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Evaluating the Social Informational Needs of Emerging Adults with Genetic Conditions

Courtney Whitmore

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EVALUATING THE SOCIAL INFORMATIONAL NEEDS OF EMERGING ADULTS
WITH GENETIC CONDITIONS

by

Courtney Whitmore

Bachelor of Science
North Carolina State University, 2017

Submitted in Partial Fulfillment of the Requirements

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School of Medicine

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Accepted by:

Jessica Fairey, Director of Thesis

Gretchen MacCarrick, Reader

Jodie Neukirch Elliott, Reader

Cheryl L. Addy, Vice Provost and Dean of the Graduate School

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DEDICATION

This project is dedicated to all of the individuals affected by a genetic condition whom are working towards a fulfilled and healthy life. Each of you have so much strength and hope, you are the future. In addition, I would like to dedicate this work to all of the healthcare providers who have afforded me world class care in my personal medical journey.

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To my family, I could not have done it without you whatsoever. You are the team a woman needs behind her to succeed. To my fiancé, Josh, you are the greatest confidante and have made my life infinitely better in every way. Thank you to all of you for believing in me and helping me pursue my dreams.

ABSTRACT

Purpose: This study evaluated the healthcare provider's role during the social development of emerging adults with a genetic condition. The study also identified the type of information and ways that these adolescents hope to receive information on the potential side effects or complications from engaging in risk taking behaviors and lifestyle choices.

Methods: Participants included both males and females aged 18-26 with achondroplasia, sickle cell disease, cystic fibrosis, or Marfan syndrome. Respondents were recruited to complete an anonymous online questionnaire via social media support groups or email notification.

Results: There were 103 total respondents that completed the questionnaire and met the study's inclusion criteria. Fifty percent or more of participants have not talked to their providers about smoking, tobacco, drug use, sexual activity, contraceptives and birth control, moving out, or independent living. Using thematic analysis of open responses, it was reported those participants having discussions were not satisfied with the age in which the topics began to be discussed or the manner of the communication. Respondents indicated this information needs to be reviewed sooner and should include the topics of mental health and specific activity and exercise restrictions.

Conclusion: These results suggest that this patient population is experiencing gaps in care when it comes to anticipatory guidance in regard to making informed decisions about risk taking behaviors and their genetic condition. The patients who reported they did receive some of this information suggested that the timing or manner in which it was discussed

with a provider was not preferred. Overall, there are improvements to be made to aid in the emerging adult population which could empower their ability to make educated decisions about their genetic condition and lifestyle, thus improving their quality of life.

TABLE OF CONTENTS

DEDICATION	iii
ACKNOWLEDGEMENTS	iv
ABSTRACT	v
LIST OF TABLES	viii
LIST OF FIGURES	ix
CHAPTER I: BACKGROUND	1
CHAPTER II: EVALUATING THE SOCIAL INFORMATIONAL NEEDS OF EMERGING ADULTS WITH GENETIC CONDITIONS	14
2.1 ABSTRACT	15
2.2 INTRODUCTION	16
2.3 MATERIALS AND METHODS	20
2.4 RESULTS	24
2.5 DISCUSSION	37
REFERENCES	42
APPENDIX A: QUESTIONNAIRE	44

LIST OF TABLES

Table 1.1 Summary of Conditions..... 1

Table 2.1 Summary of Conditions..... 16

Table 2.2 Demographics of Study’s Participants 22

Table 2.3 Mean Age Information was Provided 36

LIST OF FIGURES

Figure 2.1 Participants' Provider Types	26
Figure 2.2 Amount of Social Information	28
Figure 2.3 Transition Assessment of Participants	35

CHAPTER 1
BACKGROUND

As medicine advances, there is an increasing number of children with genetic conditions who are living longer and reaching adulthood (Romelecyk et al., 2015). This growing population is creating unique, new topics for providers to address with their patients. Genetic conditions typically onset at birth, are heritable with an increased risk to offspring, and require particular management by specialty care teams. As research continues there are frequently new genetic conditions being identified with large spectrums of symptoms, inheritance patterns, and varying prognoses and presentations.

Table 1.1 Summary of Conditions

Condition	Inheritance	Prevalence	Predominant ethnicity affected	Carrier Risk
Cystic fibrosis (CF)	Autosomal Recessive	30,000 in the US	Caucasian Ashkenazi Jewish	Caucasian: 1 in 25 African American: 1 in 61 Asian: 1 in 94 Hispanic: 1 in 58 Ashkenazi Jewish: 1 in 24
Sickle cell disease (SCD)	Autosomal Recessive	100,000 in the US	African American	African American: 1 in 10 Hispanic: 1 in 30 to 200
Marfan syndrome	Autosomal Dominant *De novo (25%)	1 in 5,000 worldwide	Panethnic	N/A
Achondroplasia	Autosomal Dominant *De novo (80%)	1 in 20,000 worldwide	Panethnic	N/A

*De novo = The (%) of cases that arise from a sporadic new mutation

Table 1.1 lists four genetic diseases that can present in many different populations, each with unique sets of symptoms, which make them applicable and appropriate for further research on the social informational needs of affected individuals. Often individuals with any of these four conditions can have life expectancies well into adulthood and typically their cognitive abilities are not affected. The symptoms of these conditions each affect different body systems, but they cover a large range of possible outcomes and symptoms for an emerging adult. This group of conditions can affect different ethnic backgrounds with varying incidences, modes of inheritance, and disease phenotypes. Collectively these conditions display a wide spectrum of phenotypes and impacts which would allow this research to have applications to other disorders beyond the scope of this study. These genetic conditions present differently from one another, but the challenges of adolescents with these diagnoses can overlap in their need for multidisciplinary care, potential medication therapy, lifestyle restrictions and/or limitations, and anticipatory guidance on activities and their health.

Individuals with cystic fibrosis (CF) are often affected by chronic excess mucus in their lungs, pancreatic insufficiency that can impact their digestive tract, and potential infertility. Due to these complications affected individuals should never smoke or be exposed to smoking of any kind as their lungs are not able to process or filter like an unaffected individual. Individuals with pancreatic involvement may require special diets and monitoring for potential development of diabetes. The majority of males with CF have congenital bilateral absence of the vas deferens (CBAVD) and thus experience infertility due to azoospermia; however, a female's reproductive anatomy is not typically impacted by the condition. Females who are able to become pregnant would be classified as high risk due to potential pulmonary exacerbations and medication dosage adjustments

required in pregnancy (Yankaskas et al., 2004). When family planning an affected individual is recommended to have their partner undergo carrier screening to assess reproductive risk in offspring. If their partner is not a carrier of CF, then their children would be carriers of the condition. If their partner is a carrier of CF, then there is a 50% chance of the pregnancy being affected by CF and a 50% chance the pregnancy is a carrier. If both individuals are affected by CF then all of their children will be affected (Yankaskas et al., 2004).

Those affected by Marfan syndrome, a connective tissue disorder, have cardiac, musculoskeletal, and ocular manifestations. The cardiac symptoms primarily put the aorta at risk for aneurysm and dissection. In addition, Marfan may cause dislocations of the eye lens, joint problems, and scoliosis. Based on these symptoms there are recommendations to avoid contact sports and certain physical activities. Specifically, activities that involve “burst” exertion, like sprinting, and intense isometric exertion, like weight lifting can be dangerous (Tinkle et al., 2013). When family planning, if an individual’s partner is not affected by the condition, there is a 50% chance of each of their pregnancies being affected by Marfan syndrome. Pregnancy can lead to significant maternal medical complications, such as rapid aortic dilation or risk for catastrophic aortic dissection. Due to the increased risks for an affected mother and her pregnancy, careful consideration should be taken to assess the individual’s risks prior to pregnancy (Tinkle et al., 2013). Often times to maintain low blood pressure and minimize stress on the arteries, angiotensin receptor blockers may be prescribed; however, this class of medications may be contraindicated when pregnant. Additionally, alcohol may intensify the effects of these medications. It is important for people to not only understand the symptoms of their condition, but also the potential side effects of medications and treatments.

Sickle cell disease (SCD) results from abnormal hemoglobin which causes blood cells to turn into a sickle shape, impeding their ability for the cells to move easily through the blood vessels. Thus, the cells cannot carry adequate amounts of oxygen to the body and the cells may build up causing slowing or blockages in those blood vessels. Lack of oxygen, reduced blood transport, and potential clotting may result in organ damage, painful crises, and an increased risk for stroke. Crisis episodes are more likely to occur with illness especially when one becomes febrile, making an emergency management plan necessary to ensure these individuals receive care in an urgent situation. One recommendation in the American Academy of Pediatrics' guidelines is to avoid reptiles as this can decrease the risk of salmonellosis, a potential hazardous infection for these individuals. Immunizations are also crucial to reduce the instances of severe illness (Sox et al., 2003). Additionally, people with sickle cell disease are at increased risk for stroke, therefore they must seek urgent evaluation for symptoms such as sudden headaches. In order to avoid exacerbations that could lead to crisis it is recommended that physical activities and sports be evaluated on a case by case basis. However, avoidance of temperature extremes and maintenance of hydration are necessary for all and often times can limit the athletic activities one can participate in. Some sports and activities may be more compatible than others, such as golf instead of basketball. High altitudes and environments with low oxygen should also be avoided. Smoking, tobacco, and cocaine can also lead to exacerbations and crises due to the reduction of oxygen and extra burden on the body (Sox et al., 2003). Ideally prior to pregnancy, an affected individual's partner should undergo carrier screening to assess whether or not there is a risk for the pregnancy to be affected by SCD. If their partner is not a carrier then their children would be carriers of the condition. If their partner is a carrier of SCD then there is a 50% chance of the

pregnancy being affected by SCD and 50% chance the pregnancy is a carrier. If both individuals are affected by SCD then all of their children will be affected.

Achondroplasia is a skeletal dysplasia that causes short-limbed dwarfism which can lead to obesity, sleep apnea, spinal stenosis, kyphosis or lordosis of the spine, and recurrent ear infections. These individuals may need to adapt their home and workplace to establish independence. Due to risks associated with spinal stenosis, activities such as gymnastics and collision sports should be avoided, and instead activities such as biking and swimming should be encouraged. Also due to the lumbosacral spinal stenosis, spinal anesthesia is not recommended (Trotter et al., 2005). When considering birth control options, oral contraceptives should not be used long term, due to the increased risk of uterine fibroids that may be further exacerbated by these medications. During pregnancy women with achondroplasia are at risk for respiratory compromise. At delivery women [may] require a cesarean section due to their small pelvic outlet (Trotter et al., 2005). In addition, recurrence risk for this condition is unique because if two individuals with achondroplasia have a child there is a 50% chance the child will be affected, a 25% chance for a lethal presentation of severe skeletal dysplasia, and a 25% chance the child will be unaffected. If only one parent is affected by achondroplasia, then there is a 50% chance each of their children will be affected by the condition and a 50% chance the child will be unaffected.

Individuals affected by any of these conditions are often able to attend college, engage in personal and romantic relationships, and participate in activities like other typical adolescents; however, they must consider their medical condition in the context of these activities. Specifically, behaviors that can impact their health such as alcohol, tobacco, and drug use, sexual activity, family planning, recurrence risks, career options,

and independent living. These topics are important, as affected individuals must understand how these actions could negatively impact their health and wellbeing prior to engaging in these behaviors.

Emerging adulthood is commonly the age that individuals must broach these activities for the first time when a large shift occurs from codependence to independence. The emerging adult is defined by various age groupings, but most often covers the period from late teens to late twenties (Bundick, 2011). It has been identified that one of the most important indicators for people to signal a successful transition to adulthood is when one accepts responsibility for their own actions and is able to define one's own beliefs and values. Previous studies have also identified additional indicators of the attainment of adult status. These indicators are the establishment of a relationship with parents as an equal adult, being financially independent, and no longer living in the parents' home (Arnett, 2001). Individuals affected by a chronic, genetic disease may also use their medical management as a signal for transition to adulthood. When one manages their own medical care and makes educated decisions during everyday life, with their condition in mind, that individual feel a greater sense of independence and empowerment.

Emerging adults typically go from relying on others to assist them in their decision-making processes to making these decisions on their own, independent of their parents. This same period is usually when these individuals may be confronted by exposures to risk taking behaviors. In a multicenter study it was discovered that individuals aged 12 to 19 with sickle cell disease or cystic fibrosis may wait until later, compared to their unaffected peers, to engage in risk taking behaviors. The study also found that 21% of teens with cystic fibrosis and 30% of those with sickle cell disease had

previously smoked. In addition, 28% and 51% of teens, respectively, engaged in sexual intercourse. (Britto et al., 1998). Due to the sensitive nature of these topics it is suggested that participation may be underreported, and these statistics are an underestimate. That leaves a large population of individuals who are engaging in risky behaviors prior to receiving potentially impactful information. This time in life can be complex and challenging for any person, but those affected by a chronic condition may find navigating these life milestones even more problematic, especially when they are not receiving the necessary information and communication to make such decisions. Young individuals with chronic illness and disability are not less likely than peers to participate in risk taking behaviors such as substance use and sexual activity. Although they are engaging in similar behaviors, often times their approach and reason for participating in risk taking behaviors is due to teens processing a potential lethal condition that could impact their lifespan and/or life outcomes. (Suris et al., 2004). Often times, these adolescents could be described as understanding and perceiving life in a unique way compared to their unaffected peers. This way of thinking may be attributed to the earlier age in which they must worry about their health and the impact it may have on their life experiences.

Decisions regarding risk behaviors may be more complicated for individuals with a chronic medical condition. The experience with these behaviors was described by a recent study, “Young adults with special needs are particularly vulnerable, especially those with a genetic disease accompanied by some level of disability.” (Dogba et al. 2014). The vulnerability of these adolescents may result from the added need to consider the risks and implications that their decisions could have on their health. In order to be empowered to make informed decisions, these emerging adults need specific, individualized medical advice about the implications of social activities. For example,

more than a third of children and half of the adults with a genetic condition reported that they had never or rarely obtained the social and emotional support they needed from a health care provider or support group (Romelczyk et al., 2015). There are major gaps in social information support inside and outside of the clinic setting. Often times the answers to the social and emotional concerns of these patients cannot be found on the internet or within a routine care setting. Rather, these concerns must be addressed through communication with educated healthcare professionals.

Lack of information is one barrier to patients receiving anticipatory guidance in regard to social developmental topics such as alcohol and substance use, sexual activity, and independent living. Each individual face unique challenges, but chronic genetic conditions tend to have overlapping social informational needs. A 2007 study reviewed the informational needs of emerging adults with phenylketonuria (PKU) or congenital adrenal hyperplasia (CAH). The study found that these individuals most desired information about how their condition affects them, the science behind their condition, and how to best manage their case (Szybowska et al., 2007). Although the study analyzed a specific population of individuals, it is predicted that these findings can carry over to many other genetic conditions. The study's main conclusion was that adolescents would like information on the genetic nature of their condition at a much younger age, with 65% of participants reporting that patients should begin receiving this information before the age of 12, and 43% reporting they believe it should begin between age 6-10. In addition, the study suggests care for these individuals could be improved by meeting their needs through addressing knowledge gaps by providing genetic counseling at an earlier age (Szybowska et al., 2007).

The lack of available information and counseling for this population may result from the sensitive nature of these topics causing uncomfortable encounters for both patient and provider. A previous study identified providers' tendencies to approach clinic appointments focused entirely on the condition by providing medically focused information, rather than focusing on the person with the condition; thus, frequently omitting social behavioral topics. Providers feel it is necessary to approach the patient with medically focused information and management of the condition. In contrast, the teen's needs include information on coping with the ramifications and impacts of chronic condition management on everyday life (Beresford & Sloper, 2003). Individuals do not typically feel comfortable to broach these topics without the provider initiating the conversation (Beresford & Sloper, 2003). Therefore, it is essential the provider integrates this information into appointments at an age prior to encountering these potential decisions. The patient may then feel empowered to make an independent and informed decision in regard to alcohol, tobacco and drug use, sexual activity, family planning, and independent living. Another possible cause for discomfort among patient and provider is the legal age for engaging in such activities and they often associate them as being 'forbidden' in nature (Beresford & Sloper, 2003). Although these topics may be undisclosed it does not mean that individuals are not engaging in these behaviors. Studies have reported the issues that adolescents feel important to discuss with a healthcare provider are not always addressed. The same study suggests this barrier is present due to: "lack of comfort level in sharing personal information, gender differences in the quality of communication between provider and patient, a parent present in the session, or simply a difference in needs and wants of adolescents versus those of the provider." (Szybowska et al., 2007).

One of the largest barriers for this group is communication, as expressing thoughts and feelings can prove to be difficult for any adolescent. This fundamental barrier is also one of the biggest influences of health outcomes. In a recent study, the ability to communicate with health care providers has been found to be one of the leading factors in the quality of life of adolescents with a chronic condition (Ma et al., 2018). Communicating openly and often with providers is an essential skill that adolescents with genetic conditions must develop in order to best manage their care. Another study looked at the communication practices of adolescents with juvenile chronic arthritis, cystic fibrosis, diabetes, epilepsy, and Duchenne muscular dystrophy (Beresford & Sloper, 2003). The study identified the common theme that adolescents are reluctant to share personal or sensitive topics and questions due to a perceived lack of interest of the providers with issues that may arise in day to day life. Also, the same group reported that they feared initiating conversations on such topics due to potentially revealing poor adherence (Beresford & Sloper, 2003). This ultimately leaves the provider responsible to engage these patients in regard to practical, behavioral, social, and emotional issues. Providers often have time constraints in clinical practice. These confinements force providers to prioritize conversations and topics to immediate medical management. Genetic counselors could appropriately address these concerns, thus improving overall communication of this crucial information.

Genetic counselors are uniquely trained in both medical knowledge and counseling techniques to best convey information at a level that could optimize communication with adolescents. An additional barrier to patient uptake of this information has been attributed to lack of access to providers, specifically genetic counselors. Often times these individuals have complex healthcare needs with many

specialist evaluations. Genetic counselors are trained healthcare professionals that may be better suited to meet these needs of adolescents. Case preparation is an essential portion of patient care that genetic counselors utilize. Prior to each routine appointment a patient's medical records are thoroughly reviewed to understand how each specialist is managing that individual's condition. This understanding allows counselors to coordinate care and adapt anticipatory guidance in light of their current medical status.

Rapport building and comfort level are continually highlighted as important factors to communicating with adolescents. Genetic counselors often utilize these two skills in conjunction with their genetic knowledge in each of their patient encounters. Communication could be improved with this age group by utilizing techniques such as allowing time to discuss concerns without a parent/guardian present, treating their concerns without judgement, giving autonomy to the individual, and use of reflection. (Callard et al., 2012). These practices could be utilized by many providers; however, genetic counselors are distinctively trained and positioned to serve the needs of this population. Genetic counselors can adapt the information and conversation to an educational level most appropriate for the patient. Often times they use visual aids which can assist in clarifying why there is increased risk in these behaviors by further explaining the specifics of their condition's mechanism. Genetic counselors training could be used to improve communication during patient encounters with emerging adults and to reduce or remove barriers to the patient obtaining pertinent information. The uptake of genetic counseling is increasing, but not fast enough to meet all of the patients' needs. Patients aged 0-17 and 18+ were surveyed, 40% and 34.2% respectively, identified having genetic counseling needs in the past year. Yet, only 69.5% and 56.2% reported receiving all the genetic counseling services they would have liked. Patients listed cost

of an appointment and doctors not knowing how to treat their condition as the top two barriers for not receiving needed genetic counseling (Romelczyk et al., 2015).

It appears that the perception of health care professionals in the transition of care of individuals with long term illness identified the most important factors in transition care initiation is not just the patient's age, but the maturity, family situations, and psychosocial factors of the patient. (Sparud-Lundin et al., 2017). Thus, the individuality of each patients' experience as they progress on the path of living with a chronic, lifelong condition is emphasized. Each patient will meet these hallmarks of transition readiness at different points, but this readiness is also directly related to providing the necessary information for a person to further develop in their own personal journey. An individual can only become adapted and mature when provided with the proper information and platform for communication. Transition care is an important aspect of an individual's medical management during the time of emerging adulthood. The study also found that providers are eager to adapt new strategies and programs to improve transition care for these individuals. (Sparud-Lundin et al., 2017).

In order to initiate new and improved practices to serve these unique needs, a better understanding of what emerging adults want to discuss during their health appointments must be obtained. According to a 2014 study in the United Kingdom there are a series of questions, known as a health professional checklist, to assess a young person's transition readiness. These questions include knowledge of condition, self-advocacy, smoking, alcohol, substance use, sexual health, and medical management (Wilson & McDonagh, 2014). The transition period is a crucial part of an individual's development of independence. The same social topics of substance use, sexual health, and independent living are part of the transition readiness assessment which indicates the

need for information on these topics in order to be prepared to be independent and self-managing one's condition.

Emerging adults with genetic conditions are a group of patients who need a provider to give them the tools and information for decision-making as they transition to independence. The provider should help the patient consider their larger healthcare context (condition's current status, development and prognosis, prescriptions, therapies, and side effects) when discussing the potential implications of social activities and behaviors. There are few studies looking at the social developmental aspects of patients with chronic conditions. Previous research has largely focused on the logistics of transition care from the pediatric to adult clinic; however, the needs of this patient population during this transition stretch far beyond attending appointments in a new clinic. To best serve this patient population, it is crucial to learn their preferred methods of communication regarding these topics. A needs assessment for preferred topics including age, setting, provider, and method to communicate should be performed. This study seeks to identify the social information needs of these individuals to help guide providers in partnering with this population to help them lead an informed, empowered, and healthy lifestyle.

CHAPTER II: MANUSCRIPT
EVALUATING THE SOCIAL INFORMATIONAL NEEDS OF EMERGING ADULTS
WITH GENETIC CONDITIONS¹

¹Whitmore, C., Fairey, J., MacCarrick, G., & Neukirch Elliot, J. To be submitted to *Journal of Genetic Counseling*.

2.1 ABSTRACT

Purpose: This study evaluated the healthcare provider's role during the social development of emerging adults with a genetic condition. The study also identified the type of information and ways that these adolescents hope to receive information on the potential side effects or complications from engaging in risk taking behaviors and lifestyle choices.

Methods: Participants included both males and females aged 18-26 with achondroplasia, sickle cell disease, cystic fibrosis, or Marfan syndrome. Respondents were recruited to complete an anonymous online questionnaire via social media support groups or email notification.

Results: There were 103 total respondents that completed the questionnaire and met the study's inclusion criteria. Fifty percent or more of participants have not talked to their providers about smoking, tobacco, drug use, sexual activity, contraceptives and birth control, moving out, or independent living. Using thematic analysis of open responses, it was reported those participants having discussions were not satisfied with the age in which the topics began to be discussed or the manner of the communication. Respondents indicated this information needs to be reviewed sooner and should include the topics of mental health and specific activity and exercise restrictions.

Conclusion: These results suggest that this patient population is experiencing gaps in care when it comes to anticipatory guidance in regard to making informed decisions about risk taking behaviors and their genetic condition. The patients who reported they did receive some of this information suggested that the timing or manner in which it was discussed with a provider was not preferred. Overall, there are improvements to be made to aid in

the emerging adult population which could empower their ability to make educated decisions about their genetic condition and lifestyle, thus improving their quality of life.

2.2 INTRODUCTION

Sickle cell disease, cystic fibrosis, Marfan syndrome, and Achondroplasia are conditions that impact a patient’s day to day life in many ways. Each of these conditions have life expectancies well into adulthood and do not typically affect an individual’s intellect. These conditions have distinct clinical presentations, but all require anticipatory guidance on activities far beyond their medications, therapies, and routine management.

Table 2.1 Summary of Conditions

Condition	Inheritance	Prevalence	Predominant ethnicity affected	Carrier Risk
Cystic fibrosis (CF)	Autosomal Recessive	30,000 in the US	Caucasian Ashkenazi Jewish	Caucasian: 1 in 25 African American: 1 in 61 Asian: 1 in 94 Hispanic: 1 in 58 Ashkenazi Jewish: 1 in 24
Sickle cell disease (SCD)	Autosomal Recessive	100,000 in the US	African American	African American: 1 in 10 Hispanic: 1 in 30 to 200
Marfan syndrome	Autosomal Dominant *De novo (25%)	1 in 5,000 worldwide	Panethnic	N/A
Achondroplasia	Autosomal Dominant *De novo (80%)	1 in 20,000 worldwide	Panethnic	N/A

*De novo = some cases can be due to a sporadic new mutation

For example, individuals with cystic fibrosis are often affected by chronic excess mucus in their lungs and pancreatic insufficiency that can impact their digestive tract. Due to these complications affected individuals should never smoke or be exposed to smoking of any kind (Yankaskas et al., 2004). An individual with Marfan syndrome primarily

experiences cardiac manifestations increasing risk for aortic aneurysm and dissection. Due to these risks, affected individuals should avoid contact sports and certain physical activities (Tinkle et al., 2013). Sickle cell disease results from abnormal hemoglobin and thus causes blood cells to form in a sickle shape which impedes their ability to travel, causing inadequate transport of oxygen throughout the body. Smoking, tobacco, and cocaine can lead to exacerbations and crises due to the reduction of oxygen and extra burden on the body, therefore these activities should be avoided (Sox et al., 2003). Achondroplasia is a skeletal dysplasia that causes short-limbed dwarfism which can lead to obesity, sleep apnea, spinal stenosis, kyphosis or lordosis of the spine, and recurrent ear infections. If a woman with achondroplasia is pregnant, she may require a cesarean section at delivery due a small pelvic outlet impeding a vaginal delivery (Trotter et al., 2005). Each of these conditions have a unique presentation, but all require specific anticipatory guidance on topics that may present themselves during emerging adulthood.

Emerging adulthood is a time in which a large shift occurs from codependence to independence. The emerging adult has varying definitions but most often covers the period from late teens to late twenties (Bundick, 2011). During this time period individuals affected by these conditions may engage in the same activities as an unaffected peer, including being able to attend college, engaging in personal and romantic relationships, and participating in activities. The main difference is that individuals with genetic conditions must consider their medical condition in the context of these activities. Activities with a potential health impact include alcohol, tobacco, and drug use, sexual activity, family planning, recurrence risks, career options, and independent living. All of these behaviors are topics that one may face in their teens and as they begin to make independent decisions. It has been identified that one of the most

important indicators for people to signal a successful transition to adulthood is when one accepts responsibility of their own actions and is able to define one's own beliefs and values. (Arnett, 2001). Individuals affected by a chronic, genetic disease may also use their medical management as a signal for transition to adulthood. When one can manage their own medical care and make educated decisions in day to day life, with their condition in mind, that individual may feel a greater sense of independence and empowerment.

Emerging adulthood is usually when these individuals are confronted by exposures to risk taking behaviors. In a multicenter study of individuals aged 12 to 19 with cystic fibrosis and sickle cell disease it was reported that 21% of teens with cystic fibrosis and 30% of those with sickle cell disease had previously smoked. In addition, 28% and 51% of teens, respectively, engaged in sexual intercourse. (Britto et al., 1998). Since this study was asking teens to report on sensitive topics it is suggested that these numbers may be underestimates. A 2004 study reported similar results; that young individuals with chronic illness and disability are not less likely than peers to participate in risk taking behaviors such as substance use and sexual activity (Suris et al., 2004). This leaves a large portion of affected individuals engaging in risky behaviors prior to receiving anticipatory guidance in the context of their specific medical condition.

Individuals with chronic illness and disability may be comparably engaging in similar behaviors as their unaffected peers, but often times their approach and reason for participating in risky activities is different. Adolescents with chronic illness tend to act in this manner due to the processing of their potential lethal condition that could impact their lifespan and/or life outcomes. (Suris et al., 2004). Frequently, these adolescents could be described as understanding and perceiving life in a unique way compared to

their unaffected peers. These differences in reasoning may be attributed to the earlier age in which they must worry about their health and the impact it may have on their life experiences.

In a recent study, the ability to communicate with health care providers has been found to be one of the leading factors in the quality of life of adolescents with a chronic condition (Ma et al., 2018). Another study looked at the communication practices of adolescents with juvenile chronic arthritis, cystic fibrosis, diabetes, epilepsy, and Duchenne muscular dystrophy. It found key themes that impact adolescent's communication with healthcare providers. Adolescents assume that providers are not interested in the issues that may arise in their daily lives. They also reported that they feared initiating conversations on such topics due to potentially revealing poor adherence (Beresford & Sloper, 2003). In order to initiate new and improved practices to serve these unique needs, a better understanding of what emerging adults want to discuss during their health appointments must be obtained.

A 2015 study surveyed individuals with genetic conditions and found that more than a third of children and half of the adults reported that they had never or rarely obtained the social and/or emotional support they needed from a health care provider or support group (Romelczyk et al., 2015). The lack of available information and counseling for this population may result from the sensitive nature of these topics causing uncomfortable encounters for both patient and provider. Furthermore, difficulty communicating and a lack of access to providers, specifically genetic counselors, are additional barriers. Genetic counselors are uniquely trained to utilize counseling techniques combined with their genetic and medical knowledge to provide tailored information and best communicate with adolescents. Interactions with adolescents

requires utilizing specific techniques such as allowing time to discuss concerns without a parent/guardian present, treating their concerns without judgement, giving autonomy to the individual, and use of reflection. (Callard et al., 2012).

The provider should develop methods to help the patient consider their larger healthcare context (condition's current status, development and prognosis, prescriptions, therapies, and side effects) when discussing the potential implications of social activities and behaviors. There is limited information on the social developmental issues of patients with chronic conditions. Previous studies have largely focused on the physical logistics of transitioning a patient's care from a pediatric-focused care pediatric to an adult-focused setting. The needs of these patients stretch far beyond coordinating the transfer of care. In order to best serve this patient population during this crucial time in life, it is essential to understand their preferred methods of communication regarding these sensitive topics. A needs assessment of preferred topics including age, setting, provider, and method to communicate should be performed. This study seeks to identify the social information needs of these individuals to help guide providers in partnering with this population to help them lead an informed, empowered, and healthy lifestyle.

2.3 MATERIALS AND METHODS

2.3.1 Participants

The Institutional Review Board at the University of South Carolina approved this study in June of 2018. Both male and female participants aged 18-26 years old with a diagnosis of cystic fibrosis, achondroplasia, sickle cell disease, or Marfan syndrome were invited to take part in the study. Exclusion criteria was applied to ensure that responses were not influenced by other individuals and to obtain feedback from those able to independently live and make decisions.

Exclusion criteria for this study were as follows:

- Participants must be able to independently answer the questionnaire
- Participants must not have a diagnosis of intellectual disability
- Participants must be able to read and write in English

There were 168 participants who started the questionnaire. Of those, 103 participants completed the questionnaire and met eligibility requirements and inclusion criteria. The questionnaire was administered to English speaking participants only as there were not accessible resources to permit interpretation to other languages.

A summary of the participant's demographics can be found in Table 2.1. The average age of the participants was 22.17 years of age. The majority of participants were female (67.96%), resided within the United States (80.6%), and reported Caucasian ethnicity (80.6%). Eighteen indicated a race other than Caucasian (17.5%), and two participants identified as more than one race (1.2%). All participants reported their highest level of education. The most common response was currently enrolled in college (37.86%) followed by completion of a college degree (29.13%), and high school diploma or equivalent (10.68%). The majority of participants identified as students (n=52) followed by employed full time (n=38). Most participants have been diagnosed with Marfan syndrome (84.47%) followed by cystic fibrosis (12.62%). Less than three percent of participants were affected by sickle cell disease and no participants reported a diagnosis of achondroplasia.

Table 2.2 Demographics of Study's Participants

Participant Characteristics	n	%
Sex	103	
Male	32	31.06
Female	70	67.96
Other	1	0.97
Residency	103	
United States	83	80.6
Outside the United States	20	19.4
Ethnicity	103	
Caucasian	83	80.6
African American	4	3.9
Asian	9	8.7
Hispanic/Latino	5	4.9
Multiethnic	2	1.2
Highest Level of Education	103	
Less than high school	1	0.97
Some high school	2	1.94
Currently in high school	3	2.91
High school diploma or equivalent	11	10.68
Some college	9	8.74
Currently enrolled in college	39	37.86
College degree	30	29.13
Graduate or professional degree	8	7.77
Occupation	128	
Participants could select more than one response		
Student	52	40.6
Employed full time	38	29.69
Employed part time	20	15.63
Disabled	10	7.81
Unemployed	8	6.25
Diagnosis	103	
Cystic Fibrosis	13	12.62
Marfan syndrome	87	84.47
Sickle cell disease	3	2.91
Achondroplasia	0	0.0

Recruitment of individuals was through support groups or foundations via Facebook, social media outlets, or email list serves. Additionally, individuals could have been recruited through support groups, website forums, and national or state level

organizations. Participants were asked to pass on the questionnaire to others whom they may know. Upon completion of the questionnaire individuals could voluntarily enter a raffle for a chance to win one \$50 Amazon gift card. The winner was randomly selected via a drawing which was held on February 11th, 2019. The winning participant received their gift card electronically within 24 hours of the drawing.

2.3.2. Materials

The study utilized an author designed questionnaire. The questionnaire was administered through an online source (SurveyMonkey.com) beginning in August 2018 and concluding in January 2019. The online questionnaire provided participants with anonymity and convenience. A consent statement was included with the online questionnaire. Completion of the questionnaire served as agreement to consent. The questionnaire took approximately twenty minutes to complete and included a total of 82 multiple choice, open response, and ranking questions. A link to the online questionnaire was posted to online support pages and included in emails. Participation was entirely voluntary and subjects could withdraw from the study at any point. Demographic information was collected as part of the questionnaire. The multiple-choice portion of the questionnaire included questions that were required in order to submit the questionnaire. Failure to answer these questions resulted in inability to submit the questionnaire.

2.3.3 Methods

Participants could follow the instructions provided to them via an online or email-based source. Subjects could complete the questionnaire at any time or place using their own internet enabled device by following the provided link. Upon beginning the questionnaire, a consent statement was present, and subjects had to read it in order to proceed. The responses to the questionnaire were then stored on the password protected

site as well as a password protected computer. Data was collected from the completed online questionnaires and responses were analyzed.

The study employed mixed methodology utilizing both qualitative and quantitative methods. Data was analyzed in January and February of 2019. The analysis was completed by the principal investigator, USC statistics, committee members, and thesis advisor. All information is now kept in secure filing cabinets or password protected computers. Quantitative data analysis was performed using SPSS statistical analysis software. Both descriptive and inferential nonparametric statistics were used. Thematic analysis of open-ended qualitative data questions was performed. One response could have multiple themes identified.

2.4 RESULTS

All 103 participants' questionnaires were considered in reporting the results and thus included in data analysis. Ninety-eight participants completed all the questions. Since no individuals reported a diagnosis of Achondroplasia, this condition is not represented in data analysis.

Forty-five participants (43.7%) indicated they had a family member or members affected by the same condition as them. If a participant responded yes, they were then asked to select the relationship they share to other affected family members. The most common family member to also be affected by the same condition of the participant were siblings ($n = 23$), followed by mother ($n = 17$) and father ($n = 16$). If the participants reported having an affected family member, they were asked to share how this had impacted their development and transition to independence. Thirty-five responses were analyzed, and the most common theme identified was that family members have had a positive impact with 12 responses. The second most common theme identified, with ten

responses was that it had a negative impact on them. Nine other responses indicated that having an affected family member has not had an impact on their transition experience.

Positive

“I’ve had a guide. It’s been very helpful to have someone show me what I need to do in order to take care of myself and stay on top of my own treatment independently.”

Negative

“We are all pretty high functioning and have known about the diagnoses my whole life so for me this is all I’ve known. However, it is an extra stressor always being concerned for each other and how things are changing for my father and brother as they age. It impacts my anxiety and depression.”

“My sister passed away from CF when I was 4. At the time, I didn’t really understand what had happened. As I grew older, my family tended to compare us often--with the implication frequently being that I was lucky to not be as sick as she had been, that I should be more consistent with my treatments like she had been, etc. I know that ultimately this came from a place of concern and love, but it was very hard as a teenager to be frequently compared to her.”

No impact

“My Grandfather, his three children and 3 out of 4 of my siblings all have Marfans. This had no impact on my transition to independence, and it had very little impact on my development.”

Figure 2.1 is a summary of the providers participants reported seeing regularly between the ages of 14 to 26. The most common type of provider seen by this age group

is a specialist physician, followed by a primary care physician and genetic counselor. Of the participants who responded, 25 have seen a genetic counselor regularly while 69 participants have not.

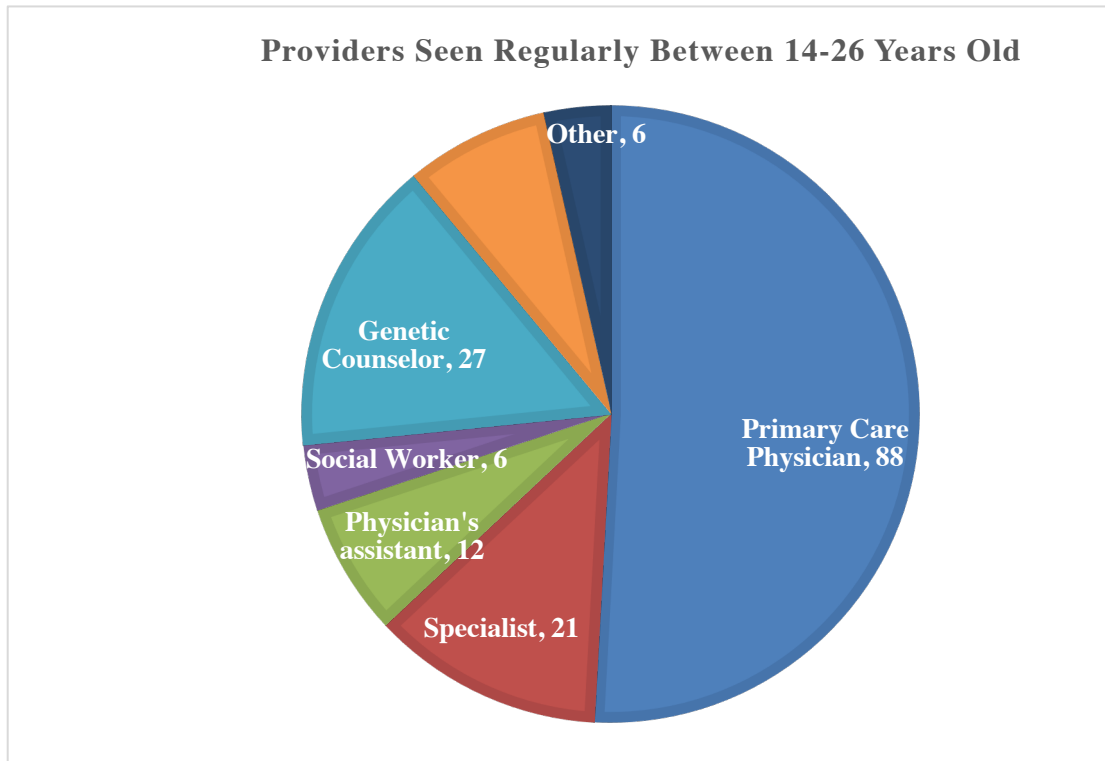


Figure 2.1 Participants' Provider Types

Thirty-nine (92.9%) participants who live in the U.S. and three (7.1%) outside of the U.S. either are still seen by pediatric providers or have providers that can see them through adulthood, while 44 (72.1%) U.S. and 17 (27.9%) outside of the U.S. participants are followed by adult care providers. There was a statistically significant difference in the likelihood of those living outside the U.S. seeing adult care providers compared to those inside the U.S. as assessed by Fisher's exact test, $p = .01$.

Forty-one (41.8%) participants are either still seen by pediatric providers or have providers that can see them through adulthood, while 57 (58.2%) participants are followed by adult care providers. The amount of social information topics reported to

have been discussed with a provider was quantified. A one-way ANOVA was conducted to determine if there was a difference in the amount of social information provided between the groups. Participants were classified into two groups: those seeing a pediatric provider ($n = 41$) and those seeing an adult care provider ($n = 57$). Those seeing a pediatric provider ($M = 18.29, SD = 2.96$) had a slightly lower mean of information provided than the mean of those seen by adult providers ($M = 19.05, SD = 2.91$), but the differences between these groups was not statistically significant, $F(1, 96) = 1.61, p = .21, \eta^2 = .02$.

There were 80 (81.6%) participants from within the U.S. and 18 (18.4%) from outside the U.S. The number of social information topics reported to have been discussed with a provider was quantified. A one-way ANOVA was conducted to determine if there was a difference in the amount of social information provided to the two different groups based on their residency. Participants were classified into two groups: those in the U.S. ($n = 80$) and those outside of the U.S. ($n = 18$). Those inside U.S. ($M = 18.43, SD = 2.93$) had a lower mean of information provided compared to the mean of information provided to those from outside the U.S. ($M = 20.11, SD = 2.61$), the differences between these groups was statistically significant, $F(1, 96) = 5.04, p = .03, \eta^2 = .05$.

Ninety-eight respondents reported if they have had conversations about specific topics with their providers. The most commonly discussed topic was recurrence risk with 77.55% of people who responded having talked about this with their provider. The least common conversation topic was moving out, with only 13.27% of participants reporting they talked about this with a provider. Figure 2.2 summarizes the number of participants reported to have had conversations about each topic with their provider.

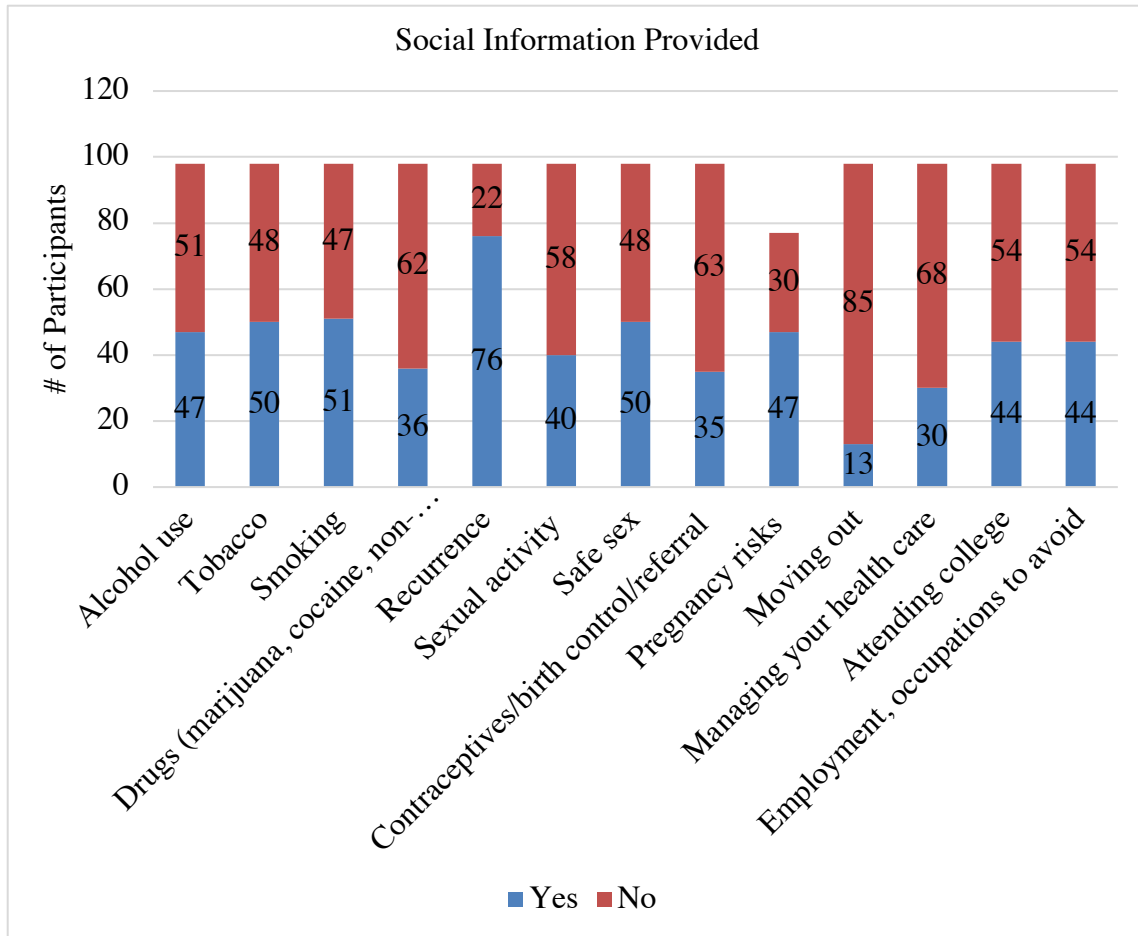


Figure 2.2 Amount of Social Information

For participants who reported having a conversation, they were asked to comment on the discussions had with their provider regarding the group of topics of alcohol, tobacco, and drug use and smoking. Fifty-two responses were analyzed, and the most common theme identified was that participants received a warning from their providers with 27 responses. The second most common theme identified, with 19 responses, was that a provider had asked if they engaged in any one of those activities. Nine other responses described a limited conversation was had in regard to these topics. Acceptable use of these substances was reportedly discussed with nine participants. Some of these themes overlap and one response can have more than one theme.

Warning

“All were just warning about effects and possible outcomes if I were to use any”

“The cardiologist simply discussed the risks involved in using any of the above mentioned.”

Asked if respondent engaged in activity

“Beyond asking me if I partake and the general "don't do it" conversation, they never really went in-depth with these topics.”

“I have always made it clear to my doctors that I have no interest in alcohol or recreational drugs, so they haven't really had to talk to me about it.”

“I've been asked questions regarding my use, which is standard medical practice anywhere. However, no I have never really discussed the specific consequences on my condition (Marfans).”

Limited

“Briefly discussed usage, if at all. Not really a pressing issue or concern, however I do not remember ever seeing any material that could be related”

“The conversations I have had with providers have been very minimal, I do not smoke, use tobacco or other drugs. My alcohol consumption is very minimal. The discussion is generally just an acknowledgement that I should continue to avoid these behaviors as I have been.”

Acceptable use

“I think the conversations were needed. I think it is embedded in my brain when I am at social events the outcome I will have when trying to drink alcohol with others and that I will have a heavier effect when drinking than others will because of my condition so it helps me limit my use.”

“My doctor explained that I could drink but because of my medication, I would feel the effects faster and with less drinks”

If the participants reported having conversation, they were asked to comment on the conversations had with their provider regarding sexual activity. Fifty-two responses were analyzed, and the most common theme identified was that participants learned of risks from their providers, with 19 responses. The second most common theme identified, with 14 responses, was that a provider had discussed recurrence risks. Fourteen responses also described a conversation on pregnancy and STD prevention. Some of these themes overlap and one response can have more than one theme.

Risks

“Just that pregnancy with Marfans is high risk.”

“I have spoken with my Marfans specialist about the effects of pregnancy on my aorta and what type of conversations we would need to have prior to my trying to conceive a child, if I ever decide to do so.”

Recurrence Risks

“Again, I have just had the generic safe sex chat with my doctor. I know the risks of// chance of passing marfans onto my future children.”

“I brought up birth control to my PCP around age 17-18. After that all providers regularly asked about sexual activity. My pulmonologist referred me to a gynecologist around 21 years old to assess for the best birth control method because of the risks of taking the pill when on antibiotics. My pulmonologist briefly mentioned needing to have my partner assessed for the CF gene prior to attempting to have kids.”

Pregnancy and STD Prevention

“Safe sex is the best and let others you commit the activity with know about my condition”

“Many conversations actually revolve around preventing the chances that my children could inherit the condition. Regarding safe sex, and when the time comes we can explore opportunities for family planning.”

If the participants reported having a conversation, they were asked to comment on the conversations had with their provider pertaining to independent living. Thirty-four responses were analyzed, and the most common themes identified in the responses were restrictions needed and limited conversations, with each of these themes classified in ten responses. A theme of overall healthy living was identified in eight responses.

Restrictions

“College was discussed in the context of daily physical activity such as increased amounts of walking or climbing stairs. I have been advised to avoid strenuous activity such as heavy lifting.”

“No strenuous/heavy lifting -- more in the scope of lifestyle (e.g. working out) rather than independent living”

Limited

“Physical restrictions- have to work with brain. Yada yada”

“There are not many resources for employment & “independent” living.”

Healthy living

“I was told to have fun at college but to be safe and listen to my body”

“We have spoken about college and how looking at colleges that are close to a hospital and have accessibility on campus”

Overall respondents were asked to comment on the transition process and information provided to them as they approached adulthood and independence. Of the 46 responses analyzed, 14 responses had a theme of lack of information and support. The second most common theme was that this was a negative or stressful experience with nine responses.

Lack of information and support

“I was given a lot of information about pregnancy and childbearing, because I’m a woman, but I didn’t receive any other information nearly as extensively. Wish I had.”

“I was never presented with any information. I knew signs and symptoms that could be red flags, having gone into the medical field, but was not given any information that could have helped me as I prepared to move to college.”

“Nothing was “provided to me” it was more of a “what questions do you have” conversation so that made it more confusing.”

Negative/stressful experience

“The transition process wasn’t easy, I had a very close bond with my pediatric team. I don’t think they handle transitioning very well.”

“It would have been nice to have more support for the transition from high school to college... and now from college to the next step”

In addition to the social topics addressed in the questionnaire participants were asked what else they believed needed to be discussed with providers, or topics missed by providers during the transition process. Twenty-four responses were analyzed. Six responses identified they desired more information in regard to exercise, which overlapped with a theme of wanting to know more about their restrictions. Another major

theme emerged, the participants believe discussions about mental health needed to be included during this transition time.

Exercise

“I think lifestyle conversations need to take place regarding diet habits, exercising, employment accommodations, disability or disability disclosure to employers, and diagnosis prognosis and what are reasonable expectations for how the condition will affect your life through adulthood.”

Mental health

“I think there should be more automatic or seamless connections between providers and social workers/therapists... like that providers for people with chronic illness are making it routine to offer to connect their patients with these supports... not just when they see signs off problems but before that more as preventative instead of as clean up...”

“MENTAL HEALTH. I believe this is the backbone to any and all other health aspects.”

If participants believed they received limited or no information on these topics in regard to their condition, they were asked to comment on the impact this has had on them. Twenty-six responses were analyzed. Fourteen responses could be classified as a negative impact or making the participant not feel knowledgeable as an adult.

Negative impact

“As I came into adulthood I had limited information about what was safe for me to do, I began to do less and less, I got into worse shape. It may have resulted in cardiac deconditioning.”

“Being in denial about my depression and anxiety certainly made an impact on my health, and my life as a whole. It took a lot of unraveling and therapy to come to terms with the fact that CF and depression in my life were closely intertwined.”

Not knowledgeable as an adult

“I don't do drugs or drink so that hasn't affected me too much, however being aware is always important. The other issues have just kind of left me feeling alone and like I have to figure things out on my own. I haven't been able to see a Marfan specialist in around 4 years and haven't been able to get the knowledge I feel I need.”

“I've had to search and find information in other places and other times I am just "winging it" and hoping for the best”

Figure 2.3 demonstrates the tasks and life milestones completed by participants. A majority of participants have moved out of their parent/guardians' home (72.82%) and manage their medications (68.93%). An overwhelming amount (90.29%) participants are able to explain their condition. A majority (54.37%) of participants do not attend their medical appointments alone.

Assessment of Transition

- Yes
- No
- No with parent/guardian
- No, I attend with a partner/spouse
- No, I bring a friend
- I am not on medication
- My parent and I co-answer
- My partner/spouse and I co-answer
- Uninsured

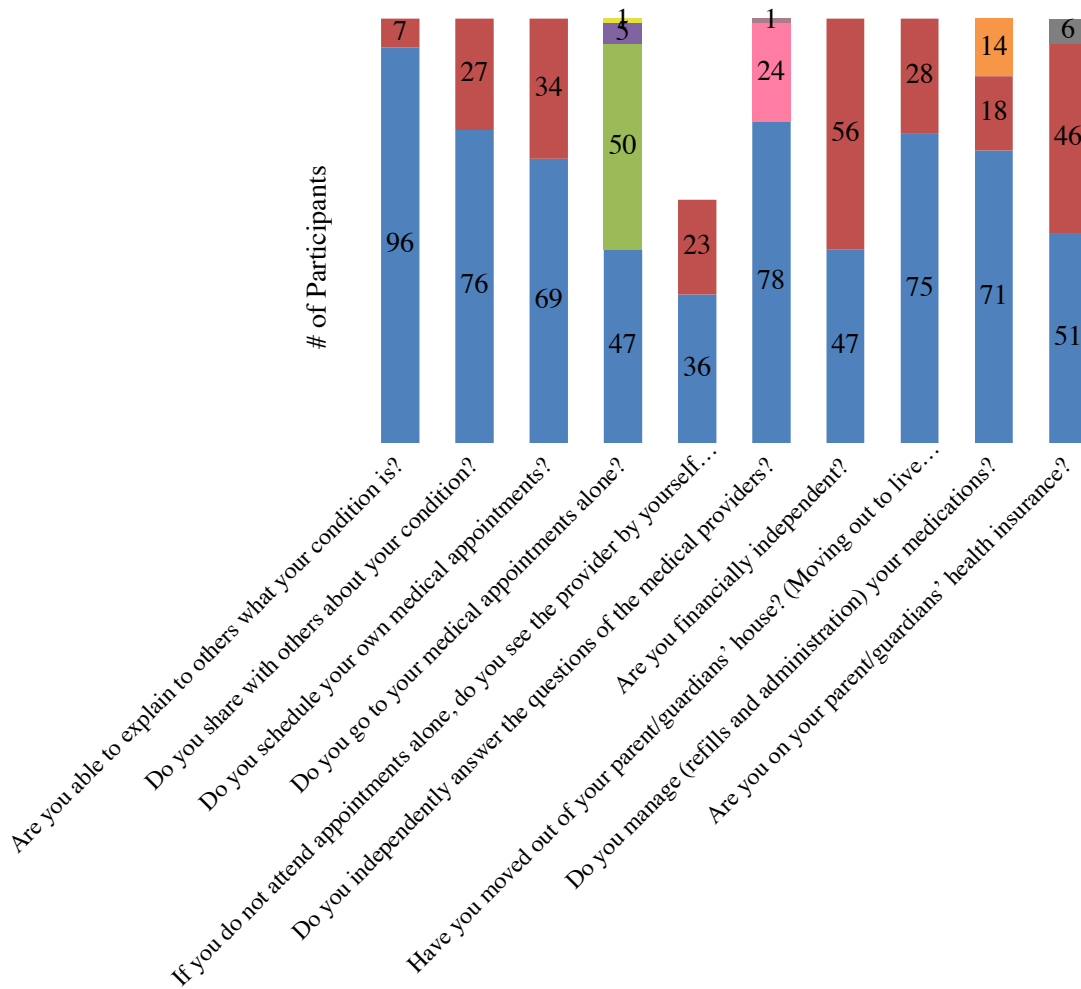


Figure 2.3 Transition Assessment of Participants

Table 2.3 reports the number of participants that provided a numerical age at which they had first discussed the indicated topic with a provider. Using this, the average age each topic began being discussed was calculated. Based on participant report the

average age was 17.91 years for alcohol, 16.4 years for tobacco, 16.32 years for smoking, and 17.07 years old for drug use.

Table 2.3 Mean Age Information was Provided

Topic	Number of participants who provided a numerical age	Average age to have first had a conversation about this topic (years)
Alcohol use	29	17.91
Tobacco	30	16.40
Smoking	30	16.32
Drugs (marijuana, cocaine, non-prescribed medications, etc.)	22	17.07

In contrast, participants were asked what age they thought receiving information about each topic would be necessary or most helpful. For alcohol, tobacco, smoking, drugs, pregnancy, recurrence risk, and sexual activity respondents thought based on the average age reported for each topic that discussions on these topics should begin at 14.6 years of age. Subjects only preferred an older age of about 16.8 years of age for discussing independent living with a provider.

Another assessment to understand how these participants feel most comfortable receiving this information was to know if they would want a parent or guardian present during conversations with providers about specific types of information. On average, 62.42% of participants indicated they would not want a parent or guardian present during conversations with their providers about any of the following topics: alcohol, tobacco, smoking, drugs, and pregnancy. When it came to the topic of sexual activity, 84.62% of participants would not want a parent or guardian present when talking to a provider about this topic. For the topic of recurrence, 54.95% of respondents wanted a parent or guardian present to discuss this, while 45.05% did not want them present. A preference for a parent or guardian to be present was indicated for conversations about independent living, with

64.84% of participants wanting a parent or guardian to be present when discussing this with a provider.

Participants were asked what factors would be important and make them most comfortable when receiving information about these topics. Two major themes appeared in the 40 responses analyzed. Sixteen responses indicated that the provider's approach and skills are essential components to delivery of this information. Twelve responses highlighted the importance of a non-judgmental provider and conversation.

Provider's skills and approach

"It coming from a provider I am comfortable with and has knowledge about my condition and background"

"To have them framed in a respectful and progressive manner, not to make the patient feel guilty, nervous, uncomfortable, or afraid as I was made to feel about the possibilities of pregnancy."

Non-judgmental

"Being able to hold the narrative, an approach without a condescending tone"

"Being given the chance to speak openly and freely without any sort of judgement about certain topics."

2.5 DISCUSSION

This study explored the amount and manner in which social information was shared with individuals with chronic genetic conditions. We also explored patient preferences on how this communication may be improved in the future. Overall, results demonstrate that there are both major gaps of knowledge and care for this patient population. Essential information pertaining to alcohol, tobacco, smoking, pregnancy risks, and safe sex was discussed at most 52% of the time. Contraceptives, sexual

activity, drugs, attending college, and employment/occupations to avoid were only covered by, at best, 44.9% of providers. At even lower rates, topics such as managing your own healthcare and moving out were discussed the least with 30% and 13%, respectively, of patients engaging with their provider (Figure 2.2). The probability of these patients receiving this information was no better than the flip of a coin. These results highlight the fact that there are many emerging adults with genetic conditions facing major decisions ill-equipped with proper knowledge to best determine a course of action for themselves in the context of their condition.

Those who did report having had conversations about these topics overwhelmingly reported the limitations and negative delivery of the information they received. There were major differences with the age these conversations took place in comparison to the age participants believed these topics should have started to be addressed. On average, 16.8 years of age was the preferred time to begin discussing matters surrounding independent living. For alcohol, drugs, smoking, and tobacco participants reported they thought on average this should start to be discussed at 14.6 years of age. However, for those who did receive this information they reported that topics were brought up much later, such as alcohol at 17.9, tobacco at 16.4, smoking at 16.3, and drugs at 17.1 years of age. Overall, when these topics are covered the participants believe it is too late by, at minimum, two years.

This study began to explore important topics to be covered by providers in order to empower this patient population to make educated decisions. However, there were topics this study did not cover that participants were interested in being addressed, such as mental health and specific lifestyle restrictions, especially pertaining to exercise. This indicates that in addition to increasing the rate at which social informational topics are

discussed, as defined by this study, providers should also incorporate talking about mental health and specific lifestyle guidelines and exercise restrictions for their patient.

Overall these individuals are relatively well versed and able to independently handle major aspects of their lives and medical care. Yet, they have not had the opportunity to discuss sensitive topics with providers. A majority (73%) of participants have moved out of their parents or guardians' home, but only 13% of them have talked about moving out with a provider. Currently, the social information provided is reactive and is not communicated proactively, when it would be most beneficial and empowering to the patient.

Emerging adulthood is a complex time, and transition care poses many unique challenges to the provider. To best improve care, not only do the rates of this information being covered need to be improved, but also the overall communication between provider and patient. It could be argued the delivery of this information is just as important, if not more important, than the actual information. Practices such as speaking to an adolescent without a parent or guardian present during the appointment could greatly change the dynamics and allow more open exchanges between provider and adolescent. Two of the most common themes identified by this population when asked what would make them most comfortable were the provider's approach and a non-judgmental attitude.

The open responses overall give us a glimpse into understanding the direct perspective of this population. When analyzed, many of these responses were polarizing and highlighted that this time is a unique experience for each individual patient. Currently, some transition tools implemented in clinic care are checklists for providers to utilize in order to address these topics. These tools can be helpful to remember to cover

these topics; however, this study highlights the personalized approach needed for each patient. Individualized care would allow providers to adapt these conversations and discussions to meet the patient needs and where they may be during their transition. Genetic counselors commonly tailor each of their appointments to meet a client's needs and their training could be greatly utilized to assist in the transition of emerging adults with genetic conditions.

This study is not without limitations. The study sample only included those diagnosed with four conditions and the number of participants representing each of these conditions was limited. It is possible that individuals with other conditions may not feel the same way. The study was also conducted and only in English; therefore participants must have had access to an internet enabled device in order to participate and been able to read and write in English. The population was overall well educated, female, and mainly of Caucasian background. However, this sample was representative of all age groups included in the study and participants reported a wide range of occupation types. Due to the nature in which the survey was distributed there was potential bias in those who would be connected to support groups and national foundation organizations.

Further research needs to be done to best understand the needs of this unique and growing patient population. A study could be completed in order to understand and explain the differences of this population within and outside of the United States. This study could potentially be repeated with populations affected by different genetic conditions such as, Neurofibromatosis type 1 or metabolic disorders, to see if similar rates of information and themes are present. Additional studies could be done to understand the provider's perspective of delivering this type of information and care. If

this information was available, tools and effective methods of improving this type of care could be developed to better meet the needs of both provider and patient.

There is still a great deal of improvement needed to serve those affected by a genetic condition in emerging adulthood. Our study hopes to assist in prompting more numerous and further discussions of these topics in order to include and empower these individuals in their daily lives.

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APPENDIX A: QUESTIONNAIRE

Thank you for your interest in this study. This questionnaire will ask you about your experience obtaining information on social behaviors and how they may impact and relate to your condition. You are being asked to participate in this research study because you have received a diagnosis of cystic fibrosis, achondroplasia, sickle cell anemia, or Marfan syndrome and are between the ages of 18-26. Your responses will be completely anonymous and not linked to identifying information. Participation in this study is completely voluntary and all responses are anonymous and will be kept confidential. Informed consent will be implied upon completion of the questionnaire, but you may stop your participation at any time or choose not to answer specific questions without any consequence, as some questions may touch on sensitive topics.

The online questionnaire should take approximately 20 minutes to complete and there will be no course credit or monetary compensation given for your participation. However, we are holding a raffle for a \$50 Amazon gift card if you would like to enter upon completion of the questionnaire. In order to enter hit submit on the questionnaire and you will be directed to opt into the raffle and an optional follow up phone interview. Both are not required for your participation and are entirely voluntary. Your time is greatly appreciated and we hope the results will further research in the emerging adult community.

If you have any questions, please do not hesitate to contact me by email at courtney.whitmore@uscmed.sc.edu or by phone at (774) 222-3674.

Thank you,

Courtney Whitmore
Master's Candidate in Genetic Counseling
University of South Carolina- Columbia

1. By clicking yes, I have read the above statement and agree to participate in this study.
 - a. Yes

Demographics

2. Age: Sliding scale of all ages between 18-26
3. I am: Male, Female, Other (Multiple Choice)
4. I live in the: United States/Not in the United States (Multiple Choice)

5. Which ethnicity do you identify most with: Caucasian, African American, Asian, American Indian, Hispanic/Latino, Hawaiian/Pacific Island, Other, please specify (Multiple Choice)
6. What is your highest level of education? (Multiple Choice)
 - a. Less than high school
 - b. Some high school
 - c. Currently in high school
 - d. High school diploma or equivalent
 - e. Some college, vocational, or trade school
 - f. Currently enrolled in college, vocational, or trade school
 - g. College degree
 - h. Graduate or professional degree
7. What is your occupation? (Multiple choice, select all that apply)
 - a. Student
 - b. Employed Full Time
 - c. Employed Part Time
 - d. Disabled
 - e. Unemployed
8. I am diagnosed with:
 - a. Cystic fibrosis
 - b. Achondroplasia
 - c. Marfan syndrome
 - d. Sickle cell anemia
 - e. Other, please specify
9. Have you received a diagnosis of intellectual disability?
 - a. Yes
 - b. No
10. Are you able to independently complete this questionnaire?
 - a. Yes
 - b. No
11. Are any of your family members affected with the same condition as you?
 - a. Yes
 - b. No
12. If yes to #11, who? (Select all that apply)
 - a. Mother
 - b. Father
 - c. Sibling
 - d. Grandparent
 - e. Cousin
 - f. Aunt/Uncle
 - g. Other
 - i. Who?
13. If yes, please share how this has impacted your development and transition to independence.

14. Are you able to explain to others (friends, coworkers, extended family) what your condition is?
 - a. Yes
 - b. No
15. Do you share with others about your condition?
 - a. Yes
 - b. No
16. Who do/did you see regularly between the ages of 14-26? (Check all that apply)
 - a. Primary care physician
 - b. Specialist physician
 - c. Nurse
 - d. Physician's Assistant
 - e. Social worker
 - f. Genetic counselor
 - g. Therapist
 - h. Other, please specify type and specialty of provider
17. Are you seen by pediatric providers?
 - a. Yes, I still see my pediatric providers
 - b. Yes, because my providers see patients through adulthood
 - c. No, I see adult providers
18. Do you independently answer the questions of the medical providers?
 - a. Yes
 - b. No, other people at appointments respond for me most of the time
 - c. My parent and I co-answer questions
 - d. My partner/spouse and I co-answer questions
19. Do you schedule your own medical appointments?
 - a. Yes
 - b. No
20. Do you go to your medical appointments alone?
 - a. Yes
 - b. No, my parent/guardian attend with me
 - c. No, I attend with a partner/spouse
 - d. No, I bring a friend
21. If no, do you see the provider by yourself for any of the appointment?
 - a. Yes
 - b. No
22. Do you manage (refills and administration) your medications?
 - a. Yes
 - b. No
 - c. I am not on medications
23. Are you financially independent?
 - a. Yes
 - b. No

24. Are you still on your parent/guardians' health insurance plan?
 - a. Yes, I am on my parent/guardians' plan
 - b. No, I have my own health insurance
 - c. No, I am uninsured
25. Have you moved out of your parent/guardian's house? (Moving out to live at college would be indicated by a yes)
 - a. Yes
 - b. No

Now thinking about your medical care and appointments please indicate if you have had any interactions with your provider about the following behaviors/topics and the potential effect they may have on your condition.

26. Have any of your providers spoken to you about alcohol use?
 - a. Yes
 - b. No
27. If yes, which provider (please specify type and speciality of the provider)? How old were you?
28. Did they explain the effects alcohol may have on your medical condition?
 - a. Yes
 - b. No
29. Have any of your providers spoken to you about tobacco use?
 - a. Yes
 - b. No
30. If yes, which provider (please specify type and speciality of the provider)? How old were you?
31. Did they explain the effects tobacco may have on your medical condition?
 - a. Yes
 - b. No
32. Have any of your providers spoken to you about smoking?
 - a. Yes
 - b. No
33. If yes, which provider (please specify type and speciality of the provider)? How old were you?
34. Did they explain the effects smoking may have on your medical condition?
 - a. Yes
 - b. No
35. Have any of your providers spoken to you about drug (marijuana, cocaine, non-prescribed medications, etc.) use?
 - a. Yes
 - b. No
36. If yes, which provider (please specify type and speciality of the provider)? How old were you?
37. Did they explain the effects drugs may have on your medical condition?
 - a. Yes
 - b. No

38. Overall, please comment on the conversations you have had with your provider regarding any of the above (alcohol, tobacco, drug use, and smoking) topics. (If you have not had a conversation please indicate N/A)
39. Have any of your providers explained the chance that if you have a child they will have your same condition (also known as recurrence risk)?
 - a. Yes
 - b. No
40. If you are a female have any of your providers discussed the potential risks associated with pregnancy?
 - a. Yes
 - b. No
41. Thinking about sexual activity, have any of your provider mentioned if you are able to engage in such behavior?
 - a. Yes
 - b. No
42. Have any of your providers spoken to you about practicing safe sex?
 - a. Yes
 - b. No
43. Have any of your providers spoken to you or referred you to a specialist about contraceptives and birth control?
 - a. Yes
 - b. No
44. Overall, please comment on the conversations you have had with your provider regarding sexual activity. (If you have not had a conversation please indicate N/A)
45. Have any of your providers spoken to you about moving out of your parent/guardians' home?
 - a. Yes
 - b. No
46. Have any of your providers spoken to you about managing or taking complete control of your own healthcare?
 - a. Yes
 - b. No
47. Have any of your providers spoken to you about attending college?
 - a. Yes
 - b. No
48. Have any of your providers spoken to you about employment and potential occupations to avoid?
 - a. Yes
 - b. No
49. Overall, please comment on the conversations you have had with your provider regarding independent living. (If you have not had a conversation please indicate N/A)

Now with these topics in mind think about how you would feel most comfortable receiving information about the following topics/behaviors and the potential effect they may have on your condition.

50. Would you ever feel comfortable being the one to initiate the conversations with a provider about these topics? Please indicate yes or no for each topic.
- Alcohol:
 - a. Yes, I would be okay to initiate a question or conversation about this topic
 - b. No, I would not feel comfortable asking any of my providers about this I would prefer they start the conversation
 - Tobacco:
 - a. Yes, I would be okay to initiate a question or conversation about this topic
 - b. No, I would not feel comfortable asking any of my providers about this I would prefer they start the conversation
 - Smoking:
 - a. Yes, I would be okay to initiate a question or conversation about this topic
 - b. No, I would not feel comfortable asking any of my providers about this I would prefer they start the conversation
 - Drugs:
 - a. Yes, I would be okay to initiate a question or conversation about this topic
 - b. No, I would not feel comfortable asking any of my providers about this I would prefer they start the conversation
 - Pregnancy:
 - a. Yes, I would be okay to initiate a question or conversation about this topic
 - b. No, I would not feel comfortable asking any of my providers about this I would prefer they start the conversation
 - Recurrence Risk:
 - a. Yes, I would be okay to initiate a question or conversation about this topic
 - b. No, I would not feel comfortable asking any of my providers about this I would prefer they start the conversation
 - Sexual Activity:
 - a. Yes, I would be okay to initiate a question or conversation about this topic
 - b. No, I would not feel comfortable asking any of my providers about this I would prefer they start the conversation
 - Independent Living:
 - a. Yes, I would be okay to initiate a question or conversation about this topic
 - b. No, I would not feel comfortable asking any of my providers about this I would prefer they start the conversation

51. Who (i.e. doctor, nurse, genetic counselor, etc.) you would feel most comfortable receiving information from about each topic in relationship to your health condition? Please rank from 1-5, 5 being the most comfortable and 1 being the least comfortable.

- Alcohol/Tobacco/Smoking/Drugs
 - a. Doctor
 - b. Nurse
 - c. Genetic Counselor
 - d. Social Worker
 - e. Therapist
- Pregnancy/Recurrence Risk/Sexual Activity
 - a. Doctor
 - b. Nurse
 - c. Genetic Counselor
 - d. Social Worker
 - e. Therapist
- Independent Living
 - a. Doctor
 - b. Nurse
 - c. Genetic Counselor
 - d. Social Worker
 - e. Therapist

52. Please indicate at what age you think each topic should begin to be discussed?

- Alcohol:
 - a. Sliding scale of ages 10-30
- Tobacco:
 - a. Sliding scale of ages 10-30
- Smoking:
 - a. Sliding scale of ages 10-30
- Drugs:
 - a. Sliding scale of ages 10-30
- Pregnancy:
 - a. Sliding scale of ages 10-30
- Recurrence Risk:
 - a. Sliding scale of ages 10-30
- Sexual Activity:
 - a. Sliding scale of ages 10-30
- Independent Living:
 - a. Sliding scale of ages 10-30

53. Please indicate if you would like a parent/guardian to be present for the conversations about each topic.
- Alcohol
 - a. Yes, I would feel most comfortable if my parent/guardian was present
 - b. No, I would feel most comfortable if my parent/guardian was NOT present
 - Tobacco
 - a. Yes, I would feel most comfortable if my parent/guardian was present
 - b. No, I would feel most comfortable if my parent/guardian was NOT present
 - Smoking
 - a. Yes, I would feel most comfortable if my parent/guardian was present
 - b. No, I would feel most comfortable if my parent/guardian was NOT present
 - Drugs
 - a. Yes, I would feel most comfortable if my parent/guardian was present
 - b. No, I would feel most comfortable if my parent/guardian was NOT present
 - Pregnancy
 - a. Yes, I would feel most comfortable if my parent/guardian was present
 - b. No, I would feel most comfortable if my parent/guardian was NOT present
 - Recurrence Risk
 - a. Yes, I would feel most comfortable if my parent/guardian was present
 - b. No, I would feel most comfortable if my parent/guardian was NOT present
 - Sexual Activity
 - a. Yes, I would feel most comfortable if my parent/guardian was present
 - b. No, I would feel most comfortable if my parent/guardian was NOT present
 - Independent Living
 - a. Yes, I would feel most comfortable if my parent/guardian was present
 - b. No, I would feel most comfortable if my parent/guardian was NOT present
54. What other things would be important to you and make you most comfortable when receiving information about these topics?
55. Please comment on the transition process and information provided to you as you approached adulthood and independence.
56. Is there other information or topics you think should be addressed during this time?
- a. Yes
 - b. No
57. If yes, please comment which topics or information also needs to be addressed or were missed during your conversation with a provider.
58. If you feel you received limited or no information about these topics in regard to your medical condition, please comment on the effect this had on you.
59. If there is any other information you would like us to know about this topic please comment here about your experience, opinions, or reflections.