THE EXPERIENCES OF ADOLESCENTS AND YOUNG ADULTS WITH LI-FRAUMENI SYNDROME SURROUNDING HEALTH CARE TRANSITION

by
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Abstract

Health care transition (HCT) is the process of adolescents and young adults (AYAs) taking an increased responsibility for health care management and decision-making and transferring from pediatric to adult providers. HCT is a complex and non-standardized process and can be further complicated when the adolescent has a chronic condition requiring ongoing medical management and bringing psychosocial demands. Li-Fraumeni Syndrome (LFS) is an autosomal dominant inherited cancer pre-disposition syndrome with a nearly 100% lifetime risk for cancer, high childhood cancer rates and a demanding recommended surveillance schedule that adds a level of chronicity to LFS even in the absence of a cancer diagnosis. Research on families with other chronic disorders has shown that patients and families often struggle with HCT, leading to decreased treatment and surveillance adherence, emergence of complications and general health deterioration as the patient ages. Although recent research shows that personalized care during transition including tailored collaboration and early planning can make HCT more effective, most research on effectiveness focuses on medical outcomes and knowledge. Little is known about the influence of psychosocial facilitators and barriers and less still is known about the lived experiences of AYAs with LFS.

For this work, semi-structured interviews with 30 AYAs with LFS were initially transcribed and coded utilizing thematic content analysis by one coder using an inductive, iterative approach. Ten transcripts chosen for maximum variability were analyzed by a second coder, and the two coders modified the codebook as necessary. One sheet of paper exercises deepened the analysis and uncovered common themes within and across codes and disparate participant experiences. AYAs discussed their healthcare management process and while many described the important role of family members, providers and other individuals during decision-making, some also discussed their independence in management. Participants described their experiences with changing healthcare management over time, and the importance of
personalized, tailored support from both providers and support people in their life. These findings suggest the importance for providers to comprehend the central role that family and others may play and the significance of providing personalized care as AYAs with LFS approach and navigate health care transition.

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**Introduction**

Health care transition – a process for adolescents and emerging adults

Individuals may undergo several transitions in healthcare providers, healthcare settings and overall management throughout their life. Within the literature, the term “health care transition” (HCT) is commonly used to specifically refer to the process of an adolescent or young adult beginning to take an increased responsibility for health care management and decision-making, roles traditionally performed by caregivers when a person is a younger child. The thesis adheres to that convention and will use HCT to refer specifically to this type of transition. In a position paper of the Society for Adolescent Medicine, Blum and colleagues described HCT as “a multifaceted, active process that attends to the medical, psychosocial, and educational/vocational needs of adolescents as they move from the child-focused to the adult-focused health care system” (Blum et al., 1993). It traditionally involves planning with the caregivers, the actual transfer of care from the pediatric medical team to the adult and integration of the patient into that new medical setting [gottransition.org]. For children diagnosed with chronic conditions, this transition process may be especially important in order to set them on a successful path towards lifelong management of their condition and its unique challenges.

The main goals of the process, as identified by the federally funded national resource on HCT, include improving the abilities of youth to manage their own health care needs and effectively utilize available services, facilitating communication and organization between pediatric and adult services, effectively transferring care, and smoothly integrating the patient into the new adult services [gottransition.org]. In a scoping review designed to conceptualize an HCT education program, Morsa and colleagues described similar aims. Their review included 20 papers, most utilizing surveys and questionnaires, to assess factors influencing HCT for young people with a variety of long-term health conditions including HIV, diabetes and
neurological disorders. The HCT aims they identified included increasing patient knowledge and skills, encouraging self-efficacy in the patient, building trust between patient and service providers and decreasing the emotional attachment to pediatric providers, if necessary (Morsa, Gagnayre, Deccache, & Lombrail, 2017). Central to understanding how to improve efficacy of HCT and reach these goals is understanding the needs of patients undergoing the process.

HCT is ideally begun early, opening conversations with adolescents before they must leave their pediatric providers. The process continues through adolescence and into the transitional life-phase of young people in their late teens and twenties termed “emerging adulthood” by Jeffrey Arnett (Arnett, 2000). According to Arnett’s framework, this phase is marked by several key elements of change and exploration, but perhaps the most complicating characteristic when considering working with this population is its heterogeneity. Arnett describes how emerging adults are less restricted by the required roles of either adolescence or adulthood, and therefore exhibit wide demographic diversity. They exhibit flux in housing, relationships, schooling, work and other activities as they develop their identities and cement their future paths. Importantly, they also display high levels of ambivalence about whether they consider themselves adults, typically giving a ‘yes and no’ style answer (Arnett, 2000). Drawing on multiple studies, Arnett found that it was not solid demographic transitions such as marriage, graduation or becoming a parent that ranked highest in these young people’s minds as steps towards adulthood. Instead, they cited personal characteristics, and more specifically “accepting responsibility for one’s self and making independent decisions” (Arnett, 2000). Healthcare management of a chronic condition may require repeated decision-making, and as emerging adults’ medical management and decision-making is not well understood, work to elucidate the unique characteristics of this process in this population is important (Liu, Calzone, Fasaye, & Quillin, 2021).
Evidence from studies of HCT and remaining gaps

Children with chronic conditions face a variety of challenging outcomes as a result of their experiences, and these needs underscore the importance of a well-organized HCT. Survivors of childhood cancer, regardless of the cause, have been shown to lack appropriate health-related knowledge and screening practices, often resulting in little cancer-related follow-up care, specific knowledge on risk reduction, or survivor-focused care (Freyer, 2010; Nathan et al., 2008; Oeffinger et al., 2004). In one study of adult survivors of childhood cancer and a randomly selected group of their siblings, the cancer survivors sustained medical complications such as decreased general health, activity limitations and persistent cancer-related pain. The cancer survivors also exhibited negative psychosocial outcomes including increased mental health concerns and anxiety (Hudson et al., 2003). Several studies have shown that young adults with chronic conditions such as organ transplants, diabetes and congenital heart disease are at risk for negative consequences including decreased treatment adherence, an interruption in follow up schedules, the emergence of complications and general health deterioration (Ferro, 2014; Pacaud, Yale, & Health, 2005; Watson, 2000; Yeung, Kay, Roosevelt, Brandon, & Yetman, 2008). The authors propose that one cause of these outcomes is a poorly managed HCT, which can be characterized by lack of trust between providers and patient or an absence of establishing the importance of routine follow-up care. These findings suggest not only the importance of provider experiences that build trust, but also the importance of maintaining connections between young adults and their care team to facilitate continued efficient health care management and decision-making.

Given the importance of characterizing effective HCT, several systematic reviews have investigated aspects of HCT. In a realist review of the literature, Kerr and colleagues collected 78 papers investigating HCT for adolescents and emerging adults with life-limiting and chronic conditions; cystic fibrosis and HIV were frequent illnesses studied, but malignant neoplasms and
cancer were also included (Kerr, Price, Nicholl, & O’Halloran, 2017). The authors’ objective was to explore how contextual factors affect the application of interventions aimed at supporting these young people through HCT. David Fryer investigated transition care specifically for young adult survivors of childhood and adolescent cancer in an effort to hypothesize relevant HCT approaches for this unique population (Freyer, 2010). Finally, Betz and colleagues performed a systematic review including 35 mostly qualitative and mixed methods studies specifically investigating the emerging perspectives of adolescents and young adults regarding HCT experiences (Betz, Lobo, Nehring, & Bui, 2013). Although these works build a foundation of knowledge characterizing and describing HCT, an evidence-based consensus regarding the best approach for an effective HCT is yet to be determined.

In their review, Kerr and colleagues identified several transition interventions that were intended to improve outcomes for patients across various studies. They fall into three main categories of beginning the process early; collaboration between patient, family, providers and agencies; and increasing the autonomy and confidence of the young adult (Kerr et al., 2017). These interventions, however, were implemented in varying combinations within different settings, which made analysis of individual effectiveness extremely difficult. A key finding of several studies was that post-transition medical outcomes are less desirable when HCT is approached rigidly, without contextual tailoring to the patient. These data suggest that person-centered HCT that tailors the process towards the individual needs of the patient, family and the structure of the health care systems involved is more effective (Freyer, 2010; Kerr et al., 2017). Several studies further identified the importance of involving a key worker who can build a therapeutic relationship with the patient which increases the young person’s confidence, can assist with agency collaboration and supports their continued engagement with services (Kerr et al., 2017). However, many settings reported in the review lacked the resources to employ such
person-centered initiatives and therefore lacked the capability to support their patients in this way.

In Betz et al.’s review specifically describing the perspectives of adolescents and emerging adults, several thematic foci emerged. Patients described not feeling sufficiently informed or prepared for changes they would encounter in adult care, such as differences in service models or insurance coverage, suggesting a lack of understanding future expectations. Also, they discussed feeling unprepared to assume role changes, such as increasing responsibility for their care (Betz et al., 2013). However, Betz and colleagues point out that most of the studies’ findings center on the actual transfer of care. They suggest it would be worthwhile to examine the young adults’ psychological and social needs related to the transition. These might include services or decision-making supports needed or how individuals acquire health-related self-management knowledge and skills. It could be similarly important to research the learning skills that individuals require to access community-based programs related to their condition or HCT itself. Morsa et al. similarly found that studies reporting on factors influencing HCT tended to focus on knowledge of illness, treatment plans and the health care system structure, which are again related to the transfer of care providers. The least well characterized factors are those relating to other relevant knowledge and skills for young people such as general knowledge of the transition process or skills needed to become successful health care managers (Morsa et al., 2017).

A final fundamental gap that emerged from these reviews is the scarcity of adolescent and young adult voices amongst HCT research (Betz et al., 2013; Forbes Shepherd et al., 2018; Morsa et al., 2017). The majority of reports currently focus on medical outcomes and transfer of care providers – treatment adherence, follow-up care, illness knowledge and observations, etc. – and not on opinions or lived experiences of the participants around the transition. Many studies utilized quantitative surveys and few followed participants long enough to fully assess
their experiences (Betz et al., 2013). Few studies explored adolescents’ or young adults’ perspectives on psychosocial factors or familial influences on HCT. An interesting point made by both Morsa et al. and Betz et al. points to the difficulty in documenting unheard adolescent voices, in contrast to young adult ones. The authors describe how adolescents have a less mature comprehension of medical system characteristics and less experience functioning as health care consumers, and may therefore be less able to judge and articulate their needs (Betz et al., 2013). Furthermore, many adolescents may find psychological or social milestones such as development of self-image and the search to understand their feelings “more useful than maintaining good health” (Morsa et al., 2017). For these reasons, obtaining emerging adult perspectives on transition may be more informative in that the experiences and opinions can be coalesced and deepened by the more mature introspections of this older population. It is important to qualitatively describe the full lived experiences and opinions of adolescents and emerging adults with chronic conditions in order to more fully understand their HCT needs.

**Li-Fraumeni Syndrome is a rare but life-altering condition**

Li-Fraumeni Syndrome (LFS) is an inherited cancer predisposition syndrome with a high risk of a variety of malignancies across the lifespan. Patients’ lifetime risk of cancer is greater than 70% for men and 90% for women (Schneider, Zelley, Nichols, & Garber, 2019). Patients are at increased risk of childhood cancers; one study showed that the average age of onset of the first LFS-associated malignancy was 17 years for men, 28 years for women when breast cancer was included and 13 years when breast cancer was excluded (Bougeard et al., 2015). Similarly, another study showed that 50% of LFS-associated cancers occurred by age 31 for women and 46 for men (Mai et al., 2016). Some high-risk cancers in childhood include adrenocortical carcinoma (develops in 6-13% of patients with LFS typically before age five
years), leukemias (median age of diagnosis at 12 years), rhabdomyosarcomas and other soft-tissue cancers (often diagnosed before age five years) (Schneider et al., 2019).

Due to these excessively high lifetime risks, a potentially burdensome surveillance program is typically recommended for patients, although it is not standardized and therefore involves some degree of collaboration between patient and provider to determine the specific regimen. In childhood, surveillance often includes bloodwork, physical examination and ultrasound every 3-4 months from birth (or diagnosis) to age 18 years and an annual neurological exam and whole-body MRI from the time of diagnosis (Schneider et al., 2019). As patients enter young adulthood around age 20, they are recommended to continue bi-annual physical exams and ultrasounds and add yearly dermatologic exams, breast exams every 6-12 months, breast MRIs and mammograms starting at age 20 and 30, respectively, as well as regular endoscopy and colonoscopy starting age 25 (Schneider et al., 2019). Women may also consider a risk-reducing bilateral mastectomy. At the same time, patients are advised to minimize exposure to carcinogens such as diagnostic and therapeutic radiation, UV radiation and tobacco. In a study of 89 children and adults with LFS who either underwent or declined regular screening via whole-body MRI, breast and brain imaging as well as blood tests and endoscopies, Villani and colleagues showed that the five-year survival rate for those who participated was 88.8% compared to 59.6% for those who declined (Villani et al., 2016). Additional studies have similarly shown benefits to patients who maintain a regular whole-body MRI screening regimen, suggesting adherence may confer significant survival benefits to patients (Ballinger et al., 2017; Kratz et al., 2017; Mai et al., 2017). Furthermore, in a mixed methods study of 17 patients with LFS, McBride and colleagues reported patients experiencing psychological benefits including decreased anxiety and feeling emotionally supported by regular whole-body MRI screening (McBride et al., 2017). Taken together, this further emphasizes the benefits of adherence to the recommended screening program.
LFS is rare, although the prevalence is not well established. Most patients diagnosed with LFS have pathogenic variants in TP53, a ubiquitous tumor suppressor gene, although about 10% of clinically diagnosed patients have an unknown genetic etiology (Schneider et al., 2019). One report estimates the frequency of TP53 germline pathogenic variants at 1/3,555 to 1/5,476 (de Andrade et al., 2019). The National Organization of Rare Disorders estimates over one thousand families with LFS exist around the world (https://rarediseases.org/rare-diseases/li-fraumeni-syndrome/). Estimates place the rate of de novo germline TP53 mutations at between 7 and 20%, meaning the majority of patients have an inherited variant. Despite its rarity, within an affected family LFS is pervasive; because it is inherited in an autosomal dominant pattern, LFS can affect many individuals across generations.

LFS is a powerful force in families that grapple with its excessive cancer burden. Patients experience high incidence of cancer in adolescence and young adulthood, continuing lifetime risk and a burdensome recommended screening regimen. Although it requires significant effort to manage the many responsibilities, patients who can maintain adherence to treatment, contact with providers and psychosocial support experience significant benefits. Successful transition from pediatric to adult health care is one important aspect of this ongoing process.

Adolescents’ and Young Adults’ Experiences with LFS

An important step in understanding the needs of adolescents and young adults with LFS during HCT would be to understand their general experiences of living with a disorder that confers such significant cancer risks. Unfortunately, there have been few studies related to AYAs’ experiences with LFS. Schultz and colleagues interviewed 46 parents of children at risk for LFS to learn their opinions on the influence of various characteristics of adolescence on
testing decisions and discussions of genetic risk with their children (Schultz et al., 2018). However, opinions from the adolescents themselves were not part of this study. Alderfer and colleagues performed a small qualitative study of twelve individuals aged 12-25 in families with LFS, exploring perspectives surrounding genetic testing as children (Alderfer et al., 2017). Findings included support for optional testing in children, given parental approval and child involvement in decision-making. Additionally, patients discussed perceived benefits and risks of testing, and all participants who had undergone testing (7/12) reported no negative impact on their life outlook, regardless of the outcome. While this study begins to explore the lived experiences of adolescents and emerging adults with LFS, its focus is centered only around genetic test decision-making and impact.

Similarly, several studies exploring family dynamics and experiences with LFS have included AYA voices as well as those of older family members. Werner-Lin and colleagues conducted family interviews with 117 family members ranging from 13-81 years old in order to describe generational experiences living with LFS (Werner-Lin et al., 2020). Their findings centered around families’ experiences of dread and anticipatory loss around cancer occurrence and recurrence, uncertainty regarding prognosis and quality of life as well as shifting family roles and dynamics. Pantaleao and colleagues’ qualitative interviews with LFS families included 62 individuals, typically across generations, ranging in age from 7-81 years (Pantaleao et al., 2020). Further, the younger generation in each family grouping was typically in the AYA range (participants ranged from 7 to 40 years). This research explored the role of “health leader”, a term used to describe members who “obtained and disseminated new health-related information, facilitated healthcare appointments and served as other members’ health of LFS expert/advocate” (Pantaleao et al., 2020). Most families discussed having one or several health leaders and discussed how this position was often occupied by the older generation. They described a gradually, often unspoken process of modulating roles within families as diagnoses,
health changes or other occurrences demanded. Parents often had ongoing involvement in their children’s care through their early 20s, and this was motivated by a desire to protect their children (Pantaleao et al., 2020). Although this family health leader study begins to describe this role within families, and the transition of the role from older to younger generations, no studies to date have directly explored experiences of adolescents and young adults with LFS around the pediatric to adult health care transition. Exploring lived experiences of this population would provide important preliminary insight into this process and provide direction for future studies.

In order to gain such preliminary insight into this population’s lived experiences, Forbes Shepherd and colleagues performed a systematic review of 39 studies representing 765 AYAs to characterize how they live with other disorders inherited in an autosomal dominant pattern, most commonly Hereditary Breast and Ovarian Cancer (Forbes Shepherd et al., 2018). Much of the research centered around genetic testing, decision-making surrounding testing and the immediate impact – both positive and negative – of receiving test results. Additional findings described young peoples’ lived experiences with inherited predispositions, including their experiences surrounding relationships and family planning, common concerns for emerging adults. Young people with inherited cancer risks were often unsure of how or when to tell partners, and women who wanted to have children felt a distressful urgency to do so before undergoing recommended breast/ovarian cancer risk reducing surgeries. Additionally, the authors reported varying awareness of alternative reproductive options like preimplantation genetic testing amongst these young patients. An important theme that Forbes Shepherd and colleagues coalesced from their study was the impact of family narratives and roles influencing the experiences of young adults with these disorders, suggesting the same may hold true for LFS.
Given the dearth of research on the lived experiences of young people with LFS, qualitative work exploring and describing those experiences is crucial. A deeper knowledge of this population’s needs, difficulties and outlook will be vital to improving their HCT experience.

**Could Genetic Counselors Improve the HCT Experience?**

The role of genetics providers, including genetic counselors, during health care transition has not been explored. However, the involvement of a genetic counselor in a patient’s care has been associated with improved outcomes in other care contexts outside of the HCT. The presence of genetic counselors in patient sessions may increase adherence, which has been used as a measure for organized and successful HCT. Rutherford and colleagues examined adherence to medical recommendations for pediatric patients after their initial visit to the general genetics clinic at Cincinnati Children’s Hospital Medical Center in 2008. Out of 198 patients 12 years and younger matched by age and sex, the authors found improved adherence when a genetic counselor was present with a medical geneticist at the visit, compared to a medical geneticist alone (Rutherford, Zhang, Atzinger, Ruschman, & Myers, 2014). The importance of addressing psychosocial needs around transition has also been emphasized in HCT reviews, and research from Forbes Shepherd and colleagues suggests genetics professionals may be more equipped to handle these needs in the clinic as well. The authors conducted a survey of 43 health care professionals including oncologists and genetic counselors exploring their experiences caring for adolescents and young adults with LFS (Forbes Shepherd, Keogh, Werner-Lin, Delatycki, & Forrest, 2019). One area explored in the survey was whether various AYA psychosocial needs including sharing information about family planning and peer support or providing support for anxiety and stress were addressed in the clinic or referred elsewhere. Genetic health professionals reported being much more likely to address these psychosocial needs in the clinic than oncology professionals.
In their systematic review of adolescent and young adult voices, Betz and colleagues found that adolescents and emerging adults desire providers who listen to them and are sympathetic, as well as knowledgeable on their condition (Betz et al., 2013). Genetic counselors are specifically trained to have depth of knowledge, empathy and person-centered counseling skills that places the patient’s experiences in the forefront. However, as late as 2013, no specific guidelines or models for working with youth in the genetics clinic existed (Duncan & Young, 2013). Werner-Lin and colleagues worked to address one aspect of this gap by providing recommendations to genetic counselors for how to support parents in discussing inherited cancer risk with their children (Werner-Lin, Merrill, & Brandt, 2018). Theories of child development, empirical evidence and clinical experience informed their recommendations that counseling be tailored to the child’s developmental, cognitive, emotional and behavioral abilities, advice that could reasonably be applied to AYAs. Genetic counselors in Australia also recently worked to address this lack of an AYA genetic counseling model, acknowledging that genetic counselors are well-equipped to address specific challenges of this population. Young and colleagues developed a framework for genetic counseling with youth by undertaking a literature review, collaborating with a multidisciplinary panel of experts in AYA oncology, genetic counseling, adolescent health, ethics and research (Young, Thompson, Lewin, & Holland, 2020). Their work suggests principles and practice points concerning youth-friendly engagement and communication, developmentally appropriate information, education and capacity as well as AYA-specific psychosocial assessments. While the framework does not address the issue of pediatric to adult transition, the communication principles could be applied to the HCT setting, suggesting that genetic counselors could play an important role.

Clearly, genetic counselors recognize the need to deliver appropriate genetics care to this age group and are working to develop appropriate frameworks and recommendations. There is a need to understand the roles that genetic counselors and other genetics
professionals play for young adults with LFS during their HCT and identify areas that these providers could further apply their specialized knowledge and skills to improve this process.

**Specific Aims**

AYA voices are underrepresented within both the HCT literature, as well as qualitative work exploring the lived experiences of individuals with LFS. In addition, as the bulk of HCT work focuses on medical outcomes and knowledge, exploring the influence of psychosocial facilitators and barriers on this process is important as well. Therefore, the specific aims of this work included:

**Aim 1.** To explore and describe how young adults with Li-Fraumeni Syndrome perceived their experiences surrounding the transition from pediatric to adult health care.

**Aim 2.** To explore how young adults with LFS describe the facilitators that helped and barriers that hindered their transition to the adult health care setting.

**Aim 3.** To explore how young adults with LFS recall the role played by genetics providers during their transition to adult health care.
Methods & Analysis

Background

For this thesis, I collaborated with an ongoing longitudinal Li-Fraumeni Syndrome study conducted by investigators within the Clinical Genetics Branch, Division of Cancer Epidemiology and Genetics, NCI (parent study; NCT01443468; www.lfs.cancer.gov). This study is one of the largest international cohorts of families with LFS and is the largest longitudinal cancer screening cohort. Additionally, the study offers genetic counseling, TP53 testing and study enrollment to at-risk family members (Werner-Lin et al., 2020). I worked with an Adolescent & Young Adult sub-study within this parent study (the AYA sub-study). The aims of the AYA sub-study include:

- understanding the ways that living with LFS impacts AYAs’ development, social/provider networks and overall wellbeing
- understanding primary informational, psychosocial, and instrumental (unmet) needs of AYAs with LFS and their families
- understanding how providers can optimally support AYAs with LFS and their families
- describing healthcare needs of AYAs with LFS, given geographic diversity and barriers/facilitators to care

Working with these investigators, I requested questions pertaining to my aims be added to the protocol, and then worked with the team to finalize these questions for implementation in phase II of semi-structured interviews planned with adolescents and young adults with LFS (phase I was completed before our collaboration). The AYA sub-study will continue beyond my collaboration with a quantitative survey and an eventual third round of interviews. The PI of the parent study is Dr. Payal Khincha, MD.
Sample

Participants were eligible for the parent study if they met one or more of the following criteria:

- A personal germline pathogenic or likely pathogenic $TP53$ variant
- A family or personal cancer history consistent with clinical criteria for LFS/Li-Fraumeni-like syndrome
- A personal history of 3 or more LFS-related primary cancers
- A personal history of adrenal cortical carcinoma or choroid plexus carcinoma at any age, regardless of family history

All participants enrolled in the parent study are part of the ‘field’ cohort. Field cohort participants receive any recommended screening they desire and are able to procure entirely from local providers and participate in the study by providing their medical information and biospecimens, completing study questionnaires and participating in interviews. A subset of the field cohort members were invited to participate in a longitudinal cancer screening study; these individuals formed the ‘clinic cohort’. Clinic cohort participants travel to the NIH annually for recommended cancer screening including whole body, brain and breast MRIs (for eligible female participants), physical exams, and psychosocial interviews. They obtain additional cancer screening such as recommended GI screening and dermatological exams, as well as any necessary interval screening from their local providers.

Participants for the AYA sub-study (and therefore the sample analyzed in this project) were recruited from the parent study if they met the following additional inclusion criteria:

- Age 15-39 at recruitment
- Field or clinic cohort
- Have a confirmed germline $TP53$ variant
Approximately 120 individuals were invited to participate in the AYA sub-study phase 2 interviews, and thirty individuals completed interviews.

**Interview Guide Development**

Through iterative rounds of development with the NIH AYA team of investigators, several questions investigating HCT were developed and embedded as section two of five, within a larger semi-structured interview guide (see Appendix 1 for the interview guide of section B "AYA Experiences with Healthcare and Transitions in Care"). These questions prompted participants to reflect on how they and their family manage their healthcare, how this has changed over time and their feelings on any changes or stability in management. They also prompted participants to reflect on who they turn to during decision-making around managing their healthcare, and how these people provide support. Lastly, the questions asked participants what advice they would want to give, either to providers or family and friends supporting a teen with LFS as they grow into an adult. Additional subjects explored within this interview guide and not included in the analyses for this project included the following:

- Section A: Daily life with LFS during the COVID-19 Pandemic
- Section C: Intimate relationships and communication
- Section D: Family formation
- Section E: Perspective on LFS and life

**Data Collection**

Interviews were conducted by phone by four interviewers (CW, CR, PB and AS) between March and June 2021. Interviews were an average of 78 minutes long and ranged from 42 to 129 minutes. After obtaining verbal consent, interviews were audio recorded and transcribed by
an external service; participant names were anonymized by substituting participant-selected pseudonyms. All personal identifying information was removed from transcripts and they were stored on a secure, password-protected NIH computer, as was the MaxQDA analysis file.

Analysis

Transcripts were uploaded into MaxQDA, qualitative analysis software that facilitated the coding and analysis process. During initial readthroughs the primary coder, MS, read the first two sections of the transcripts for context and updated background information on participants. Section B of interview transcripts were then coded and analyzed initially by MS using thematic analysis which allows for identification of common patterns and themes among the interviews. Interviews were analyzed in order to describe the ways that participants managed their healthcare, their experiences with providers and others involved in their healthcare, the perceived and predicted change over time of this process, as well as advice that participants would give to providers and other people supporting AYAs with LFS. Several a priori codes were initially created based on these topics from the interview guide, such as “management process”, “decision-making support”, and “change in management over time”. The codebook was expanded as codes emerged from the data, utilizing primarily an in vivo coding approach, such as “NIH and provider expertise”, “tailoring of care and support” and “satisfaction”. The preliminary codebook was modified and revised as new codes became apparent, and previously coded transcripts were re-coded to incorporate any new codes. Eventually, code saturation was reached and no additional concepts were identified through analysis of additional transcripts or subsequent re-readings (Hennink, Kaiser, & Marconi, 2017). MS then applied the codebook to the entirety of the thirty transcripts, without adding to it or modifying codes, to ensure themes from the section on healthcare transitions that may have been discussed elsewhere in the
transcript were captured. Codes not discussed in this work, many of which were applicable elsewhere in the transcript, will be addressed in a separate publication.

At this point, a second coder (MF) utilized the completed codebook to code section B only in a single transcript, then three additional transcripts and finally an additional six for a total of ten transcripts (one third of the total transcripts). At each break point, MS and MF discussed their coding strategy and consensus coding was reached, code definitions were modified, and several codes were merged (e.g. the individual code “differences between adult and pediatric health care setting” was merged into the “change in healthcare over time” code).

After coding was completed, ‘One Sheet of Paper’ (OSOP) exercises were performed on each code, grouping coded segments into themes, noting similarities and discrepancies or unique usages of the code (Ziebland & McPherson, 2006). This process allows nuances of the code to be retained and revealed as participants’ experiences are further clarified and grouped. The coded segments were reviewed for appropriate quotations representing the rich variation amongst the themes and topics noted in this work.

Interview questions uncovered diverse topics of conversation among AYAs, not all of which are discussed within this work. This paper will focus on AYAs experiences managing their healthcare, including interactions with genetics and other providers, the supports they turn to during decision-making, and how they have experienced or predict change in these areas over time.
**Results**

**Study Population Characteristics**

As was true for the participants in the parent study from which they were recruited, our 30 study participants were primarily a White and well-educated group, consisting of more females than males (n = 21 and n = 9, respectively) (Werner-Lin et al., 2020). At interview date, they ranged from 18 to 41 years old, with more than two-thirds of participants in their 20s and 30s (n = 25). They underwent genetic testing, and were given a genetic diagnosis of LFS, across a range of ages; the youngest by far was two years old, several additional participants were under age 18 and most were in their 20s. More than two thirds of participants had a previous cancer diagnosis (n = 22), and greater than two thirds had inherited LFS from a parent (n = 25), versus presenting as a *de novo* case (n = 2). Ten out of the thirty were field cohort members, engaged in remote monitoring from the NIH team, and twenty were clinic cohort members, traveling to the NIH for regular screening. For a full summary of study group characteristics, see Table 1.
Table 1 - Demographics and Summary of Characteristics of AYA Study Sample

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Number of participants</th>
</tr>
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<tbody>
<tr>
<td>Age at interview date</td>
<td>18-21: n = 3</td>
</tr>
<tr>
<td></td>
<td>22-29: n = 10</td>
</tr>
<tr>
<td></td>
<td>30-39: n = 15</td>
</tr>
<tr>
<td></td>
<td>&gt;39: n = 2</td>
</tr>
<tr>
<td>Age at time of genetic testing</td>
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<tr>
<td>(genetic diagnosis of LFS)</td>
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</tr>
<tr>
<td></td>
<td>19-29: n = 18</td>
</tr>
<tr>
<td></td>
<td>30+: n = 4</td>
</tr>
<tr>
<td></td>
<td>Unknown: n = 1</td>
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<tr>
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</tr>
<tr>
<td></td>
<td>No: n = 8</td>
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<tr>
<td></td>
<td>Clinic: n = 20</td>
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<tr>
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<tr>
<td></td>
<td>de novo: n = 2</td>
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<tr>
<td></td>
<td>Unknown: n = 3*</td>
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<tr>
<td>Children</td>
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</tr>
<tr>
<td></td>
<td>No: n = 21</td>
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<td>Sex (M/F)</td>
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<tr>
<td></td>
<td>Females: n = 21</td>
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<td>Reported Race (note numbers exceed 30 because some participants reported multiple races)</td>
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<tr>
<td></td>
<td>Asian: n = 2</td>
</tr>
<tr>
<td></td>
<td>Black or African American: n = 1</td>
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</table>

*inherited vs. de novo status could not be confirmed for these individuals due to a lack of parental testing and/or limited family history.

To protect the identity of participants, they chose or were assigned pseudonyms.

Pseudonyms of participants along with individual characteristics can be found in Table 2.
<table>
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<th>Pseudonym</th>
<th>Age</th>
<th>Sex</th>
<th>Age of genetic testing</th>
<th>Inherited vs. de novo LFS</th>
<th>Field vs. clinic cohort</th>
<th>Cancer diagnosis (y/n)</th>
<th>Children (y/n)</th>
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Summary of Findings

When participants spoke about managing their healthcare over time, they specifically described who they rely on for decision-making support, and many spoke about how and why these supports offered unique assistance. Many participants relied on various family members, both in the presence of generational experience with LFS and when this was not present in the family structure. Participants shared their experiences, both positive and negative, with providers; many spoke about the importance of the provider’s depth of knowledge of LFS, and how effort and seriousness on the part of the provider was crucial to building trust. Participants also spoke about the importance of tailoring care so that AYAs are treated as individuals with a consideration of unique life situations when providing care, both in the context of their desire to be heard and their changing needs over time. When we asked participants to reflect on changes in their health care management over time, many noted consistency and lack of foreseeable changes in the future. Some mentioned barriers to changes, either in the past or predicted for the future, and a few discussed how changes in their family structure affect their healthcare management.

Sources and Mechanisms of Decision-Making Support

When we asked participants to discuss who they relied on during decision-making regarding managing their healthcare, they talked about different types of decisions. Some specifically mentioned decisions about following screening regimens to manage cancer risk, some spoke about managing cancer diagnoses, some spoke about other aspects of healthcare including family planning or advance care planning and some spoke generally about their decision-making process. Participants often identified supports based on what their own needs were, including information and expertise, emotional connection, motivation, or a sounding board space for discussion. Many participants relied on close family members including their
parents, spouses, partners, or siblings. Others relied on additional family members including grandparents, aunts, and uncles. Participants talked about various providers on whom they relied, including primary care providers and specialists like oncologists, cardiologists, palliative care specialists, counselors, and genetic counselors, in addition to the NIH team of providers. Several participants also discussed their self-reliance and independence, indicating they didn’t rely on others for decision-making support.

**Self-reliant Decision Making**

The few AYAs who expressed their self-reliance mentioned a possible decision-making support person to whom they could turn, but clearly stated they felt the process was more independent than joint. Mary, 28, mentioned that her partner might have “a bit more of a say” regarding decision-making about her care because she’s a doctor, however Mary still explained that her decision-making is “mostly me; yeah, it’s basically me”. She implies some dependence on her partner by saying “mostly” me but then repeats and seems to instead confirm that she feels she is decision-making alone. George, 36, similarly described dealing with decision-making or times of uncertainty “pretty much” himself. He recalled leaning on his ex “a little bit” in the past, but generally described himself as “pretty much on my own”. Rick, 41, also mentioned the option to consult with certain family members for information, but clearly stated that during times of decision making regarding his LFS care he relied on “myself”. In contrast to the ways that decision-making supports are described below, these AYAs mentioned a person with whom they might consult, yet they described their decision-making process primarily as independent.

**Information and Expertise on LFS**

Many participants sought out decision-making supports for their ability to provide information based on prior experience or expertise with LFS. Many AYAs spoke specifically about relying on various family members for information, and some spoke about the role providers play in this area as well. When participants discussed which family members they
relied upon for decision-making support, they often valued prior experience with LFS in some capacity. One participant noted “[my family members have] lived through it, whether it’s from the patient aspect or the caregiver aspect” (Chelsea, 20). These experiences allowed her family members to understand and sympathize with what she may be experiencing and strengthened their ability to provide information and decision support. Chelsea continued to discuss her thankfulness that her mother and grandmother are “so vocal because they know how to handle this best”. Sophie, 35, similarly noted that since her sister has LFS, she is “very aware” and “concerned about the issues”, making Sophie feel comfortable taking her sister’s advice. However, she went on to mention that not all family members’ experiences align with hers. Her cousins who have LFS approach it with a different mindset; Sophie remarked:

*The cousins that have Li-Fraumeni syndrome, they – I feel like they don’t see it the same as my sister and I see it. So, they may be less proactive, and more willing – or used to be more willing to just listen to a doctor, even though the doctor may not know what’s best.*

Sophie is outlining characteristics that are important to her during decision making that she shares with sister, but not her other affected relatives, including being proactive and knowledgeable. Clearly, not all experiences with LFS lead to the same management approach, and some AYAs who rely on family members seek out those members who align with their understanding of LFS.

Additionally, some family members may utilize additional resources, like LFS support or advocacy groups, which an AYA themselves may be less interested in engaging with personally. Rick, 41, described such a situation:

*I might talk with my mother or Aunt, because both of them have [LFS] and I think they stay a little more in the loop…reading all the updates and new releases. And they’re in*
the Facebook groups and stuff, you know. So, if I had any questions, I’d probably ask one of them.

Here, Rick demonstrates his understanding that support and advocacy groups can provide useful information and insight but feels less inclined to engage himself since he has family members to whom he can turn that are active in these groups.

Some family members could supply information not only from their experience with LFS, but also from their medical expertise as part of the healthcare field. Bonnie, 39, explained why she could turn to her mother before she died for information support saying, “I used to talk to my mom quite a bit, her being a nurse and I’m a nurse…she likes to read a lot of studies, too, so she was just very helpful”.

When speaking about medical providers specifically, participants discussed a range of decision-making support often related to the level of trust in that provider and the provider’s perceived knowledge base with regard to LFS. Carrie, 33, described feeling fortunate that she could “trust what my doctors have told me, and I figure they’ve kept me alive this long, so I should probably listen to them.” Lauren, 39, called her oncologist her “go-to” because of her eagerness to listen to any health concern Lauren has. She also described her primary care doctor as her “quarterback”, helping to organize and manage the many aspects of her healthcare.

AYAs talked about receiving guidance and clarity from leading LFS and cancer centers across the nation, including the NIH providers. Chelsea, 20, described how, although she relies on her family, they in turn are guided by the LFS team at NIH. Sophie, 35, described how helpful it has been that the NIH team has “made the path pretty clear of like what you need to do, and the care you need to get as far they know, and then they let you know”.
In contrast to using provider insight for eventual family or personal decision-making, one participant described family decision-making discussions as a precursor to a provider’s final choice. Cindy, 30, explained:

*When I first was diagnosed with breast cancer, [my parents] kind of discussed the surgery plan with my doctor in China. Of course, the final decision is from the provider, the doctor, but my parents were involved in that discussion.*

Cindy is suggesting here that although her parents and herself were involved in discussions with the doctor, the doctor’s knowledge and expertise contribute to the importance of allowing him or her the final decision in surgery planning.

**Emotional or Life-stage Connection**

AYAs relied on decision making support people based on aspects of their relationship with these people including emotional connection and similarity in life stage. Consistency was also mentioned by some AYAs as an important aspect of this relationship. Veronica, 25, spoke about the ongoing support from her aunt who has “just always been there for me”. Several participants discussed the empathy and concern they receive during decision-making, and Sophie, 35, discussed the “deeper level of empathy and concern” that she feels with her sister, compared to other relatives with whom she’s not as dependent during decision-making. Bonnie, 39, discussed how her husband specifically plays a “calming supportive role” during the decision-making process.

Ben, 18, discussed how he considers himself more of an emotional than strictly logical person, and described how generally having space to discuss his emotions is helpful during decision-making. He distinguished the different roles his parents play based on their specific strengths during that process:
[I turn to] my dad, as well, but he’s really logical and as in near-minded, and there’s nothing wrong with that, but emotions aren’t always his strong suit. Yeah. It helps me to talk to, like, my mom or grandma just because I am a bit more emotional, so to look at it through that perspective really appeals to me and helps to make decisions.

In addition to emotional connection or consistency, AYAs sometimes talked directly or indirectly about how life stage affected whether they turned to family members for various types of decision-making support. Silver, 33, described the task of writing advanced care documents and how involved her siblings were, none of whom have LFS themselves. She talked about her siblings’ support and willingness to “take on specific assignments” in a way that differed from her parents’ knowledge of her “general intentions”. Working together through this difficult decision-making process, Silver described how “they really helped me think creatively about like how to combine these versions in a way that I actually felt comfortable with…actually made it something I felt good about”. Perhaps she and her siblings’ more similar life stages or different emotional connection made this project for Silver easier to complete than with her parents.

It is not only siblings that can share appropriate life stage experience with AYAs. Ben, 18, discussed beginning to have discussions about family planning decisions with family members, and distinguished the utility in speaking to his parents about that, compared to his younger brothers.

I’ve talked about [family planning] with my parents a little bit, not my brothers. My brothers, they’re in a very different stage of life. I’m not, like, trying to say I’m more mature and everything, but they’ve got a lot of growing up to do.

Ben hesitates to describe himself as more mature than his younger brothers but recognizes that they may have less pertinent thoughts and advice on family planning decisions due to their life stage than he might receive from discussions on this topic with his parents.
A “Sounding Board”

A few participants discussed how their decision-making supports helped them talk through their available options allowing for clarity of direction, and typically they referred to family members as providing this type of support. AYAs described these supports as “a sounding board” who provide comfortable space to deliberate decisions. Ashley, 32, described the decision-making support she receives from her oncologist, husband and dad:

They’ve been fantastic. It’s a good just talking through, sounding board. I’ve never ever felt like pressured on making a decision immediately. It's just, here's your options, this is what I would recommend, and talk about it and tell me what you want to do. So I feel really comfortable with that.

Ashley is suggesting not specifically the availability of expertise or emotional support here, but instead values the space to talk over options and time to reach a decision with which she feels comfortable. She appreciates how these people don't pressure her into making a decision before she's ready.

Jenny, 23, similarly talked about turning to family members for space to consider options and took this support one step further by inviting those who provide such space to attend appointments with her. She explained that she doesn’t take notes in appointments, but that often her mother will attend appointments “so that she has all the information and that two people are listening, and then you can talk about what the doctor has told you”. This second set of ears not only helps to collect information but is important because her mother then provides time and space to talk about all the options, presumably so Jenny can feel comfortable with the final decision.

Along these lines, Bonnie, 39, described how her husband is “supportive and he looks at the big picture”. Although she did not elaborate, perhaps she’s indicating here that her own
thoughts tend to hone in on specific choices or options, and her husband’s ability to provide space for considering the entire picture give her clarity. These data suggest that AYAs with LFS value nonjudgmental assistance when considering the myriad options and decisions they face as they manage their condition.

**Motivational Support**

Although only one participant discussed receiving motivational support specifically, it was crucial to him so worthy of inclusion here. Allan, 28, spoke about how his mental health counselor provides essential motivational support both before and after decision-making regarding his healthcare. Knowing that he tends to procrastinate and avoid dealing with “the bigger picture things”, Allan described his reliance on his counselor:

> I don’t think [my counselor is] very knowledgeable on LFS, but just as far as helping me kind of find the motivation and the reasoning for why I should make certain decisions. And the importance of me following through with certain actions and things like that I think has been… You know, it’s kind of like NIH people will tell me I should do something, and then she’ll be the one that makes sure I get it done.

Allan clearly states that his counselor is not providing LFS expertise, and also does not indicate relying on their assistance talking through options. Instead, he speaks about how his counselor pushes him not only to *make* a particular decision, which appears to sometimes seem less pressing to Allan, but also then to *follow through* on his decision. He’s receiving information and advice from the NIH team, but his counselor provides the push for Allan to follow through and obtain the care he needs.
Provider Experiences

Participants discussed their experiences with a variety of providers, both positive and negative. Many had positive experiences at the NIH, or with local providers, but participants also expressed disappointment or frustration with various provider experiences. Typically, they discussed providers in general, but sometimes certain specialties were mentioned, including oncologists and genetics providers. Common themes emerged as participants discussed their experiences with providers including: the significance of LFS expertise, the importance that providers “take LFS seriously”, the effect that the provider-patient relationship can have on patient experiences and AYAs’ perceptions that general provider knowledge about LFS is increasing over time.

Providers’ LFS Expertise

Several participants recalled experiences specifically with genetics providers, and there was wide variability in the positivity of these experiences. Often a genetic counselor is the provider who delivers the results of genetic testing and the official diagnosis of LFS. Several AYAs recalled disappointing experiences with genetic counselors at their initial LFS diagnosis. Silver, 33, explained that her genetic counselor:

had never heard of Li-Fraumeni but was diagnosing me with it. It was not a good situation. She did not know anything about the disorder. And in fact, told me several things about it that turned out to be false. So, that was bad.

One of the false pieces of information she recalled receiving was the etiology of the name itself, “Li-Fraumeni” syndrome, and although Silver didn’t expand on other pieces of false information, she clearly was disappointed by her counselor’s knowledge base on LFS. Sophie, 35 recollected her disappointing diagnosis experience and recalled feeling that, instead of being the patient, she and her family “counselling [the “genetic person”] on what Li-Fraumeni was”.

30
Sophie’s experience again suggests the frustration that AYAs with LFS feel when their genetics provider doesn’t provide a solid evidence and knowledge base during the crucial time of diagnosis.

Fortunately, Sophie described later finding a new genetic counselor at a different institution that she appreciated and thought “was amazing”. Other AYAs indicated their generally positive experience with genetics providers when asked what advice they would want to give these providers as they worked to support teens with LFS. Mr. T, 36, felt they could continue to “keep up the good work”. Carrie, 33, appreciated the LFS expertise amongst her dedicated genetic oncology team. She felt they were aware of the current recommended screening guidelines and always kept her up to date, both of which instilled confidence in her.

When discussing the NIH provider team, several participants described the feeling that “nobody understands [LFS] like NIH does” (Chelsea, 20). They described the “peace of mind” they find being followed by the “best doctors pretty much in the country, if not the world” (Dylan, 29). They commented on the depth of knowledge they find with NIH providers, a depth they don’t always find elsewhere. Additionally, they commented on the availability of resources and the reliability of the NIH team. Bart, 35, explained:

\[
\text{I can pick up the phone anytime I want and call someone from the NIH...somebody answers that phone, and they provide guidance as to what they think I should do, and they listen to me. I think that's valuable.}
\]

The value to Bart lies not only in the expertise and guidance he receives from the NIH team, but also the dependability. He is appreciative that he can contact his NIH team whenever the need arises, and confident that a capable provider will listen to him and assist with his needs. Other participants described how the guidance they received from the NIH team made their path
forward clear or removed the need for additional research or information seeking on their or their family’s part.

Participants spoke about the importance of provider expertise in the context of adding or removing providers over time. Some of this discussion was around specialist expertise, such as the need to add gastroenterologists when colonoscopies were recommended. The importance of LFS expertise was also discussed though, as AYAs recalled partnering or disengaging with providers based on their skill level. Veronica, 25, explained:

*it’s just really grasping what their knowledge is, and their understanding, and their care for like how they’re treating me and how they see my syndrome. If they’re pursuing for myself or for something else?*

Veronica describes that she’s evaluating providers based on their expertise and understanding of LFS, as well as their motivation. If they make it clear they are using their knowledge to place the patient first and care for her, Veronica feels more comfortable partnering with them. Recollections like these suggest that these AYAs know they deserve to be treated carefully as a unique individual and are dedicated to finding providers who are knowledgeable about LFS.

**Providers’ Vigilance**

A specific way that several participants talked about their provider’s lack of LFS expertise was in the perception that providers either weren’t being vigilant or weren’t taking LFS seriously. Dylan, 29, mentioned how her ob-gyn was the first provider to take it seriously since she’s been telling providers about her condition. “She did her research on it after the first time I met her”, in comparison to every other doctor Dylan had visited to whom she had to explain her condition. This lack of effort and inability to take LFS seriously were lamented by other AYAs as well. Jackie, 37, discussed the implications should her child test positive for LFS and how the need for vigilance will factor into her decisions regarding providers.
That will be a conversation we have. You know, what are you willing to do? And how, you know – I don’t want someone who who’s going to be like, oh, well, it’s fine, it’s nothing. Like, we’ll just monitor it for a while. I want someone who’s going to advocate for my child and be like, hey, listen, you know? This—we need to be running tests that we maybe wouldn’t normally run for an average child.

Jackie is expressing her concern that a provider who doesn’t take LFS seriously might utilize a “watch and wait” mentality with minor health concerns in her child. Jackie knows from experience that vigilance required to manage LFS and emphasizes the importance of finding a provider who understands this and will follow up with needed tests to ensure her child’s well-being. Similarly, Sophie, 35, described how following-up affects the quality of care with her own screening. She discussed the importance of finding providers who would investigate suspicious findings more so than might be done in the general population. She feels radiologists tend to lean towards delivering false negatives, since most of the population is not going to have cancer after suspicious imaging, but she needs to find providers who know that with LFS “you’ve got to be searching for the problems”.

AYAs could distinguish within their provider a comprehension that LFS must be taken seriously separate from an expertise in LFS itself. Ashley, 32, discussed struggling to deal with her LFS diagnosis after the survival mode of managing her cancer diagnosis had dissipated. She discussed how her oncologist was most helpful at that time “because he was the one that found the study at NIH for me to just say, this is a way you can deal with it.” She continued:

*Him finding that study really helped alleviate so much pressure from me because I was like, these guys are going to tell me everything I need to know. I don’t have to research it.*
Ashley seems to be expressing her gratitude that her provider grasped the seriousness of her diagnosis and, knowing he could not provide the level of care she required, worked to find an appropriate care team for Ashley. She could then release the assumed demand to research and develop an LFS expertise herself, as she could depend on her new care team for this knowledge base.

**Personal Connection**

A few participants discussed their experiences with providers in terms of a relationship and the connection they felt through that relationship. Participants discussed working to maintain connections with providers they trust, and one discussed yearly flights back to her home country for screening because of the positive connections she has built with those providers over the years. Several AYAs called their NIH team of providers a “second family” (Chelsea, 20 and Allan, 28). After calling out the importance of genetics providers “being honest and caring and listening to [AYAs’] concerns”, Sophie, 35, summed up the difference she’s experienced by saying:

> There’s a different level of understanding. And a different level of concern. Even though I still love the genetic counselor at [local institution], I think there’s just a little bit more understanding at the NIH.

Nora Radner, 36, mentioned how she collects information from her genetic counselor, but specifically and warmly described the relationship she has with her also:

> I have a really good relationship with my genetic counselor. She’s been wonderful and so helpful and accommodating whenever I have a concern and helping me just understand.

Highlighting the importance of a relationship with providers beyond information gathering, Karen, 29, talked about differences she’s felt in connection between pediatric and
adult providers. She recalled that her pediatricians spent ample time in appointments with her and took time to understand her as a person. She contrasted this to the “world of difference” she felt with adult providers who seem like they are in and out of her appointment in “a minute and a half sometimes”. Karen doesn’t emphasize that these shorter appointments leave her lacking information but seems to imply the lack of connection and time is what she misses the most. These data suggest a personal relationship and positive rapport weigh heavily during decision-making for some AYAs as they choose providers with whom to work.

**Increasing Provider Knowledge Over Time**

Optimistically, AYAs see hope for more positive experiences in their future as several discussed improvements over their time of interacting with providers. One participant mentioned preventative cancer care in general improving over time, as providers were more aware of exposures to avoid and negative impacts of screening using radiation. Another participant mentioned the “hundreds of doctors and scientists” that are working to better understand cancer and its causes (Carrie, 33). Participants felt that general knowledge of members of the medical profession regarding LFS was improving as well. Silver, 33, said:

*But I do feel like I’m gradually noticing this transition where just like my providers are kind of better prepared and I’m like oh thank God, they did not just walk in like not knowing anything about me because like the doctors who walk in without having like read anything or even knowing what conditions I have are just like, those appointments aren’t useful.*

Clearly, Silver has experienced ineffectual appointments in the past due to providers’ lack of expertise, but fortunately she has experienced this less as time goes on, perhaps indicating general awareness of LFS increasing amongst her providers. Another participant recalled an appointment where a medical student was present who had recently learned about LFS and could immediately describe its characteristics. She compared this to her mother’s recollections
of medical school when LFS had been a single line in a textbook. Dylan, 29, felt this experience was a “good sign for maybe future doctors” suggesting a similar perception that general provider expertise on LFS was increasing over time.

Tailoring Care and Support

The concept of tailored care, which aims to improve patient outcomes by taking their “individual needs and preferences into account” during treatment, is not a novel idea. It is, however, not universally utilized nor standardized in its implementation (Dekkers & Hertroijs, 2018). When asked what advice they would give to providers or family members supporting a teen with LFS, participants often described the importance of tailoring care. Generic advice wasn’t as valued or appreciated by participants as individualized and personalized support. They often pointed out that everyone is unique and noted how crucial it was for providers and other support people to embrace that concept. Travis, 28, noted:

*I think depending on the teenager – and I work with teenagers all the time every day – is that everyone deals with things differently. So I mean, just based on the kid, their response to [an LFS diagnosis] can be completely different.*

Ashley, 32, echoed these sentiments when she stated:

*Everybody’s different. How I process a long-term diagnosis is very different than how somebody else will, right? So understanding that as well.*

These AYAs recognize that their own experience and adaptation to their diagnosis will inevitably differ from anyone else’s. They’re emphasizing the importance of considering each person as an individual and that tailoring care to that individual’s needs and preferences is crucial for optimal support. Two common themes emerged as AYAs spoke about tailoring of care: the need to be heard and AYAs’ changing needs over time.
**Need to be Heard**

Nora Radner, 36, explained the need to be heard as she described the process of deciding which kinds of support to lend someone with LFS. She spoke about considering that individual’s unique needs, rather than what the support-giver would prefer to do. She explained:

*You also just have to take into account, you know, what is going to be the most helpful to the person than what is going to make you feel good. So, I think trying to be as supportive and helpful, but take your cues always from the person that is living it and going through it.*

AYAs with LFS desire to be heard and have their cues noticed and addressed by those that are supporting them. Silver, 33, discussed the wide range of issues faced by AYAs with LFS and that support should be lent where the AYA requires it. She explained that AYAs with LFS must integrate their condition into many aspects of their lives including discussions with romantic partners, friends, and work colleagues and how to decide who is trustworthy when sharing information. Additionally, Silver mentioned implications with relationships between siblings and other family members. She gave the example that considering only fertility issues and family planning, for example, restricts the conversation. She summed up this concept saying, “the issues facing young adults are not – it’s not just whether we have kids and how we have kids. It’s like life is a lot more complicated than that”. Neither of these AYAs discussed tailoring care in the context of individualizing cancer treatment or surveillance, but rather emphasize the general importance of considering an individual with LFS and their unique life situation and listening to how they describe their experience and needs.

Importantly, one AYA described her frustrations at not being heard by her oncologist. Wanda, 32, explained that the oncologist continued discussing hysterectomy when she had “made it clear [she] wasn’t ready for that”. Wanda’s situation may be somewhat unique for AYAs with LFS, as hysterectomy is not typically a recommended surgery for reducing cancer risk in
these individuals, but it relates to the importance of being heard by providers. Wanda spoke about how she clearly understood the risks and benefits of postponing surgery and described finally discontinuing with this provider because the surgery “wasn’t something that I wanted to keep talking about, and they weren’t getting that.”

**Changing Needs Over Time**

Participants also discussed tailoring care as it related to changing needs over time. All information and support related to LFS is not relevant at any given time, but instead should be shared when appropriate for the AYA by taking cues from that individual. Chelsea, 20, noted how as she has aged discussions have covered more preventative measures, including surgeries, as well as lifestyle modifications, family planning and more future considerations. Similarly, Wanda, 32, provided the example of not pushing hysterectomy discussion on an 18-year-old since its “probably not going to be on their radar”.

Nora Radner, 36, described generally how living with LFS is “an evolving thing” and was grateful for her genetic counselor checking in periodically as her priorities changed and the different implications of living with LFS became more or less salient over time. She explained first feeling that her cancer diagnosis was “the most challenging thing”, and then years later realizing that her condition and the “implications of living with LFS” would actually be more challenging. She appreciated her genetic counselor being “available and accessible” as her priorities and challenges changed over the years. Similarly, Veronica, 25, spoke about how her need for information gathering changed over time as she was able to learn independently about her condition. She described a shift as she was able to “really get into it on my own and do the research I wanted to understand it”.

Part of AYAs’ changing needs over time relate to their evolving self-motivation and acceptance. AYAs with LFS acknowledged times in their lives when information was provided that they weren’t ready to accept. Mari, 29, described her journey to acceptance: “I’ve always
been very, very hardheaded. But not until I came to my senses, I was able to accept the help that I needed”. Allan, 28, recalling times that resources or supports had been available to him, described how he struggled to utilize them, and advised:

*Having resources available is definitely I think something that I appreciate and can benefit people. But there is also that level of desiring that resource.*

He implies with this statement that as an AYA’s needs change over time, their interest in a resource may change over time as well and they may become more ready to accept and utilize that resource. He recalled in detail a time in his own life that his lack of acceptance reduced his engagement in counseling, and therefore its effectiveness was greatly diminished. He was assigned a counselor in middle school when his mother was ill with a terminal cancer diagnosis, and for some time after her death. He described being pulled from his favorite classes (art, gym, etc. that were deemed more appropriate to be absent from) and feeling called out as a “special case” instead of spending time with his friends. He explained these factors caused him to “put absolutely the worst effort into it” and acknowledged that he “wasn’t getting anything out of it because it wasn’t something I felt I wanted”. Even though Allan’s caregivers and providers connected him with counseling, he was either unable or unwilling to explain why the situation was not ideal for him, and therefore why he was not benefiting from it. Importantly, Allan compared this situation to his satisfaction with his current counselor, with whom he was able to engage services when and as he preferred. He explained “the difference was that it was something that I immediately sought out once I was kind of having these depressive thoughts”.

These acknowledgements indicate the need for providers or other support people to tailor their care and recognize that as some AYAs with LFS work towards acceptance, their changing needs might enable them to better utilize diverse resources or support.
Change or Consistency in Healthcare Management Over Time

Given the variability of life situation amongst the AYA population and the many life changes occurring during that period, we asked participants to reflect on how their care had evolved as well as how they might expect it to evolve in the future. Many spoke in general terms about how they didn’t feel their management process had changed, nor did they predict changes for the future. Several participants discussed the impending end of the ability to obtain screening as part of the NIH study, necessitating future changes in management, especially concerning insurance coverage. A few participants described how aspects of their family situation changing affected how they managed their healthcare.

Consistency in Healthcare Management for Some

Participants who discussed consistency in their healthcare management over time discussed this both in relation to their medical appointments and their steady management plan and supports. Those that mentioned consistency often didn’t elaborate but stated that they didn’t notice any changes from the past, and several didn’t predict changes to management in the future either. Rob, 20, mentioned that he was “pleased” with how he and has family have consistently managed his healthcare and couldn’t think of any changes that might happen in the future. Veronica, 25, discussed the “steady, steady support” of her aunt and Lauren, 39, agreed that consistently over time, her oncologist, partner or her sister have primarily been her support.

The few AYAs who managed their healthcare independently also predicted little change in the future of their management. George, 36, mentioned how conversations with his family members haven’t changed over time because he’s “the only one dealing with [my LFS]. I’m kind of alone on that front”. He goes on to mention however, that he does “feel slightly lonely in terms of those interactions sometimes” suggesting that his current level of independence may not be entirely comfortable for him. It’s unclear what prevents George from reaching out for support as
he manages his healthcare over time despite these lonely feelings, yet he doesn’t predict that changing in his future.

**Barriers Related to Change in Healthcare Management Over Time**

AYAs discussed several barriers to changes in healthcare management over time. Multiple AYAs discussed the experience of recommended screening guidelines changing over time and one participant particularly noted how it can be challenging to keep pace with the modifications. Another noted the frustrating process of transferring extensive medical records as a significant barrier to changing providers or modifying care locations.

An oft-mentioned barrier AYAs discussed was the ongoing challenge of insurance coverage as they managed their healthcare over time. They noted that while providers in general seemed to become more aware and knowledgeable about LFS over time, understanding from insurance companies seemed to lag and lack of coverage continued to present hurdles. Jackie, 37, spoke about her hope that “once research [from the longitudinal NIH study] is published, it’ll make it easier to get insurance companies to cover the cost of screenings”. Carrie, 33, mentioned the necessity of advocating for herself to get required pre-authorizations as part of ensuring proper coverage for her care.

Several members of the clinic cohort who receive their regular screening as part of the NIH study expressed concern for their future ability to obtain care when NIH’s screening activities cease. Carrie, 33, was thankful for her and her husband’s comprehensive coverage, but noted that “if something ever happens that I didn’t have insurance, we wouldn’t be able to get the care that I need”. Another predicted that he likely would obtain MRIs less frequently than recommended due to a high deductible on his insurance plan (*Rick, 41*). Allan, 28, also a clinic cohort member, discussed his difficulties utilizing his insurance to obtain local screening when the COVID-19 pandemic prevented him from traveling to the NIH. Several times he returned to the fact that his fear of “ending up with financial burden” had hindered and demotivated him...
during his quest to receive local care. He expressed how “overwhelming” it is to navigate the health insurance industry and his appreciation of providers or staff that have assisted him in this process. Despite these few positive experiences, Allan continued to express concern:

That’s something that always worries me, is there some day when I’m going to end up with something in my mailbox that says I owe an ungodly amount of money for something I wasn’t aware of. And it’s kind of where my ignorance of my insurance coverage kind of haunts me at times.

Not only does confusion about coverage prevent Allan from moving forward with health care procedures, but he also worries that, even when he moves forward, his coverage will be lacking and he’ll be saddled with an unpayable bill. This concern is not only in the back of his mind, but actually “haunts” him. AYAs with LFS require comprehensive insurance coverage for their myriad health care needs, and frequently return to the frustration or fear involved in obtaining this coverage over time.

*Changing Family Situations*

AYAs discussed how changes in their family situation or life stage affected how they manage their healthcare. Carrie, 33, described how having children changed how she and her husband experienced the significant time spent traveling that was required for her screening.

And before, it was just a lot easier to pick and go…so sometimes [my husband and I] would just leave in the middle of the night and make it there just on time for the appointment. So, we would do it in one day. Well, it was fun. We just kind of listened to music, talked, podcast, and it was kind of our time together…Whereas now…we have four kids at home… And so, obviously, that has changed how we have to do things. We can’t just drive in the middle of the night. That’s a lot harder on the kids than it is on us. That’s not fun for them. So, trying to just plan.
Whereas once Carrie and her husband could turn this necessary travel time into enjoyable time spent together, now it’s not a family-friendly activity. Further, Carrie’s last statement seems to imply the extra planning involved with childcare, in addition to losing the time spent with her husband. All these experiences contributed to a changed sense of managing healthcare as Carrie’s family situation evolved over time.

Another AYA discussed her changing family situation related to attending medical appointments independently. Veronica, 25, was diagnosed with LFS as a young teenager after her mother’s passing from an LFS-related cancer. She described how her father attended appointments with her for several years, but eventually stopped when she was around 16 years old.

*I think he really wanted to be there for me at first because my mom had just passed away. And I was still a child, and he was like trying to grasp it himself and understand it himself. Because I mean the love of his life, he had been with my mom for I think it was 16 to 20 years. So, he had just lost her. And finding out that his oldest daughter had this syndrome that could give her cancer as well, I think it was a big process for him to grasp and understand it... And he could potentially lose his daughter to cancer as well...So, I think like him just grasping on and holding on, going to appointments was maybe more was for him than for me. Because the process he was going through.*

Veronica mentions elsewhere that part of her father’s attendance cessation was for practical reasons, like limiting work absences, but her insight here delves deeper into her father’s motivations. She is musing about whether his attendance was part of his emotional processing of both his wife’s passing and his daughter’s diagnosis and perhaps less for the benefit of supporting his teen daughter. As Veronica aged, she felt she “was coming into her own” and was comfortable attending appointments alone and updating her father afterwards.
Taken together, these experiences support the important notion that transformation in AYAs’ family and life stages may trigger change in how they are managing and receiving health care.
Discussion

An important contribution of this work is increasing the visibility of AYA voices within studies of health care transition. Several reviews of the topic identified this as a fundamental gap in need of focus (Betz et al., 2013; Forbes Shepherd et al., 2018; Morsa et al., 2017). The thirty participants covered nearly the entire AYA age range and had a spectrum of differing life experiences with LFS – including differences in inheritance pattern, severity of disease course, cancer diagnoses, management process and life stage. Although each brought a unique voice and contribution, several themes emerged from the data as participants discussed their experiences managing their healthcare and advice for supporting young people with LFS. These common themes highlight several important topics and add to the crucial presence of AYAs’ voices in the literature.

One major theme arising from this work is the importance of family members’ support and involvement as AYAs with LFS manage their healthcare over time. Not all participants talked about family support, but those who did often described the nature of support provided in similar ways. Many AYAs with LFS valued information and expertise, often from a family member’s personal experience with LFS, or experience supporting someone with the condition. Some spoke about family providing a sounding board for the AYA to consider their options and make choices with which they were comfortable. Others received support and clarity based on their emotional connection or similar life stage. Importantly, not all AYAs with LFS spoke about crucial decision-making support, from family members or other individuals, and instead talked about their self-reliance and independence managing their healthcare over time. Still, the importance of family support for many AYAs with LFS, yet the variability within how this support enabled decision-making, were both important factors to emerge from the data.

A second common theme to arise was the stability of healthcare management within the group. Little change over time was discussed or predicted for the future by many participants.
As touched on above, this is likely due in large part to the unique nature of our cohorts’ involvement with the NIH study. Some participants specifically recalled seamless transition between NIH providers based on need, and others called out the reliable support and presence of the team. Our AYAs’ perceptions of stability may also be in part due the fact that many were diagnosed in young adulthood, rather than childhood, when they may have already transitioned to managing many aspects of their healthcare. These possibilities will be explored further in the next section of this work. When participants did discuss predicted changes in their future healthcare, such as the end of the screening activities within the NIH study, they tended to speak about the logistics of receiving continued care and monitoring. Some spoke about barriers to managing their health care over time, including insurance coverage hurdles and the significant cost of receiving care that isn’t covered. Other participants reflected on how their changing life situations affected how they receive care.

Several findings in this work indicate that AYAs with LFS may benefit from encouragement of the same goals for successful HCT that Morsa and colleagues discussed in their scoping review of AYAs with different conditions (Morsa et al., 2017). One aim for AYAs undergoing HCT that these researchers identified was increasing patient knowledge and skills. Our AYA participants discussed several sources they consulted to gather information as well as more generally an increase in knowledge about LFS across their healthcare providers over time. They spoke about positive experiences with providers who were knowledgeable about LFS and took their concerns seriously. They also relied on family members with personal or familial LFS experience or support groups as additional information sources, with an intention to improve their understanding and ability to confidently make decisions and manage their healthcare going forward. These varied information sources are crucial to support AYAs’ developing knowledge base around their condition which in turn may enable a smoother and more efficient HCT process. Crucially, AYAs in our study did not often discuss difficulties they faced in increasing
their LFS knowledge, but this would be important to explore in order to understand and eventually ease barriers they may face during this time.

A second HCT aim for AYAs that Morsa and colleagues identified was encouraging self-efficacy amongst AYAs (Morsa et al., 2017). Along with discussing their increasing knowledge and ability to seek out required information and resources, participants discussed learning to advocate for themselves over time, especially in regard to provider selection. They mentioned understanding what was required to efficiently manage LFS and learning how to seek out providers who had a similar understanding. AYAs mentioned striving not to partner with providers who wouldn’t be vigilant, for example by ordering follow-up testing in young children. AYAs mentioned seeking out support to specifically increase their self-efficacy as well, whether that was utilizing counselors to ensure they initiated and followed through on decision-making or bringing support people as a second set of ears to appointments. These highlight some methods AYAs in our study are using to increase their self-efficacy and may represent steps towards taking responsibility. Arnett’s framework on emerging adults describes the emphasis that they place on taking responsibility as a benchmark on the road towards adulthood (Arnett, 2000). If empowerment and increasing self-efficacy enable AYAs to accept responsibility, perhaps targeted interventions towards these areas could support AYAs that aim to increase their responsibility over healthcare management.

As participants discussed their experiences with providers, the importance of providers making an effort to build their expertise when seeing a patient with a rare condition such as LFS became clear. Participants spoke about feeling pleasant surprise and increased trust when providers were knowledgeable about LFS or showed an effort to learn and improve. Some spoke about noticing a general increase in knowledge amongst their providers as time has passed. Separate from their provider’s knowledge of LFS, some AYAs spoke about the importance of their provider getting to know them as a unique individual, one behavior that
enables tailored care from providers. AYAs’ discussions on the topic implied several important practice implications, including the significance of tailored care, which will be discussed in a later section of this work.

**Limitations**

The findings from this work are not representative of all adolescents and young adults with LFS. There were few adolescents among the thirty participants that completed interviews (n=3, <21 years old) and therefore adolescent views may not be well represented within this work. Participants were well-educated and had the important advantage of connection to the NIH via participating in the longitudinal parent study. Furthermore, two thirds of the study population were members of the clinic cohort, receiving their annual screening at the NIH, often with multiple family members attending together. Due to this participation, our population had access to a high level of support, including expert providers and genetic counselors, and those within the clinic cohort had their recommended screenings performed free from worries of insurance coverage. This level of connection and availability of resources likely provides a much different experience than other individuals with LFS have and therefore is likely to affect how participants discussed health care transition, management and decision-making. Some participants mentioned the seamless transition between NIH providers, again suggesting an experience with HCT amongst our population that may differ from the general population. In addition, more than two thirds of the study population (n=21) were diagnosed with LFS at age 21 or above; these participants likely had a different experience with HCT than those who had a diagnosis in childhood and presumably began extensive screening at that point. Lastly, very few of our participants had *de novo* cases of LFS (n=2) and it is likely that this study was not able to fully describe the experiences of individuals without a strong family history of the condition. Although *de novo* LFS cases are thought to represent less than 20% of all cases, and therefore
may be difficult to engage in research, it could be important to attempt purposive sampling of this population as well (Schneider et al., 2019).

Even with these limitations, this work presents an initial foray into understanding the experiences of AYAs with LFS regarding this transitional period of care, which is an important step.

We expect these findings to be transferable in some ways to other populations with inherited predisposition to cancers in the AYA period including hereditary diffuse gastric cancer, Neurofibromatosis, Gorlin syndrome and APC-associated polyposis conditions. These conditions could be diagnosed in young children and would require surveillance beginning in the childhood years, similar to LFS. In addition, like LFS they may not require daily interventions, as opposed to other childhood chronic disorders like diabetes or cystic fibrosis but would nonetheless require important health care decision-making and management skills. They are also inherited in an autosomal dominant pattern, establishing the possibility of generational family experiences that were often discussed by participants in this study. The rarity of LFS may make it difficult to purposively sample individuals with distinct characteristics, as discussed above, so future studies may benefit from including populations that face similar healthcare burdens in order to capture the desired demographics. We would not expect these findings to be transferable to individuals with other health care needs that were not included in our population, including those with intellectual disability or physical disabilities like blindness.

**Future Directions**

Future studies to build upon this work should attempt to purposively include individuals with characteristics discussed above that were not captured in this study. It will be important to describe the experiences of individuals with diverse racial and ethnic backgrounds as well as individuals with a wider range of educational experiences. Future studies should focus on
including more individuals diagnosed with LFS as a child/adolescent, more individuals representing *de novo* cases and more individuals in the adolescent age range. Although not explicitly investigated within this work, it is likely that individuals with a personal cancer diagnosis in childhood experience and manage their healthcare in different ways than those without one. It could be important to specifically explore these differences in future studies. Furthermore, engaging AYAs without access to expert leadership and organization through the NIH will be important to describe the range of HCT experiences given the likely varied availability and quality of local care centers.

As expected, and in large part likely due to their association with the NIH and age of LFS diagnosis, our participants did not often discuss traditional HCT experiences. When it was mentioned, smoothness and effectiveness from the NIH team seemed to create an easy experience for participants who aged out of providers or certain health care settings. However, some participants did discuss more traditional HCT experiences, including unfamiliar adult care settings, changing long-standing providers, changes in family members accompanying to appointments and insurance coverage changes. Delving more deeply into these experiences with targeted questions, as well as capturing a study population more likely to have experienced this, would be fruitful aims for future studies.

Regardless of the population studied, several areas of inquiry related to the themes in this work would be important to pursue. AYAs were eager to give advice for supporting their peers with LFS, and recalled experiences that were helpful, or harmful, in their own pasts. Further qualitative work to explore this more specifically could be fruitful and lend insight towards understanding how AYAs process their diagnosis, how they integrate it within the broader context of their often-complicated lives, and how their diagnosis impacts their development and wellbeing. AYAs discussed their decision-making supports, and described leaning on providers, experienced family members or support groups for information and a
sounding board. It would be important to understand the full range of information seeking this population performs, including additional sources and methods they may employ. Additionally, it would be important to understand how AYAs integrate this information, including whether and how they distinguish between sources.

Previous works on HCT have identified successful interventions in other populations, including engaging the family and starting the process of HCT early (Freyer, 2010; Kerr et al., 2017). Since many of our study participants discussed the importance of family members in the ongoing management of their healthcare, this intervention seems particularly important for individuals similar to our study population. Even participants with *de novo* cases of LFS discussed relying on family members including parents and siblings during decision-making and managing their healthcare. Future studies could explore how best to intentionally use family-based interventions with similar populations in order to facilitate HCT. Similarly, although previous studies have shown the importance of tailoring care towards the individual needs of each patient and their unique situation (Freyer, 2010; Kerr et al., 2017), and our participants often discussed this concept as well, the application of this concept is not standardized among HCT. Future studies could explore which aspects of tailored care appeal to AYAs, the areas in which it is most desired and how it is helpful during HCT and beyond. In this way, again, tailoring care could then be applied during HCT in the way that most facilitates the process.
**Practice Implications**

A crucial theme arising from this study with implications for providers is the emphasis participants placed on tailoring care. Several previous reviews investigating HCT interventions found that a rigid approach to HCT, without contextual tailoring to the patient, leads to less desirable post-transition medical outcomes (Freyer, 2010; Kerr et al., 2017). The data from these reviews support a person-centered approach that treats each individual as a unique person with unique needs, management process and health care structure. The findings from this study reinforce these conclusions. Participants were asked to provide advice for family, friends or providers supporting AYAs with LFS and many advocated for tailored care given that everyone will process their diagnosis differently and require different supports. In describing their management process and decision-making support needs, there was not a “one-size-fits-all” answer. Health care providers need to be aware of each patient’s unique situation in order to deliver person-centered care that is applicable to that individual’s life. An awareness of what their management process looks like, who they are relying on for decision-making, experiences that have shaped their past management, and their predictions for future modulations all should be incorporated into true tailored care. Furthermore, participants echoed the importance of a key worker (or “quarterback”) who can help coordinate care and ease the many complicated burdens this population faces.

Related to tailored care is the importance for providers to appreciate the mindset of an AYA as they’re working to integrate LFS into their life. Several AYAs talked about the need to reach a level of acceptance or desire before they could access available resources. While this is an important concept that deserves further work, these data suggest it would be prudent for providers to assess their patient’s level of acceptance and how it might be hindering or facilitating adaptation. Working to support and encourage acceptance could be more crucial
than specific resources provided, as it will not only enable those resources to be used, but also possibly motivate the AYA to seek their own resources, as some of our participants discussed.

In their scoping review of HCT goals, Morsa and colleagues described the importance of building trust between patient and providers both before and throughout the HCT process (Morsa et al., 2017). Participants spoke about trust when describing providers with whom they had positive relationships, including through successful cancer care from oncologists or obtaining screening from trusted providers. They discussed the clear and reliable guidance from LFS experts, including those at the NIH, allowing them to trust the path that was laid for them. Participants also discussed negative experiences with providers who either didn’t have a depth of knowledge about LFS, didn’t exert effort to learn about their patient’s unique situation or gave them false information about the condition. These experiences erode trust, impel the AYA to continue the often-difficult process of seeking out more trustworthy providers and may lead to diminished follow-up care. Providers need to be aware of the trust they can engender when approaching individuals with knowledge, effort and flexibility, and the importance this trust can have in maintaining positive experiences as AYAs manage their healthcare over time, including through HCT.

Although our participants did not often discuss genetic counselors specifically, these practice implications certainly apply to these providers as well. The few disappointing experiences mentioned by our AYAs with genetic counselors present a sobering reminder that each meeting contributes towards building and maintaining trust between patient and provider. Other participants mentioned the solid relationships they have developed with trusted genetic counselors both at the NIH and locally. Many of our participants discussed the importance of knowledgeable providers who listen to their unique needs and as genetic counselors are trained to provide medically expert and person-centered tailored care, they may be poised to fill this desired role. As genetic counselors work to support AYAs experiencing HCT, it will be important
to continue exploring AYAs’ experiences, opinions and needs during the process and incorporating these into the broader context of both their healthcare management and lives.
Appendix 1: Interview Guide for Section 2 of 5

**SECTION B: AYA EXPERIENCES WITH HEALTHCARE AND TRANSITIONS IN CARE:**
People experience a multitude of life changes as they move through adolescence into young adulthood. We’d like to hear about how your LFS care has changed over time from childhood until now.

**Management of LFS care**

1. Tell me about how you and your family manage your LFS right now.
2. How has that changed over time since your childhood?

<table>
<thead>
<tr>
<th>If it has changed:</th>
<th>If it has <strong>not</strong> changed:</th>
</tr>
</thead>
<tbody>
<tr>
<td>a) Can you tell me about changes that felt natural or easy? Why do you think they felt that way to you?</td>
<td>a) How do you feel about the way you and your family manage your care? <em>(e.g. pleased, comfortable, discontent, wishing for change?)</em></td>
</tr>
<tr>
<td>b) How about changes that worried you or felt challenging? Why do you think they felt that way?</td>
<td>b) Are there aspects you anticipate changing in the future?</td>
</tr>
<tr>
<td>c) If you feel there are more changes that may happen in the future, can you tell me about them?</td>
<td>c) When might that happen?</td>
</tr>
<tr>
<td>d) Which of the changes either excite or worry you?</td>
<td>d) Which of the changes either excite or worry you?</td>
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**People involved in LFS care**

3. Can you tell me about people, activities or supports that were helpful to you as aspects of your LFS management changed? *(or are helpful to you now if aspects haven’t changed)*
4. Who do you rely on during times of decision-making in your LFS care and how do they help you? *(e.g. providers, genetic counselors, specific family members, support groups, spiritual leaders)*
5. How has that changed over time?
6. In what ways has relying on these people been helpful or not helpful to you over time?

**Ideal world of LFS care changing over time**

7. What advice would you want to give to providers and family members about how to best support a teen with LFS as they grow into adulthood?
   a. What advice would you want to give genetics providers about supporting teens with LFS as they grow into adulthood?
Bibliography


Duncan, R. E., & Young, M.-A. (2013). Tricky teens: are they really tricky or do genetic health professionals simply require more training in adolescent health? *Personalized Medicine, 10*(6), 589-600.


