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Classifying Married Adults Diagnosed with Alpha-1 Antitrypsin Deficiency Based on Spousal Communication Patterns Using Latent Class Analysis: Insights for Intervention

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Abstract

Married adults are increasingly exposed to test results that indicate an increased genetic risk for adult-onset conditions. For example, a *SERPINA1* mutation, associated with alpha-1 antitrypsin deficiency (AATD), predisposes affected individuals to diseases such as chronic obstructive pulmonary disease (COPD) and cancer, which are often detected in adulthood. Married adults are likely to discuss genetic test results with their spouses, and interpersonal research suggests that spouses' communication patterns differ. Latent class analysis was used to identify subgroups of spousal communication patterns about AATD results from a sample of married adults in the Alpha-1 Research Registry (N = 130). A five-class model was identified, and the subgroups were consistent with existing spousal-communication typologies. This study also showed that genetic beliefs (e.g., genetic stigma), emotions, and experiences (e.g., insurance difficulties) covaried with membership in particular subgroups. Understanding these differences can serve as the foundation for the creation of effective, targeted communications interventions to address the specific needs and conversational patterns of different kinds of couples.

Keywords

Genetic test results; spousal communication; latent class analysis; alpha-1 antitrypsin deficiency (AATD)

Genetic tests can allow people to learn whether they carry a mutation that predisposes them to specific health conditions, before they experience symptoms. For example, the presence of a mutation in *SERPINA1* leads to alpha-1 antitrypsin deficiency (AATD), which predisposes affected individuals to diseases such as chronic obstructive pulmonary disease

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(COPD), emphysema, cirrhosis, and lung or liver cancer (Laurell & Eriksson, 1963; Sharp, Bridges,Krivit, & Freier, 1969).

AATD has been described as an under-recognized (Stoller et al., 2005) but common (American Thoracic Society, 2003) inherited, monogenic disorder caused by a mutation in the *SERPINA1* gene located on chromosome 14 (14q31-32. 3). It affects approximately 1 in 2,000 to1 in 5,000 people (Stoller & Aboussouan, 2012). By itself, AATD is not a disease; however, this condition—in conjunction with environmental factors, such as smoking (Tanash, Nilsson, Nilsson, & Piitulainen, 2010)—predisposes individuals to chronic obstructive airway diseases and chronic liver diseases (American Thoracic Society, 2003).

Because the presentation of AATD-related symptoms mimics other conditions like asthma, five to eight years often pass between onset of symptoms and an AATD diagnosis (Stoller et al., 2005). The delay creates uncertainty regarding the diagnostic process surrounding AATD (Sandhaus, 2010). The American Thoracic Society and European Respiratory Society now specifically recommend testing all individuals diagnosed with COPD for AATD (ATS/ERS Statement, 2003). Additionally, the prognosis for AATD is highly variable: some people who are homozygotes or compound heterozygotes for the deficiency manifest no symptoms, while some carriers do experience symptoms (Wienke, 2012). AATD is often described as autosomal recessive; however, autosomal codominant with reduced penetrance may be more accurate. This further contributes to the uncertainty associated with this disorder. Using Rolland and William's (2005) typology, AATD fits into the class of genetic conditions in which onset of clinical symptoms is in adulthood, likelihood of development is variable, and treatment/lifestyle modification can alter the onset or progression of clinical symptoms. Other conditions fitting into this category are BRCA mutations for inherited breast and ovarian cancers.

Alpha-1 antitrypsin (AAT) is a serine protease inhibitor that is primarily produced in the liver. It then gets secreted into the bloodstream and travels to the lungs, which it protects from damage caused by neutrophil elastase released during periods of inflammation. Individuals that are deficient in AAT, therefore, have a higher risk for lung damage. Currently, approximately 120 alleles have been described (Stoller & Aboussouan, 2012). There are several "normal" alleles, represented by the letter M. The most common allele associated with AATD is the Z allele, which originated in Scandinavia and accounts for 95% of recognized cases. Phenotypes are categorized to describe the protein made using a Pi (protease inhibitor) system. Those that are homozygous for the M allele (PiMM) make normal amounts of serum AAT (20-53 μM or ~80-220 mg/dl determined by nephelometry). Deficiency alleles produce serum level of AAT less than 20 μM and some have reduced function. There are also rare variants, as well as null variants that result in little to no protein production. The Z allele creates an AAT protein that polymerizes and accumulates inside the cells of the liver. With only about 15% of protein being released into the bloodstream, the remaining 80-90% can cause liver damage (ATS/ERS Statement, 2003).

AATD is primarily thought of as a predisposition to liver disease (hepatitis, cirrhosis and hepatocellular carcinoma) and lung disease (early onset emphysema, COPD, chronic bronchitis, adult onset asthma and bronchiectasis) but there are other minor clinical

symptoms described including necrotizing panniculitis and Wegener's granulomatosis (ATS/ERS Statement, 2003). While the clinical lung and liver symptoms typically present when the person is in the 3rd or 4th decade of life, some children can have signs of liver disease such as jaundice after birth. The majority of PiZZ children are clinically healthy throughout childhood, although 2.5% have severe liver disease that can lead to transplant and/or death (ATS/ERS Statement, 2003). While PiZZ is more strongly linked to severe symptom manifestations (i.e., wheezing, shortness of breath, chronic bronchitis, lung or liver deterioration), those with heterozygous results (e.g., PiMZ) can also develop serious symptoms through environmental exposure (e.g., pollutants) or health behaviors, such as smoking (Klitzman, 2009; Tanash, Nilsson, Nilsson, & Piitulainen, 2010).

Advocacy groups like the Alpha-1 Foundation have formed around AATD, created materials to help those with AATD, and lobbied for the Genetic Information Nondiscrimination Act (GINA). People with AATD face potential genetic stigmatization, and the outcomes (COPD, lung cancer, etc) have noted stigmas. Similarities between AATD and other genetic conditions and the unique features of AATD make it a highly heuristic case for analysis. When considering the psychosocial impact of genetic disease on a person as well as the married couple as a unit, genetic conditions may be categorized based on age of onset, modes of inheritance, severity, and whether or not testing and treatment are available (Lerman et al, 2002). Genetic testing is now available clinically and through direct-to-consumer companies for common, adult onset conditions like emphysema, breast cancer and Alzheimer's disease to allow adults to learn whether or not they carry a mutation in a gene that would predispose them to developing symptoms. Since testing cannot tell if a person will (or will not) develop symptoms, there is a concern that testing asymptomatic individuals can cause stigmatization and negative feelings such as anxiety, guilt and fear (Bank et al, 2004; Kenen & Schmidt, 1978; Lerman et al, 2002; Rahman, 2012). Furthermore, these conditions can be both inherited and sporadic, or they may be caused by mutations in other genes for which testing is not currently available (Bayraktar, 2013; Rahaghi, 2012; Rahman, 2012). The uncertainty associated with the nuances of these conditions may cause much of the stress and anxiety felt by individuals when they have had testing and by their unaffected spouses (Bank et al, 2004; Kenen & Schmidt, 1978; Lerman et al, 2002).

While AATD is similar to conditions like BRCA 1/2, it is also distinct in that it affects both men and women equally and the symptoms are more stigmatized. Breast cancer is most often a spontaneous condition, while emphysema and cirrhosis are often linked to environmental causes. Those affected by AATD may feel others are less sympathetic because they "caused" their illness. On the other hand, people who are not yet symptomatic have a source of control over whether or not they will develop symptoms. Like BRCA 1/2, those with a positive test can make lifestyle changes and have the advantage of regular screening and early intervention to improve health outcomes. For individuals who are already symptomatic, treatment is available, although the effectiveness is currently under debate (Stoller & Aboussouan, 2012). Identifying the specific negative outcomes for AATD patients (and spouses) that have undergone testing is certainly an area where research is needed. It is important to begin by addressing the way that patients discuss genetic testing

and genetic diseases with their spouses before specific interventions can be developed to assist these individuals in coping with their diagnosis.

People may be tested for AATD in a range of circumstances: on preconception/ prenatal carrier screening panels, in the process of diagnosing and treating health conditions related to AATD, in a battery of tests to diagnose something else, and to learn about inherited conditions. An increasing number of people are diagnosed with AATD in the context of married relationships (Coors, Levinson, & Huitt, 2008; Lazarin et al, 2013). Married adults are likely to discuss genetic test results with their spouses (Koehly et al., 2003), and couples can differ widely in their conversation patterns (Fitzpatrick, 1988; Koerner & Fitzpatrick, 2002).

Spousal Communication Profiles

According to Fitzpatrick (1987, 1988), the differences in spousal communication patterns appear on a daily basis, and when grappling with emotional or conflictual situations, such as discussing a serious diagnosis like AATD and deciding to whom the diagnosis should be disclosed (e.g., family, physicians, insurance providers). Fitzpatrick (1988) theorized and found empirical support for four spousal communication patterns. *Traditionals* engage in regular, positive communication and are highly interdependent, valuing togetherness over autonomy. *Independents* value both their relationship and individual autonomy. *Separates* avoid interactions and interdependence. Last, Fitzpatrick argues for *Mixed* couple types in which the partners represent the blending of two different types (e.g., traditional husband and independent wife).

Predicting Spousal Communication about AATD

This study investigates whether married adults may present different types of spousal communication patterns that correspond with existing family research. This kind of audience segmentation is a critical step toward developing a targeted, effective communication intervention (Slater, 1996), such as to support couples making AATD-related decisions, because it allows for targeting messages to particular subgroups. In addition to identifying profiles of spousal communication, it may be useful to identify variables predicting the odds of showing one form of spousal communication in comparison to another. This study investigates emotional, timing, testing, and stigma-related covariates.

The Alpha-1 Foundation's website (www.alpha-1foundation.org) emphasizes that most people diagnosed with AATD have a partner, and that one way couples manage the uncertainty and emotions associated with AATD is through shared information-seeking. Receiving test results for AATD can be shocking and upsetting (Dohany, Gustafson, Ducaine, & Zakalik, 2012; Klitzman, 2009; Lippi, Favaloro, & Plebani, 2011); emotional states may predict more spousal conversations and interdependent decisions. That said, the amount of time a person has been living with a diagnosis of AATD varies widely among persons diagnosed with AATD. The difference in the amount of time a person has been diagnosed with AATD may predict the amount of spousal communication and interdependent decision-making. For example, a newly diagnosed person may be faced with decisions about whether or not to disclose the AATD diagnosis to family members and

whether or not to make these disclosure decisions independently or with his or her spouse. Conversely, a person who has been diagnosed with AATD for a decade or more may have already made their disclosure decisions. On the other hand, if a diagnosed person has been diagnosed with AATD for a long time but has not been married to his or her spouse for a long period of time, they may perceive that their spouse has little information about AATD and an inability to fully discuss the diagnosis and its implications. Marital length may also predict spousal communication patterns: some kinds of couples, such as Traditionals, may remain married longer than other couple types because Traditionals place a high value on marriage (Fitzpatrick, 1984). Thus, emotional states, time since diagnosis, and marriage length may be associated with the strength of desire to communicate as a couple about AATD.

The testing, treatment, and stigma may also predict spousal communication patterns. Many adults who are tested for AATD do so because they are already experiencing symptoms, such as shortness of breath, wheezing, and chronic bronchitis (Kelly, Greene, Carroll, McElvaney, & O'Neill, 2011; Stoller & Aboussouan, 2009). During this diagnostic process, when traditional treatments are not effective (e.g., asthma medications are not decreasing asthma-related symptoms), patients can have a high degree of uncertainty about the cause of their symptoms until an AATD diagnosis is confirmed, and not talk until there is more information. Additionally, modified labeling theory (MLT; Link, Cullen, Struening, Shrout, & Dohrenwend, 1989) has shown that labeling people with health conditions can lead to negative outcomes. According to MLT, the more people believe they will be devalued and discriminated against, the more they may feel threatened when communicating with others. For people diagnosed with AATD, beginning augmentation therapy—the administration of an IV-infused protease inhibitor that replaces the antitrypsin not present in the bloodstream —may serve as a labeling moment and increase the likelihood of such persons having difficulties in getting and maintaining health insurance coverage. Couples, then, may manage uncertainty about genetic risks, potential for genetic stigmas, and worries about insurance discrimination by avoiding communication that could yield undesirable information (Yaniv, Benador, & Sagi, 2004).

Finally, much has been researched about differences between males and females in regard to spousal communication (Ball, Cowen, & Pape-Cowen, 1995; Noller, 1993; White, 1988). For this reason, gender differences were explored.

The Present Study

To integrate the work in genetic and spousal communication, this study investigates how married adults tested for AATD talk with their spouses and identifies how many audience segments (also called subgroups or classes) may exist based on spousal communication patterns. Specifically, we address several questions: Do they talk frequently about AATD with their spouse (i.e., Traditionals or Independents), or do they feel as if they are already talking too much about the diagnosis (i.e., Separates)? Do married, diagnosed adults differ in their desire for interaction when discussing AATD? Similarly, we assess whether diagnosed, married adults decide with their spouse whether or not to tell other family members, physicians, or insurance providers about the AATD diagnosis (i.e., Traditionals), or if they

make these disclosure decisions autonomously (i.e., Independents or Separates). The research questions are:

RQ1: Does a latent class structure representing the heterogeneity of spousal communication about AATD correspond with Fitzpatrick's typology (1988)?

RQ2: How well do covariates (positive affect, negative affect, time since diagnosis, time with spouse, Augmentation therapy, insurance discrimination, genetic stigma, and gender) predict membership in spousal communication profiles?

Methods

Procedures

An institutional review board approved the study. Participants were recruited through the Medical University of South Carolina (MUSC) research registry. The registry includes 1788 members who provided email addresses and indicated willingness to be contacted for research. The recruitment invitation (provided via email) told registered members that the study was interested in married couples' experiences with the AATD diagnosis, and provided the link to access our online questionnaire. Of the 1788 members, 219 started the survey. After giving consent, participants were asked whether they had a partner who could also complete the survey. Those without partners (n = 40) were sent directly to a thank you page, and were not able to answer questions in the survey. Of the 179 remaining members who started the survey, 130 completed it. We do not know how many of the registered members are married, but the response rate (179/1788 or 10%) is likely an underestimate.

Married participants were asked to report their marriage date and then state where they currently reside, which was used to link couples' responses (a dyadic analysis of the spousal communication of 50 couples appears in Smith, Wienke, & Coffman, 2013). Participants were then asked to complete measures related to genetic beliefs, stigma, genetic testing, couple communication, emotions related to the AATD diagnosis, marital quality, health insurance coverage, and current health status and behaviors. The survey was piloted with ten adults who were not members of the registry, but were involved with AATD-related activities (e.g., education). Based on their feedback, we adjusted a few items in the spousal communication scales to make them clearer. Participants in the pilot, on average, completed the survey in 20 minutes.

Latent Class Analysis (LCA) Indicators

Seven indicators were included in the latent class profiles (see Table 1 for descriptive statistics of indicators). The items were created by the research team, and were then piloted with a small group of adults (who were not in the registry, but participated in Alpha-1 related activities) before sending out the survey. Measures with multiple items, current spousal communication and desired spousal communication, were dichotomized at the mid-point of the scale (i.e., 1-2.99 recoded to 1, and 3-5, recoded to 2). Decision items were recoded so that decisions made by both spouses together (*my spouse and I together*, recoded to 2) were separated from decisions made by one spouse or other people (*me*, *my spouse*, *me and someone else* and *someone else*, recoded as 1).

Current spousal communication—Six items (Smith, Wienke, & Coffman, 2013) were used to measure the frequency of conversations spouses currently have regarding AATD-related information (e.g., *I have talked with my spouse about how the AATD results make me feel*). The response options were strongly disagree, disagree, neutral, agree and strongly agree, which were later coded for analysis (1 = *strongly disagree* to 5 = *strongly agree*). Responses were averaged into one score (α = .84), with higher scores indicating more frequent current AATD-related couple communication.

Desired spousal communication—Six items (see Appendix) were used to measure how much the person diagnosed with AATD desired to have conversations with his/her spouse regarding AATD-related information (e.g., *I want to talk with my spouse about how the AATD results make me feel*; 1 = *not at all* to 5 = *very much*). Responses were averaged into one score (α = .89), with higher scores indicating stronger desire for AATD-related couple communication.

Discrepancy between current and desired couple communication—The average score for current couple communication was subtracted from the average score for current couple communication to create the discrepancy score.

Decision to tell family—One item was used to measure who decided whether or not to tell family members about the AATD diagnosis (*me, my spouse, my spouse and I together, me and someone else, someone else*).

Decision to tell insurance provider—One item was used to measure who decided whether or not to tell insurance providers about the AATD diagnosis (*me, my spouse, my spouse and I together, me and someone else, someone else*).

Decision to tell physicians—One item was used to measure who decided whether or not to tell physicians about the AATD diagnosis (*me, my spouse, my spouse and I together, me and someone else, someone else*).

Covariates—Eight covariates were included in the latent class profiles. All continuous responses were marked on five-point scales (see Table 2 for means and correlations). Binary covariates were coded so that 0 = *no* and 1 = *yes*. Continuous covariates (negative and positive affect, genetic essentialism, marital satisfaction, and secrecy) were standardized as suggested by Lanza et al. (2007) to allow the LCA procedure to generate standardized logistic regression coefficients, which facilitate their interpretation.

Tested when sick—Participants were asked to identify which of these reasons best applies to their reason for getting the AATD test: prenatal testing, family member tested positive, and the respondent was symptomatic. Those identifying themselves as symptomatic before testing occurred were coded as sick before testing (1); everyone else was coded as not (0).

Negative affect—Seven items were used to measure negative emotional responses to the AATD diagnosis (e.g., *anger, frustration, fear;* 1 = *not at all* to 5 = *very much*). Responses

were averaged into one score (α = .82), with higher scores indicating stronger negative emotions regarding the AATD diagnosis.

Positive affect—Three items were used to measure positive emotional responses to the AATD diagnosis (e.g., *relief, happy, hopeful;* 1 = *not at all* to 5 = *very much*). Responses were averaged into one score (α = .67), with higher scores indicating stronger positive emotions regarding the AATD diagnosis.

Time since diagnosis—The number of years that participants had been diagnosed with AATD was used to measure time since diagnosis. This ranged from 0 to 41 years (*M* = 9.47, *SD* = 8.05).

Time with spouse—The number of years that participants had been married to their spouse was used to measure time with spouse. This ranged from 0 to 58 years (*M* = 27.23, *SD* =14.40).

Augmentation therapy—Augmentation therapy was measured by one item indicating whether or not (i.e., *yes* or *no*) participants had been on augmentation therapy in their lifetime.

Insurance difficulties—Insurance difficulties was measured by one item indicating whether or not (i.e., *yes* or *no*) participants experienced difficulties in obtaining health insurance coverage.

Genetic stigma—As seen in Smith et al, 2013, eight items based on Link and colleagues (1989) were used to measure genetic stigma beliefs (e.g., *Most people would feel that being diagnosed with a genetic mutation is a sign of personal failure*). The response options were *strongly disagree*, *disagree*, *neutral*, *agree* and *strongly agree*, which were later coded for analysis (1 = *strongly disagree* to 5 = *strongly agree*), with higher scores indicating stronger beliefs that AATD is a stigmatized condition.

Gender—Participants' indicated their biological sex, and this was coded as 0 for males and 1 for females.

Analysis Plan

Latent class analysis (LCA) is used to empirically test whether people fall into mutually exclusive and exhaustive subgroups (Lanza et al., 2007). LCA, conceptually, is similar to other latent variable models such as factor models, in that it attempts to capture latent constructs from measurable variables. LCA is used when the latent construct is categorical (Collins & Lanza, 2010). PROC LCA (Collins & Lanza, 2010) requires categorical observed variables to measure the categorical latent variable. It provides two kinds of parameters: the likelihood of providing a particular response to a measured variable conditional on the set of classes, and the likelihood of membership in a latent class. LCA also provides goodness-of-fit indicators for models, which are used to determine the best number of classes (e.g., a three-class or four-class model).

LCA also allows one to test whether other variables predict the odds of membership in one class relative to another. In the context of this study, the LCA procedure can show the optimal number of different classes of couples based on their spousal communication patterns about AATD, and the covariate analysis can suggest particular needs or concerns associated with each class. LCA, then, provides an empirical basis for understanding how many different interventions may be needed for married adults based on their communication profiles and profile-related variables.

Results

Participants

Married adults diagnosed with AATD who are members of the research registry at the Medical University of South Carolina participated in this study (N = 130, 55% female, 48% employed, 94% White, 78.5% ZZ and 21.5% MZ phenotype). Participants on average were 56.79 years old (*Mode* = 60, SD = 10.67, *Minimum* = 29, *Maximum* = 81). Of the 130 registered members in the study, 45% reported being symptomatic before they were tested for Alpha-1; the rest reported other reasons, such as a diagnosed family member (42%) and prenatal testing (6%). Of note, although participants could mark multiple reasons, no overlap was observed in marking symptomatic reasons and prenatal or positive-family reasons (4% did mark both of these last two reasons).

Descriptive Statistics

As shown in Table 1, married adults tested for AATD varied in their reported decision-making and communication patterns with their spouses. Over half reported frequent discussions of the genetic diagnosis (61.5%) and desires for more conversations about it (58.5%). Indeed, over half, wanted to talk more about the diagnosis than they were doing currently (56.9%). More registered members reported making decisions about telling other family members together (61.5%), than telling insurance (56.2% decided together), or telling physicians (43.1% decided together). Of note, those who reported not making decisions together typically reported that they or their spouse made the decision (family disclosure: 29% me or 8% spouse; insurance disclosure: 28% me or 11% spouse; and physician disclosure: 43% me or 13% spouse). Very few people reported making decisions with someone other than their spouse or that the decision was made by someone other than their spouse.

As for the covariates (see Table 2), the sample was split in the context for their AATD testing: 41% of the married adults were sick when they were tested for AATD; a different 42% were tested after hearing about another family member’s diagnosis. Some (21.5%) reported difficulties getting health insurance; of these, 61% reported that it was related to the AATD diagnosis. Of note, many covariates were not significantly correlated. Marital satisfaction was also measured and was found to be extremely high (M = 4.42, SD = 0.84), and unrelated to the covariates. Thus satisfaction was not included in the analysis.

Spousal Communication Classes

To address RQ1, Proc LCA (Lanza et al., 2007) was used to calculate fit indices for two- to seven- class models using 500 sets of random starting values for each test (see Table 3 for fit indices). The five-class model was selected because it had the lowest Akaike's Information Criterion (AIC) and Bayesian Information Criterion (BIC) scores (Collins & Lanza, 2010). These results answer RQ1: latent classes of married adults do exist based on their spousal communication patterns.

LCA generates two parameters to characterize these underlying classes of spousal communication (see Table 3). The first is the likelihood of membership in a class; the second is the likelihood of reporting a particular answer (code 2 in Table 2) within a class. Three classes (labeled as Separates, Independents, and Traditionals) closely reflected the couple communication profiles described by Fitzpatrick (1988) and Koerner and Fitzpatrick (2002). One (labeled herein as Conformers) was close, and the last (labeled herein as Self-Determiners) was new.

Respondents in the class labeled Self-Determiners (20%) were unlikely to have frequent conversations about AATD or a desire for more AATD-related conversations with their spouse, but they desire to talk more than they do. Self-Determiners were also unlikely to make disclosure decisions as a couple for family, insurance providers, or physicians. Separates (17%), labeled based on Fitzpatrick's description (1998), also were unlikely to talk or want to talk with their spouses about AATD, and, in fact, talk more than they desire, and were unlikely to make disclosure decisions together. Conformers (14%) were also unlikely to talk or want to talk with their spouses about AATD, and talked more than they desired. However, Conformers were likely to make disclosure decisions together, deciding as a couple whether or not to disclose the AATD diagnosis to family, insurance providers, and physicians. Respondents in the class labeled Independents (20%), based on Fitzpatrick's description (1988), reported frequent conversations and desire for more conversations about AATD with their spouses, and they talked less than they desired. Independents were unlikely to make disclosure decisions as a couple. Finally, Traditionals, the largest class (30%), reported having frequent current communication about AATD and a high desire for AATD-related conversation, and talked less than they desired. Additionally, Traditionals were likely to make disclosure decisions as a couple, which fits Fitzpatrick's description (1988).

Covariate Analysis

To answer RQ2, the covariates (tested when sick, time since diagnosis, time with spouse, negative and positive affect, augmentation therapy, experience with insurance difficulties, and genetic stigma, and gender) were explored. The Self-Determiners class was used as the reference class. All covariates were significant at $p < .05$. Results appear in Table 5.

In comparison to Self-Determiners, those who were tested for AATD when they were already sick were more likely to be Separates and Conformers and less likely to be Independents or Traditionals. In comparison to Self- Determiners, Conformers were more likely, whereas Separates, Independents, or Traditionals were less likely, to have a negative

emotional response to the AATD diagnosis. Conversely, compared to Self-Determiners, Separates and Conformers were more likely and Independents or Traditionals were less likely to have a positive emotional response to the AATD diagnosis. Compared to Self-Determiners, those who have lived with the AATD diagnosis longer were more likely to be Separates and less likely to be Conformers, Independents, or Traditionals. Finally, compared to Self-Determiners, those who have been married to their spouse longer were more likely to be Independents or Traditionals and less likely to be Separates or Conformers.

Compared to Self-Determiners, those who had been on augmentation therapy were more likely to be Traditionals and Independents and less likely to be Separates or Conformers. Similarly, compared to Self-Determiners, those who perceived that AATD carries a social stigma were more likely to be Traditionals and Independents and less likely to be Separates or Conformers. Finally, compared to Self-Determiners, those who had experienced difficulty obtaining health insurance coverage were more likely to be Separates, Traditionals, or Independents and less likely to be Conformers.

Lastly, in comparison to Self-Determiners, females were more likely to be Independents, while males were more likely to be Separates, Conformers, or Traditionals.

Discussion

This study used LCA to identify subgroups of married persons diagnosed with AATD based on their self-reported spousal communication patterns and to identify covariates that predicted membership in one spousal communication profile rather than another. From the analysis, five classes emerged based on spousal communication patterns related to disclosure decision-making and discussing the AATD diagnosis. Three classes closely reflected the couple communication profiles described by Fitzpatrick (1988) and Koerner and Fitzpatrick (2002); one was close, and the last was new. Additionally, membership in the five classes was predicted by covariates associated with being sick while receiving genetic news, affective responses to AATD, time with diagnosis and spouse, genetic stigma and insurance discrimination, and gender.

Spousal Communication Patterns

Three of the classes found in this study closely represent those described by Fitzpatrick (1988): Separates, Independents, and Traditionals. According to Fitzpatrick (1988), such communication patterns reflect how spouses respond to conflict, in this case how spouses communicate about the incurable genetic diagnosis of AATD. Similar to Fitzpatrick's (1988) descriptions, Separates in the present study demonstrated autonomous decision-making and an avoidance of conversation about AATD; Independents demonstrated a willingness and desire to communicate about AATD but preferred autonomous disclosure decisions; and Traditionals demonstrated strong current and desired communication about AATD and interdependent disclosure decisions. In previous research, Fitzpatrick (1984) finds that 60% of couples fit into these three profiles; our results predict that 67% of the sample fit into these profiles, which is quite similar to the previous research.

A fourth class, Conformers, reflected the communication pattern labeled Protective in Koerner & Fitzpatrick (2002), in which couples place a high value on congruity between the partners and less value on open communication. In this study, the Protectives class has been relabeled Conformers due to their high degree of conformity orientation, as described by Koerner and Fitzpatrick (2002), to avoid confusion with patterns that could protect their health. Finally, a new class emerged that had not been represented in these typologies: Self-Determiners. This class was very similar to Fitzpatrick's (1988) Separates; however, Self-Determiners differed from Separates in their desire to talk about AATD. Separates reported talking more with their spouses about AATD than they desired, while Self-Determiners did not. The findings, overall, resonated well with previous couple communication profiles.

The covariate analysis sheds light on differences among the classes that may make membership in each class more likely. Conformers, as compared to Self-Determiners, were far more likely to have been tested when sick, to have more positive and negative emotional reactions to the AATD diagnosis, to be newly diagnosed and to be newly married. (Of note, older age was also associated with more years since diagnosis and longer marital length.) This combination of experiences may imply that Conformers are still using single-person mechanisms to manage their illness, emotions, and decisions related to AATD diagnosis in their new relationships, and using communication to synchronize their feelings with their spouse, which corresponds with their highly interdependent decision-making (Fitzpatrick, 1987). This class, compared to Self- Determiners, is also more likely to be male, which is in keeping with Koerner & Fitzpatrick's (2002) Protective profile, in which important decisions—such as deciding with your spouse to whom to disclose the AATD diagnosis—would be made interdependently to foster congruence between the spouses for the protection of the family.

Separates, compared to Self-Determiners, were also more likely to have been tested when they were sick, and they, too, have been with their spouse for a short time. However, Separates (as compared to Self-Determiners) have been living with the AATD diagnosis for a longer time, and report more positive affect toward the diagnosis. For Separates, then, it is possible that the length of time they have been living with the AATD diagnosis may change their communication needs; in other words, they are not managing negative emotions associated with being newly diagnosed or undertaking information-seeking efforts like those who have just learned about the genetic condition. Separates also reported not making interdependent disclosure decisions, and this, too, may be a result of the length of time they have been living with AATD. Self-Determiners may have made AATD-related disclosure decisions prior to the start of their relationships, and therefore, disclosure is not a salient issue for these couples.

These findings suggest that the timing of clinical onset and learning of a genetic-based risk for a genetic condition may be very important for understanding family communication around genetics. Rolland and Williams' (2005) work on the family systems genetic illness (FSGI) model provides a helpful explanation for the differences between families reacting to genetic information in comparison to medical information. Our study, then, suggests that we need to also consider when these two worlds overlap for those experiencing a chronic condition and then learning of their genetic risk for it. One concern raised in studies of the

psychological effects of genetic testing is whether some people get tested without careful consideration of the consequences of testing (Lerman, Croyle, Tercyak, & Hamann, 2002), which may be particularly relevant for symptomatic persons.

Labeling and stigma theory

Both Independents and Traditionals, compared to Self-Determiners, are more likely to experience insurance discrimination and perceive a genetic stigma. Stigma is "an attribute that is deeply discrediting" that reduces an individual from a "whole and usual person to a tainted, discounted one" (Goffman, 1963, p. 3). Independents and Traditionals, then, compared to Self-Determiners, are more likely to view having AATD as something disgraceful that would discredit them in society. These two classes are also more likely to have been on augmentation therapy, compared to Self-Determiners, because they would have been diagnosed with AATD before augmentation therapy was prescribed (Link et al., 1989; Smith, 2007, 2011). Both Independents and Traditionals, compared to Self-Determiners, are more likely to have experienced difficulties obtaining insurance, which is one hallmark of discrimination against groups with stigmatized health conditions (Link et al., 1989). Both Independents and Traditionals were more likely than Self-Determiners to be asymptomatic when they received their genetic information. It is also possible that married adults managing the information about a genetic risk for a future condition may be concerned and aware of possible societal and discrimination consequences for themselves and their families, while those who are sick may need to manage the emotions of a diagnosis and treatment for a serious chronic condition.

Study Limitations and Research Recommendations

By choosing people who are part of the Alpha-1 Research Registry at the Medical University of South Carolina, it is possible that these findings may not be broadly generalizable. Also, by solely focusing on AATD, our findings may not be generalizable to other genetic conditions. Future studies could sample persons diagnosed with AATD outside of the research registry or choose a comparison group (e.g., BRCA1/2) to explore any differences that exist between the groups. In addition, participants completed questions about their spousal communication before they answered the questions about the covariates. It is possible that there is an order effect, which should be addressed in future studies.

Our findings are limited to the perceptions of one of the partners in each couple: the sample in this study only represents persons diagnosed with AATD, but not their spouses. Future studies may examine both partners' impressions of the couple's communication patterns in a latent class analysis. With this approach, it would then be possible to analyze the findings in regard to Fitzpatrick's (1988) mixed-couple types, to determine whether partners blend two different preferred types of communication (e.g., Separate husband and Traditional wife). Further, it is possible that some of our participants are married to people who also have this genetic mutation, which may affect their communication as well.

An additional limitation is that our study focused on married adults, which would have excluded those in long-term relationships in other forms (de facto, common law, etc). Married relationships have different legal implications from other forms of long-term

relationships, which may influence the number and types of subgroups based on communication patterns within other forms of relationships. The spousal communication patterns may also represent broader family communication patterns; these family patterns vary in how open or closed the communication is among family members (Koerner, LeRoy, & Veach, 2010). Other forms of committed and family relationships should be studied in the future.

Practical Implications for Alpha-1 Organizations

A practical outgrowth of this analysis would be the development of targeted communications material that Alpha-1 organizations such as the Alpha-1 Foundation or Alpha-1 Association could provide to couples managing an AATD diagnosis (Slater, 1996). Comparable communications audience segmentation has been done regarding the dissemination of genetic/genomic information within families (e.g., BRCA1/2 mutations; Koehly et al., 2009), with a similar call for tailored communications interventions targeted at differences in family communication. Although much research exists regarding Fitzpatrick's typologies, the authors know of no communication interventions that have been grounded in these typologies. Therefore, an innovative approach to designing a tailored intervention for couples dealing with AATD-related communication and possibly other genetic conditions would be to base such an intervention on the couple profiles found by Fitzpatrick and the present study. In addition, genetic counselors may benefit from having a means by which to classify spouses seeking their guidance. For instance, they could tailor their content and delivery for their clients who are processing the information more on their own or more in coordination with a partner, who may or may not be present.

Although five classes emerged from this analysis, it is possible that one type of communications materials would benefit couples in two or more classes. For example, Conformers and Separates are both likely to be symptomatic (i.e., they were tested when sick), and they are dealing with both positive and negative emotional responses to the AATD diagnosis. Though their time with the AATD diagnosis differs, their relationships with their spouses are relatively short. People in these classes may be trying to handle illness, a new relationship, and their emotions at the same time. Communications materials that address these needs (e.g., how to live with symptoms related to AATD, how to get treatment, how to deal with emotional battles associated with the diagnosis) may be pertinent to people in these classes. Conformers and Separates perceived less genetic stigma, and communications materials that help people cope with AATD-related stigma might cause unnecessary stress (i.e., alerting these people to a possible stigma they had not previously been focusing on).

Conversely, Independents and Traditionals, who are likely to be asymptomatic and not feeling emotions related to AATD, would most likely not benefit from the same materials. Independents and Traditionals are more likely to have been married longer, and therefore may have more couple-oriented rather than single-oriented communication strategies. These two classes are also far more likely to view AATD as a stigmatized condition—they have likely been on augmentation therapy and therefore are "known" (i.e., labeled) as having AATD, and they are more likely to have experienced insurance difficulties.

Communications materials targeted to these two groups should focus more on health insurance support and the effective management of AATD-related stigma than on symptom management and emotional needs. That said, Separates and Traditionals are argued to continue sharing facts with each other; Independents are thought to share affective communication (Fitzpatrick, 1984, 1987). The balance of emotions and facts in the materials may need to differ as well.

For all of these couple classes, however, targeted materials should not focus on changing the communication dynamic of the couples. Though Fitzpatrick (1988) asserts that different communication patterns can lead to martial dissatisfaction, our findings suggest martial satisfaction is high across all couple communication classes. Therefore, such targeted communication interventions should not seek to change the couples' communication patterns but instead address the specific needs of the couples in each of the classes.

Conclusions

Increasingly, people in the U.S. are being tested for genetic conditions such as AATD while they are in committed relationships (Coors et al., 2008), and they are likely to discuss genetic test results with their spouses (Koehly et al., 2003). Such couples may benefit from targeted communications materials to help support them as they talk about the genetic diagnosis and make disclosure decisions. Because spousal communication patterns vary (Fitzpatrick, 1984, 1987, 1988), it is important for those communications materials to address the relevant needs of particular couples. This study suggests that married adults do vary in their spousal communication patterns related to AATD and that different subgroups have different AATD-related concerns. Indeed, it is possible that these subgroups extend to couples in committed relationships who are not married. Understanding these differences can serve as the foundation for the creation of effective, targeted communications interventions to help couples manage the AATD diagnosis well.

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Appendix

Scale for Desired Spousal Communication

Response options were *not at all, a little, somewhat, moderately,* and *very much*.

1. I want to talk with my spouse about what Alpha-1 is.
2. I want to talk with my spouse about how to treat conditions related to Alpha-1.
3. I want to talk with my spouse about how the Alpha-1 results make me feel.

4. I want to talk with my spouse about whether to share genetic results with insurance companies.
5. I want to talk with my spouse about changing behavior (such as drinking, eating, exercise, or smoking) in order to avoid health conditions related to Alpha-1.
6. I want to talk with my spouse about who else we will tell about the Alpha-1 diagnosis.

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Table 1

*Indicators for Latent Class Analysis (*N = *130)*

Indicators	Code	Label	n	%	*M*	*SD*
Current talk	1	Infrequent	50	38.5	2.45*	0.50
	2	Frequent	80	61.5	3.95*	0.58
Desire conversation	1	Low	54	41.5	2.22*	0.65
	2	High	76	58.5	4.15*	0.64
Talk-Desire Difference	1	Desire more than talk	74	56.9	−.52	0.52
	2	Talk more than desire	56	43.1	0.74	0.63
Decides to tell family	1	Not us	50	38.5	n/a	
	2	My spouse and I together	80	61.5	n/a	
Decides to tell insurance	1	Not us	57	43.8	n/a	
	2	My spouse and I together	73	56.2	n/a	
Decides to tell physicians	1	Not us	74	56.9	n/a	
	2	My spouse and I together	56	43.1	n/a	

*Significantly different from the mid-point of the scale (3) at $p < .05$.

Table 2

Descriptive Statistics and Correlations among Covariates (N = *130)*

	M	*SD*	1.	2.	3.	4.	5.	6.	7.	8.
1. Tested when sick	0.45	0.50	--							
2. Negative affect: AATD	2.15	0.81	−.03	--						
3. Positive affect: AATD	2.04	0.97	−.03	.03	--					
4. Time since diagnosis	9.47	8.05	−.06	−.12	−.07	--				
5. Time with spouse	27.23	14.40	.09	.00	.12	.11	--			
6. Augmentation therapy	0.36	0.48	.31*	−.03	−.02	.15	.12	--		
7. Insurance difficulties	0.22	0.41	−.03	.15	.08	−.10	−.11	−.04	--	
8. Genetic stigma	2.51	0.70	.12	.39*	.12	−.05	.01	−.03	.08	--
9. Gender	0.58	0.50	−.12	.15	−.09	−.01	−.06	−.17	.00	−.16

* $p < .05$

Table 3

Model Fit Information for Comparison of Latent Class Models

Number of classes	G^2	AIC	BIC	*df*
2	125.82	151.82	189.1	50
3	75.3	115.3	172.66	43
4	46.07	100.07	177.5	36
5	**25.94**	**93.94**	**161.44**	**29**
6	20.82	102.82	220.39	22
7	15.21	111.21	248.85	15

Note. Boldface type indicates the selected model. AIC=Akaike's Information Criterion; BIC=Bayesian Information Criterion; *df*=degrees of freedom.

Table 4

Item-Response Probabilities for Five-Class Model Given Latent Class Membership

	Self-determiners	Separates	Conformers	Independents	Traditionals
	19%	17%	14%	20%	30%
Frequent current talk	0.01	0.38	0.49	**0.99**	**0.97**
High desire for conversation	0.47	0.01	0.02	**1.00**	**1.00**
Talk more than desired	0.01	**0.98**	**0.99**	0.32	0.20
We decide to tell family	0.23	0.43	**0.99**	0.34	**1.00**
We decide to tell insurance	0.39	0.16	**0.99**	0.25	**0.93**
We decide to tell physicians	0.20	0.15	**0.66**	0.12	**0.85**

Note. Percentages reflect the number of participants likely to be in each profile. Cells contain the likelihood of agreeing with the concept. Likelihoods over 50% appear in bold.

Table 5

Covariate Analysis with Self-Determiners as the Referent Class

	Separates		Conformers		Independents		Traditionals		
	OR	***B***	***OR***	***B***	***OR***	***B***	***OR***	***B***	**LL²**
Tested when sick	**1.52**	0.42	**97.27**	4.58	0.24	−1.42	0.08	−2.53	13.45
Negative affect: AATD	0.27	−1.30	**7.07**	1.96	0.21	−1.57	0.58	−0.54	18.25
Positive affect: AATD	**1.19**	0.17	**3.88**	1.35	0.64	−0.44	0.73	−0.32	11.79
Time since diagnosis	**1.94**	0.66	0.43	−0.85	0.48	−0.73	0.53	−0.64	11.14
Time with spouse	0.31	−1.18	0.34	−1.07	**1.63**	0.49	**1.58**	0.46	11.74
Augmentation therapy	0.14	−1.96	0.03	−3.48	**3.05**	1.11	**6.27**	1.84	13.39
Insurance difficulties	**4.74**	1.56	0.00	−7.28	**1.82**	0.60	**3.23**	1.17	11.23
Genetic stigma	0.38	−0.96	0.08	−2.51	**1.31**	0.27	**1.17**	0.16	11.00
Gender	0.02	−3.97	0.00	−6.57	**1.00**	0.00	0.85	−0.17	20.44

Note. All covariates significant at $p < .05$. OR = Odds ratio; B = beta estimate.