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Genetic Counseling for Alcohol Use Disorder: Assessment of Need in Affected and At-Risk Populations

by

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Bachelor of Arts
Indiana University, 2013

Submitted in Partial Fulfillment of the Requirements

For the Degree of Master of Science in

Genetic Counseling

School of Medicine

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Dedication

To my wonderful classmates and friends. I have been so fortunate to get to know all of you over the last two years. Thank you for being my family away from home and for your constant support, encouragement, and laughs. I am so proud to call you all my colleagues, and I will miss you all as we go forward and do great things.

To my family- Mom, Dad, and Haley. Thank you for always believing in me and encouraging me to chase my dreams. I would not be where I am, or who I am, without you. I love you all to the moon and back.

To my fiancé and best friend, Max. Thank you for your endless support, smiles, laughs, and encouragement. You have been the best teammate and hand to hold throughout this adventure, and I cannot wait for the many adventures ahead of us. I love you.

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Abstract

Introduction: Alcohol use disorder (AUD) is highly heritable, yet there has been no investigation regarding the possible benefits of genetic counseling for AUD. This study assessed the beliefs individuals with and at risk for AUD have regarding recurrence risk and etiology of AUD, how the presence of the condition in themselves or their family history has affected their lifestyle decisions, and potential benefit from AUD genetic counseling. **Methods:** An online questionnaire was distributed through social media to support groups for AUD inviting adults 18 years and older with a personal or family history of AUD. **Results:** Of the 122 individuals who completed the online questionnaire, 60% of participants perceived a potential benefit of AUD genetic counseling. Participants reported a wide range of estimated recurrence risks for first-degree relatives (5% to 100% for a child; 0% to 80% for a sibling). The most common recurrence risk estimate was 50%. Respondents expressed the most concern for their children developing AUD. Concern level did not influence their perceived benefit of AUD genetic counseling, yet those who felt genetics to be an important cause of AUD were more likely to perceive a benefit from AUD genetic counseling ($\rho = .19$, $df = 120$, $p = .019$). Participants reported many areas of their lives to be affected by AUD. In general participants recognized the multifactorial nature of AUD but seemed to lack a clear understanding of recurrence risk. **Discussion:** Based on the responses of the participants in the current study, genetic counseling could be beneficial to those with or at risk for AUD. Genetic counseling for this patient population could help clarify recurrence risk misconceptions and facilitate a

clearer understanding of the environmental and biological factors that influence the development of AU. Further studies are warranted to evaluate the outcomes of AUD genetic counseling with respect to patient understanding, lifestyle modifications and psychological adaptation. Genetic counselors should be encouraged to reach out to this population.

Table of Contents

List of Tables

List of Figures

List of Abbreviations

AA.....Alcoholics Anonymous

ADHD.....Attention Deficit Hyperactivity Disorder

AUD.....Alcohol Use Disorder

GC.....Genetic Counseling

NSGC.....National Society of Genetic Counselors

PTSD.....Post Traumatic Stress Disorder

OCD.....Obsessive Compulsive Disorder

Chapter 1. Background

1.1 Alcohol Addiction

Alcohol is a psychoactive drug that has been consumed for millennia (Mandelbaum, 1965). Renowned scholars as early as Aristotle have observed and made note of alcohol drinking habits running through families (Warner & Rosett, 1975). While alcohol is a drug that has been used throughout societies for relaxation and enjoyment, it is also highly addictive. Alcohol use disorders (AUDs) are defined in the Diagnostic and Statistical Manual of Mental Disorders as "a problem pattern of alcohol use leading to clinically significant impairment or distress" (American Psychiatric Association, 2013). Both alcohol abuse and alcohol dependence fall under this diagnosis. Until 2013 the American Psychiatric Association categorized alcohol abuse and dependence as two separate diagnoses. Abuse is less severe and does not involve the addiction or compulsion to use alcohol, nor does it result in withdrawal symptoms. Alcohol dependence, however, involves the compulsion to use, and abstaining from use causes withdrawal symptoms in the affected individual (American Psychiatric Association, 2000). The etiology of AUD is complex and multifactorial, with genetics and environment of the individual both contributing significantly to the development of the disease.

Individuals begin consuming alcohol for a variety of reasons including cultural acceptance, curiosity, societal pressures, or purely for enjoyment. However, alcohol consumption when not monitored by the individual has the possibility to develop into addiction and/or dependence and ultimately into an AUD.

Alcohol intake causes a variety of physiological changes within the body including an increased release of serotonin that can result in intoxication (National Institute of Alcohol Abuse and Alcoholism, 2010). When alcohol is consumed in overabundance, we can see adverse affects on the body including but not limited to tachycardia, nausea, headache, tremors, and fatigue (Trevisan, Boutros, Petrakis, & Krystal, 1998). Chronic overconsumption of alcohol can result in reduced brain mass, cardiomyopathy, heart arrhythmias, cirrhosis, and chronic pancreatitis, among many other medical and psychological problems (National Institute of Alcohol Abuse and Alcoholism, 2010). After an extended period of overconsumption of alcohol, some individuals can go through alcohol withdrawal. Symptoms of alcohol withdrawal come in stages, with the first stage manifesting similarly to a hangover, but escalating within one to five days to possible hallucinations and seizures or other neurological concerns (Manasco, Chang, Larriviere, Hamm, & Glass, 2012).

AUDs wreak havoc on an individual physically, mentally, and emotionally. However, the effects of an AUD are not isolated to the affected individual. A study by Bouchery and colleagues (2011) uncovered that excessive drinking cost the U.S. $223.5 billion in 2006. The majority of this cost came from lost productivity with healthcare and criminal justice costs also contributing. Additionally, alcohol overconsumption is responsible for 79,000 deaths annually in the United States (Bouchery, Harwood, Sacks, Simon, & Brewer, 2011).

Reich, Earls, & Powell (1988) compared the home environments of the children in families with and without alcohol addiction and found overall more adverse environments when one or more parents are addicted to alcohol. Specifically,

significantly more abuse, marital conflict, and parent-child conflict was found. Also, children were less likely to view their parents as positive role models if they were in a family with alcohol addiction. Children of people with alcohol addiction are also found to be at higher risk for developing many psychiatric conditions such as attention deficit hyperactivity disorder, conflict disorders, oppositional defiant disorder, and major depressive disorder (Hill et al., 2009; Reich, Earls, Frankel, & Shayka, 1993).

There is no cure for alcohol addiction. There has been a wide range of treatments used with variable rates of success. The history of alcohol addiction treatment has been long and varied. It was not until the end of the late 18th and early 19th centuries that alcohol addiction came to be seen as a habit or disease as opposed to a sin. This paradigm shift away from viewing alcohol addiction and "drunkenness" as a sin opened the door for conversations about medical treatments for the addiction (Edwards, Marshall, & Cook, 2003). By the mid-19th century temperance societies were beginning to form in larger cities. These societies promoted a large self-help aspect to regaining sobriety and their ideals are thought to have heavily influenced the Alcoholics Anonymous program. However, the temperance societies quickly were abandoned in favor of institutionalized treatments for alcohol addiction, in which addicted individuals were admitted for anything ranging from five years to life in an environment heavily dependent on their social class but always without access to alcohol (Edwards et al., 2003).

Alcoholics Anonymous (AA) began in 1935 by two men in Ohio. This treatment program focuses on character and spiritual development and operates on two main premises: one, that the key to recovery is one turning over their will and life to a higher power, and two, that recovery requires the recognition of powerlessness in regards to

alcohol. The AA approach has individuals work through 12 steps, or levels, to focus on taking personal responsibility for their sobriety (Huebner & Kantor, 2011). AA has been met with wide amounts of success and is still a very commonly sought out program for those addicted to alcohol. Since the introduction of AA into society, many other "step" programs have been utilized to aim to treat alcohol addiction. After the utility of these group programs was recognized, many institutions paired them with residential programs and family involvement to attempt to maximize success (Edwards et al., 2003).At their core, however, all behavioral therapeutic approaches to treat alcohol addition utilize general behavioral principles such as reward and punishment alongside concepts from both social learning theory and cognitive behavioral therapy.

Physical treatments have been just as common throughout history as behavioral approaches. In centuries past, electroconvulsive therapy, brain operations, cannabis, and carbon dioxide injections have been used in attempts to cure alcohol addiction (Edwards et al., 2003).More recently medications have been utilized to help facilitate the recovery of those with AUDs. Disulfiram, known under the drug name Antabuse, was introduced in the 1950s to clinical practice. It, along with newer anti-craving medications acamprosate and naltrexone, cause extremely toxic physical reactions when mixed with alcohol resulting in alcohol consumption to becoming an adverse activity (Center for Substance Abuse Treatment, 2009; Edwards et al., 2003).

Large-scale studies looking at the effectiveness of different treatments for AUDs have shown that no single treatment strategy or medication is completely effective across all individuals. A treatment that may work for one individual is not guaranteed to work for another (Huebner & Kantor, 2011).This fact leads to the complexity of treating AUDs.

Not only must an individual be motivated to seek and commit to a treatment plan, but additionally the team of healthcare providers responsible for guiding the treatment must identify the treatment type(s) that will be most effective for that individual (National Institute of Alcohol Abuse and Alcoholism, 2014).

1.2 Genetics of Alcohol Addiction

AUD can be seen aggregating in families. It is a highly heritable condition, with 40-60% of the likelihood to develop AUD due to genetic factors (Agrawal & Lynskey, 2008; Kendler K., Heath A., Neale M., Kessler R., 1992). It is estimated that there is a fourfold increase in risk for children of individuals with AUDs over the general population to develop an alcohol addiction (Russell, 1990). Using the current population frequency in the United States, children of those with AUDs have approximately a 28% chance to develop the same condition. Given the increased frequency of relatives of individuals with AUDs to develop the same addiction, research into the genetic causes of alcohol addiction has been a growing field.

Recent research has attempted to find an association of multiple genes with the occurrence of AUD and has found several copy number variants (CNVs) and single nucleotide polymorphisms (SNPs) that associate with the condition (Iyer-Eimerbrink & Nurnberger, 2014). Possibly the most well-studied genes pertaining to risk for AUD are the *ADH* and *ALDH* gene families. Both of these gene families are heavily involved in alcohol metabolism, with genes in the *ADH* family encoding the enzyme alcohol dehydrogenase which catalyzes the rate-limiting step for ethanol metabolism and genes within the *ALDH* family coding for acetaldehyde dehydrogenase, the next enzyme in the metabolic pathway (Edenberg, 2007). Some of these genes within the *ADH* and *ALDH*

families have different alleles causing variation in the rate at which the enzymes metabolize ethanol or acetaldehyde and can thus affect a person's drinking amount and therefore habit (Edenberg, 2007).

Certain variants within the *ALDH2* gene, such as the ALDH2*2 allele, have been associated with increased sensitivity to alcohol and adverse reactions to alcohol consumption. These adverse side-effects of consumption include flushing and nausea as a result of excessive acetaldehyde accumulation due to decreased *ALDH2* activity caused by the genetic variant (Stamatoyannopoulos, Chen, & Fukui, 1975; Wolff, 1972). The variants responsible for these adverse reactions are more common in individuals of East Asian descent and are thought to lead to decreased alcohol consumption in individuals and an overall lower rate of alcoholism within the race (Goedde, Harada, & Agarwal, 1979).

A variation within the *ADH1B* gene, the ADH1B*2 allele, has also been associated with lower frequency of AUDs (Muramatsu et al., 1995). Studies have not correlated this polymorphism as strongly with negative side effects of drinking like those experienced by individuals with the ALDH2*2 allele. However, many overall studies have found that individuals with the ADH1B*2 allele consume less alcohol, which could contribute to the lower frequency of AUDs within these individuals (Carr et al., 2002; Muramatsu et al., 1995; Shea, Wall, Carr, & Li, 2001)

Many other genes appear to be involved with the development of AUDs, however, none as strongly as the genes discussed above. Many of these lesser-implicated genes play a role in neurotransmitter response and/or the reward system within the brain. Recently ten polymorphisms within the CPNE5 gene, which encodes for a calcium-

dependent lipid-binding protein expressed during neural development in mice, were associated with alcohol dependency within the Caucasian population. However, this recent study has yet to be replicated (K. S. Wang, Zuo, Pan, Xie, & Luo, 2015). Additionally, variants within the muscarinic acetylcholine receptor gene *CHRM2* have been associated with alcohol dependency along with major depressive disorder (Wang & et al, 2004). There have been well over 50 different genes located within various neurotransmitter, drug metabolism, and other pathways that have been associated with alcohol dependency and/or addiction (Iyer-Eimerbrink & Nurnberger, 2014). Many of these genes, however, lack strong associations and their studies have either not been replicated or have had mixed results upon replication.

Currently there are a limited number of SNPs that are strongly associated with the development of or protection from AUDs, all of which are within the alcohol-metabolizing enzyme genes. Many genome-wide association studies have looked for other SNPs associated with AUDs. However, the only consistent findings have been in those previously described genes (Hart & Kranzler, 2015). The present thought is that many different SNPs have small individual effects on developing the condition, but cumulatively create a larger influence to develop the condition. Research is ongoing to further uncover genetic contributions to the development of AUDs.

Learning more about the genetic components of AUDs opens the door for more targeted treatments, particularly in the realm of pharmacogenetics. Currently there are three medications approved by the FDA to treat AUD. The oldest medication Disulfiram, causes unpleasant physical responses (such as nausea, vomiting, dizziness, etc.) when taken alongside alcohol. It works by inhibiting the metabolism of acetaldehyde, creating

excessive buildup of the toxin similar to individuals carrying the ALDH2*2 allele (Chen, Ferreira, Gross, & Mochly-Rosen, 2014; Wright & Moore, 1990). Naltrexone, an opioid receptor antagonist, blocks μ-opioid receptors which in turn seems to reduce the reinforcing effects of alcohol, alcohol cravings, and feelings of intoxication after alcohol consumption (Rösner et al., 2010; Williams, 2005). Acamprosate, which was approved solely on its efficacy, is thought to alter NMDA receptor, a glutamate receptor, composition and possibly indirectly effects $GABA_A$ receptors therefore reducing relapse risk. (Kalk & Lingford-Hughes, 2014; Rösner, Hackl-Herrwerth, Leucht, Lehert, et al., 2010). While these medications all seem to have potential to be effective in reducing AUDs, studies have shown that as many as 70% of individuals treated with either acamprosate or naltrexone do not respond to the medication (Jones, Comer, & Kranzler, 2015). Understanding why certain individuals do not react to these medications from a genetic standpoint will help targeted treatment and ensure that individuals with AUD are receiving treatment that has the potential to be effective.

Genetic variation within the alcohol metabolism genes has been shown to have protective effects against developing AUD; however, there is little evidence showing that these same variations affect medication response (Jones et al., 2015). SNPs within many genes involved with neurotransmitter responses and some within opioidergic genes have appeared to be possible candidates for moderating drug response from initial findings in recent studies. Despite these numerous promising initial studies, none of the candidate SNPs have generated replicable findings across studies (Jones et al., 2015). Better understanding of the genetic contributions to AUDs will enable researchers to identify new drug therapies to target specific pathways and to identify genotypes that would be

the best candidates for new and current treatments. This will allow physicians to incorporate personalized treatment plans into the care of their patients with AUD leading to increased response rates, decreased relapse rates, and an increase in the number of sober days for their patients.

1.3 Psychiatric Genetic Counseling and the Genetic Counseling Profession

The National Society of Genetic Counselors defines genetic counseling as the process of assisting people to comprehend and adapt to the medical, psychological, and familial implications of genetic contributions to disease (Resta et al., 2006). Genetic counseling includes interpretation of medical and family histories to provide risk information; education about disease management, prevention, inheritance, and testing, providing resources pertaining to the family's condition; and counseling to assist in informed decision-making and adaptation. Despite decades of research supporting psychiatric conditions as heritable and the idea of psychiatric genetic counseling being in existence since the 1970's, only recently has there been research looking into the potential for utilizing genetic counseling as a component of care for the psychiatric patient (Tsuang, 1978).

Psychiatric genetic counseling is not just addressing risk and recurrence within the family. It can also be used for individuals to alleviate guilt, shame, and stigma through addressing etiology of the condition, along with helping people to better understand the cause of psychiatric disorders and how genes and environment work together to contribute to these conditions. Additionally, psychiatric genetic counseling can facilitate a discussion around strategies to protect one's mental health and provide

both support and counseling around issues that arise due to guilt, shame, and stigma. (Austin & Honer, 2008; Costain, Esplen, Toner, Hodgkinson, & Bassett, 2014)

Research has shown that individuals both diagnosed with and at risk for psychiatric conditions desire genetic counseling (Costain et al., 2014; Lyus, 2007; Quaid, Aschen, Smiley, & Nurnberger, 2001). Inglis and colleagues studied the effects of genetic counseling for psychiatric conditions and found an overwhelmingly positive response. They found that genetic counseling for psychiatric conditions significantly increased patient empowerment and that individuals affected with a psychiatric condition scored significantly higher post-session on the Illness Management Self Efficacy Scale post-session (Inglis, Koehn, McGillivray, Stewart, & Austin, 2014) which is an instrument used to measure an individual's confidence in managing their personal illness. Similarly, Costain and colleagues focused on genetic counseling outcomes among individuals with schizophrenia and their family members and found similar results. They found that all individuals benefited from genetic counseling and showed increased knowledge and decreased sense of stigma, and that the majority of participants endorsed genetic counseling for the population (Costain et al., 2014).

Genetic counseling has just recently begun to be applied to psychiatric conditions. However, healthcare providers well established in mental healthcare are still on the fence about involving genetic counselors in their field. In a recent study the majority (82 of 113) psychologists indicated that it would be beneficial for genetic counselors to be included in psychiatric care (Thompson, Hamilton, & Hippman, 2015). However, an alternative study performing qualitative interviews with ten psychiatrists revealed that the majority of these psychiatrists seemed to lack the acceptance of adding additional

clinicians in the field of psychiatry and were skeptical about psychiatric genetic counseling and how it was different from the services they are already providing (White, Youngbloom, Austin, & Thompson, 2015).

Genetic counseling has been utilized as a service in other areas of healthcare for much longer than in the psychiatric sector and outcomes of genetic counseling have been observed in other fields of healthcare for many decades. Very few early studies specifically aimed to investigate the outcome of genetic counseling; however, many studies within the prenatal and preconception field recognized the patient benefit in the form of increased understanding and ability to make better informed decisions (Lubs & Travers, 1981; Sahney, Weiss, & Levin, 1982; Tercyak, Johnson, Roberts, & Cruz, 2001). Additionally, pediatric genetic counseling has been shown to bring significant increases to patient treatment adherence (Rutherford, Zhang, Atzinger, Ruschman, & Myers, 2014), reduce anxiety, and to encourage open dialogue within families about pediatric genetic conditions. (Kladny, Williams, Gupta, Gettig, & Krishnamurti, 2011).

Studies assessing outcomes of genetic counseling have been conducted within the field of cancer genetics as well, and the findings remain consistent with the conclusions of studies performed in other genetic counseling settings. These studies have looked at patient knowledge and illness-related anxiety comparing individuals receiving genetic counseling and those who did not. Within these studies, genetic counseling has been shown to significantly improve patient knowledge and understanding of their condition and risks, lower their illness-related anxiety, and increase perceived personal control around the condition (Braithwaite, Emery, Walter, Prevost, & Sutton, 2006; Meiser et al.,

2001; Mikkelsen, Sunde, Johansen, & Johnsen, 2009; Pieterse, Ausems, Van Dulmen, Beemer, & Bensing, 2005).

1.4 Rationale

Previous research has shown significant patient benefit from genetic counseling for a variety of conditions. Additional research has shown that there is desire for and benefit from genetic counseling in individuals affected by psychiatric conditions. However, there are no studies that have looked at the perceived benefit of genetic counseling among those affected by and at risk for alcohol addiction, nor how this population perceives recurrence risks of the condition. Nor have their been studies investigating how the presence of AUD in their family/personal history has affected at-risk or affected individuals' lives or how concerned they are about themselves and their relatives developing the condition. It is very possible that this patient population could benefit from genetic counseling regarding AUD. This study aimed to investigate the understanding of recurrence risk and etiology of AUD among affected and at-risk individuals while investigating how their personal/family history of AUD has impacted their lives and their level of concern around developing the condition. Additionally, this study examined the participants' experience with genetic counseling and their perceived benefit of AUD genetic counseling to further investigate the possible need for genetic counseling within this population.

Chapter 2. Genetic Counseling for Alcohol Use Disorder: Assessment of Need in Affected and At-Risk Populations[1]

2.1 Abstract

Introduction: Alcohol use disorder (AUD) is highly heritable, yet there has been no investigation regarding the possible benefits of genetic counseling for AUD. This study assessed the beliefs that individuals with and at risk for AUD have regarding recurrence risk and etiology of AUD, how the presence of the condition in themselves or their family history has affected their lifestyle decisions, and potential benefit from AUD genetic counseling. **Methods:** An online questionnaire was distributed through social media to support groups for AUD inviting adults 18 years and older with a personal or family history of AUD. **Results:** Of the 122 individuals that completed the online questionnaire, 60% of participants perceived a potential benefit of AUD genetic counseling. Participants reported a wide range of estimated recurrence risks for first-degree relatives (5% to 100% for a child; 0% to 80% for a sibling). The most common recurrence risk estimate was 50%. Respondents expressed the most concern for their children developing AUD. Concern level did not influence their perceived benefit of AUD genetic counseling, yet those who felt genetics to be an important cause of AUD were more likely to perceive a benefit from AUD genetic counseling ($\rho = .19$, $df = 120$, $p = .019$). Participants reported many areas of their lives to be affected by AUD. In general participants recognized the multifactorial nature of AUD but seemed to lack a clear

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understanding of recurrence risk. **Discussion:** Based on the responses of the participants in the current study, genetic counseling could be beneficial to those with or at risk for AUD. Genetic counseling for this patient population could help clarify recurrence risk misconceptions and facilitate a clearer understanding of the environmental, biological, and genetic factors that influence the development of AUD. Further studies are warranted to evaluate the outcomes of AUD genetic counseling with respect to patient understanding, lifestyle modifications and psychological adaptation. Genetic counselors should be encouraged to reach out to this population.

2.2 Introduction

Alcohol use disorders (AUDs), including alcohol addiction and dependence, are highly prevalent psychiatric conditions with an estimated 7% of adults affected within the United States (National Institute of Alcohol Abuse and Alcoholism, 2015). AUDs, like other mental health conditions, can be seen aggregating in families. It is a highly heritable condition with 40-60% of the likelihood to develop AUD due to genetic factors (Agrawal & Lynskey, 2008; Kendler K., Heath A., Neale M., Kessler R., 1992). It is estimated that there is a fourfold increase in risk for children of individuals with AUDs over the general population to develop an AUD during their lifetime (Russell, 1990).

Treatments and remedies for AUDs have been in existence for as long as alcohol addiction itself. From the antiquated institutionalization of addicts to the more modern 12-step programs and medications, it has become clear over time that no single treatment strategy or medication is completely effective across all individuals. A treatment that may work for one individual is not guaranteed to work for another (Huebner & Kantor, 2011). It seems that the best treatment plan for AUDs involves a motivated patient and a

combination of biological and psychological therapies put together by a team of healthcare providers individually for that patient (Anton et al., 2006).

At its core, genetic counseling is the process of assisting people to comprehend and adapt to the medical, psychological, and familial implications of genetic contributions to disease. Genetic counseling services have been utilized in the prenatal and pediatric settings for decades, and more recently has moved into helping families understand the impacts of familial cancers. Utilizing genetic counseling for psychiatric conditions has been discussed for decades (Tsuang, 1978), yet only recently has its practice been studied. Genetic counseling for psychiatric conditions can help individuals to alleviate guilt, shame, and stigma through addressing etiology, along with helping people to better understand how genes and environment work together to contribute to these conditions. Additionally, psychiatric genetic counseling can facilitate a discussion around strategies to protect one's mental health and provide both support and counseling around issues that arise due to guilt, shame, and stigma (Austin & Honer, 2008; Costain et al., 2014).

Previous studies have shown that genetic counseling has resulted in an increase in patient understanding and knowledge regarding conditions and risks, an increase in the ability to make better-informed decisions, increased patient treatment adherence, increased perceived personal control, and a decrease in anxiety (Braithwaite et al., 2006; Kladny et al., 2011; Lubs & Travers, 1981; Meiser et al., 2001; Mikkelsen et al., 2009; Pieterse et al., 2005; Rutherford et al., 2014; Sahney et al., 1982; Tercyak et al., 2001). Recent studies investigating the effects of genetic counseling for psychiatric conditions have found similar results: significantly increased patient empowerment, increased

knowledge, a decreased sense of stigma, and an overall benefit across participants (Costain et al., 2014; Inglis et al., 2014).

To the best of our knowledge, published literature has not yet investigated the possible desire for or the utility of genetic counseling for individuals coping with or at risk for alcohol addiction. An understanding of how these populations perceive risk and etiology of AUDs and how AUDs in a family history can affect lifestyle decisions may be helpful for genetic counselors and other healthcare providers who serve these populations to address these areas of concern with their patients.

2.3 Materials and Methods

2.3.1 Participants

Individuals who are affected by or at risk for AUD were invited to participate in the study. The primary author defined 'at risk' as an individual having one or more first-degree relatives with an AUD. Additionally, spouses of individuals with an AUD were eligible to participate in the study. Only adults aged 18 or older were eligible to participate. Individuals were invited using the social media websites Reddit and Facebook. A short message regarding the study and inviting interested individuals to participate in the study (Appendix A) was posted in forums specifically focusing on alcohol addiction recovery and family support. Additionally, if the group was private requiring membership, the principle investigator reached out to a group leader with a message (Appendix B), requesting that the leader share the study information with the groups' members.

2.3.2 Survey Tool

Data was collected from September 2015 through January 2016 using an online questionnaire with a variable number of questions depending on participant responses. The questionnaire (Appendix C) was created and hosted using www.surveymonkey.com. The questionnaire included demographic questions and quantitative questions investigating the participants' experience with alcohol addiction, their family history, their risk perception regarding inheritance of AUDs, and their beliefs about the causes of AUDs. Participants were asked about their level of concern regarding family members and were given the opportunity to explain their personal reasons with an open-ended text box. In addition, participants' experiences with genetic counseling were explored and a brief video explaining addiction genetic counseling was viewed before inquiring about their perceived benefit of genetic counseling for AUDs. A transcript of the video can be found in Appendix D.

The questionnaire utilized skip logic to ask participants details to their answers for certain questions and to avoid participants being required to elaborate on certain areas that were not applicable. The questionnaire had a maximum of 43 questions.

2.3.3 Data Analysis

Descriptive statistics were primarily utilized for this study, as this study aimed to gauge participants' understanding of the familial risk and causes of AUDs as well as their feelings of the possible benefit of genetic counseling. Microsoft Excel was utilized to store and organize the data as well as to perform the descriptive statistical analyses. SPSS software, Version 23, was additionally used for back-up data storage and to run the

statistical analyses, including Spearman's Rho for correlation analyses and multiple independent sample t-tests.

2.4 Results

2.4.1 Demographics

A total of 155 individuals began the online survey, which was distributed via support groups on social media websites. Uncompleted surveys were discarded from data analysis leaving 122 total responses. Participants were primarily college educated and Caucasian, and there was a near even balance of males and females (Table 2.1).

2.4.2 Benefit of AUD Genetic Counseling

After viewing a description of genetic counseling for addictions, 73 individuals (60%) felt that they would experience some amount of benefit from genetic counseling (Table 2.2). The majority of respondents in this category (n = 40; 33%) reported they would somewhat benefit from addictions genetic counseling, while 28 (23%) felt they would experience a moderate benefit and five individuals (4%) felt they would extremely benefit from addiction genetic counseling. When comparing demographics to perceived benefit of AUD genetic counseling, the majority of the demographic groups seemed to mimic the overall finding of AUD benefit from the entire study population (Table 2.2). Demographic categories, personal or family history of other mental health disorders, and a personal as opposed to a family history of AUD did not appear to influence perceived benefit of AUD genetic counseling.

2.4.3 Perceptions of AUD Causes

The large majority of respondents indicated more than one cause to be an important contributor the development of AUD. Nearly three quarters of respondents

Table 2.1: Demographics and Perceived Benefit of AUD Genetic Counseling

			Perceived benefit of AUD GC			
			Yes		No	
	Percentage (%)	Frequency (N = 122)	Percentage (%)	Frequency (n = 73)	Percentage (%)	Frequency (n = 49)
Gender						
Male	48%	59	48%	35	49%	24
Female	52%	63	52%	38	51%	25
Highest Completed Education*						
High School	17%	21	16%	12	18%	9
Associate's Degree	11%	13	11%	8	10%	5
Bachelor's Degree	47%	57	45%	33	49%	24
Postsecondary School	25%	31	27%	20	22%	11
Race*						
Caucasian	92%	112	88%	64	98%	48
African American	1%	1	0%	0	2%	1
Asian	2%	2	3%	2	0%	0
Hispanic	2%	2	3%	2	0%	0
Native American	1%	1	1%	1	0%	0
Other	3%	4	5%	4	0%	0
AUD Diagnosis						
Yes	38%	47	38%	28	39%	19
Active	4%	5	4%	3	4%	2
Attempting Recovery	12%	15	12%	9	12%	6
Recovered	23%	28	22%	16	24%	12
No	62%	75	62%	45	61%	30

* Percentage totals may not equal 100% due to rounding

Table 2.1: Demographics and Perceived Benefit of AUD Genetic Counseling (cont.)

			Perceived benefit of AUD GC			
			Yes		No	
	Percentage (%)	Frequency (N = 122)	Percentage (%)	Frequency (n = 73)	Percentage (%)	Frequency (n = 49)
Family History of AUD						
Yes	81%	99	82%	96	80%	3
Fist Degree Relative	61%	60	51%	37	47%	23
Second or Third Degree Relative	83%	82	73%	53	59%	29
Spouse	6%	6	5%	5	4%	1
No	19%	23	18%	21	20%	2
Mental Health History						
Personal History**	45%	55	41%	52	51%	3
Family History in Relatives(s) with AUD**	43%	52	45%	50	39%	2

*Percentage totals may not equal 100% due to rounding
**Reported mental health diagnoses include depression, anxiety, schizophrenia, bipolar disorder, PTSD, panic disorder, OCD, ADHD, and autism spectrum disorders.

Table 2.2: Perceived Benefit of AUD Genetic Counseling

	Frequency (N = 122)	Percentage (%)
Extremely	5	4%
Moderately	28	23%
Somewhat	40	33%
No, I do not think I would benefit	49	40%

(74%; n = 90) thought that genetics/family history was an important contributor to the development of AUD (Figure 2.1). Seventy-four individuals (61%) thought that traumatic life experiences and how your body reacts to alcohol were important contributors. Three individuals marked traumatic life experience and/or lack of self-control as the only cause of AUD, while four individuals said the only cause was how your body reacts to alcohol.

When comparing the perceived benefit of AUD genetic counseling against perceived attributed causes of AUD, there were some trends observed.
Mean attribution scores, representing how strongly the participants felt each cause contributed to the development of AUD, were calculated for each etiology utilizing responses to the Likert scale given in the questionnaire (Table 2.3). Participants overall reported that genetics/family history played a large role in the development of AUD score while participants felt that spending time around others with an AUD was a much smaller contributing factor.

Utilizing a Spearman's Rho correlation, participants who attributed the development of AUD highly to genetics/family history were more likely to report a perceived benefit of genetic counseling ($\rho = .19$, $df = 120$, $p = .019$) than those who did not see it as a contributing factor. In contrast, participants who attributed the

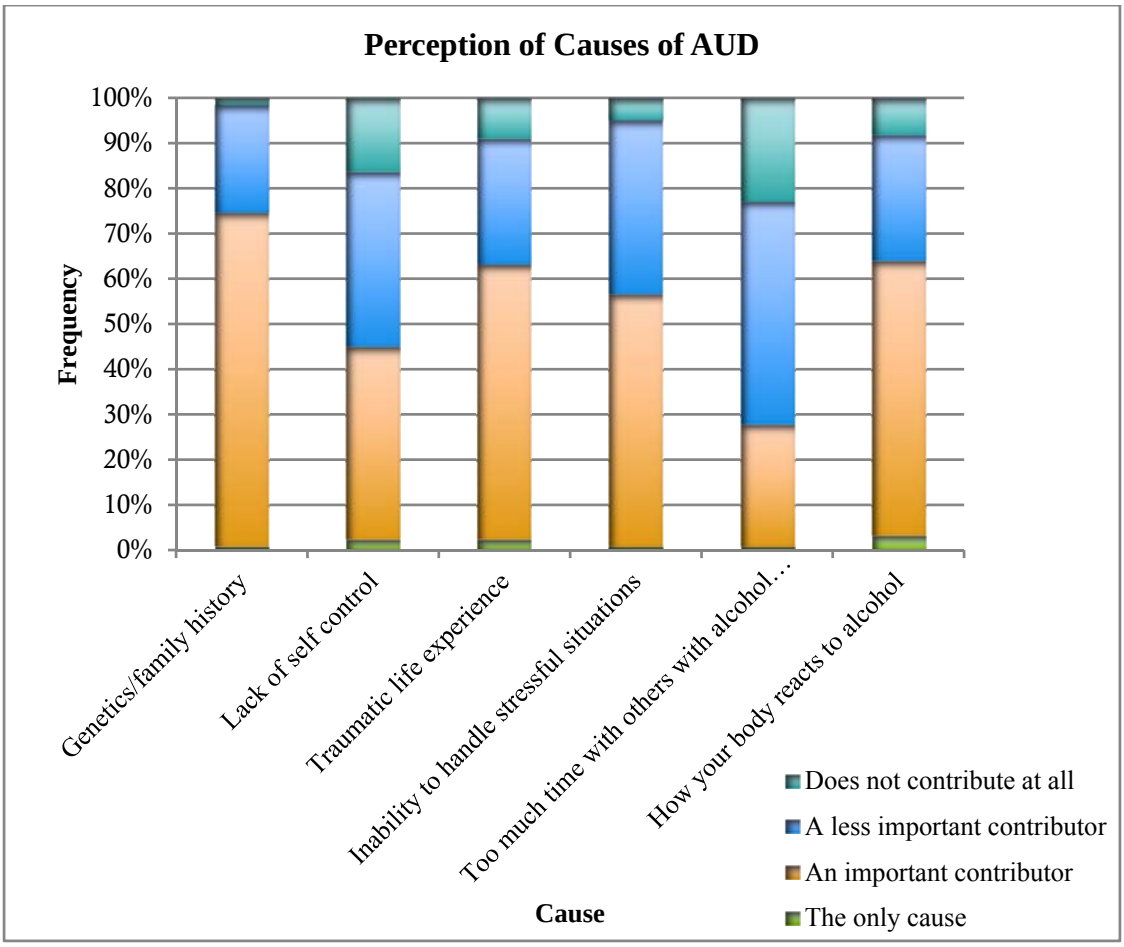

Figure 2.1: Perceptions of AUD Cause

Table 2.3 Mean Attribution Score of AUD Cause and Perceived Benefit of AUD Genetic Counseling*

		Perceived Benefit from AUD GC	
Cause	Total (N = 122)	Yes (n = 73)	No (n = 49)
Genetics/Family History	2.74	2.81	2.63
Lack of self control	2.31	2.22	2.45
Traumatic Life Experience	2.57	2.52	2.63
Inability to Handle Stressful Situations	2.52	2.44	2.65
Spending Too Much Time Around Others with AUD	2.06	2.05	2.06
How Your Body Reacts to Alcohol	2.59	2.55	2.60

*Mean attribution scores calculated from four-point scale responses. 1 = does not contribute at all, 2 = a less important contributor, 3 = an important contributor, 4 = the only cause

development of AUD more to an individual's lack of self-control were more likely to report no perceived benefit of AUD genetic counseling (ρ = -.15, *df* = 120, p = .049). No significant correlations were found between perceived benefit of AUD genetic counseling and any other perceived causes.

2.4.4 Recurrence Risk Perceptions

Respondents had largely varying responses when asked about the recurrence likelihood of AUD for a child of someone with the condition. Out of the 122 individuals, two reported that they did not know what the recurrence risk was and one individual gave a range of likelihood, for which the average was calculated and used for that response. Answers ranged from 5% to 100% (Figure 2.2), with an average answer of 40%. The most frequent answer was 50% (24%; *n* = 28). Thirteen individuals (11%) thought the chance of someone with AUD having a child who developed AUD later in life was 70% or higher.

When asked the same question but in regard to the likelihood of a sibling of an affected individual developing AUD, the answers were also quite varied. Out of the 122 individuals, two reported that they did not know and one individual gave a range of likelihood, for which the average was calculated and used for that response. The responses ranged from 0% to 80%, with an average of 29% (Figure 2.2). The most frequent answer was 50 % (15%; *n* = 8). Six individuals (5%) thought the chance of someone with AUD having a sibling with an AUD was 70% or higher. The estimates of mean recurrence risk were then further divided into males and females, as well as their perception of the benefit of AUD genetic counseling. These estimates can be seen in Table 2.4.

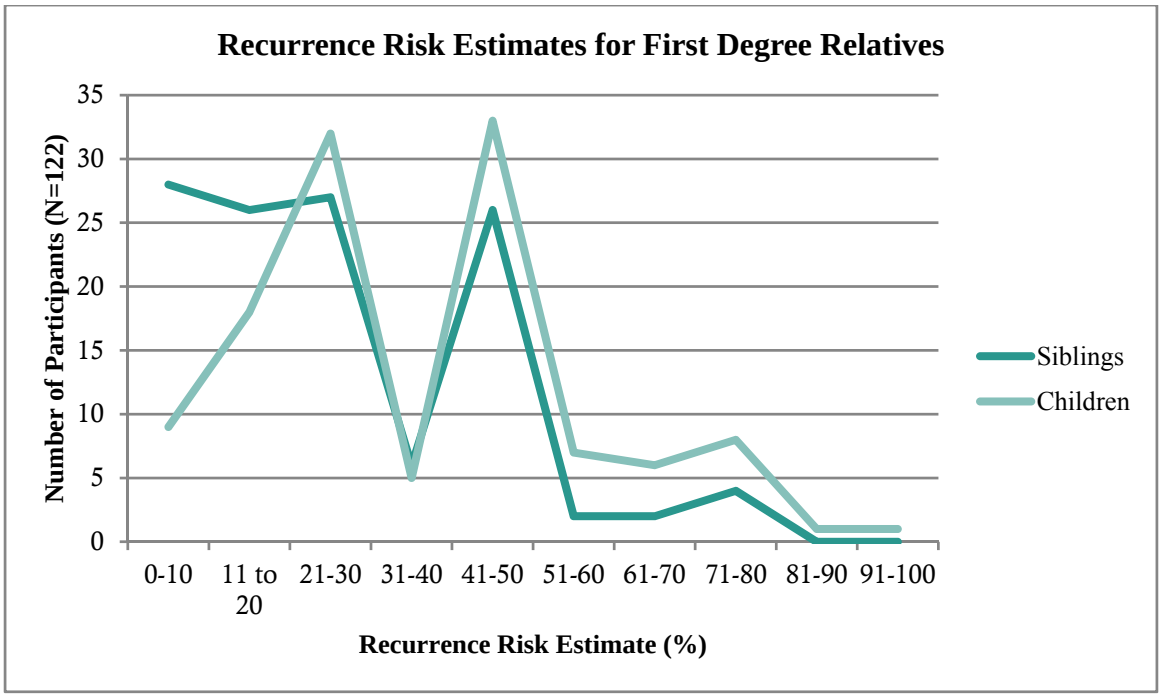

Figure 2.2: Estimated Recurrence Risk for First Degree Relative

Table 2.4: Mean Recurrence Risk Estimates for Males and Females

	Perceived Benefit from AUD GC	
Recurrence Risk Estimate Mean For Children	**Yes**	**No**
Total	40%	39%
Male	39%	28%
Female	41%	47%
Recurrence Risk Estimate Mean For Sibling	**Yes**	**No**
All	30%	25%
Male	32%	18%
Female	28%	30%

2.4.5 Concern around AUD

Nine (12%) respondents without AUD (*n* = 77) were extremely concerned about developing the condition, whereas thirty-one (41%) respondents without AUD were not concerned at all. Of all 122 participants, 14% (*n* = 17) reported that they were extremely concerned about their children developing AUD and an additional 25% (*n* =

30) reported that they were moderately concerned. Thirty-five (29%) were not concerned at all about their children developing the condition. When asked about level of concern regarding siblings developing AUD, 4% (n = 5) of participants reported being extremely concerned, while 58% (n = 71) were not concerned at all (Table 2.5).

Table 2.5: Participants' Concern for Developing AUD

	Frequency	Percentage (%)
Concern for Self	(n=77)	
Extremely	9	12%
Moderately	16	21%
Somewhat	21	27%
Not concerned at all	31	40%
Concern for Children	(N=122)	
Extremely	17	14%
Moderately	30	24%
Somewhat	40	33%
Not concerned at all	35	29%
Concern for Siblings	(N=122)	
Extremely	5	4%
Moderately	21	17%
Somewhat	25	21%
Not concerned at all	71	58%

Mean concern scores were calculated by averaging Likert scale responses to questions regarding concern. Overall mean concern scores for the cohort were highest in reference to participants' children and lowest in reference to their siblings (Table 2.6). However, when independent sample t-tests were performed to see if there was a relationship between participants' concern for their children (t = 1.57, df = 120, p = ns) or their siblings (t = -.63, df = 120, p = ns) and their perceived benefit of genetic counseling there were no significant differences relative to their perceived benefit of genetic counseling. Mean scores for self were calculated from the 75 individuals who reported they did not have a personal diagnosis of AUD. The mean concern scores for

the entire cohort as well as those that did and did not perceive a benefit to AUD genetic counseling can be seen in Table 2.6.

Table 2.6: Mean Concern Scores for Developing AUD and Perceived Benefit of AUD Genetic Counseling*

		Perceived Benefit from AUD GC	
	Total (N = 122)	Yes (n = 73)	No (n = 49)
Concern for Self	2.04	2.33	1.61
Concern for Children	2.24	2.36	2.06
Concern for Siblings	1.67	1.63	1.73

*Mean concern scores calculated from four-point scale. 1 = Not concerned at all, 2 = somewhat concerned, 3 = moderately concerned, 4 = extremely concerned

2.4.6 Lifestyle Effects of AUD

The majority of individuals (61%; n = 75) reported that the presence of AUD in their personal or family history has not affected their decision to have children. An additional seven individuals (6%) reported that they were unaware of AUD in the family when they were family planning. Five individuals (4%) report that their decision has affected their family planning by either choosing to adopt children or to not have children. The remaining 34 respondents (28%) said that they are not sure if their personal/family history of AUD will influence their family planning, as they are not ready for children yet.

The most commonly affected area of participants' lives was their drinking habits. Fifty-two (43%) individuals said that their family/personal history of AUD has extremely influenced their drinking habits, while another 35 (29%) said that their drinking habits were moderately influenced. The next most influenced area was how the respondent copes with stress. Forty-two individuals (34%) reported their coping mechanisms have been extremely influenced, while an additional 37 (30%) reported them being moderately influenced. The least variation in responses was seen in regard

to if their personal or family history of AUD influenced with whom the participants spend their time. This factor was extremely affected in 21% of respondents, moderately affected in 26% of respondents, somewhat affected in 25% of respondents, and not affected in 28% of respondents. Leisure activities and diet/exercise shared similar numbers of individuals reporting an extreme influence with 30% (n = 37) and 30% (n = 36) respectively. Religious and social views were the least likely to be influenced, with only 9% (n = 11) and 12% (n = 15) of respondents reporting AUD extremely affecting these two areas of their lives (Figure 2.3).

2.4.7 Personal Experience with Genetic Counseling

Less than half (41%; n = 50) of respondents reported they had heard of genetic counseling before the survey. Of those 50 individuals, 82% (n = 41) of them reported that they knew what genetic counseling was, yet only two individuals had experienced genetic counseling. Both individuals reported that they had prenatal genetic counseling during their pregnancies and in neither session was the individual's personal or family history of AUD addressed during the genetic counseling session.

2.4.8 Seeking Advice from Healthcare Providers

Only one quarter (n = 31; 25%) of participants reported that they had sought advice regarding AUD risks or protective strategies from a healthcare provider. (Table 2.7). The most common provider to be consulted was a therapist or counselor (71%; n= 22). Other providers consulted were Alcoholics Anonymous or spiritual leaders. When asked what healthcare providers they would consider consulting for AUD related advice around things such as risk factors or risk-reducing strategies, the most common

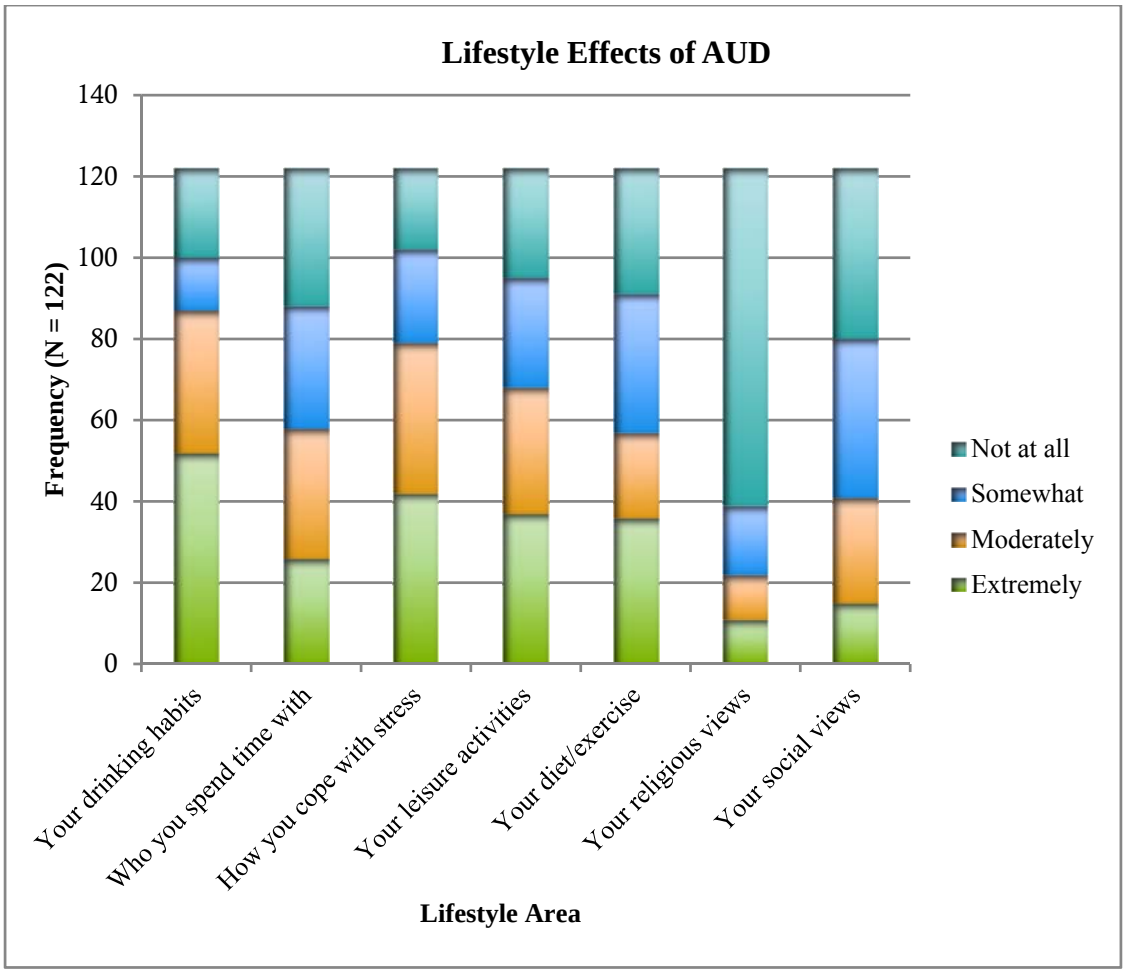

Figure 2.3: Lifestyle effects of AUD

Table 2.7: Healthcare Professionals Sought for AUD Advice

	Have Sought Advice		Would Seek Advice	
Healthcare Professional	Frequency (*n*=31)	Percentage (%)	Frequency (*n*=122)	Percentage (%)
Family doctor	18	58%	75	63%
Specialist doctor	10	32%	44	37%
Therapist/counselor	22	71%	91	77%
Nurse	2	7%	17	14%
Genetic Counselor	1	3%	18	15%
Other (please specify)	3	10%	3	3%

provider considered was a therapist or counselor (77%, *n* = 91). Eighteen (15%) respondents said they would consider asking a genetic counselor.

2.5 Discussion

Despite never experiencing genetic counseling for AUD, 60% felt that genetic counseling for AUD would potentially be beneficial. Individuals were significantly more likely to find genetic counseling beneficial when they attributed genetics/family history to be a larger cause of AUD. In contrast, individuals were less likely to report a perceived benefit of AUD genetic counseling if they attributed a lack of self-control to be a higher cause of AUD. The participants' sex, reported level of concern about risk for first-degree relatives, or their estimated recurrence risks did not appear to influence the perceived benefit of genetic counseling.

With few exceptions, the participants in this study recognized the multifactorial etiology of AUD and very few participants claimed genetics or a specific environmental factor to be the only cause of AUD. Participants' beliefs of cause did appear to play a role in their perceived benefit of AUD genetic counseling in regards to genetics/family history and lack of self control. Individuals who attributed the development of AUD more to lack of self-control may have felt that a conversation with a genetic counselor would do little to instill self-control in an individual. In contrast, those who felt that genetics/family history contributed highly to the development of AUD would perceive a benefit of discussing genetic contributions to AUD with a genetic counselor.

There was a wide range of estimated recurrence risks for first-degree relatives within the study group, suggesting that this group does not have a complete understanding of the recurrence risks of AUD. They perceived the risk for children of affected individuals to develop AUD (40%) to be higher than the risk for siblings of

affected individuals (29%). Extrapolating from the current population frequency of 7% for AUD (National Institute of Alcohol Abuse and Alcoholism, 2015) and the reported four-fold increased risk for children of individuals with AUD (Russell, 1990), the recurrence risk of AUD for children is approximately 28%. This study group perceived the recurrence risk for child to be higher than this estimated empiric recurrence risk, further suggesting that this group does not have an adequate understanding of risk. Also, the participants reported the highest level of concern about recurrence for their children to develop AUD compared to the level of concern for themselves or their siblings. This may be an important piece of information for genetic counselors and other healthcare providers to address when working with individuals with a personal or family history of AUD.

To better understand the overall impact a personal or family history of AUD has had on participants' lives, this study inquired into the level of impact a personal or family history of AUD had on a variety of lifestyle aspects. The majority of participants who had children or were considering having children reported that their reproductive decisions were not influenced by AUD, but five participants reported that their decision was affected that they either decided not to have children or to adopt. Participants reported that the two most common areas of their lives to be affected by AUD were the way they treated alcohol and the way that they cope with stress. This study did not explore specifically how these areas of lifestyle were impacted, and it is possible that the presence of AUD in a personal or family history impacted each participant very differently.

Overall, this survey population on average overestimated recurrence risk for children. While some individuals grossly overestimated recurrence risk for children and siblings, still others grossly underestimated the risk. Genetic counseling would help to clarify the recurrence risk for patients. Moreover, genetic counseling may help patients to gain a better understanding of the role different environmental and biological factors have in the development of AUD and would prompt them to consider how they can adjust some of the environmental factors (ways to deal with stress, a healthy social environment) and make behavior modifications to reduce their risk of developing AUD. By gaining a more balanced understanding of the factors involved in the development of AUD, patients may find a reduction in their guilt and shame around their personal or family history of AUD and an overall reduction of their stigma of the condition.

Future larger scale studies should include both individuals at risk for or affected with AUD as well as a more general population to assess understanding of etiology and recurrence risk. This would redress a limitation of the current population, as it is rather homogenous, being primarily Caucasian, well educated and potentially highly motivated individuals identified through AUD resources. Larger studies would provide more insight into possible factors that influence the perceived benefit of AUD genetic counseling. Pilot studies looking at the outcome of AUD genetic counseling can provide insight with respect to perceived benefits and/or behavior modifications. Additionally, investigating attitudes towards AUD genetic counseling among practicing genetic counselors and their comfort level with addressing AUD would be beneficial to understand what education needs to be provided to the profession. Finally,

education to this population regarding genetic counseling services would increase awareness and possibly uptake.

2.6 Conclusions

To our knowledge, this is the first study to explore the perceptions of recurrence risk and etiology as well as the possible benefit of genetic counseling among individuals at risk and affected by AUD. Based on the findings of this study, a personal or family history of AUD would be an appropriate indication for genetic counseling. While none of the participants previously had genetic counseling for AUD, the majority of the participants felt that genetic counseling for their personal or family history of AUD would be beneficial. Genetic counseling for this population could help clarify their misconceptions about recurrence risk and facilitate a better understanding of the many causes of AUD. Genetic counseling may also lessen the impact of AUDs on an individual and family by encouraging risk reduction strategies and reducing the psychological burden of the condition by addressing the guilt, shame, and stigma of AUDs. Further research is warranted into the outcomes of AUD genetic counseling and an increased effort for awareness of the availability of genetic counseling is needed for this population

Chapter 3. Conclusions

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Appendix A. Public Forum Invitation

Hello,

I am a graduate student doing research on alcohol addiction in hopes of shedding insight into new counseling methods. I am looking for individuals who would be willing to take a brief anonymous survey to answer a series of questions regarding their understanding of and experiences with alcoholism. There is a chance to win an Amazon gift card if desired.

I am hoping to survey both individuals who have or had an addiction to alcohol and their family members including spouses.

Many thanks in advance to all of you who decide to participate. Feel free to PM me if you have any questions!

To participate in this brief survey, follow this link:
https://www.surveymonkey.com/r/WCG3KM6

Appendix B. Private Group Leader Outreach

Hello NAME,

I hope this e-mail finds you doing well. My name is Fayth Kalb, and I am a graduate student at the University of South Carolina. I found your page through the GROUP NAME HERE that you are an administrator of.

I am currently doing research on alcohol addiction in hopes of shedding insight into new counseling methods. I am looking for individuals who would be willing to take a brief anonymous survey to answer a series of questions regarding their understanding of and experiences with alcoholism, and I was wondering if you would be willing to share my survey with your group members, as I am wanting to take the insights of both individuals with alcohol addiction and their family members into account during my research. I am not sure what your group's policy is on research studies being shared, but I would truly appreciate it if you would consider sharing my survey. There is additionally an opportunity for participants to win a $25 Amazon gift card if they choose to.

The short 5-10 minute survey can be found at the following link:
https://www.surveymonkey.com/r/WCG3KM6

Thank you so much again for your consideration of sharing my project. By having the thoughts and feelings of both individuals with alcohol addiction and their family members included in my research I am hoping to reach well-balanced and insightful conclusions for my research.

If you have additional questions or would like more information for my study, please let me know- I would be happy to share more!

Best,
Fayth

Appendix C. Survey Tool

1. How do you identify?
    a. Male
    b. Female
    c. Other
2. How old are you?
3. What is your highest level of education you completed?
    a. Middle school
    b. High school
    c. Associate's degree or trade school
    d. Bachelor's degree
    e. Postsecondary degree
4. What race are you?
    a. Caucasian
    b. African American
    c. Asian
    d. Hispanic
    e. Native American
    f. Other
5. Have you ever been diagnosed with alcoholism?
    a. Y/N
    b. If yes, are you active, attempting recovery, or recovered?
6. If you have not been formally diagnosed, do you identify as a person with alcoholism?
    a. Y/N
7. Do you have a relative or relatives with a history of alcoholism?
    a. Y/N
    b. Which family member(s) have had alcoholism? (choices will be given)
8. Do you have any other psychiatric conditions?
    a. Y/N
    b. Check all that apply: depression, anxiety, bipolar, schizophrenia, PTSD, panic disorder, other substance addiction, other (please specify)
9. Does your family member(s) with a history of alcoholism have any other psychiatric conditions?
    a. If yes, who? (choices will be given)
    b. Check all that apply for each: depression, anxiety, bipolar, schizophrenia, PTSD, panic disorder, other substance addiction, other (please specif

10. How much do you think each of the following factors causes alcohol addiction?
    a. Genetics/family history
        i. Extremely agree, somewhat agree, somewhat disagree, strongly disagree
    b. Lack of self control
        i. Extremely agree, somewhat agree, somewhat disagree, strongly disagree
    c. Traumatic life experiences
        i. Extremely agree, somewhat agree, somewhat disagree, strongly disagree
    d. Inability to handle stressful situations
        i. Extremely agree, somewhat agree, somewhat disagree, strongly disagree
    e. Spending too much time around others with alcohol addiction
        i. Extremely agree, somewhat agree, somewhat disagree, strongly disagree
    f. How your body reacts to alcohol
        i. Extremely agree, somewhat agree, somewhat disagree, strongly disagree
11. What is the likelihood of a child of someone with alcohol addiction developing the same addiction? (eg. 10%, 50%, etc.)
12. What is the likelihood of a sibling of someone with alcoholism developing the same addiction? (eg. 10%, 50%, etc.)
13. If you don't have a diagnosis of alcohol addiction, how concerned are you about developing it?
    a. Extremely | Moderately | Somewhat | Not concerned at all
    b. If extremely/moderately- what factors make you concerned?
14. How concerned are you about your children developing alcohol addiction?
    a. Extremely | Moderately | Somewhat | Not concerned at all
    b. If extremely/moderately- what factors make you concerned?
15. How concerned are you about your sibling(s) developing alcohol addiction?
    a. Extremely | Moderately | Somewhat | Not concerned at all
    b. If extremely/moderately- what factors make you concerned?
16. Has having alcohol addiction personally or in the family influenced your decision to have children?
    a. Yes- I've chosen to not have children
    b. Yes- I've chosen to have less children
    c. Yes- I've chosen o have more children
    d. Yes- I've chosen to adopt children
    e. No- it hasn't affected my decision to have children
    f. No- I'm not ready for children
17. How has having alcohol addiction personally or in the family affected the following-
    a. Your drinking habits
        i. Extremely | Moderately | Somewhat | Not at all
    b. Who you spend time with
        i. Extremely | Moderately | Somewhat | Not at all
    c. How you cope with stress
        i. Extremely | Moderately | Somewhat | Not at all
    d. Your leisure activities
        i. Extremely | Moderately | Somewhat | Not at all
    e. Your diet/exercise
        i. Extremely | Moderately | Somewhat | Not at all

f. Your religious views
    i. Extremely | Moderately | Somewhat | Not at all
g. Your social views
    i. Extremely | Moderately | Somewhat | Not at all

18. Have you looked for advice on topics such as risk factors or risk-reducing strategies regarding alcohol addiction in your family from a healthcare provider?
    a. Y/N
    b. If yes, check all that apply: your family doctor, a specialist doctor, therapist/counselor, nurse, genetic counselor
19. What healthcare providers would you consider talking to about risk factors and risk-reducing strategies for alcohol addiction?
    a. Check all that apply: your family doctor, a specialist doctor, therapist/counselor, nurse, genetic counselor
20. Before completing this survey, had you heard of genetic counseling?
    a. Yes, move to question 21
    b. No, move to question 27
21. Do you know what genetic counseling is?
    a. Yes, move to question 22
    b. No, move to question 27
22. Explain what genetic counseling is
23. Have you had genetic counseling?
    a. Yes, move to question 24
    b. No, move to question 27
24. What was the reason for your genetic counseling?
    a. Something was found on a prenatal test (ultrasound, blood work, etc.)
    b. I have a personal or family history of cancer
    c. I have a personal or family history of a genetic condition
    d. My child has a genetic condition
    e. I had a child when I was over 35 years of age
    f. Fertility trouble
    g. I underwent genetic testing
    h. Other (please explain)
25. Please describe your experience with genetic counseling.
26. Was your personal or family history of alcohol addiction addressed?
    a. Y/N

PROVIDE VIDEO ON WHAT ADDICTION GENETIC COUNSELING IS

27. Do you think you would benefit from addictions genetic counseling?
    a. Extremely | Moderately | Somewhat | No I do not think I would benefit
28. Would you like the chance tow in a $25 Amazon.com gift card?
    a. Y/N
29. Please enter the following information and then click ‘done’ to complete the survey
    a. Name
    b. E-mail address
    c. Mailing address

Appendix D. Video Transcript

Genetic counselors work with individuals or families to help them better understand the causes of conditions that occur or can run in families. Because people often may not understand the causes of conditions or may feel guilty or afraid that they may have caused the illness that they or their loved one has, genetic counselors are also able to provide education, support and counseling around these thoughts and feelings.

In genetic counseling related to addictions, the genetic counselor will talk with you to understand your medical and family history. They can then use the information you give them to provide you with personalized information about the possible genetic and non-genetic causes of illness in your family. The genetic counselor can also work with you to help you better understand what you can do to manage your condition, or reduce the risk of onset or relapse. If you are interested, genetic counselors can provide you with personalized information about the chances for others in the family to be affected. People can find genetic counseling helpful in coping with the affects alcohol addiction has had on you and your family as well as dealing with the guilt and stigma that can come along with mental health conditions.

To get in touch with a genetic counselor in your area to see if addiction counseling is a service they provide, you can visit www.nsgc.org

Appendix E. Additional Data

Table E.1 Participants' Mental Health Diagnoses

	Percentage (%)	**Frequency**
Yes	**45.1%**	**55**
Depression	34.4%	42
Anxiety	27.9%	34
Bipolar Disorder	5.7%	7
Schizophrenia	0.8%	1
Post Traumatic Stress Disorder	7.4%	9
Panic Disorder	0.0%	0
Other	8.2%	10
No	**54.9%**	**67**

Table E.2 Mental Health Diagnoses in Family Members with AUD

	Depression	Anxiety	Bipolar disorder	Schizop-hrenia	PTSD	Panic disorder	Other addiction
Yes (*n*=52)							
Mother	11	7	2	1	2	1	1
Father	17	6	4	1	4	0	3
Brother	6	5	5	0	3	0	5
Sister	2	4	0	0	3	3	0
Grandparent	12	3	2	1	1	0	1
Aunt	8	8	5	0	1	2	5
Uncle	8	4	5	2	1	1	6
Spouse	3	1	0	0	0	0	1
Son	0	0	0	0	0	0	0
Daughter	0	1	0	0	0	1	0
Grandchild	0	0	0	0	0	0	0