The Impact of Early-Onset Hereditary Essential Tremor on Affected Individuals: A Qualitative Study

Master’s Thesis

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Master of Science

by

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ABSTRACT

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A thesis presented to the Department of Biological Sciences, Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

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Essential tremor (ET) is a non-life-threatening progressive neurological disorder that causes postural and kinetic tremor in the hands and other body parts. Age of onset, degree of tremor severity, and body parts affected are all highly variable, and an estimated 10 million Americans are affected. The genetics of ET is currently not well understood, but many individuals have a positive family history with an autosomal dominant pattern of inheritance. ET is often regarded as a disorder of the elderly, but with peaks in tremor manifestation in the second and sixth decades of life, many young individuals are also affected. Few studies have addressed the younger ET population and issues facing these individuals. In our study, eight individuals with early-onset (tremor onset before age 30) hereditary essential tremor participated in audiotaped interviews to further explore the impact of this condition on their lives. The format of the interview was based on a semi-structured interview guide that prompted discussion of life issues. The three main themes derived from the interviews included difficulties in social
situations, restrictions and limitations, and adaptation, coping, and acceptance. ET had a significant impact on social life, interactions with strangers, functionality in daily living, careers and hobbies, relationships with friends and family, and overall attitude. Positive and negative experiences with medical providers and support groups were also described. Our study provided a detailed look into the lives of young individuals with ET, and the findings suggest that there are many functional and emotional issues surrounding this condition. The degree to which these issues impact those with ET may vary based on the individual’s age and age of tremor onset. Medical providers need to be aware of the multidimensional effects of ET, and genetic counselors may be able to assist in providing support and resources to individuals with ET.

Keywords: essential tremor; early-onset disorder; emotional issues; physical disability
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INTRODUCTION

Essential tremor (ET) is a progressive neurological disorder that causes an involuntary, rhythmic trembling in the hands and other body parts of affected individuals. ET is the most common type of abnormal tremor in adults, and an estimated 10 million Americans are affected by this condition. Classic ET is characterized by postural and kinetic tremor. In patients with advanced ET, intention tremor, rest tremor, and impaired tandem walking may also develop. In 95% of patients, tremor is apparent in the upper limbs; however, the face and jaw, voice, tongue, trunk, and lower limbs can also be affected. The tremor may gradually increase in amplitude over time and spread to other body parts. Age of onset, degree of tremor severity, and body parts affected are all highly variable (Aridon et al., 2008; Deuschl and Elble, 2009).

The genetic component of ET is currently not well understood. Approximately 50-70% of individuals with ET have a family history of the disorder. Linkage studies have identified four candidate loci associated with ET in some large families, and these include \textit{ETM1} on 3q13, \textit{ETM2} on 2p24.1, a locus on 6p23, and most recently \textit{LINGO1} on 15q24.3. However, genetic heterogeneity is evident in that only a small number of families are linked to these loci. It is likely that many genetic and non-genetic factors play a role in ET manifestation. In families in which multiple members are affected, the mode of inheritance appears to follow an autosomal dominant pattern and tremor onset typically occurs at an earlier age (Aridon et al., 2008; Deng et al., 2007; Louis and Ottman, 2006).
Treatment for ET is available, but the efficacy of treatment options varies among individuals. Propranolol and primidone are two of the most effective medications available, yet only 50% of patients experience some tremor improvement from these options. These medications rarely completely suppress tremor and average tremor reduction is about 50%. Also, these medications are most effective for hand tremor; patients with tremor in other body parts may not benefit from use of propranolol or primidone. Botulinum toxin injections may be used in cases of vocal cord tremor and hand tremor, but when used for hand tremor, an overall weakness in the wrist or fingers usually results. Stereotactic surgery has been shown to be the most effective treatment for ET, but it is typically only performed for those who fail to respond to other treatment options. Of the two surgical options available—deep brain stimulation (DBS) and thalamotomy—DBS produces the best results with the fewest side effects. About 70-90% of patients experience tremor improvement in the limbs and other body parts after DBS. Like all ET treatments, however, not all patients will experience tremor reduction after DBS, and the efficacy of the procedure may decrease over time in some patients. A unique feature of ET is that drinking alcohol has been shown to temporarily alleviate tremors. Unfortunately, there appears to be a rebound effect following the period of tremor reduction. Individuals may also use alcohol as a form of self-medication which can result in an alcohol addiction (Elble and Deuschl, 2009; Louis et al., 2010; Mostile and Jankovic, 2010).

Historically, ET has been termed “benign tremor.” This name, however, is no longer considered appropriate. Although non-life-threatening, several studies have shown that ET can have a significant impact on an individual’s functional and emotional well-
being. Described as purely a movement disorder, some degree of functional disability is expected in affected individuals; however, a number of non-motor features appear to be additional sources of disability.

Several studies have described the disability experienced by ET patients in carrying out activities of daily living. Koller et al. (1986) and Louis et al. (2001) assessed the difficulty in performance of tasks including handwriting, drinking liquids, fine manipulations, dressing, and eating in individuals with ET. Most patients had difficulty with these tasks and many had to adopt new strategies or completely discontinue tasks. For example, some individuals could only drink using a straw, and others abandoned writing activities. The tasks that subjects had the most difficulty with were drinking, writing, pouring, and signing one’s name. Overall, 75% of individuals with ET reported significant disability caused by the condition. These studies highlight the fact that normal, everyday activities can become a challenge to individuals with ET (Koller et al., 1986; Louis et al., 2001).

Other studies have found that the impact of ET can extend beyond functionality to affect individuals both mentally and emotionally. Busenbark et al. (1991) used the Sickness Impact Profile to evaluate ET patients in 12 categories of dysfunction spanning both physical and psychosocial issues. Overall, the scores in the psychosocial categories for the ET group differed more from the control group than in the physical categories, suggesting that the psychosocial impact of ET on individuals may be more significant to patients than the physical impact (Busenbark et al., 1991). In 2006, Lorenz et al. looked at the impact of ET on the quality of life and personality profile of patients with ET. Overall, the patients with ET scored lower in all eight areas of a health-associated quality
of life survey compared to the control population. The lowest scores were in the areas of role limitations due to physical symptoms and mental well-being. In the responses from ET patients, the mental categories differed more from the control population than the physical categories. Like the study by Busenbark et al., this study showed that the most significant burden of ET in this population was caused by psychological and psychosocial factors rather than functional disability (Lorenz et al., 2006).

Embarassment has long been thought to be a cause of distress and social phobia for affected individuals, and a handful of small studies have noted embarassment in patients. Louis and Rios (2009) assessed the prevalence of embarassment in patients with ET, showing that even among mild cases of tremor, almost one-half of patients reported embarassment. Women were twice as likely as men to feel embarassment regardless of tremor severity. In terms of age of onset, with each 10-year reduction in the age patients first noticed tremor, the likelihood of embarassment increased by 20%. Overall, this study demonstrated that embarassment is a common finding among individuals with ET and may be a source of disability, inhibiting individuals from participating in social activities (Louis and Rios, 2009).

ET is a unique disorder. It is defined by its motor effects, yet many individuals experience significant physical, psychological, and psychosocial sequelae. Although the impact of ET in some may be minimal, the progressive nature of ET and the lack of optimal treatment options can also increase disease severity in individuals who initially had mild symptoms. ET is often regarded as a disorder of the elderly, but many young individuals are affected. Peaks in ET manifestation appear in the second and sixth decades of life (Hawkins-Walsh, 2003). Previous research has tangentially addressed
onset age, but has not focused on young individuals with ET and the effect of this disorder on their lives. Tremor, as shown by many of the studies described, can cause both functional and emotional difficulties that may interfere with occupational tasks or cause social phobia. Manifesting symptoms of ET at a younger age may compound issues surrounding lifestyle choices and interactions with others, and the impact on this population needs to be studied.

Patients desire tailored information, shared decision making, emotional support, and empathy from their healthcare providers (van der Eijk et al., 2011). In order to offer this, it is important for healthcare providers to have a good overall understanding of the impact of disease on affected individuals. Previous studies have shown the many ways in which ET can cause distress and disability, but an in-depth study of the main issues from an affected individual’s perspective has yet to be performed. By interviewing individuals with early-onset ET about the effect it has had on their lives, the implications of this disorder will further be characterized in a novel way. The goal of this study is to use the experiences shared by the participants to develop a better understanding of the impact of this condition and determine ways for medical professionals to provide better care and support for individuals with ET.
METHODS

The study design described was approved by the Brandeis University Institutional Review Board.

Study Population

We recruited participants for this study through essential tremor support groups in Arizona and Virginia. A recruitment notice (see Appendix A) was provided to the leaders of these support groups, and they were asked to distribute the information to their members at their meetings or by email. The support group leaders then provided us with the contact information of any interested individuals, or the individuals were encouraged to contact us directly. We then performed a brief phone questionnaire (see Appendix B) to assess eligibility of the study candidates. Subject inclusion criteria included the following: the individual must be 18 or older, must speak fluent English, must personally have essential tremor, must have first noticed symptoms of essential tremor before age 30, and must have at least one other family member with essential tremor.

Eight respondents who expressed interest were eligible for study participation, and interviews were scheduled at their convenience. A copy of the informed consent form (see Appendix C) was mailed to each of the participants and collected prior to the interviews. In-person interviews were scheduled for four of the eight respondents, and phone interviews were scheduled for the remaining four respondents. During two of the in-person interviews, an affected parent of the participant was also present during the interview and provided additional commentary.
Of the eight participants we recruited for this study, five live in Arizona and the remaining three live in Virginia. Six participants are female and two are male. The participants represent a wide range in ages from 32 years old to 77 years old. All of the participants experienced symptoms of tremor before age 30, but the age when they first noticed symptoms spanned from before ten years old to the late twenties. All of the participants had at least one family member with essential tremor, and some had a very extensive family history which included multiple relatives. Table I shows further information about the participants.

**Interviews**

We designed a semi-structured interview guide with open-ended interview questions (see Appendix D). The main questions on the interview guide focused on the participant’s personal and family history of essential tremor, relationships with friends, families, and others, career and reproductive choices, coping strategies, and experiences with medical professionals. Question design was based on previous literature regarding essential tremor and the author’s personal experiences with essential tremor. Though the same interview guide was used for each participant, the number and type of questions asked of each participant were dependent upon the issues explored during the interview and the completeness of the answers. Questions were adapted based on thoughts and experiences expressed by the participant, but the overall interview content remained consistent.

Interviews lasted approximately 45 to 60 minutes and all interviews were audiotaped. We did not mention participant names during interviews or in interview notes.
in order to protect participant confidentiality. Study files were labeled with identification numbers and were kept in a secure location only accessible to the authors.

Data Analysis

The audiotaped interviews were transcribed by a confidential transcriptionist and the authors. We used the qualitative analysis software Atlas.ti (Version 5.0) to code the transcripts and organize the data into themes that described the experiences of living with early-onset hereditary essential tremor. The transcripts were read through line by line and open coding was used to identify significant issues facing the participants. The codes and key quotes were then grouped into themes. These themes consisted of broad topics that were emphasized in the interviews and highlighted the most commonly expressed experiences and views.
<table>
<thead>
<tr>
<th></th>
<th>Number</th>
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<tr>
<td>Female</td>
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<td></td>
</tr>
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<td>37.5</td>
</tr>
<tr>
<td>50-69</td>
<td>3</td>
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</tr>
<tr>
<td>70 and older</td>
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</tr>
<tr>
<td>Virginia</td>
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</tr>
<tr>
<td>3-5</td>
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<tr>
<td>Greater than 5</td>
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<tr>
<td>20-30 years old</td>
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</tr>
<tr>
<td>Legs</td>
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<tr>
<td><strong>Current treatment</strong></td>
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<tr>
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<td>37.5</td>
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<tr>
<td><strong>Diagnosis of ET by neurologist/PCP</strong></td>
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</tr>
<tr>
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<td>0.0</td>
</tr>
<tr>
<td><strong>Referral to genetic counselor for ET</strong></td>
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</tr>
<tr>
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<td>0.0</td>
</tr>
</tbody>
</table>

Table 1. Demographics of the participants (N = 8).
RESULTS

Themes

The eight interviews provided an in-depth view into the experiences and issues of individuals living with early-onset ET. Using codes to highlight and categorize the participants’ remarks, we analyzed the eight transcripts and identified the following recurring themes:

1. Difficulties in social situations
2. Restrictions and limitations
3. Adaptation, coping, and acceptance

Each theme is an overarching issue that encompasses several subthemes which are shown in Table 2. We will discuss each theme and subtheme by describing the participants’ responses, including quotes that illustrate the impact of ET on their lives.
<table>
<thead>
<tr>
<th>Themes</th>
<th>Subthemes</th>
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<tbody>
<tr>
<td>Difficulties in social situations</td>
<td>Embarrassment and self-consciousness</td>
</tr>
<tr>
<td></td>
<td>Reduced social activity</td>
</tr>
<tr>
<td></td>
<td>False assumptions and labels</td>
</tr>
<tr>
<td></td>
<td>Lack of understanding from others</td>
</tr>
<tr>
<td>Restrictions and limitations</td>
<td>Struggle with everyday activities</td>
</tr>
<tr>
<td></td>
<td>Impact on hobbies and career</td>
</tr>
<tr>
<td></td>
<td>Impact on choice to have children</td>
</tr>
<tr>
<td></td>
<td>Ineffective treatment options and care</td>
</tr>
<tr>
<td>Adaptation, coping, and acceptance</td>
<td>Strategies to manage tremor</td>
</tr>
<tr>
<td></td>
<td>Support from family and friends</td>
</tr>
<tr>
<td></td>
<td>Support groups</td>
</tr>
<tr>
<td></td>
<td>Positive outlook</td>
</tr>
</tbody>
</table>

Table 2. The subthemes categorized under the three major themes: Difficulties in social situations, Restrictions and limitations, and Adaptation, coping, and acceptance.
THEME: DIFFICULTIES IN SOCIAL SITUATIONS

All participants described feelings of discomfort when in public places due to their essential tremor. ET is a visible condition that subjects expressed can evoke stares and questions from strangers. Concerns about what others may think or assume was a cause of embarrassment, self-consciousness, and reduced social activity for some. The lack of understanding from others and false assumptions that they may make was described as very frustrating and an everyday challenge.

Subtheme: Embarrassment and self-consciousness

Each of the participants described instances throughout their lives where they felt particularly embarrassed or self-conscious. These events were often times when they were the focus of attention and their tremors were visible to many people. Two interviewees described being in a wedding party and the embarrassment they felt when their shaking was visible to the entire crowd of guests. One interviewee said, “I had to carry a bouquet down the aisle. That was horrible because these flowers were just quaking and shaking.” Another interviewee commented:

One time when I was in college, I was an usher at a wedding and I had to light a candle, and I couldn’t light it. I was shaking so much. I couldn’t even do it with two hands. One of the other ushers had to come over. That’s embarrassing.

Events during which tremors were apparent to a large group of people were very memorable for the participants. For another interviewee with a leg tremor, embarrassment felt due to her tremor caused her to stop participating in swimming competitions. She said:

When I needed to get out of the pool, I couldn’t stand straight up and come out of the pool on my own. I’d have
to sit down and let my legs relax before I could stand, but even then, it was still a very visible tremor that everyone at the swim meet was able to see…when you’re in a swimsuit, you can’t exactly wear baggy clothes to hide it.

There was also a common feeling of stress and pressure when being in public places and needing to perform a task. Many interviewees stated that the added stress causes their tremors to increase in severity. Several participants described the check-out line in a store as a particularly stressful situation that can cause self-consciousness. Interviewees know that signing a check or using the pinpad is a challenge for them, and they feel stress because they do not want to be a burden to the rest of the people behind them in line. One participant said:

> When I’m in a store with a lot of people and I’m checking out and have to sign the charge slip, I get so shaky. I feel that the pressure makes it so much worse. I feel like I’m holding up the line because I’m so slow, or I’m afraid that they’ll say that the signature was unacceptable.

Other social situations like eating in restaurants or simply meeting new people were a cause for self-consciousness and stress. This self-consciousness stuck with some participants and became a constant concern in these situations. For one interviewee, a handshake became a cause of embarrassment when he learned his tremor could be felt by the opposite person:

> He came in to say goodbye and I shook his hand and he said “Why are you shaking so much?” And I never thought anything of it, and I thought to myself, “When I shake people’s hands do I shake all the time?” I must, and then I started to think that. Nobody’s ever said that to me but I thought, “When I shake people’s hand do I shake?” That’s embarrassing. So now that’s another thing…a negative thought to store away.

*Subtheme: Reduced social activity*
For some participants, the embarrassment and self-consciousness felt when in these social situations caused them to avoid public places and activities altogether. Some individuals personally felt that it was easier to stay home than go out and be around others. One participant who was interviewed with her mother present said the following:

I think what’s interesting is it’s very, in my opinion with my mother, she’s very self-conscious about it, and she’s become much less active as far as going places. We don’t go out to eat. We don’t go to do things like that because she is so self-conscious about the head shaking, or the hands shaking, or the voice shaking. And talking on the phone, she’s convinced that nobody can understand her and that nobody can read her writing. And it’s not true.

Several interviewees spoke of the challenge of having ET at a young age and its effect on social activity. An interviewee with a nephew in college said, “[he] has made the comment that there were activities he didn’t join in because he got tired of people asking questions about it.” How others react to the tremor—whether with curiosity and questions or with discomfort and avoidance—aﬀected the participants’ willingness to be social. Another interviewee in her early 30s said:

But, the illness overall probably keeps me from being able to meet a wide variety of people, and the tremors and all that I think do keep people a little bit at bay. I think when you go out, I just think people are maybe a little less likely to approach you. They see something’s wrong because you’re shaking…and they don’t want to approach you. So, that’s a strange thing. I think there’s a herd type mentality. When there’s a weak one, everybody knows it. She’s sick or something’s off. That’s the weird part of socializing with it because it becomes noticeable. If somebody spends at least 20 minutes with you, they see the hands and they figure it out.

Apprehension felt when meeting new people was a common thread throughout the interviews. The methods used to cope with that apprehension varied among the
participants, but for these few, choosing to avoid social situations was one of their main coping strategies.

*Subtheme: False assumptions and labels*

False assumptions made by others regarding the reason for their tremor was particularly upsetting and hurtful for several of the participants. There was a variety of labels that strangers gave the participants out of ignorance. One of the most common assumptions made about the participants was that they had a drinking problem. Two interviewees talked about being pulled over by a police officer who then assumed their shaking meant that they were drunk. One interviewee described her experience: “[The police officer] said, ‘Why are you shaking?’ I said, ‘It’s a neurological condition. It’s called essential tremor.’ But he was ready to pull me out of that car and do a DUI. That’s frustrating.” Another interviewee described experiences of college students she knows with ET:

> The young people I know that have this who are in college get accused of that all the time. You know, their professors will say, “Why were you partying last night when you knew you had a test today?” or “You’re shaking so much right now. You should have stayed home and taken care of yourself last night instead of going out and partying.” And they say that’s so hurtful because they don’t even drink at all. That’s a very cruel thing to say and people shouldn’t pass judgment like that. It’s like they say, until you’ve walked a mile in their shoes…

Another assumption that people make is that an individual with ET is shaking because he or she is a very nervous person. Three participants described having others assume they were nervous, and for two participants, this assumption really affected their self-image. As one interviewee described:
People would always say, “Why are you so nervous? Your hands are shaking.” And I just assumed I was nervous. Instead of thinking there was anything wrong, I thought, “Get yourself together. You must be too shy, so work on your shyness”…and that’s the thing that was hard for a long time was that assumption in my 20’s and earlier that I was nervous…just a really nervous person and shy and I’m thinking, “But I’m not that nervous.”

For the interviewees who always considered themselves to be nervous people, the essential tremor diagnosis was particularly helpful because it gave them an explanation for why they shook. As one interviewee said, “Once it was identified, it was a lot easier.” However, the label of “a nervous person” really affected the self-esteem and self-image of these individuals throughout a significant part of their lives.

Subtheme: Lack of understanding from others

Essential tremor is a very common condition, and yet many interviewees commented about the limited public awareness of ET. Two participants stated that they do not discuss ET with strangers unless they are asked about it. One interviewee commented that she tries not to pay attention to others staring at her, and “if they say something about it I’ll tell them, but if they don’t then I don’t.” Two participants said that they use the comments people make about their tremors as a chance to educate. One interviewee said, “Usually I want to tell them what it is and am very open about telling them, but the majority of people didn’t know or never heard of it.” Several interviewees commented that there is more public knowledge about Parkinson’s disease and that they often compare ET to Parkinson’s to explain the condition to others.

Others interviewees described some very hurtful things others said out of ignorance. Repeatedly hearing these comments or being asked questions can be tiresome and frustrating. As one participant said:
And you know, people don’t understand the comments they make. I work at a drive-thru and people don’t understand that, when I have the speakers on, I can hear everything they say. The comments that people make are really…some days I get annoyed by them because I hear it so many times. Other days I just think that they don’t know what they’re saying, they just don’t understand it.

These comments can become exhausting and make it more difficult to use as opportunities for education. Even one participant who advocated for increasing awareness of essential tremor and talking openly to others about the condition said that some days can be very hard:

If people have been especially cruel or thoughtless, it gets to the point where I don’t want to have to explain this again. I know that it’s something that fascinates them watching it, but I really just get tired discussing it. But, I try to be polite and understand that people are curious.

**THEME: RESTRICTIONS AND LIMITATIONS**

The interviewees spoke of feeling restricted and limited in certain aspects of their lives due to essential tremor. The physical impact of ET affected their ability to perform everyday activities and continue with their hobbies and careers. ET was a consideration for some in their decision to have children. Additionally, the lack of effective treatment options available to individuals with ET limits their abilities further.

**Subtheme: Struggle with everyday activities**

All of the participants discussed difficulties performing everyday tasks that are simple, “no-brainers” to others without ET. Eating was the most commonly described activity that people struggled with. One interviewee noted:

There have been times when I haven’t been able to get food into my mouth on a fork or hold a glass. I couldn’t hold a coffee mug probably right now, even though I’m not shaky.
It would just make me shake, the weight of it, so I avoid that.

Another interviewee said that her struggle with eating certain foods was causing her to lose weight to the point that her doctor became concerned. She said:

To be honest, there are a lot of days where eating is so much of a hassle, trying to get the fork or spoon from point A to point B, that I pick and choose. I eat things like oatmeal because oatmeal will stick to the spoon. I will have a Slim Fast because I can put a straw in it and drink it. I make a lot of choices if I go out to eat, which I don’t do very often anymore. My choices are not what I’m hungry for, but what on the menu is easiest to handle.

Every participant talked about their frustrations with eating, and all but two participants also described writing as a challenge. One interviewee said that “writing is the hardest thing.” Another interviewee described that her writing worsened as the severity of her tremor progressed:

The tremor just got worse to the point where I was not happy with what I was able to do. My writing became erratic. I could be writing and suddenly it was like my hand was possessed. It goes up and down with these scribbly movements.

Two individuals said that typing was easier than hand-writing, and that as computers have become more and more common, their ET has become less of a burden. However, two interviewees noted that typing was just as difficult if not more so. One interviewee stated, “I can’t use the computer. [I] can’t use the mouse. I would love to do my writing, but I can’t write.”

This inability to write or type can have a big impact on hobbies and jobs, as will be discussed later.
Dressing and applying makeup were also frequently brought up as challenges. These tasks involve fine-motor movements with which the interviewees struggled. An interviewee who went on to have the deep brain stimulation treatment noted, “It was interfering with everyday things…I couldn’t put on earrings. You could just forget buttons or snaps. It was getting to the point where it was frustrating and I had to do something.” The female interviewees discussed having their husbands help them with earrings and jewelry, but one interviewee noted she was no longer able to wear contacts. Another interviewee said, “Applying makeup is an extreme sport. When I put eyeliner on, sometimes I have to put it on and take it off three or four times before it’s something where I can leave the house.”

Daily tasks such as eating, writing, and dressing can be very difficult for individuals with ET. One interviewee focused on how these “simple tasks become really big deals” that require thought and strategy to perform them. She said:

> Everyday tasks are…I mean, putting a key in the door has become a real problem. I have to hold one hand on the lock and hold my other hand with it to put the key in the door. Simple, simple things are so frustrating. These are things that people take for granted…everything I do has to be a deliberate thought process in order to minimize the amount of tremor.

> Essential tremor has a significant impact on these daily activities, and the hardship experienced by affected individuals can be significant.

**Subtheme: Impact on hobbies and career**

The functional disability associated with ET extends from daily activities to the hobbies and careers of affected individuals. Every participant discussed the effect of ET on a hobby they enjoy. For some, their tremor caused them to completely discontinue
their hobby. One interviewee described his passion for writing. He said that if he could
do anything, he would want to write a short story. However, his tremor makes it
impossible for him to write or type. He said, “I would love to be able to write, but there’s
no way I can do it. I have a lot of things I would like to say, but I can’t get them out.”
Another interviewee who used to work and build with wood said he can no longer do it
because of his tremors: “I used to make stuff out of wood when I was in Texas…I used to
be able to hold really small pieces and sand them, but I can’t do it anymore.”

However, not all of the participants were forced to give up on their hobbies. For
many, their tremors made their hobbies more difficult, but they were able to continue
with a little creativity. One interviewee commented:

I used to do beadwork and I’d work with little seed beads
and then I realized that I couldn’t do it anymore…but now
I’m going to have to reshape how I do that because I love
doing beadwork and making jewelry. I just have to learn
how to do it differently. I need to use bigger stones. I’m
starting to get more into working with stones versus beads,
so that will allow me a little bit more grasp on each piece
and longer needles. So, I had to drive the car around and do
it in a different way, and I’m learning that I don’t want to
give up that part of my life.

Although most interviewees were negatively affected in their abilities to continue
their hobbies, one participant noted that her tremor actually benefitted her. As a painter,
she found that painting provided a way to calm her tremor, but even on days when she
was very shaky, her tremor created a unique paint stroke that worked well in her
paintings. She said, “I learned fast that I was actually good at painting…and it’s been
great for tremors because it puts them in their place. But also, the more I concentrate, the
better I get the fine lines, and essential tremor is actually a boon instead of a hindrance.”
Five of the eight participants also discussed the impact of ET on their careers. These individuals noted that their tremor interfered with their tasks at work. Two participants who worked as secretaries said that their ET affected the types of jobs they applied for. One interviewee said, “I limited myself to entry-level jobs like filing and things, especially as I was getting older and my tremors were worse.” Another participant who was a teacher in her 30s said that her inability to write impacted the types of assignments she gave her class:

What I noticed was that my handwriting disappeared. I would be writing something and all of a sudden, no matter how much I tried to hold then pen, it would just totally disappear. And it usually disappeared at about 10 o’clock in the morning when I was grading papers. There was no pain or anything. I would just look and realize that I couldn’t write anymore. I had stacks of papers, and as I started teaching and essential tremor started taking over my life, I quit assigning term papers because I couldn’t grade them.

An interviewee who worked as a realtor remembered trying to hide his tremors in front of his clients. He said, “I used to have to go to people’s houses and sign papers and I used to be able to cover...I’m sure they saw the tremors but I covered them up pretty well. If you saw me put the paper down here [on my lap], I would be trying to hide it.”

The effect of stress on job performance was also a common topic of discussion. One interviewee described her job as a cashier in a small but busy shop at age 16. She said, “We did long lines on a Saturday and I would be going as fast as I could, but the stress really activated the tremors. I would come home and I was really wound up. That’s when it was very, very noticeable.” Another interviewee previously worked as a bank teller but now works in a café. She noted the difference in her tremor severity at her two work places and felt that stress made all of the difference. She said, “If your job is
stressful, it’s definitely going to affect your performance. The stress is going to bring on the tremors ten-fold.”

Overall, ET affected the participants’ abilities to enjoy their hobbies and perform at work. Their difficulties stemmed from lack of functionality but also included embarrassment and stress.

Subtheme: Impact on choice to have children

The participants were asked if they considered the possibility of passing on ET to their children and if that had any influence on their choice to have children. Of the eight participants, four had children while four did not. Two participants with children said that, before they had children, they had never thought about the risk that their children may also have ET. However, these two participants said that had they known, it would not have prevented them from having children. One interviewee said:

I never thought about it. And I don’t think it ever would have stopped me. It’s serious, but not serious enough to not give life. To me, it wouldn’t even come into the picture. Even if I knew what I do today, it wouldn’t have interfered with anything.

The other participant said that, because “there are all degrees” of severity of ET, he would still have had children had he known their risk.

Of the four interviewees with children, all four also had children who were affected with ET. Two of the participants expressed a lack of concern for their children because their children’s tremor severity was minimal compared to their own. One participant stated, “As far as my kids are concerned, they have tremors now and then, but they’re nothing compared to what my brother and I had. So, they don’t discuss it and it doesn’t interfere with anything they’re doing at this time.” Another interviewee had a
daughter in elementary school who just began to notice tremors in herself. However, the interviewee felt that her daughter has a good support system in place and that they talk to each other openly about ET. She said:

I don’t think it scares her. When she noticed it in her hands, she came to me about it and said, “Is this how you always feel?” and I said, “Yeah. Right now, you only see it when you’re nervous, or scared, or sick. You may eventually have it full time, but you may not and you’ll just have them when you’re nervous or scared”…but luckily with her having a teacher with it, me having it, and her grandmother having it, I don’t think it’s going to faze her when it does turn full time.

Although most of the participants agreed that essential tremor would not have kept them from having children, two participants without children stated that it would have been a concern for them. One interviewee reflected on her own experience with ET at a young age and knowing the difficulties of being a child with ET. She said:

I think about the younger generation—kids in high school and elementary school. Kids are so mean and cruel. They make all these thoughtless comments. My niece’s son who is six years old is going to have a tough time. He’s going to have a really tough time…self-esteem is so hard to keep when you’re a teenager anyway; you’re all over the place. To deal with something like this makes it a lot worse.

Subtheme: Ineffective treatment options and care

The lack of effective treatment options for essential tremor is another limiting factor that was discussed by the interviewees. Many expressed frustration with the little success they have had with medications. One individual said, “Well, they started me off years ago with a very low dose, and it helped. But, unfortunately as you know, essential tremor is something that gets progressively worse as you age, so they had to keep bumping up the dose.” Another individual received no help from one of the most
common medications prescribed for ET and finally opted to proceed with deep brain stimulation. She stated:

I started taking [propranolol] back in the 80s and it just never worked well. They kept increasing the doses and still it did not do anything for me. So I just said, ‘Enough of this.’ I decided to have the surgery and I went to the neurologist.

Five participants discussed the “horrible” side effects associated with the available medications. As one interviewee noted, “The side effects are worse than the shakes, so it’s easiest to just accept the shakes.” Another interviewee with little hope in medications said that the side effects made them even less worthwhile: “I think that may be why I don’t even try out the treatments. It’s not going to stop it; it’s not going to cure it, so why bother with the side effects? I might as well find ways of dealing with the tremor on my own.” One tactic that two interviewees mentioned for dealing with the tremors was drinking alcohol. As one interviewee said, “I do my bills, write checks and such after I had a glass of wine at night.” Although alcohol has been shown to subdue tremors, this effect can also lead to alcoholism. An interviewee described her father’s ET and his use of alcohol:

Unfortunately, so many people who drink alcohol become alcoholics because it will slow the tremor down for four or five hours, but then you get the rebound tremor which is much worse. So what do you do? You drink more alcohol. My father became a very, very bad alcoholic because he realized that alcohol would slow the tremor down. There are many, many people who have essential tremor who become alcoholics because alcohol works. I don’t drink because I’m afraid of it. I saw what happened to my dad and my family.

Another interviewee shared that he was an alcoholic is his 20s, and he attributed part of the reason to ET. He said:
But I’ll tell you another thing—I’m giving you my innermost secrets here—I drank a lot. I drank a lot to cover up my [tremors] and I became an alcoholic. Now I’m not saying that was the only reason, but that had a lot to do with it. If I were going someplace, I would take a few drinks. Fortunately I haven’t had a drink in 33 years, but it’s amazing, you cover up things.

In addition to poor treatment options, the participants also had varied experiences with medical providers. One interviewee was very skeptical about how much medical providers can help with his ET. He said, “You know, it’s ridiculous to try to work on tremors because it’s a waste of everybody’s time…at this stage of the game, nothing can help.”

One participant said that she tried several neurologists and had bad experiences until she found her current neurologist who has ET herself. She said:

The one I’m seeing now is the third or fourth one. The first one, I didn’t feel he thought too much about the essential tremor. He wasn’t really serious about it. I went there for about a year and a half and I was on a very high dose of propranolol which did absolutely nothing as far as I’m concerned. Then I went to another guy and I wasn’t that pleased with him either. Then I saw an article in the paper about a neurologist that I’m seeing now. She was in research and she specialized in essential tremor and Parkinson’s. I thought to myself, “this is the person for me.” So I went to her, and come to find out, her mother has it and she has it herself. So she was compassionate-she knew what people went through. She lived through it for all of her life. I was very comfortable with her and I continue to see her today. She was the one that encouraged me, also, to go on to have surgery.

Two additional participants noted that the neurologists who diagnosed their condition did not spend time explaining the condition to them. One interviewee described the session in which she was diagnosed:

All the doctor had me do was hold a pen upright and draw a
spiral. The original one that I drew was perfect on the right side and really squiggly on the left side. Each circle, each spiral was squiggle, perfect, squiggle, perfect. He looked at that and he said, “You have essential tremor.” I said, “What’s that?” He said, “Well, it’s also called familial tremor. You should go back in your family and find out who had a shaking disorder.” And I said, “Oh, ok.” And that was it.

The other interviewee said, “It’s not explained to you. It’s really not. It’s just “shaky.” That’s all they tell you.”

Additionally, of the eight participants interviewed, none had ever been referred to a genetic counselor for their essential tremor. Most interviewees discussed seeing their primary care provider and their neurologist for their care.

**THEME: ADAPTATION, COPING, AND ACCEPTANCE**

The final key theme identified throughout the interviews concerns adaptation, coping, and acceptance. Participants discussed many creative strategies they have developed to manage their tremor and adapt to everyday challenges. Coping was assisted by support from family, friends, and support groups, but the role of these support systems was variable. Finally, the positive outlook and attitude about the condition was a very common theme throughout all of the interviews.

*Subtheme: Strategies to control tremor*

All of the participants had developed tricks to make performing daily tasks with ET a little easier. These tricks related to many of the activities that they struggled with including eating, writing, and dressing. One interviewee noted that writing with certain pens is easier for her: “When you’re writing, you have to have a non-slip cushion so you can grab onto it. When I have to sign, I like to have a pen with a good grip on it. I don’t like these very thin ballpoint pens that you buy in the store.” Another interviewee
purchased a program that helps individuals with tremors use the computer:

I also got this special computer program that disregards extra taps when you’re using a mouse. That has greatly helped because I was all over the screen clicking everywhere. It slows it down and disregards if it taps too quickly. The other thing I have is a voice-activated program that lets me dictate to the computer and it types it out for me. It takes some time for it to get used to your voice, so there are some things I have to correct by hand, but it still helps a lot.

Several interviewees had tips to help with eating. Many discussed using a straw to drink beverages and having weighted utensils with thick handles. One interviewee said, “When I go to restaurants, I’ve been starting to take my own fork and spoon.” Another interviewee said, “I found out that if you have a cup of tea or coffee, you keep the teaspoon in the cup when you’re walking across the floor because that keeps the coffee from shaking.”

For one participant, bathing had been a problem that caused her tremor to increase in severity. She said, “I would say one of my tricks is doing the showerhead that pulls down and taking a bath and then doing my hair and stuff that way because it will affect my arms and hands if I have them up for a long time so being able to sit down in the bath and do it is probably one of my tricks.”

Additionally, one interviewee described taking yoga classes and the benefit she receives from them. She said, “I do yoga…after the yoga class, [my tremors] may be worse, but the next day I’ll hardly notice my tremors at all.”

There were many strategies and devices that the participants used to help with the activities they struggled with. These strategies were their way of adapting and continuing with the activities they need to do.
Subtheme: Support from family and friends

The participants described their experiences with support from both family and friends. As a requirement for study participation, all interviewees had at least one other family member with essential tremor. The participants were asked about discussions they had had with their families. Two participants stated that they frequently spoke with other affected family members who were very supportive because they could understand and relate. One participant commented:

I feel like we all certainly understand each other. When I speak with my nieces who I’m very close with, I’ll ask them, “How were you on a scale of 1 to 10 today?” They’ll talk about how, you know, it was particularly bad today, or I was hurried today or stressed today or very tired today. The tremors were worse. So, I try to pass along some things that I have gotten out of my research in essential tremor that I think may help.

Three participants said that they and their families joke around about ET together. As one interviewee said, “And I mean, we laugh because when we used to go out for dinner as a family, people would just look at us and think, “What in the world goes on in that house?” These participants used joking and laughing as a way to make light of the condition and cope.

Some interviewees noted that after their initial diagnosis, they discussed ET with their family members, but have not discussed it since. Two participants said that this was because their family members were less affected than they, so they did not bring it up. Others did not have a good relationship with their family members and so they did not talk about it again. Two participants said that ET is “just accepted” so they do not openly discuss it.
The interviewees’ relationships with friends were very supportive. Many participants said that they openly discuss ET with their close friends and that they are always willing to help. One participant said, “Well with friends, I talk about it all the time. When we go out and if I’m shaking, they’ll make a comment that I’m shaking and they always want to help. They always say, ‘let me do that, let me do this.’” Another interviewee said, “They’re just friends. And if I’m having a bad day they’ll carry the coffee for me.”

One participant noted that her friends have been very supportive and, though much has changed with the onset of her tremors, they still see her as the same person. She said:

> It’s strange to me because they still see me as that girl down in the canyon and I think “Oh, no. Everything’s all different. I’m all shaky, and my head’s moving. I’m not the same.” But they take me as the same and they take care of me…you want them to still need you as a friend and realize that you’re still there. You’re still the same person, and I think you’re afraid you’ll scare them.

**Subtheme: Support groups**

The individuals that participated in this study were contacted through support group directories; however, not all of the participants were active members. Half of the interviewees had attended a support group meeting once and had not visited since. The remaining half frequently attended the meetings.

The interviewees had both positive and negative experiences with support groups. Two participants shared that they felt like they could relate to others at the support group meetings and not feel embarrassed or different. As one participant said, “It’s like walking
into a room of people who are the same as you and knowing that ahead of time.” Another participant described the support group as a positive experience:

[It is] something where these people could get together and have some place soft to fall. They would accept it. No one would question what was happening. When they are with these people, they are in a comfort zone where they don’t have to worry if they splash or spill or their writing isn’t good.

Three participants also commented on the quality of the information at support groups. They noted that they learned a lot of information from attending the meetings. As one participant said, “We had wonderful speakers—we had an acupuncturist, a dietician, a massage therapist, a pharmacist—and we had lots of discussions about what we found has helped make different tasks easier…if anybody came across an article about tips for shaking, we’d talk about those.” This same participant also commented on the nature of the meetings. She said:

We did not like to focus on all the negative things. I found that focusing on negatives like shaking, shaking, shaking was really bad. I focused more on positives and the things we could do to improve our day. I felt like it’s a negative thing anyway, and if we just sat there and said, “Woe is me,” that wasn’t being supportive. That was detracting rather than adding to the support issue of it.

Although many of the interviewees had positive things to say about support groups, some discussed negative experiences they have had. One interviewee stated that she found the meetings to be depressing:

You know, I never went to the essential tremor support group. At the time they were forming it, I was interested in it. But my experience with support groups is that I’m usually more depressed when I come out of them than when I go in, and I don’t need to be any more depressed than I already am.
Another participant in her 30s said that the meeting she attended had mostly members who were older than she. She found it difficult to get support from these individuals. She said, “I didn’t see anybody that I could relate to, and they were all in totally different stages of their lives and they’re not informing me of anything that’s helping me to understand the disease. So, it doesn’t matter that they shake as much or that they look similar.”

When asked about the value of support groups, one additional participant stated that he did not see the point in attending. He argued that it would not benefit him and, if he has made it this far in his life without going, he did not need it. He said:

I’ve never been in any of it. I just live with it. I’ve never, like you say, join a group, why? I couldn’t help anybody else out. What can I help anybody with? And they certainly couldn’t help me because I have no worries. I have no hang-ups about it. I’ve gotten along all these years. I mean, I’ve gotten to my work with it, and I’ve gotten to my family with it. People look at you and you shake, so, I’ve gotten through that.

*Subtheme: Positive outlook*

Although much of the interviews were focused on hardships and challenges associated with ET, all of the participants showed a positive attitude and acceptance regarding the condition. Four of the participants compared ET to another condition in order to say that “it could be worse.” For example, one interviewee said, “I think of Christopher Reeves and his injury. He would have traded places with me any day, and I try to keep that in mind. There’s always someone worse.” Another interviewee commented:

I’ve never thought of this as a terrible handicap, ever...because, all you have to do is look around. It doesn’t take long to look around and see that this is a very minor...I
have a good friend of mine, a very good friend of mine, who lost his leg to diabetes, and just the other day lost his other foot. Tremors aren’t very bad when you think of those things.

Other conditions that the participants compared ET to were Alzheimer’s disease and Multiple Sclerosis. Two participants also made comments that everyone has good traits and bad traits, and ET is just one of the bad traits that they have. As one individual said, “I thought it was just like anything else. You get some good things, and you get some bad things. You get some good genes, and you get some bad ones. Hope you get more of the good than the bad.” The other participant said, “I hope [ET] is our worst one. I hope we don’t have any really bad ones.”

Three participants were hopeful that scientific discoveries would soon lead to the development of treatments and eventually a cure for ET. One participant said, “I so desperately hope that, before I die, there will be some breakthrough where people will be more quickly diagnosed, there will be more effective treatments.” Another interviewee said, “We’re just on the cusp of it. And hopefully there will be treatments. I’m hoping.”

Four participants also commented that ET was “just a part of life” and something that they needed to “learn to live with.” An interviewee stated, “Yeah, you know it’s my cross to bear. It is what it is. You can’t let life get you down. You’ve got to just keep on going, and I’ve enjoyed life far too much to let something like this really get to me.” Another interviewee said, “I’m thick-skinned. If someone doesn’t like it, well I’m the one who has to live with it, not them. If they can’t accept it, well, that’s the way it goes.”

These statements showed the ways in which the interviewees were coping with ET and moving forward. Although ET can be a constant, day-to-day challenge, the
comments from the participants in this study illustrate how they were positive and creative and refused to be beaten by this condition.
DISCUSSION

In this exploratory study, we interviewed eight individuals with early-onset hereditary essential tremor about the impact this condition has had on their lives. The themes and subthemes we identified highlight the key issues and common topics discussed during the interviews. Speaking with these individuals and hearing their stories enabled us to capture an insightful and detailed glimpse into their everyday lives. Through these interviews, it became clear that individuals with ET have many challenges—both physical and emotional—and that the impact of this condition on quality of life should not be overlooked.

DIFFICULTIES IN SOCIAL SITUATIONS

As shown in the study by Louis and Rios (2009), embarrassment is common among individuals with essential tremor. Embarrassment and self-consciousness were recurring themes throughout the interviews. Participants felt anxiety when in public places and around large groups of people or strangers. They did not want their essential tremor to be noticed, and several interviewees said that they could often “feel” people staring at them. The embarrassment experienced by the interviewees can reach levels that cause social phobia. Participants discussed avoiding social situations altogether to reduce distress caused by embarrassment. There appears to be a sense of isolation caused by feeling “different” that can lead to actual isolation, and this may have a significant impact on self-esteem and overall happiness.
Greater embarrassment has been shown to be associated with a younger age of tremor onset, and is also more prevalent in females (Louis and Rios, 2009). Given that only two males participated in this study, gender differences in embarrassment were difficult to identify. The two males differed from each other in the amount of time they devoted to describing embarrassment. A larger sample of male participants would be necessary to closely examine gender differences in embarrassment. Also, although all individuals involved in this study noticed symptoms before age 30, there was a wide range of onset ages including as young as six or seven years of age. The two individuals (one female and one male) with tremor onset under ten years of age spent more time discussing embarrassment and false assumptions in our interview compared to the other interviewees. However, current age also seemed to impact embarrassment level. The youngest interviewee in her 30s spent much time talking about how others see her and its effect on her social life. It may be that certain social aspects including dating and identity formation are of greater importance for younger individuals; therefore, the younger interviewees or those with tremor onset at younger ages had a greater struggle with embarrassment and self-consciousness. Medical providers need to be aware of the prevalence of embarrassment and its potential side-effects, such as isolation—especially when the patient is young or first noticed symptoms at a young age. Even with appropriate treatment that reduces tremor, embarrassment may still linger and continue to cause anxiety (Louis and Rios, 2009). Comfort in social situations and experiences with embarrassment are issues that medical providers should address with their patients in order to better assess the psychological impact of ET and address the patients’ needs.
Also related to social setting, several interviewees remarked that they were frustrated with the public’s ignorance about ET and the misconceptions that result. Strangers would often falsely label the subjects as drunk or nervous. These assumptions were distressing, and trying to correct these assumptions and educate people about ET on a daily basis became tiresome for some interviewees. Several interviewees noted that many people understand or have at least heard of Parkinson’s disease, but essential tremor, a much more common condition also causing tremors, is not well-known. Some interviewees discussed the need for increased public awareness. The interviewees hoped that greater public knowledge about ET would make their tremors less unusual, and the hurtful comments and assumptions made out of ignorance would be reduced. Healthcare providers should discuss comfortable and concise ways of explaining ET to strangers. Providers may also give patients educational materials about ET that they can hand out to others. Developing a strategy for what to say when false assumptions or questions arise may be helpful in reducing embarrassment and anxiety.

RESTRICTIONS AND LIMITATIONS

The constant struggle with basic, everyday activities was a focus of all the interviews as has been noted in the current literature. In the studies performed by Koller et al. (1986) and Louis et al. (2001), the researchers set up tasks including pouring, writing, and eating, and observed the difficulty experienced by each interviewee. In this study, interviewees discussed their daily challenges with these, and other, simple tasks. Being able to perform ordinary activities requires extra thought, time, and effort by the participants. This can not only be frustrating, but can also affect desires and choices. As one interviewee pointed out, she eats foods that are easiest to eat rather than foods she
would like to eat. This everyday annoyance can wear on the individuals and greatly affect their behavior.

Interviewees also discussed the impact of ET on their hobbies, careers, and family planning decisions—issues that have not been previously reported in the literature. Impact on both career choice and family planning decisions are salient issues in the early-onset population.

Activities and jobs requiring fine motor skills became very difficult if not impossible for the interviewees. One interviewee who always wanted to write stories felt his dreams will never be realized because of ET. Certain hobbies became very challenging for many participants. The careers of most interviewees were also affected. Some limited the jobs they applied for due to their ET, while others found themselves constantly hiding their tremor from others in the workplace. An undesirable job or inability to enjoy hobbies may have a significant impact on an individual’s quality of life. Unhappiness in these areas may translate to dissatisfaction in other areas of their lives.

The effect of ET on reproductive choices was also examined. There was a consensus among the participants that essential tremor would not have prevented them from having children, though some participants without children said their ET would have been a concern during family planning. Reasons others were not concerned included the variability in severity and that ET was “not serious enough to not give life.” Two other participants said they had never considered that their children may be at risk for ET before planning their families. These findings show that ET did not play a large role in reproductive decisions for the participants. This may be, in part, due to the fact that at this point, the genetics of ET is not fully understood and genetic testing is not available.
Should prenatal genetic testing one day be an option, it would be interesting to see if opinions would change.

Other limitations for individuals with ET include the lack of effective treatment options and satisfactory medical care. Interviewees discussed their unsuccessful trials with medications and the medications’ unmanageable side effects. As a result, many forgo treatment and choose to live with their tremor. Two interviewees spoke of the risk of alcoholism related to the un-prescribed treatment of tremors with alcohol and this treatment’s potential to become dangerous. Healthcare providers should be aware of the risk of alcohol addiction and ask ET patients about their alcohol consumption. Without promise of effective treatment options, some interviewees became skeptical of any type of care for ET. One interviewee felt that seeing his neurologist was “a waste of everybody’s time.” Three individuals discussed negative experiences with their neurologists due to lack of knowledge, not treating ET as a serious condition, or not properly explaining ET to them.

Additionally, none of the eight interviewers had ever been referred to a genetic counselor for their ET. In 2002, a study by Watner et al. found that only 24% of participants affected with ET thought that it was a heritable condition, and 44% thought it was not likely that their family members may develop the disease. Medical providers may not be educating patients about the genetics of ET because it is perceived as a “benign” condition and therefore the information seems less important, or because an associated genetic defect has yet to be discovered. Medical providers may also be unaware of the genetic basis themselves. With the increasing amount of genetic research surrounding ET, it is vital that patients are educated so they can better understand the risk to family
members, possible mode of transmission, and the potential for genetic testing in the
future. Genetic counselors are trained to provide this information to patients and explore
other psychosocial issues surrounding their condition. A referral to a genetic counselor
after a new diagnosis may be helpful to individuals with ET to ensure they receive this
information and to discuss some of the main issues surrounding the condition. This
information is especially important in family planning decisions, and genetic counselors
may be able to interpret family history and present a possible mode of inheritance
(usually AD with variable penetrance).

ADAPTATION, COPING, AND ACCEPTANCE

With the lack of effective treatments available to them, all of the participants had
tricks they employed to manage their tremor. In order to maintain their abilities, they had
to adapt and develop new strategies to perform tasks such as eating and writing. These
strategies were helpful for everyday activities as well as in their hobbies and careers. All
of the interviewees spent time sharing their inventive means of working around their
tremor. Although tasks were still burdensome, they were able to alleviate some
frustration by creating these original tricks. The participants were able to adapt, and
others with ET would benefit from learning about some of these strategies.

Support from both family and friends was also a topic of discussion in all of the
interviews. Some participants spoke of ET often with their family members because, as
individuals also affected with ET, they could personally understand the burden of the
condition. Others would joke about ET together with their family as a coping mechanism.
However, some rarely discussed ET with family because it was simply an “accepted” part
of the family and not a constant cause of concern. All experiences described with friends
were supportive. One interviewee described her fear that friends would see her or treat her differently than before her tremors progressed, but that fear was never realized. The participants stated that friends were helpful when their tremors were especially challenging and that they did not feel the need to hide their tremors around them.

The interviewees’ comments about their experiences with ET illustrate the utility of support systems for individuals affected with this disorder. In any situation where an individual faces a difficulty, having a support system in place can be of great benefit. Medical providers should make it a practice to ask about the support systems in their patients’ lives. Family and friends can be supportive and necessary for coping. If an individual does not have family or friends to talk to about their difficulties with ET, a referral to a support group would be recommended as a place to openly talk, share strategies, and heal.

Because the participants were recruited through support groups, they have all experienced at least one support group meeting. The majority described positive experiences in their support group encounters. Interviewees felt that being surrounded by others with the same condition was comforting because they had no reason to feel embarrassed in the group. They also stated that everyone in the support group understood what they were going through. Others described the meetings as informative and commented on the fact that they focus on positives and methods of coping rather than negatives. At the same time, there were some individuals who did not find support groups helpful. One individual did not return to her support group because she was younger than the rest of the members and felt out of place. Young individuals with ET may not find support groups comforting because these groups primarily consist of older individuals.
who are at a much different stage in their lives. The issues that people of dissimilar ages deal with may differ greatly. Support group leaders should be sensitive to age differences, and medical providers, especially those caring for young individuals with ET, should be aware that support groups may not be helpful to all people. At the same time, support groups should be discussed as an option with all affected individuals and their feelings about attending should be explored.

The final key theme identified throughout the interviews was a positive outlook present in all participants. While a previous study by Louis et al. (2007) has shown a higher rate of depression in individuals with ET, several interviewees felt that there were many worse conditions than essential tremor, and said that they felt lucky that this is all they have. They compared ET to other conditions including Parkinson’s disease, Alzheimer’s disease, and Multiple Sclerosis. Many were hopeful that research would soon identify the cause of ET and effective treatments would soon follow. Several interviewees also expressed that they had accepted ET and learned to make the best of it. We are not able to determine from this study, however, whether this positive attitude is present in all individuals with ET or whether it may be more predominant in the early-onset population. Most of the individuals interviewed had developed ET decades earlier and, therefore, have had a long time to cope and come to accept the condition. In their minds, they have made peace with their tremor although it continues to be a frustration for them. Individuals with a more recent onset of tremor or diagnosis may not have yet developed this positive outlook. Medical providers need to be aware that all individuals with ET may not have reached a point where they have accepted it, and the coping and psychological state for all patients should be evaluated appropriately.
IMPLICATIONS FOR PRACTICE

The results of this study better define and characterize the main issues in the lives of individuals with ET. There are many emotional challenges that these individuals struggle with, and they may not have access to all of the appropriate resources needed for coping on a day-to-day basis. The insight provided by our participants is particularly valuable to healthcare providers, as it may assist them in providing quality patient-centered care to individuals with ET. Additionally, there is a role for genetic counselors and sub-specialty healthcare providers in helping with the coping process by discussing potential challenges with the individual, facilitating support groups, and providing additional resources.

LIMITATIONS AND FUTURE STUDIES

This was an exploratory qualitative study designed to identify the key issues facing individuals with essential tremor. The interviewees generously shared their experiences so that others can have a better understanding of what it means to have ET. However, the sample size was small, consisting of only eight participants. Only two participants were male, and other demographic information including race, socioeconomic status, and level of education was not obtained. Therefore, the results of this study may not represent the views and experiences of all individuals with early-onset hereditary essential tremor and generalizability is limited. This study can be used to guide further research into the various issues identified and discussed.

Participants were obtained through support groups which may have a caused a sample bias. The individuals who attend support groups may be more severely affected, while mildly affected individuals with ET would be less likely to attend support groups or
even accept the fact that they might need help. We also noticed a difference between our experiences with the phone interviews compared to the in-person interviews. Participants that we interviewed face-to-face were much more open to sharing their stories. We were less able to build a rapport with the interviewees we spoke with over the phone, and so they may have felt less comfortable discussing such personal experiences with us. Lastly, as with all qualitative studies, there is the limitation of recall bias. It is possible that the interviewees’ memories and emotions have changed as time has passed.

Continued research in this population is warranted. Further studies should include a larger sample size incorporating individuals who do not attend support groups. They should include a broader range of ages. Additionally, a qualitative study focusing on individuals without the early-onset type of ET would allow a comparison of the main issues between these two groups.

PERSONAL REFLECTIONS

As a genetic counseling student and an individual with a personal and family history of essential tremor, this project was of interest to me on both a personal and professional level. I have grown up with essential tremor and several of my family members are also affected. I have always felt lucky that my tremor is mild; however, there are times when it will become more noticeable. As a cashier in high school, I knew customers were staring when I was particularly stressed and my tremor was more pronounced. In my college genetics class, a professor told me to cut down on the caffeine when I was unable to pipet solution into a small vial. My interest in a career in research declined when I realized my ET was affecting my lab work, and I knew the tremor would only get worse with time.
My grandfather’s ET is severe, and I have long feared tremor progression in myself. Currently, my ET does not affect my day-to-day life—but when will it? It is something that I know I will face one day and there may not be effective treatments available to me. I have discussed ET with some of my family members, but I had rarely mentioned it to anyone else before this project. I have also never met another person with this condition outside of my family, and I was interested in hearing about other people’s perspectives and experiences. After reviewing the literature, I felt that individuals with early-onset ET were under-represented, and I was curious to see if they experienced different issues than individuals with later-onset ET.

Interviewing the participants was an enlightening and rewarding process. Many of the comments I heard were thoughts or feelings that I have also had many times before. It felt good to be able to connect to the participants in a way I can connect with few others. I also heard some remarks that were unexpected or were simply a different way of looking at things. I feel that I have learned so much from these individuals and have gained a new perspective on this condition. Each person I interviewed was more seriously affected than I, but I was so impressed by their positive attitudes. The challenges they face on a daily basis are like nothing I have ever experienced, and yet they are pushing onward. They shared many of their hardships with me, but they refused to dwell on the negatives. Instead, they laugh, they hope, and they take each day one at a time. I was most inspired by their attitudes, and I thank all of the participants who so willingly shared their stories with me.
CONCLUSIONS

Individuals with early-onset hereditary essential tremor were interviewed about how this condition has impacted many different aspects of their lives. Some themes that emerged include difficulties in social situations, restrictions and limitations, and adapting, coping, and acceptance. This study is important because it is the first detailed look into the everyday lives of individuals with this condition. Relationships, major life decisions, self-esteem, and attitude can all be affected by ET. Understanding the issues is the first step for medical providers to offer higher quality and more personal care. ET is a lifelong condition for which many individuals do not have access to effective treatment, and they require continued support as they cope throughout their lifetime. Onset of ET can occur during a wide range of ages, and medical providers need to be aware that different issues may be more emphasized during different life stages. Effective counseling approaches may vary based on the age when tremor was first noticed. Genetic counselors may be more adept at recognizing the unique challenges facing each individual and providing the appropriate resources and care. It is our hope that this study emphasized that there are many key issues facing this population. Medical providers have a duty to be aware of these issues when caring for individuals with essential tremor.
REFERENCES


APPENDIX A

Recruitment notice

Do you have a personal and family history of essential tremor?

I am a graduate student in the Genetic Counseling Program at Brandeis University and I am seeking volunteers to participate in a qualitative research project. The goal of this study is to explore how early-onset familial essential tremor impacts the lives of affected individuals. My interest in this topic stems from the fact that I have essential tremor, and I am eager to learn about the experiences of others with this condition.

Participation in this research study is open to all individuals who:

- are 18 years of age or older
- first noticed symptoms of essential tremor before age 30
- have at least one other family member who has essential tremor

Participation in this study is voluntary. Following a brief phone or email questionnaire to assess eligibility, participants will be asked to take part in one audiotaped face-to-face or telephone interview that will last approximately one hour. I hope to have all interviews completed by January 10, 2010.

All identifying information of the participants will be kept confidential and will be destroyed after completion of the study. Identifying details will be changed to protect the privacy of the participants.

If you are interested in participating in this study, please contact me by email Lauren Ahles at lnahles@brandeis.edu by December 30, 2010.

I appreciate your willingness to participate in this study and look forward to hearing from you.

Sincerely,

Lauren Ahles
Genetic Counseling Graduate Student
Brandeis University
Waltham, MA
APPENDIX B

Eligibility screening tool

Name:
Location:
Phone Number:

1. Are you over 18 years old?

2. Have you been diagnosed with essential tremor?
   - If yes, at what age were you diagnosed?
   - If yes, how long have you been followed by the neurologist/PCP who diagnosed you?

3. At approximately what age did you first begin to notice symptoms of essential tremor in yourself?

4. Could you briefly describe your family’s history of essential tremor and which relatives of yours have essential tremor?

5. Would you be willing to meet for an in-person interview lasting for about an hour?
   - If unable to meet in person, would you be willing to participate in a telephone interview lasting for about an hour?
APPENDIX C

Informed consent form

BRANDEIS UNIVERSITY
DEPARTMENT OF BIOLOGY
GENETIC COUNSELING GRADUATE PROGRAM

Informed Consent to Participate in Research

The Impact of Early-Onset Hereditary Essential Tremor on Affected Individuals: A Qualitative Study

Student Researcher: Lauren Ahles
Principal Investigator: Gretchen Schneider

INTRODUCTION
Lauren Ahles is a graduate student in the Genetic Counseling Program at Brandeis University. She is conducting a research study to learn more about the issues facing individuals with early-onset hereditary essential tremor. Gretchen Schneider is a Professor of the Practice at Brandeis University and Co-Director of Clinical Training for the Brandeis University Genetic Counseling Graduate Program.

You are being invited to participate in this study because you have a personal and family history of essential tremor.

Your participation in this study is completely voluntary. You should not feel any pressure to participate. You can decide to stop taking part in this research study at any time for any reason.

Please read all of the following information carefully and ask any questions that you have about this research study. Do not sign this consent form unless you understand the information in it and have had your questions answered to your satisfaction.

If you decide to take part in this research study, you will be asked to sign this form. You will be given a copy of the signed form. You should keep your copy for your records. It has information, including important names and telephone numbers, to which you may wish to refer in the future.
PURPOSE OF STUDY
The purpose of this study is to explore the experiences of individuals who (1) have at least one other family member who has essential tremor, and (2) first noted symptoms of tremor in themselves before the age of 30. We hope to better understand the impact and experiences of individuals with early-onset hereditary essential tremor. It is our hope that the experiences shared by participants in this study will be useful in educating genetic counselors and other health professionals about the impact that essential tremor can have on the lives of individuals with this condition.

PROCEDURES TO BE FOLLOWED
You will be asked to participate in an audiotaped face-to-face or telephone interview lasting approximately one hour. During this interview, you will be asked questions regarding your experiences as an individual with essential tremor.

RISKS
Participation in this study presents no more than minimal risk. However, it is possible that by taking part in this interview, you may experience thoughts or feelings that are upsetting to you. Should that occur, Gretchen Schneider is available to talk with you.

BENEFITS
There will be no direct benefit to you for your participation in this study. We hope that in the future, information obtained from this study will help us gain a better understanding of the psychosocial and emotional experiences of individuals with early-onset hereditary essential tremor.

ALTERNATIVES
An alternative is to not participate in this research study.

PRIVACY AND CONFIDENTIALITY
All records containing identifying information, such as names, email addresses, telephone numbers, and home or work addresses will be kept strictly confidential during the study. All study related documents and materials (including consent forms, interview transcripts, and audiotapes) will be kept in a secure location accessible only to the student researcher, and any databases containing identifiers will be password protected using a password known only to the student researcher. Transcripts, interview notes, and audiotapes will be labeled with a coded ID number, which will be assigned to you upon enrollment into the study. If you are quoted or referred to in any written or oral reports of the study, you will be given an alternate name. You will never be referred to by your real name or any other identifying information in any written or oral reports based on the interview.
PAYMENT
You will not be paid to participate in this study.

COST
There will be no cost to you to participate in the study, other than the time it takes to conduct this interview.

WHOM TO CONTACT
If you encounter any problems related to study participation or have questions about the study, you may contact the student researcher, Lauren Ahles, at lnahles@brandeis.edu or (602) 558-4909.

You may also contact the Brandeis University Faculty Sponsor for this project, Gretchen Schneider, at gretchen@brandeis.edu or (781) 736-3108.

If you have questions about your rights as a research study subject, contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8136.
PARTICIPANT’S STATEMENT

I have read this consent form and have discussed with Lauren Ahles the procedures described above. I have been given the opportunity to ask questions, which have been answered to my satisfaction. I understand that any questions that I might have will be answered verbally or, if I prefer, with a written statement.

I understand that my participation is voluntary. I understand that I may refuse to participate in this study. I also understand that if, for any reason, I wish to discontinue participation in this study at any time, I will be free to do so.

If I have any questions concerning my rights as a research subject in this study, I may contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8136.

I have been fully informed of the above-described study with its risks and benefits, and I hereby consent to the procedures set forth above.

I understand that as a participant in this study my identity and data relating to this research study will be kept confidential.

Please indicate your willingness to be audiotaped by initialing here: __________

____________________________________________
Date

____________________________________________
Participant’s Signature

____________________________________________
Participant’s Name (Please Print)

____________________________________________
Date

____________________________________________
Student Researcher’s Signature
APPENDIX D

Interview guide

Participant Name:
Date:
Time:
Location:

Background Information from Eligibility Screening
Diagnosed with ET: No Yes Age of diagnosis: _____
Age when symptoms developed: ______
Relatives with ET: ________________________________________________

I. Introduction and Background
   a. Conduct informed consent process and discuss confidentiality
   b. Let the participant know that he/she can take a break or terminate the interview at any time for any reason
   c. Explain the presence and purpose of recording equipment and note-taking
      i. With your consent, I will be audiotaping our interview so that I don’t miss anything. I may also take a few notes during the interview. Afterward, the interview will be transcribed by a confidential transcription service and your name will not be associated with it. Any potentially identifying information that might come up as we talk will be deleted from the transcript before data entry.
   d. Briefly describe the research aims and purpose of interviews
      i. The aim of this study is to learn more about the experiences of individuals with early-onset hereditary essential tremor. I will be asking you questions about various ways in which essential tremor has or has not impacted your life. Any experiences you can share will be valuable.
   e. Any questions?

II. Questions
   a. I know we spoke a little about this on the phone, but can you describe your family’s history of essential tremor?
      i. Which family members have essential tremor?
      ii. Is there variation in severity in different individuals?
   b. Could you describe when you first noticed symptoms?
      i. How old were you?
ii. In which body parts do you experience tremor?
iii. How did you learn that essential tremor was the explanation for these symptoms?
iv. Have new symptoms developed since your original symptoms?
v. Can you describe how your symptoms have progressed?
vi. (If individual was formally diagnosed by a neurologist/PCP) Can you describe the diagnosis process?

**c. What have conversations about essential tremor been like in your family?**
   i. Do you openly discuss essential tremor with your family?
   ii. Do you feel more or less comfortable talking about essential tremor with certain relatives?
   iii. How has having other affected individuals in your family altered your perceptions of ET or impacted you?

**d. What have conversations about essential tremor been like with people outside of your family?**
   i. Do you share that you have essential tremor with your friends?
   ii. Do you tell people who you’re unfamiliar with that you have essential tremor?
   iii. Are there any situations in which you feel uncomfortable or embarrassed because of your tremor?

**e. How has essential tremor affected or not affected your choice in having/not having children?**
   i. Do you feel that essential tremor is genetic or not?
   ii. What feelings do you have about the possibility of passing essential tremor on to your children?
   iii. We still don’t know enough about the genetics of essential tremor to test for it in our DNA, but if someday a test was offered that identified essential tremor before birth, would you consider it during a pregnancy?

**f. How has essential tremor influenced or not influenced your choice of career?**
   i. Did you feel limited in any way in potential careers due to essential tremor?
   ii. Were you ever unable to perform your job due to essential tremor?
   iii. Have you ever changed jobs or were you ever let go from a job due to essential tremor?
   iv. Outside of your career, has ET affected the activities or hobbies you participate in?

**g. How do you cope with essential tremor in your day-to-day life?**
   i. Are you undergoing any treatments for essential tremor?
   ii. How successful have treatments been for you?
   iii. Have you ever had or considered surgery to treat ET?
   iv. Do you have any strategies of your own that help to reduce the tremor?
h. **How has your experience been with your PCP/neurologist regarding essential tremor?**
   i. Have you received psychological support from your PCP/neurologist?
   ii. Have you ever been referred to a genetic counselor to discuss essential tremor?
   iii. How has your experience been with your ET support group?

III. Closing comments
   a. Thank the participant
   b. Ask them how the interview experience was for them?
   c. Remind them that Gretchen Schneider is available as a resource if the experience any emotional distress following the interview
   d. Reassure participant of confidentiality of responses
   e. Offer to send a copy of the abstract to the participant when the study is complete