptoms of FTD begin? _____ OR circle: Not Applicable

14. Have you had genetic testing for FTD? Please circle your response.

1. Yes
2. No
3. I'm not sure

15. If you have had genetic testing, what was the result? Please circle your response.

1. Showed that I have FTD (positive result)
2. Did not show that I have FTD (negative result)
3. I am not sure what the test found
4. Not applicable, I did not have genetic testing
5. I do not know/remember if I have had genetic testing

16. If you had genetic testing that showed that you have FTD (a positive result), do you recall which gene the change was found in? Yes [If so, which one(s)]_____ No Not Applicable

17. At what age did you learn the results of your FTD genetic testing? OR circle: Not Applicable

18. How many of your family members have had any type of dementia? _____

19. Which family members have had any type of dementia? Circle all that apply.

1. Parent 2. Child 3. Sibling 4. Aunt/Uncle 5. Cousin 6. Grandparent 7. Other 8. None

20. How many of your family members have had FTD? _____

21. How many of your family members have had Amyotrophic Lateral Sclerosis (ALS or Lou Gehrig's) or a combination of FTD/ALS? _____

22. Which family members have had FTD or FTD/ALS? Circle all that apply.

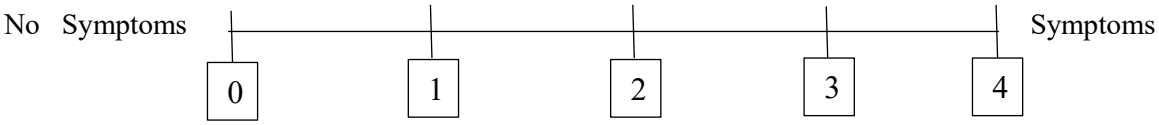
1. Parent 2. Child 3. Sibling 4. Aunt/Uncle 5. Cousin 6. Grandparent 7. Other 8. None

23. How many close friends do you have with any type of dementia? _____

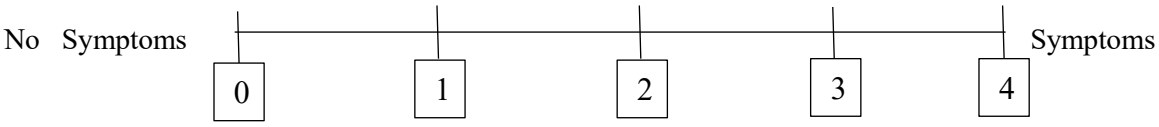
24. Do you have any other medical conditions that impact your daily life?

Yes (If so, please describe) _____ No

25. Please tell us how sure YOU are about whether or not you currently have symptoms of FTD. Please circle the number that YOU feel best represents your experience. For example, select "0" if you are sure that you have not had any bvFTD symptoms, "4" if you are sure that you have had symptoms, or somewhere in between if you are not certain about whether you have had symptoms.



26. Please tell us how sure YOUR DOCTOR is about whether or not you currently have symptoms of FTD. Please circle the number that you think YOUR DOCTOR feels best represents your experience. For example, select "0" if your doctor is sure that you have not had any bvFTD symptoms, "4" if your doctor is sure that you have had symptoms, or somewhere in between if your doctor is not certain about whether you have had symptoms.



REVISED TELEPHONE COGNITIVE SCREEN (T-CogS)

Participant ID Number: _____ Date of interview: _____

Award 1 point for each correct response. Award 0 points for each incorrect response. Mark the total points per question to the left of the numbered item.

ORIENTATION:

25. What is the year?_____

26. What season of the year is it? _____

(During March, winter or spring is acceptable; during June, spring or summer is acceptable; during September, summer, fall, or autumn is acceptable; during December, fall, autumn, or winter is acceptable).

27. What is the date or day of the month? *(+/- 1 date is acceptable)*_____

28. What is the day of the week?_____

29. What is the month?_____

30. Can you tell me where you are right now? For instance, what state are you in?_____

31. What county are you in?_____

32. What city/town are you in? _____

33. IF IN A PRIVATE HOUSEHOLD, ASK:

What is the address you are at?_____

Correct street name and house number must be given.

Zip code is not necessary for a correct response.

OR

IF INSTITUTIONALIZED, ASK:

What is the name of the place where you are staying?_____

Name of institution must be given to receive credit.

34. What is your telephone number (there/at home or where you usually can be reached)?_____

REGISTRATION:

35. I am going to name three objects. After I have said them, I want you to repeat them. Remember what they are because I am going to ask you to name them again in a few minutes.

The three objects are: “Apple”, “Table”, and “Penny”. Could you repeat the three objects for me?

The words should be read at a rate of 1 per second, speaking clearly and audibly. You are allowed to read the words only once before scoring.

Score on first trial

Apple _____ Table _____ Penny _____

Repeat the three words until:

1) the subject correctly repeats all three or

2) 3 total trials have been presented (including initial presentation).

Number of trials:_____

ATTENTION:

36. Now, I am going to give you a word and ask you to spell it forwards and backwards. The word is WORLD. Spell WORLD forwards.

(If the subject is unable to spell the word, spell it out loud, and ask the subject to repeat the spelling. Continue until it has been spelled successfully or until you have spelled it to the subject three times.

Now spell the word WORLD backwards: ___ ___ ___ ___ ___

D L R O W

Score 5 points for a correct sequence. Count 1 error for each omission, letter transposition (switching adjacent letters), insertion (inserting a new letter), or misplacement (moving W,O,R,L,D *by more than one space).*

RECALL:

37. Now, what were the 3 objects I asked you to remember?

(This should be administered as soon as the “world backwards” item is completed. Cueing is allowed if the subject is not able to recall words, but credit is not given for any word recalled after a cue).

*Apple*_____ Table_____ Penny_____

LANGUAGE:

38. I would like you to repeat a phrase after me exactly as I say it. The phrase is: "No ifs, ands, or buts".

It is very important to speak loudly and enunciate clearly as you read this phrase. One repetition of the phrase is permissible if it is clear that the phrase was not adequately heard. Otherwise, repetition is not allowed.

39. Tell me, what is the thing called that you are speaking into as you talk to me?

Correct: Telephone, Cell Phone, Hand-held, Mobile Device, etc…

40. Now I'd like you to do these three things. Say hello, tap the phone to a table 3 times, then say I'm back.

Hello_____ Tap 3 times_____ I'm back_____

Total Score (26 Max):_____

Comments:

*Revised appendix for: Newkirk LA, Kim JM, Thompson JM, et al. Validation of a 26-Point Telephone Version of the Mini-Mental State Examination. J Geriatr Psychiatry Neurol 2004; 17(2): 81-7.

Bottom Row Corrected 1/11/2005. Items 39 and 40 revised 2018 to match current telephone terminology. Formatting updated 2/10/2019.

Revised Table of In-Person and Telephone Cognitive Screen Conversions Based on Regression Equations*

In-Person Cognitive Screen Score	Predicted Telephone Cognitive Screen Score	Telephone Cognitive Screen Score	Predicted In-Person Cognitive Screen Score
0	0	0	6
1	0	1	7
2	0	2	8
3	0	3	9
4	1	4	10
5	2	5	10
6	3	6	11
7	4	7	12
8	5	8	13
9	6	9	13
10	7	10	14
11	8	11	15
12	9	12	16
13	10	13	16
14	11	14	17
15	12	15	18
16	13	16	19
17	14	17	20
18	15	18	20
19	16	19	21
20	17	20	22
21	18	21	23
22	19	22	23
23	20	23	24
24	21	24	25
25	22	25	26
26	23	26	27
27	24		
28	25		
29	26		
30	26		

Insight Probe

Insight Probe

Immediately following the completion of the T-CogS, participants will be asked the following.

1. How well do you think you answered the questions that I asked you?

2. The highest score you can get for answering all of the questions I just asked you correctly is a 26. The lowest score for answering none of the questions correctly is a zero. What do you think your score was?

Evaluation:

Question 1: Compare to LD's assessment of performance written in comments about the T-CogS, and MMSE categorization of the numerical value (ex: cognitively normal, mild cognitive impairment, etc.).

Question 2: Record the discrepancy between the actual score and the participant's guess of the score.

Appendix III: Interview Materials

At-Risk Interview Guide

Interview Guide – Presymptomatic

Hello. This is Laynie Dratch from Johns Hopkins. I'm calling for (name of participant) for our scheduled interview. Am I speaking with (name of participant)?

Are there people around you right now? Do you feel okay talking to me? Is anyone with you during this interview? Are you at home or in another location?

Before we begin the interview, I would like to ask you a few questions that I ask every participant to assess your thinking. Is this okay with you?

[Perform T-CogS and Insight Probe]

Thank you for answering those questions. Now we can begin the interview. I want to remind you that if you are not comfortable answering a question, you do not have to answer it. You may also ask for a break or stop the interview completely at any time. Do you have any questions? I want to make sure you are in a comfortable place before starting the interview. Do you feel okay talking to me where you are?

Self-Assessment:

The goal of the study is to learn about the experiences of bvFTD. We are interviewing people who have symptoms of bvFTD, and people who do not yet have symptoms. Which category do you feel like you belong in: someone with or without symptoms?

Okay, thank you. Because the study is about FTD, most of the questions will focus on your FTD experiences. But, the research team recognizes that you are a person with other life experiences, and you are more than just your FTD. Let's talk first about your life beyond FTD.

Tell me about yourself. If you were meeting someone for the first time, what would you want someone to know about who you are?

- About your personality?
- About your career and other responsibilities?
- About what you like to do for fun? Your hobbies or activities that you do?
- About your relationships with your family and friends?

Thank you. Now that I have learned a little about you as a person, I will ask you some questions about your experiences with FTD.

FTD Journey

What brought you to the [*FTD clinic* OR *research study OR support group*]?

Please tell me about the onset of [*family member's*] FTD.

- When and how did [family member's] symptoms begin?
- Who first noticed the symptoms?
- What symptoms did you notice?
- What thoughts went through your head when you started noticing symptoms?
- How would you describe how you were feeling at that time?

Please tell me about the genetic testing process.

- What made you decide to get the testing?
- What were you expecting the results to be?
- How did you feel between when you had the testing and when the results came back?
- How did it feel when the results came back? What went through your mind?

What has it meant to you to learn about the genetic/inherited nature of FTD?

- What, if anything, has it changed for you? For your family?

Tell me about how things have changed for you since you learned about your results.

- What are some of the biggest changes in terms of [relationships with people, outlook on life, daily activities]?
- How has the knowledge of risk of developing symptoms affected your life?
    - Physical symptoms? Behavioral symptoms? Cognitive symptoms? Emotional symptoms? Changed personality?
    - Tell me about a time when anticipating [*mentioned symptom*] has affected your life.
    - How have the potential for these symptoms affected your career? The activities you enjoy doing? Who you spend time with?

What have you been told about your risk of developing symptoms from [name of FTD provider]?

- What are your reactions to that information?
- How does it fit with how you thought or felt about your risk?

Personhood, Appearance to Self and to Others, Identity

You've told me about how [*changes above*] have changed for you. Besides these changes, how has FTD changed the way that you think about yourself?

- **The way you feel about yourself?**

Tell me about a time when you told someone about your FTD risk. What was it like? How did they react? How did you feel?

What changes, if any, have you noticed with your relationships with people close to you (care partner, family, friends, etc.)?

- What changes, if any, have you noticed in the way people treat you?
    - In the way people talk to you?
- What changes, if any, have you noticed in the way you treat other people?

Many people at risk of developing FTD have experiences or interactions with others that stand out to them because they were especially positive or especially negative.

- **Tell me about a time when someone made you feel bad about yourself.**
    - What do you worry about when you interact with other people?
- **Tell me about a time when someone made you feel good about yourself.**
    - What do people do that makes you feel valued? Accepted? Understood?

Tell me about an important decision that you had to make recently. How satisfied are you with how much you got to decide for yourself? How do you feel about whether other people listened to what you had to say?

- Tell me about a medical decision that had to be made. What was your role in making the decision? In participating in the appointment?
- Tell me about a time when you had to decide about an activity. What was your role in making the decision? [*employment, hobbies, driving]*
- Who helps you with decision making? How do they help? How do they include you? Please give me an example of a time someone helped you make a decision.
- To what extent do you feel like other people understand you?

How has FTD changed your sense of independence?

- How do [potential] changes in your employment impact your sense of independence?
    - How have financial changes contributed?
- How do [potential] changes in your driving relate to your sense of independence?

What do you think [care partner, friends, family] would say about how your FTD results have affected you?

- How would your [care partner, friends, family] say you have changed as a person since your testing?
- What do you think [care partner, friends, family] would say are your biggest challenges?
- How do you think your [care partner, friends, family] would say your results have positively impacted you?

Coping and Management: Facing Challenges, Finding Successes

What is a bad day for you like? What are the biggest challenges about living with your risk?

- What is hardest for you, and why? What do you wish you could do but cannot?
- What feelings does this bring up for you?

What strategies do you use to manage [any of the challenges mentioned above]?

- What are things that you do that help make [above challenges] less challenging?

What is a good day for you like? What are the most important things that you have learned from living with your FTD risk?
Who helps you with your FTD risk? With emotional support? With physical things? With appointments?

How do you get support?

- Who do you go to when things get tough? Who do you talk to about your challenges? How does this [*person or provider or organization*] help you?
- What is good about getting support this way? What is bad about it?

How do you feel about the amount of support you are getting?

- Tell me about a time when you wish that you had more support.

We talked earlier about your initial thoughts and feelings about the genetic testing results. We've also talked about how you are thinking and feeling about them now. Tell me about how you have gotten from your initial reactions to where you are now. What has helped with that? What has made it difficult?

How have you dealt with stressful situations in the past? Is that different from how you deal with stress now? If so, how?

Future Orientation

What do you think life will be like for you in the next year? In the next five years? How might it be different from your life now?

What worries do you have about the future? What challenges do you think you will face?

What are your expectations, hopes, and dreams? How have those changed over time?

Mental Health

Some people living with risk of FTD experience depression. Tell me about your experiences with that.

Some people living with risk of FTD feel lonely. Tell me about your experiences with loneliness.

Conclusion

What do you think is most important for other people to understand about living at risk of FTD?

- What is the most important thing for a [a health care provider, care partner, family member, friend, colleague] to understand?
- What would you say to another person who was just given testing results that showed high risk of FTD? What advice would you give them?

I'd like to ask you to be part of my research team for these next few questions. Imagine you are now the person in charge of an interview, and the person you are interviewing was recently given testing results that show high risk of developing FTD.

- **What questions would you ask them? What would you want to know about their experience? What do you think would be important to know about them?**

Is there anything that you think I should have asked you but didn't?

- Is there anything that we didn't talk about that you feel is important and you would like to share?

How do you feel about being in this study?

We are now at the end of our interview. The rest of the research team and I know that sharing your experience of living at risk for FTD can be difficult and emotional. We want you to know how much we value your responses and hearing your story. Thank you for your time and for your answers. Do you have any questions, comments or final thoughts?

Thank you for completing the interview. I will send you the $20 gift card soon.

Would you like me to send you a summary of the results when I finish the project?

[if yes] Great. I will need to keep your contact information to be able to send you the results. Your contact information will not be linked to any other study information. Is this okay with you?

[if no] Okay. I will go ahead and remove your contact information from the study database after I mail your gift card.

Thank you again and take care!

Diagnosed Interview Guide

Interview Guide – Symptomatic

Hello. This is Laynie Dratch from Johns Hopkins. I'm calling for (name of participant) for our scheduled interview. Am I speaking with (name of participant)?

Are there people around you right now? Do you feel okay talking to me? Is anyone with you during this interview? Are you at home or in another location?

Before we begin the interview, I would like to ask you a few questions that I ask every participant to assess your thinking. Is this okay with you?

[Perform T-CogS and Insight Probe]

Thank you for answering those questions. Now we can begin the interview. I want to remind you that if you are not comfortable answering a question, you do not have to answer it. You may also ask for a break or stop the interview completely at any time. Do you have any questions? I want to make sure you are in a comfortable place before starting the interview. Do you feel okay talking to me where you are?

Self-Assessment:

The goal of the study is to learn about the experiences of bvFTD. We are interviewing people who have symptoms of bvFTD, and people who do not yet have symptoms. Which category do you feel like you belong in: someone with or without symptoms?

Okay, thank you. Because the study is about FTD, most of the questions will focus on your FTD experiences. But, the research team recognizes that you are a person with other life experiences, and you are more than just your FTD. Let's talk first about your life beyond FTD.

Tell me about yourself. If you were meeting someone for the first time, what would you want someone to know about who you are?

- About your personality?
- About your career and other responsibilities?
- About what you like to do for fun? Your hobbies or activities that you do?
- About your relationships with your family and friends?

Thank you. Now that I have learned a little about you as a person, I will ask you some questions about your experiences with FTD.

FTD Journey

Please tell me about the onset of your FTD.

- When and how did your symptoms begin?

Who first noticed your symptoms? Was it you or someone else? [*If someone else, was it a family member, doctor, friend, etc.*].

- What symptoms did you notice? What did other people notice or tell you?
- What thoughts went through your head when [*you or others*] started noticing symptoms?
- How would you describe how you were feeling at that time?

What brought you to the [*FTD clinic* OR *research study OR support group*]?

Tell me about how things have changed for you since you started to have symptoms.

- What are some of the biggest changes in terms of [cognitive ability, physical ability, relationships with people, outlook on life, daily activities]?
- Besides [*symptoms discussed above*], what other symptoms do you have?
- How have the symptoms affected your life?
    - Physical symptoms? Behavioral symptoms? Cognitive symptoms? Emotional symptoms? Changed personality?
    - Tell me about a time when [*mentioned symptom*] has affected your life.
    - How have the symptoms affected your career? The activities you enjoy doing? Who you spend time with?
- How have the symptoms changed from when they started until now?

Tell me about the time between when your symptoms became noticeable and when you were able to get a diagnosis.

- What was it like to not have a diagnosis?
- What changed after getting a diagnosis in terms of [your thoughts about your symptoms, relationships with people, outlook on life, daily activities]?

What have you been told about your symptoms from [name of FTD provider]?

- What are your reactions to that information?
- How does it fit with how you thought or felt about your symptoms?

Personhood, Appearance to Self and to Others, Identity

You've told me about how [*changes above*] have changed for you. Besides these changes, how have your symptoms or diagnosis changed the way that you think about yourself?

- **The way you feel about yourself?**

Tell me about a time when you told someone about your FTD diagnosis. What was it like? How did they react? How did you feel?

What changes, if any, have you noticed with your relationships with people close to you (care partner, family, friends, etc.)?

- What changes, if any, have you noticed in the way people treat you?
    - In the way people talk to you?

What changes, if any, have you noticed in the way you treat other people?

Many people with FTD have experiences or interactions with others that stand out to them because they were especially positive or especially negative.

- **Tell me about a time when someone made you feel bad about yourself.**
    - What do you worry about when you interact with other people?
    - Tell me about a time when someone was embarrassed to be with you. How did you feel?
- **Tell me about a time when someone made you feel good about yourself.**
    - What do people do that makes you feel valued? Accepted? Understood?

Tell me about an important decision that you had to make recently. How satisfied are you with how much you got to decide for yourself? How do you feel about whether other people listen to what you had to say?

- Tell me about a medical decision that had to be made. What was your role in making the decision? In participating in the appointment?
- Tell me about a time when you had to decide about an activity. What was your role in making the decision? [*employment, hobbies, driving]*
- Who helps you with decision making? How do they help? How do they include you? Please give me an example of a time someone helped you make a decision.
- To what extent do you feel like other people understand you?

How has FTD changed your sense of independence?

- How do changes in your employment impact your sense of independence?
    - How have financial changes contributed?
- How do changes in your driving relate to your sense of independence?

What has it meant to you to learn about the genetic/inherited nature of FTD?

- What, if anything, has it changed for you? For your family?

What do you think [care partner, friends, family] would say about how your symptoms have affected you?

- How would your [care partner, friends, family] say you have changed as a person since your symptoms started or since you got your diagnosis?
- What do you think [care partner, friends, family] would say are your biggest challenges?
- How do you think your [care partner, friends, family] would say your symptoms have positively impacted you?

Coping and Management: Facing Challenges, Finding Successes

What is a bad day for you like? What are the biggest challenges about living with your symptoms?

- What is hardest for you, and why? What do you wish you could do but cannot?

- What feelings does this bring up for you?

What strategies do you use to manage [any of the challenges mentioned above]?
- What are things that you do that help make [above challenges] less challenging?

What is a good day for you like? What are the most important things that you have learned from living with your symptoms?

Who helps you with your FTD diagnosis? With emotional support? With physical things? With appointments?

How do you get support?
- Who do you go to when things get tough? Who do you talk to about your challenges? How does this [*person or provider or organization*] help you?
- What is good about getting support this way? What is bad about it?

How do you feel about the amount of support you are getting?
- Tell me about a time when you wish that you had more support.

We talked earlier about your initial thoughts and feelings about the genetic testing results. We've also talked about how you are thinking and feeling about them now. Tell me about how you have gotten from your initial reactions to where you are now. What has helped with that? What has made it difficult?

How have you dealt with stressful situations in the past? Is that different from how you deal with stress now? If so, how?

Future Orientation

What do you think life will be like for you in the next year? In the next five years? How might it be different from your life now?

What worries do you have about the future? What challenges do you think you will face?

What are your expectations, hopes, and dreams? How have those changed over time?

Mental Health

Some people with FTD experience depression. Tell me about your experiences with that.

Some people with FTD feel lonely. Tell me about your experiences with loneliness.

Conclusion

What do you think is most important for other people to understand about living with FTD?

What is the most important thing for a [a health care provider, care partner, family member, friend, colleague] to understand?

- What would you say to another person who was just diagnosed with FTD? What advice would you give them?

I'd like to ask you to be part of my research team for these next few questions. Imagine you are now the person in charge of an interview, and the person you are interviewing was recently diagnosed with FTD.

- **What questions would you ask them? What would you want to know about their experience? What do you think would be important to know about them?**

Is there anything that you think I should have asked you but didn't?

- Is there anything that we didn't talk about that you feel is important and you would like to share?

How do you feel about being in this study?

We are now at the end of our interview. The rest of the research team and I know that sharing your experience of living with FTD can be difficult and emotional. We want you to know how much we value your responses and hearing your story. Thank you for your time and for your answers. Do you have any questions, comments or final thoughts?

Thank you for completing the interview. I will send you the $20 gift card soon.

Would you like me to send you a summary of the results when I finish the project?

[if yes] Great. I will need to keep your contact information to be able to send you the results. Your contact information will not be linked to any other study information. Is this okay with you?

[if no] Okay. I will go ahead and remove your contact information from the study database after I mail your gift card.

Thank you again and take care!

Interview Summary Sheet

Interview Summary Sheet

Participant ID Number: _____
Participant type: Symptomatic At-Risk
Date of interview: _____
Interview start time: _____ Interview end time: _____
Interview type: Telephone In-Person
Interview location: _____

Context of interview (setting, mood, unique situations):

Adverse events?

Interview question(s) most responsive to:

Interview question(s) least responsive to:

Overall impression of interviewee's psychological state:

Overall impressions of interview:

Categories or major themes in interview:

New or different information (from previous interviews):

Suggestions for subsequent interviewees:

Appendix IV: Recruitment Materials

Recruitment Prompts

Email or Mail Prompt

Hello *(name of potential participant),*

This is [*name of recruiter*], a research coordinator in Dr. Grossman's office at the Penn FTD Center. I am contacting you because we have a Masters student, Laynie Dratch, who would like to interview you for her thesis research project. She is studying how people like you have experienced (*living with or living at risk for)* FTD.

Laynie would like to contact you to tell you more about her project and how you can participate. If you want Laynie to contact you, I need your permission to give her your name, email address, phone number, and mailing address. After talking with Laynie, you can decide whether or not you want to participate in her study. You would receive a $20 gift card after participating in the research project. Not every interested person will be able to join the study, but you might be a good fit.

If it is okay for Laynie to contact you, she would like me to send you a packet of three short forms for the study. Would you prefer this to be via email, fax, or mail?

Please let me know if it is okay to give Laynie your name and contact information. If it is okay, please tell me how you want me to send you the study packet.

Thank you for your consideration!
Best, [*name of recruiter*]

B. Email or Mail Prompt For When Permission is Received

Hi *(name of potential participant),*

Thank you for responding and I am happy to hear that you are interested in joining Laynie's study! I will send you the study packet by [*method requested*] soon.

In this packet, there are three short forms:
1. A description of the study and consent information. Please look over this form if you have time. Laynie will review this with you in detail over the phone. You keep this form.

2. A short demographics questionnaire. Please answer these questions if you have time. Laynie will ask you for the answers on the phone. You keep this form.

3. A HIPAA authorization form. It is important that you send this signed form to Laynie as soon as possible. Laynie will not contact you until she has received this form. Laynie's contact information is on this form. This is the only form that you need to sign and return to Laynie.

Please let me know if you have any questions or if you do not receive the packet.
Best, [*name of recruiter*]

C. Telephone Format

Hi! This is [*name of recruiter*], a research coordinator in Dr. Grossman's office at the Penn FTD Center. I am calling for *(name of potential participant),* is this the correct person? Hi, *(name of potential participant).*

I'm calling to talk to you about an FTD research study that you might be interested in.

We have a Masters student, Laynie Dratch, who is interested in interviewing you for her thesis project. She is doing a project about how people like you have experienced (*living with or living at risk for)* FTD.

Laynie would like to contact you to tell you more about her project and how you can participate. If you want Laynie to contact you, I need your permission to give her your name, email address, phone number, and mailing address. After talking with Laynie, you can decide whether or not you want to participate in her study. You would receive a $20 gift card after participating in the research project. Not every interested person will be able to join the study, but you might be a good fit.

Do you want Laynie to contact you for her research project?

If they say no: No problem. Thank you for consideration and have a wonderful day!

If they say yes: Great! Is it okay for me to give Laynie your contact information? *(Participant provides verbal confirmation…)* Thank you. I will note your permission in your records.

Laynie would like me to send you a packet of forms on her behalf. Do you want me to send it to you through email, fax, or postal mail?

Okay, great. I will send you the packet by [*method requested*] shortly. In this packet, there are three forms. The first form is a written version of the study details and consent information. Please look over this form if you have time. Laynie will review this with you in detail over the phone. The second form is a short demographics questionnaire. Please answer these questions if you have time. Laynie will ask you for the answers on the phone. The third form is a HIPAA authorization form. It is important that you send this signed form to Laynie as soon as possible. Laynie will not contact you until she has received this form from you. Laynie's contact information is written on this form. The only form that you need to sign and return to Laynie is the HIPAA authorization form.

Do you have any questions? [Answer questions]

Great, Laynie will contact you as soon as she receives the signed HIPAA authorization form. Feel free to reach out to myself or Laynie with any questions. Thanks and have a wonderful day!

D. In-Person Format

I wanted to mention an FTD research study that you might be interested in.

We have a Masters student, Laynie Dratch, who is interested in interviewing you for her thesis project. She is doing a project about how people like you have experienced (*living with or living at risk for)* FTD.

Laynie would like to contact you to tell you more about her project and how you can participate. If you want Laynie to contact you, I need your permission to give her your name, email address, phone number, and current mailing address. After talking with Laynie, you can decide whether or not you want to participate in her study. You would receive a $20 gift card after participating in the research project if you decide to take part. Not every interested individual will be able to participate in the study, but you might be a good fit!

Are you interested in Laynie contacting you for her research project?

If they say no: No problem.

If they say yes: Great! Is it okay for me to give Laynie your contact information? *(Participant provides verbal confirmation…)* Thank you. I will note your permission in your records.

Laynie would like me to give you a packet of forms on her behalf. Here is that packet of materials [*hand participant packet*]. In this packet, there are three forms. The first form is a written version of the study details and consent information. Please look over this form if you have time. Laynie will review this with you in detail over the phone. The second form is a short demographics questionnaire. Please answer these questions if you have time. Laynie will ask you for the answers on the phone. The third form is a HIPAA authorization form. It is important that you send this signed form to Laynie as soon as possible. Laynie will not contact you until she has received this form from you. Laynie's contact information is written on this form. The only form that you need to sign and return to Laynie is the HIPAA authorization form.

Do you have any questions? [Answer questions]

Great, Laynie will contact you as soon as she receives the signed HIPAA authorization form. Feel free to reach out to me or Laynie with any questions.

*Note: The recruitment prompts included in this document are those from the University of Pennsylvania. Parallel versions of these prompts existed for other recruitment sites.

Recruitment Letter

VOICE Of bvFTD [Voices Of Individuals: Challenges and Experiences of bvFTD]

Dear (name of potential participant),

You may be able to join a research study about living with or at risk for behavioral variant frontotemporal dementia (bvFTD). The study is run by the Johns Hopkins Bloomberg School of Public Health, the National Institutes of Health and the University of Pennsylvania. This is an interview study. The goal of the study is to learn how bvFTD impacts your day to day life, how you think about yourself, and what challenges you face. You might be able to join the study because you either have been diagnosed with bvFTD or were found to have a genetic variant associated with bvFTD. The study team at [name of site] thought you might be a good fit for the study.

The study team hopes this study will help to learn more about the needs of persons at risk or in the early stages of bvFTD. The study team hopes what they learn can lead to better future FTD care.

If you want to participate in the study, you will have at least two phone calls from the study. During the first phone call, which will take about 15 to 20 minutes, you and the researcher will review the study in detail together as part of the consent process. The researcher will talk about what you are asked to do as part of the study, and potential risks and benefits of joining the study. The researcher will ask you some basic questions about yourself and your understanding of the study as part of the consent process, and to make sure that you are eligible for the study. The study cannot include every interested person, so the study team wants to make sure that each person included is a good candidate for the project.

If you are a good match for the project based on that conversation, then you will schedule another phone call. During that next call, the researcher will ask you a few more questions about your thinking, and you will also complete the interview. The interview questions will be about your experiences living with or at risk for bvFTD. Laynie Dratch, a genetic counseling graduate student at Johns Hopkins, will interview you. The interview will last about 30 to 60 minutes.

The study team will send you a $20 gift card to thank you for your time if you participate in this interview study. It is your decision whether or not you want to join the study. You can discuss the study with your family or people that support you if you want to. This might help you decide if you want to join the study. You can share the consent form with them if you want.

If you are interested in participating or want more information, please contact Laynie Dratch. If you do not want to join the study, please contact Laynie Dratch to let her know. Thank you for your time and consideration. The study team looks forward to hearing from you.

Sincerely,
[Name of provider or site]

Laynie Dratch, B.A.
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JHU/NHGRI Genetic Counseling Training Program
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Recruitment Post

VOICE Of bvFTD [Voices Of Individuals: Challenges and Experiences Of bvFTD]

The **VOICE Of bvFTD** study is currently seeking volunteers to learn more about experiences of living with or at risk for developing behavioral variant frontotemporal dementia (bvFTD) as part of a multicenter research project on bvFTD. The study will involve interviews to help us learn as much as possible about living with bvFTD. We hope that this will guide future research, resource development, and clinical practice.

The study is called **VOICE Of bvFTD**. You must be 18 years of age or older and speak English to participate. You may be able to take part in this study if you are:

- A person who has bvFTD

OR

- A person who is at risk of developing bvFTD in the future because of an identified disease-causing change in a gene that is known to cause bvFTD, such as the *C9orf72* gene

The goal of the VOICE Of bvFTD project is to better understand how people perceive life with or at risk for bvFTD. We want to know how the disease affects your day to day experiences, how you think about yourself, and how you overcome challenges.

What's involved for a research participant?

If you participate, you will have at least two phone calls from us. During the initial phone call, which will take about 15 to 20 minutes, we will ask you some basic questions about yourself and we will review the study together as part of the consent process. During another call we will ask some questions to assess your thinking and complete the interview, which will last about 30 to 60 minutes. There are no physical or medical procedures included in this study. The consent process, screening, interview scheduling, and the interview itself will take place over two to three phone calls, which may occur over several weeks. You will be given a $20 gift card for your time if you participate in this interview study.

Contact information

If you are interested in this study or have any questions, please contact the VOICE Of bvFTD team by email at **laynie.dratch@nih.gov** or by phone at (**301-827-5029**).

Recruitment Flyer

VOICE Of bvFTD [Voices Of Individuals: Challenges and Experiences Of bvFTD]

We are currently seeking volunteers to learn more about experiences of living with or at risk for developing behavioral variant frontotemporal dementia (bvFTD) as part of a multicenter research project on bvFTD. The study will involve interviews to help us learn as much as possible about living with bvFTD. We hope that this will guide future research, resource development, and clinical practice.

The study is called **VOICE Of bvFTD**. You may be able to take part in the VOICE Of bvFTD study if you are:

- A person who has bvFTD

OR

- A person who is at risk of developing bvFTD in the future because of an identified disease-causing change in a gene that is known to cause bvFTD

The goal of the VOICE Of bvFTD project is to better understand how people perceive life with or at risk for bvFTD. We want to know how the disease affects your day to day experiences, how you think about yourself, and how you overcome challenges.

What's involved for a research participant?

If you participate, you will have at least two phone calls from us. During the initial phone call, which will take about 15 to 20 minutes, we will ask you some basic questions about yourself and we will review the study together as part of the consent process. During another call we will complete the interview, which will last about 30 to 60 minutes. There are no physical or medical procedures included in this study. The consent process, screening, interview scheduling, and the interview itself will take place over multiple phone calls, which may occur over several weeks. You will be given a $20 gift card for your time if you participate in this interview study.

Contact information

If you are interested in this study or have any questions, please contact the VOICE Of bvFTD team by email at laynie.dratch@nih.gov or by phone at (301-827-5029).

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Curriculum Vitae

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EDUCATION & HONORS

Johns Hopkins University, *ScM Candidate,* Baltimore, MD — January 2020
- Trainee of the National Human Genome Research Institute, National Institutes of Health (NIH)
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Colgate University, *Bachelor of Arts,* Hamilton, NY — May 2017
- Major: Behavioral Neuroscience; Minor: Psychology
- Summa cum laude, GPA: 3.95 / 4.00
- Dean's Award for Academic Excellence (All Eligible Semesters)
- Service Award in Psychology
- The William E. and Nellie K. Edmonston Neuroscience Award
- Charles A. Dana Scholar: Awarded for superior academic achievement and demonstrated leadership
- Liberal Arts Core Curriculum Essay Competition Winner
- Phi Eta Sigma: National honor society for first-year college and university students

CLINICAL ROTATIONS IN GENETIC COUNSELING

Developmental Neurogenetics, Kennedy Krieger Institute, Baltimore, MD — Fall 2019
Adult Neurogenetics, The Hospital of the University of Pennsylvania, Philadelphia, PA — Summer 2019
Neurogenetics, The Johns Hopkins Hospital, Baltimore, MD — Spring 2019
Cancer Genetics, Greater Baltimore Medical Center, Towson, MD — Spring 2019
General and Cancer Genetics, Walter Reed National Military Medical Center, Bethesda, MD — Fall 2018
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Intramural Researcher, Thesis Student — Spring 2018 – Present
National Human Genome Research Institute, NIH, Bethesda, MD
Advisor: Dr. Lori Erby, PhD, ScM, CGC, "*'It takes away what made a person that person'*: Reconstructing identity in response to genetic risk of behavioral variant frontotemporal dementia"

Thesis Student — Spring 2017
Department of Psychology & Neuroscience, Colgate University, Hamilton, NY
Advisor: Dr. Scott Kraly, PhD, Co-researcher: Connor Dufort, "Neonatal clomipramine treatment influences voluntary exercise and anxiety behavior in adolescent rats"

Research Intern — Summers 2015, 2016
Penn FTD Center, Penn Medicine, Philadelphia, PA
Principal Investigator: Dr. Jeffrey Phillips, PhD

Research Assistant — Spring 2014 – Fall 2015
Department of Psychology & Neuroscience, Colgate University, Hamilton, NY
Professor: Dr. Neil Albert, PhD

PUBLICATIONS

Phillips, J. S., Da Re, F., **Dratch, L.**, Xie, S. X., Irwin, D. J., McMillan, C. T., ... & Grossman, M. (2018). Neocortical origin and progression of gray matter atrophy in nonamnestic Alzheimer's disease. *Neurobiology of aging*, *63*, 75-87.

ACADEMIC PRESENTATIONS

National Human Genome Research Institute, Symposium, *Poster* Fall 2019
VOICE of bvFTD - Voices of Individuals: Challenges and Experiences of Behavioral Variant FTD

National Institutes of Health, Clinical Conference, *Speaker* Fall 2019
Huntington Disease Genetic Testing in Minors

Johns Hopkins School of Medicine, Genetics Careers Panel, *Speaker* Winter 2019

National Institutes of Health, Clinical Conference, *Speaker* Winter 2018
Facilitating Family Communication in Genetics: The Role of the Provider?

Colgate University, Thesis Presentation, *Poster* Spring 2017
Neonatal Clomipramine Treatment Influences Voluntary Exercise and Anxiety Behavior in Adolescent Rats

TEACHING EXPERIENCE

DNA Day Ambassador Spring 2018 - Present

- Teach high school students in Baltimore and Philadelphia about DNA Day and genetic counseling, and engage them in science lab work and case examples

Verbal SAT Coach and College Mentor Fall 2016- Spring 2017
Let's Get Ready, Hamilton, NY

- Taught, mentored, supported, and served as a college guidance counselor as part of an SAT tutoring program to assist students through the college application process

LEADERSHIP EXPERIENCE

Student Representative Fall 2014 – Spring 2017
Department of Psychology & Neuroscience, Colgate University, Hamilton, NY

Executive Board Member (Vice President, Professor of the Year Chair) Spring 2015 – Spring 2017
Phi Eta Sigma Honor Society, Colgate University, Hamilton, NY

Student Mentor Spring 2017
Impulse, Colgate University, Hamilton, NY

- Founding member of a club that provides resources and support for women and underrepresented populations in the sciences

ADDITIONAL EXPERIENCE

Volunteer Fall 2017 - Present
NIH Children's Inn, Bethesda, MD

Foster Volunteer Fall 2017 - Present
City Dogs Rescue & City Kitties, Washington, D.C.

Zumba Fitness Instructor December 2014 - Present

Signature Programs Intern Summer 2016 – Spring 2017
Center for Career Services, Colgate University, Hamilton, NY