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## The Psychosocial Effects Of The Li-Fraumeni Education And Early Detection (Lead) Program On Individuals With Li-Fraumeni Syndrome

Jessica L. Ross

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THE PSYCHOSOCIAL EFFECTS OF THE LI-FRAUMENI EDUCATION AND EARLY  
DETECTION (LEAD) PROGRAM ON INDIVIDUALS WITH LI-FRAUMENI  
SYNDROME

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A

THESIS

Presented to the Faculty of  
the University of Texas  
Health Science Center at Houston  
and  
The University of Texas  
MD Anderson Cancer Center  
Graduate School of Biomedical Sciences  
in Partial Fulfillment  
of the Requirements  
for the Degree of

MASTER OF SCIENCE

by

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Houston, Texas

May 2016

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Li-Fraumeni syndrome (LFS) is a hereditary cancer syndrome that leads to an increased risk of multiple cancers. In the past five years new screening protocols have been developed that provide improved screening options for individuals with LFS. However, very little has been published on the psychosocial impact of these screening protocols. The goals of this study were to determine how participation in screening impacts individuals psychosocially, to examine the benefits and drawbacks of screening, and to evaluate possible barriers to continued screening. This qualitative study consisted of phone interviews with 20 individuals that took part in an LFS screening program at M.D. Anderson Cancer Center. Data analysis showed that benefits of screening include early detection, peace of mind, centralized screening, knowledge providing power, and screening making LFS seem more livable. Perceived drawbacks included logistical issues, difficulty navigating the system, screening being draining, and significant negative emotional reactions such as anxiety, fear, and skepticism. Regardless of the emotions that were present, 100% of participants plan on continuing screening in the program. Our data indicates that the perceived benefits of screening outweigh the drawbacks of screening. Individuals in this screening program appear to have improved psychosocial well-being because of their access to the screening program.

## TABLE OF CONTENTS

Approval Page.....	i
Title Page.....	ii
Acknowledgements.....	iii
Abstract.....	iv
List of Tables.....	vi
Introduction .....	1
Methods .....	3
Results .....	4
Discussion .....	14
Appendix A.....	19
References .....	21
Vita .....	23

## LIST OF TABLES

Table 1: Participants' (n=20) Demographic Characteristics at Time of Study .....	5
Table 2: Perceived Benefits of Screening .....	9
Table 3: Perceived Drawbacks of Screening .....	13

## Introduction

Li-Fraumeni Syndrome (LFS) is a rare hereditary cancer predisposition syndrome that is caused by mutations in the *TP53* gene.<sup>1,2</sup> Classically, this cancer syndrome has been associated with sarcomas, pre-menopausal breast cancer, brain cancer, adrenocortical carcinomas, and leukemia.<sup>2</sup> However, in recent years, more cancer types have been found to be associated with LFS, including colon, pancreatic, stomach, renal cell carcinomas, endometrial, ovarian, prostate, lung, and skin cancers.<sup>3</sup> Overall, by the age of 50, there is a 68-93% risk of developing cancer and within 18 years of their first diagnosis, at least 15% will develop a second primary, 4% develop a third primary, and 2% develop a fourth.<sup>4,5</sup> Moreover, young children may also be affected by cancers.<sup>4</sup>

The risk and occurrence of these multiple cancers creates not only a large physical burden on these individuals, but also a large psychosocial burden. For individuals that are going through the genetic testing process for LFS, 23% have clinically relevant distress that is caused by higher levels of cancer worry and greater perceived risks.<sup>6,7</sup> In addition, because 80-93% of individuals with LFS inherit it from one of their parents, the prevalent cancer history in the family also is a factor leading to this increase in overall distress.<sup>7,8,9</sup> Individuals with multiple cancers in their family due to LFS have been found to be “psychologically fragile” and experience fear of passing on “doom and death” to their future children.<sup>10</sup> It has also been reported that individuals with LFS feel that their diagnosis is always at the back of their mind and the anxiety and distress that can be associated with the diagnosis can increase dramatically during a personal or family medical crisis.<sup>11</sup>

In order to address the large concern for these individuals’ cancer risks as well as their family members’ risks, one of the most important steps for people with LFS is to be on a cancer screening regimen. The topic of screening methods for LFS patients dramatically changed in



2011 when a study performed by Villani et al. (2011) showed significant survival benefits for individuals following a new screening protocol.<sup>12</sup> A combination of ultrasound, urinalysis, blood tests, breast mammograms, colonoscopies, annual brain MRIs, and rapid whole body MRIs (WB-MRI), were used to screen these patients. Under this screening regimen, cancers were detected early in asymptomatic individuals and after 100 months of follow-up, all individuals in the screening group were living. Conversely, individuals that chose not to undergo screening had a survival rate of only 20% after 60 months.<sup>12</sup> Another study used a similar WB-MRI technique to screen children with hereditary cancer syndromes and found a sensitivity of 100% for detecting cancers, providing support that WB-MRIs could also help detect cancers in children with LFS.<sup>13</sup> These studies have allowed for new screening modalities to be offered to individuals with LFS. Before this study was published, guidelines produced by groups such as The National Comprehensive Cancer Network (NCCN), only provided LFS patients with specific screening recommendations for breast cancer and colon cancer.<sup>14</sup> However, with the new findings, the 2015 and 2016 NCCN guidelines adopted additional recommendations, including WB-MRI.<sup>15, 16</sup> The expansion of these guidelines may offer more screening options and may potentially improve the clinical picture and prognosis of LFS.

Based on the highly significant survival benefit and cancer detection demonstrated by Villani et al., M.D. Anderson Cancer Center initiated a screening program for LFS patients similar to that offered by Villani et al (2011).<sup>12</sup> Through their Cancer Prevention Center, in 2013 the **Li-Fraumeni Education and Early Detection (LEAD)** program, was initiated. The goal of this program is to provide screening and education to individuals with LFS in an effort to improve their survival and medical management.

Although the main goal of the LEAD program is to increase survival in patients with LFS, the psychosocial impact of undergoing novel comprehensive LFS screening is largely

unknown. Previous research on screening for Li-Fraumeni syndrome, prior to the introduction of comprehensive screening, showed that individuals felt screening helped provide early detection and a sense of control and security.<sup>17</sup> In contrast, there has been one case study reviewing the experience of one individual in a comprehensive screening program. This individual reported that she experienced “exhaustion” with the number of times she underwent screening and the number of cancer diagnoses she received.<sup>18</sup> At this time, no other psychosocial studies have been performed to examine these effects across more extensive LFS populations. The aims of this study were to identify the psychosocial impact of LFS comprehensive screening options on individuals with LFS and determine what factors may influence screening adherence.

## **Methods**

This study was approved by the Institutional review boards at the University of Texas Health Science Center at Houston (HSC-MS-15-0410) and the University of Texas MD Anderson Cancer Center (BS99-038).

### Participants

Individuals were eligible for this study if they were 18 years or older, English-speaking, had a germline *TP53* mutation, and completed at least one LFS screening visit at MD Anderson Cancer Center. A total of 34 participants were initially eligible for this study.

### Study Design

Potential participants received a study invitation, followed by a phone call from study personnel to determine interest in participating. After informed consent, participants completed semi-structured interviews by phone or in person that included questions regarding their experience with LFS screening. Interview questions addressed emotional reactions to screening

and test results, satisfaction and perceived efficacy of screening, drawbacks of the screening process, future screening intentions, and financial and logistical implications of screening participation. Interviews were audiotaped and transcribed using Adept Word Management professional transcription services.

### Analysis

Qualitative analysis was guided by the grounded theory approach using ATLAS-ti Scientific Software Development GmbH<sup>19</sup>. Each transcript was coded and analyzed in order to determine overarching themes. A preliminary codebook was created by the primary author (J.R.) and reviewed with a second coder (R.Y.). The primary and second coders reviewed and coded five transcripts until an inter-coder reliability of greater than 80% was reached. Coding discrepancies were discussed until a consensus was reached. The primary author coded the remaining transcripts. Codes were grouped into the following topics that reflected common themes: benefits and drawbacks of participating in LFS screening and plans for future participation.

### **Results**

A total of 34 individuals met the eligibility criteria of our study. Of these, 10 (29%) did not respond or could not be reached by phone, 4 (12%) declined participation, and 20 (59%) consented. Demographic characteristics of the participants are shown in Table 1. The final sample included 16 females and 4 males which matched the gender distribution of the eligible population. Most participants had at least one previous cancer diagnosis. Most also had at least one WB-MRI as part of their LFS screening, while four had other screening tests, such as a brain MRI.

**Table 1:** Participants' (n=20) Demographic Characteristics at Time of Study

<b>Characteristic</b>	<b>n</b>	<b>%</b>
<i>Mean Age (range)</i>	39.1 (18-61)	
18-25	3	15%
26-33	4	20%
34-41	5	25%
42-49	3	15%
50-57	2	10%
57-64	3	15%
<i>Ethnicity</i>		
Caucasian	12	60%
Hispanic	4	20%
African American	2	10%
Other	2	10%
<i>Gender</i>		
Females	16	80%
Males	4	20%
<i>Screening History</i>		
Previous WB-MRI	16	80%
Previous brain MRI only	4	20%
<i>Personal Cancer History</i>	17	85%
Mean number of cancers (Range)	2 (0-6)	
<i>Reproductive History</i>		
Number with children	11	55%
Mean number of children (Range)	1.1(1-3)	

## *Aim 1: Perceived Benefits of Participation in the LEAD Program*

### ***Early Detection***

Most participants indicated that early detection of cancer is a benefit of LFS screening and they perceived that if they were to develop cancer, it would be caught early and with a better outcome. Some reported that early detection was the sole reason they participated in screening. When thinking about what would happen if the screening program didn't exist it was indicated that, *“Early detection is key to like – I guess fighting off cancer. So I don't know how you'd do it without screening.”*

### ***Peace of Mind***

Most individuals also expressed that having access to screening has given them peace of mind, which they attributed to the lessened worry they now have for their cancer risk and to knowing more about their current health status. Specifically, one person explained that individuals with LFS worry that every illness they have may be a sign of cancer and that screening has helped mitigate those feelings: *“Actually, the screenings – when they told me, you know what, you're good. Everything's fine. And later on, if I get like a flu or something, I'm fine because I already know my screenings are good.”* When asked how screening affects their feelings about their LFS cancer risk, one person said, *“I think it's lessened my feelings about feeling worried about future cancer. I know – I mean, I'd be naïve to think it's not – that it couldn't happen to me again, because I know darn good and well it could. But, like I said, the goal is to catch things early before it's too late.”*

### ***Centralized Screening***

Participants valued having screenings centralized in one location. Prior to an organized screening program, many expressed that LFS is *“so rare, most doctors haven't heard of it.”*

They indicated how difficult it was to organize screening for multiple cancers on their own: *“it was just you or going with—you know—each department. It was a little bit overwhelming.”*

These individuals feel that a centralized screening program provides a place for keeping track of all their screenings and assurance that the screening protocol is being followed.

Respondents also valued having health care providers who were knowledgeable about LFS and who could advocate the need for screening. They noted the burden and difficulty of having to explain LFS to different health care providers who often were not knowledgeable about the syndrome, and the need to act as their own advocate in regard to their LFS-related preventive care. A participant stated that the screening program enabled affected persons to *“sit in the back seat, instead of always in the driver’s seat.”*

### ***Knowledge is Power***

Of the 20 individuals interviewed, 16 (80%) of them discussed that the information gained from screening is beneficial. For some, the information about their current health status was most important while others thought that, *“being a little bit more educated about – you know—what you put in your body and what you expose yourself to,”* was an important piece of information gained. Regardless of what pieces of information each participant felt were most powerful, many expressed that if they didn’t have access to the LEAD program they would be losing vital information. As one lady explained, *“I would feel like I’d be in the dark, like I wouldn’t know anything.”*

### ***LFS is more Livable***

It was reported that screening makes LFS feel more livable. Prior to participation in the LEAD program, a diagnosis of LFS felt like they were being told *“you have this horrible disease and you’re going to die of cancer.”* However, with the LEAD program and the set

screening protocol that goes along with it, there is hope that LFS will be more livable as this woman indicated: *“Just knowing that it’s not a death sentence—you know—and that it is livable for a lot of people.”*

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**Table 2: Perceived Benefits of Screening**

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<b><i>Themes</i></b>	<b><i>Participant Responses</i></b>
<b>Early Detection</b>	“I think the biggest thing was I knew it was going to help catch things early, and I felt that was – with the way cancer treatment is now, it’s all about catching things before they’ve progressed.”
<b>Peace of Mind</b>	“The program definitely gives me more peace of mind. Like I know I still have a significantly higher chance of getting cancer than the most average person. But I have more confidence that if I do, it will be more manageable”  “I just feel more confident. I feel I have more peace of mind and even for my child. I feel like it’s very good for her, and she knows it now”
<b>Centralized Screening</b>	“Just the overall screening and not having to take care of it yourself. It’s allowed me to step back from being my own doctor. I didn’t go to med school, I don’t know things, and people with Li-Fraumeni should not have to know as much as they do because it makes it hard to just live life. So I try and leave that to other people, and the screening has allowed me to really step back and just enjoy being me.”  “Just literally having it all under one roof, and having one person who really knows—what’d y’all call it? The syndrome? The disease? Someone who knows all about it.”
<b>Knowledge Is Power</b>	“The only way to know is to screen. And that is the most beneficial. That’s the reason I do it.”  “So, this to me is knowledge is power. You know, the more I know, the better off I am.”
<b>LFS is More Livable</b>	“I have much more confidence that if I continue doing this, that it’s more likely for me to live a long and healthy life”.

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## *Aim 2: Perceived Drawbacks of Participating in the LEAD Program*

### ***Logistical Issues***

There were a number of logistical issues that were seen as drawbacks to the screening process. For some, the time commitment was burdensome. Many individuals had to travel across the country, come in for multiple visits, and miss work in order to fulfill the screening protocol. Regardless of the participant's situation, the majority felt like the time commitment was burdensome and often times expensive. As one person indicated, *"I can't drive it. I'm not capable of driving all that way. So, I have to fly, which means scheduling and money and—you know—we're on a fixed income, so it's a burden. But I'm doing what I can to stay alive."*

The biggest logistical issue, however, involved insurance and whether it would cover the recommended screening. A large proportion of the participants (n=9, 45%) expressed that they currently have concerns about insurance coverage. One individual had recently been denied coverage by their insurance to get a WB-MRI and had a significant emotional reaction to this process: *"I got sick from it. I was depressed. I was—I got really sick."* Another group of individuals (n=8, 40%), explained that although they haven't had coverage concerns yet, they felt that insurance difficulties may prevent them from getting screening in the future. The remaining three individuals felt that insurance coverage was not a concern at this time.

### ***Organization and Navigation within the Program***

Participants felt that the organization of the program needed improvement. The lack of organization was noted in terms of communication errors between the LEAD program and the patient, scheduling problems, and lack of knowledge about the program by other physicians throughout M.D. Anderson. However, many of those that felt the organization needed improvement also expressed that they have seen improvements from the start of the program in 2013, till they were interviewed in January 2016.

Other individuals expressed that trying to navigate the program on their own was difficult. Whether it be finding the correct location or knowing who to call with their questions, this was a significant barrier to successful screening. One individual expressed concern for other participants, *“But I know other people in this—they’re going to give up and quit.”*

Although there are challenges to navigating your way through the program, one lady expressed improvement in this, *“I think your first time there, it’s a little confusing at times—which I think anytime you go someplace new is. But I think the second time is a lot easier.”*

### ***Draining***

The screening process was noted to be both physically and emotionally draining. From a physical standpoint, many of the screening techniques, particularly the WB-MRI, was said to take a long time. When asked how the participant felt during their screening visit, one person expressed, *“That I want to get out. That I have to go to the restroom or I’m just stiff. I think most of the time, the first twenty minutes, I’ll fall asleep. And once I get up, I’m like, oh, man. I’m still here.”*

Others expressed that from an emotional standpoint, *“there are still times you just get tired of it and you just kind of don’t want to do it anymore.”* Often times coming in for screenings also brought up old memories as one individual indicated, *“If you had something detrimental happen or take a trip down memory lane or – it might get you in the gut a little bit.”*

### ***Negative Emotions***

The vast majority of participants (n=18, 90%) expressed negative emotions throughout the screening process, with three main emotions being most prevalent: anxiety, fear, and skepticism. The most reported negative emotion was anxiety, or as many of the interviewees explained, *“We call it scan-xiety.”* This so called “scan-xiety” is often related to the uncertainty

of what will be found during screenings. For some, this anxiety is so severe that they report taking medication to control it. While some experience anxiety prior to screening, others report that waiting for the results is the worst part *“worse than when they actually give you the results. It’s just—it makes you crazy.”* Although the anxiety associated with the initial screening process can be very intense, around half of these individuals felt the anxiety decreased with additional screenings.

Half of the participants also expressed experiencing fear related to the screening process, which was often related to claustrophobia during the MRI’s, as one person expressed, *“I have to be strapped down.”* Another cause of fear was the presence of inconclusive or benign findings on the screening results as another person explained, *“I was so scared, and I thought I had breast cancer. But no, it was just like a little mass of fat.”*

A small proportion of the participants also experienced skepticism surrounding whether the screening results would be correct or whether the doctors themselves would be correct. When asked how it feels to get a normal screening result, one participant indicated, *“I feel relieved, but I feel like ‘are they sure?’ Like I need to read it myself, like I’m going to find something they didn’t.”*

Although there were significant negative emotions for many individuals, there were some individuals that expressed not having negative emotions related to screening. These people felt like a *“rare breed”* because of their lack of emotions related to screening. Some individuals actually felt the opposite of the majority, explaining that without screening they experience *“panic attacks”* because they feel that *“if I get a cough, I think I have like throat cancer. Or if my head is hurting or my eyes are hurting, something’s going on with me. Something’s back.”*

**Table 3: Perceived Drawbacks of Screening**

<b><i>Themes</i></b>	<b><i>Participant Responses</i></b>
<b>Logistical Issues</b>	<p>“But some people may have to pay way more than that, so I think cost is really, really prohibitive for some people being able to do all the screenings—just because it’s—imaging is expensive.”</p> <p>“I mean the time because it’s a field trip every time you go to Anderson.”</p> <p>“My insurance doesn’t cover my whole-body MRI. So I haven’t had one of those yet.”</p>
<b>Organization and Navigation within the Program</b>	<p>“It seemed a little bit disorganized at first, but I think it was just—maybe I was one of the first patients that was in it. But I think that has gotten better.”</p> <p>“I think the harder thing for me is knowing who to get in touch with to figure things out. That’s a little confusing, I will say. Because I know some people do just some things in their local area and then some things out. I wish there was a social worker at my full program that I could kind of contact to ask questions.”</p>
<b>Draining</b>	<p>“They can be kind of draining, in terms of like energy and emotionally sometimes. Just you know, knowing that you have the condition and that you have to go do it.”</p>
<b>Negative Emotions</b>	<p>“It was just the anxiety that you go through every time you’ve got to go do your screening. It’s just like, ‘Oh, my God. What are they going to find now?’”</p> <p>“I think that a lot of the anxiety has subsided and kind of the nervousness and the fear of the unknown has subsided. And now they’re familiar and I know the drill.”</p> <p>“I take a Xanax in the morning, just because—you know—being in enclosed spaces really bothers me”</p>

### *Aim 3: Plan for Future Screening*

All of the individuals that were interviewed expressed that they would like to continue screening within the LEAD program. Some of the reasons given for why they would like to continue included “*I think it would be foolish of me not to*” and “*I want to stay alive.*”

A few individuals discussed that a family member has chosen not to continue screening in the LEAD program. Reasons given for a family member not continuing screening included being physically and emotionally tired of going through the screening process, moving away from M.D. Anderson, and lack of communication between the individual and the LEAD program.

We also asked each participant what potential barriers may prevent them from continuing screening. Most individuals expressed that loss of insurance coverage was the largest barrier. Another barrier included moving to new locations where they may not have direct access to screening. This concern was particularly poignant among young individuals who may decide to move away from home because of college or future jobs. One young adult explained, “*But if I ever want to maybe transfer out of state or just anything—anything that’s out of Houston, that kind of scares me, because I’m like, “What am I supposed to do?” Like all my testing and everything is here.*” Finally, another concern for continuing screening was the uncertainty about reclassification of their familial *TP53* mutation. One individual expressed that if their mutation was reclassified as a variant of unknown significance (VUS), they may not continue screening.

## **Discussion**

This study, to the author’s best knowledge, is the first multi-participant study looking at the psychosocial effects of a novel comprehensive LFS screening program. Similar to findings in a previous study looking at screening prior to the comprehensive protocols, we also found

that LFS screening provides patients with a sense of security and participants felt that the largest benefit of screening was early detection of cancers<sup>17</sup>. However, our study was found to be in contrast to a previous case study that expressed concern that comprehensive screening may lead to testing fatigue as well as significant emotional strain on patients.<sup>18</sup> Instead, our study showed that the benefits gained from screening significantly outweigh any perceived drawbacks. Although screening increased some negative emotions such as the so called “Scan-anxiety”, fear associated with screening, and skepticism centered on the accuracy of the tests and doctors, there were also a number of negative emotions that were eliminated because of screening. Participants felt less worried about their own cancer risk, less worried about their family’s cancer risk, were less fearful about their diagnosis of LFS, and they became less focused on their own mortality. They also expressed that LFS in general feels less overwhelming and more livable with the LEAD program in place. These sentiments are supported by the fact that 100% of participants plan on continuing screening within the LEAD program.

The information we gained in this study contrasts with that found in a review article by Gopie et al. (2012) which found increased distress and lower quality of life in individuals receiving screening for hereditary cancer syndromes.<sup>20</sup> The LFS patients in this study underwent screening prior to 2012 which consisted of only breast MRIs and targeted screening dependent on family history. They were not getting comprehensive screening similar to the screening performed in the LEAD program. Prior to the LEAD program and other similar programs, there may have been significant distress and lower quality of life. With our study, however, although it was not measured directly, we have heard from patients that there is actually lowered distress and an expected increase in quality of life from participating in comprehensive screening. This shows that comprehensive screening programs are vital to patients’ well being.

Screening through the LEAD program has also provided reassurance about participants' current health status. Prior to screening, many people expressed that they felt like every lump, bump, bruise, headache, or illness was a sign of cancer. However, individuals enrolled in the LEAD program now rely on their normal screening results as reassurance that these findings are not signs of cancer. This has allowed people with LFS to live their lives more normally, without the constant mental draw of worrying that everything is a sign of cancer. The LEAD program has taken significant weight off of the patients' shoulders and allowed them to be less focused on their diagnosis and more focused on everyday life.

### *Implications*

Many comments made by participants can be used not only by M.D. Anderson to help grow and develop the LEAD program, but also by centers around the world that are in the process of trying to implement improved screening for their patients with LFS. A significant proportion of individuals stated that they would like to see more programs developed throughout the country as well as in other countries as some have family members that do not have direct access to screening because of their location. Testimonies such as these support the need for easier access to screening programs not just in the United States, but worldwide. Currently there are other groups both in the United States and overseas that utilize similar screening protocols to what is offered in the LEAD clinic, such as the University of Utah, the National Cancer Institute, the National Institute of Health, the Dana-Farber Cancer Institute, the Gustave Roussy Institute in Paris, the Institute of Cancer Research in Surrey, United Kingdom, and the Australasian Sarcoma Study Group in Victoria, Australia. It will be important to review data from these other sites once it becomes available to compare how it may be different or similar to our findings so that others can continue to learn how to develop improved screening programs across the world.

It is also important to remember that even though there are significant benefits to screening, there are still notable drawbacks that limit participation. In particular, the significant negative emotions that arise around the screening process need to be evaluated for not only by physicians but also by genetic counselors, nurse practitioners, and other staff interacting with these individuals. For some, referrals to social workers or psychologists may be necessary to help manage these emotions. All healthcare providers that interact with these individuals need to be aware of these emotions and help patients navigate these potentially difficult times.

Finally, many of the individuals interviewed desired improvements to insurance coverage. Many people stated that screening is the only thing keeping them alive and when screening wasn't covered by insurance, they expressed significant negative emotional reactions. While insurance coverage may be out of the hands of health care providers, we and other groups hope that by providing more research based evidence showing that screening is necessary for these patients' physical and emotional well-being, the insurance companies will follow suit and insurance coverage will no longer be a barrier to screening.

### *Study Limitations*

One of the limitations of this study includes its small sample size. Although our sample size appears small, we obtained a 59% response rate from the eligible participants. Of those that we were able to reach by phone, 83% chose to participate. Since this condition is rare, it is challenging to observe a large LFS population at one cancer center. Despite the small sample size, when analyzing the data, we reached saturation in terms of identifying new themes. Therefore, we feel that while having a larger sample size would be desirable, it would not change our findings significantly.

Another possible limitation of the study is the amount of time that elapsed between a participants' screening appointment and our interview. For most individuals their last screening



was weeks or months prior to our interview. This time period may have affected how well the participant was able to remember their emotions surrounding that screening experience. It is also important to note that most of these individuals received normal screening results at their last visit, which may have led to a more positive memory of the screening experience than if a cancer was identified at their last visit. However, the one individual that did have cancer found at their last screening visit did not feel negatively towards the screening process.

Finally, there were four individuals that declined to participate in our study. Those individuals may represent a unique perspective that was not captured in our study. Some of these individuals may have had negative experiences with the screening program that they did not feel like sharing with providers at the same institution as where their screening occurred. These individuals may also be those that are less likely to continue participating in screening in the future, in which case we would not have an accurate representation of their perspective.

### *Future Directions*

Although our study directly investigated individual's experiences with LFS and participation in the LEAD program, many people expressed that there is a large familial impact as well. It appears that family members are impacted both by the participants' diagnosis of LFS as well as their participation in screening. Further research should investigate how family members are impacted by these events since familial support and interactions are so important to many cancer patients.<sup>11, 21</sup>

It would also be important to talk with individuals who have quit screening or who declined initial participation in screening. These individuals were not captured in our study and it would be helpful to explore their motivations so that we can understand how to improve our screening clinic to include as many high risk LFS individuals as possible.

## Appendix A: Qualitative Interview Guide

### Prior to participation in screening program:

1. Do you have any children? If so, how many do you have and how old are they?
2. Tell me about your experience with Li –Fraumeni Syndrome?  
*Probe:* personal experience? family experience?
3. What led you to get genetic testing?  
*Probe:* Did you have concerns about testing? Was it an easy choice? Decision for children to get testing?
4. How did you first talk to your family members about Li-Fraumeni Syndrome?  
*Probe:* How did you talk to your children about Li-Fraumeni Syndrome? Did this work well?
5. How did you find out about the Li-Fraumeni Syndrome screening program?  
*Probe:* doctor? family member? other?
6. What influenced your decision to participate in the screening program?  
*Probe:* role of family or others in decision-making? expected benefits from participating (e.g. reduction in uncertainty, feelings of control over cancer risk, relief from anxiety)? Potential concerns about screening participation (e.g., worries or anxiety around screening, burden in terms of travel, cost, other)?
7. What made it easy for you to participate? What made it difficult to participate?
8. What were your expectations about participating in screening?  
*Probe:* expectations about information, knowledge of health status, emotional reactions, health care services? **What were some of your emotional feelings about participating in screening?**
9. Tell me about your experiences in getting health care related to Li-Fraumeni Syndrome prior to starting the screening program?  
*Probe: Tell me about your experiences seeking care- positives and negatives, prior to screening at M.D. Anderson* experiences with health care providers? Experiences in seeking care related to possible symptoms or early detection/screening? communication with providers about cancer risk?
10. How well-prepared did you feel for your appointment(s)?  
*Probe:* satisfaction with information received? Seek any other information on your own? Scheduling, travel, other logistic issues?

### Post-screening:

11. Do you go to screening visits alone or do people come with you? What happens for you or your family around testing time?

*Probe: Emotions? Traditions?*

12. How many screening appointments have you had?
13. Please tell me about your experience with the actual screening appointment?  
*Probe: What emotions did you have during your screening visits? What was going through your head?*
14. Please tell me about the experience while waiting for the results of your tests?  
*Probe: length of wait time, emotional reactions, concern or worry about results?*
15. Please tell me about your experience when you received your test results?  
*Probe: what was your reaction when you received an abnormal finding? what was your reaction to receiving normal results? **On any of your screenings did you ever have any unexpected non-cancerous findings?** What was your reaction to any non-cancerous findings (suggest examples)?*
16. Please tell me what you feel are positive or beneficial aspects of Li-Fraumeni Syndrome screening?
17. Please tell me what you feel are negative aspects or drawbacks of Li-Fraumeni Syndrome screening?
18. Thinking about your expectations of Li-Fraumeni Syndrome screening prior to undergoing the tests, please tell me how your experience compared to those expectations?  
*Probe: gains/losses in relation to expectations?*
19. What are your thoughts about having Li-Fraumeni Syndrome screening again?  
*Probe: short-term intentions, long-term intentions, financial/other support concerns*
20. How has undergoing Li-Fraumeni Syndrome screening affected your feelings about your risk for developing cancer?  
*Probe: Have they increased, decreased, stayed the same?*
21. How have your views on Li-Fraumeni Syndrome changed since undergoing this screening?  
*Probe: Do you feel better, worse, or the same about the disease?*
22. Tell me about your family's reactions or responses to your undergoing screening?  
*Probe: family members' interest in and/or experiences with screening?*
23. What advice do you have for families that are recently diagnosed with Li-Fraumeni Syndrome?  
*Probe: For screening? How to tell family members?*

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## VITA

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