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Congruence-Incongruence Patterns in Alpha-1 Antitrypsin Deficiency Couples' Genetic Determinist Beliefs and Perceived Control over Genes: Implications for Clinical and Public Health Genomic Communication

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Abstract

Genomics makes possible the isolation of multiple genes as co-factors that increase, but do not determine, risk for many adult-onset medical conditions, including alpha-1 antitrypsin deficiency (AATD). Those diagnosed with an adult-onset medical condition, such as AATD, are often married and make decisions about testing and care as a couple. We examined genetic essentialist and threat beliefs, focusing on beliefs about the genetic contribution to disease susceptibility and severity, as well as perceptions of control related to genes and health for married couples (N =59), in which one spouse has been tested for genetic mutations associated with AATD. The intraclass correlation for spouses' beliefs about genetic essentialism was strong and statistically significant, but the associations for their other beliefs were not. Incongruence between AATD participants and their spouses regarding genes' influence on disease severity directly related to incongruent perceptions of control and genetic contribution to disease susceptibility. Results revealed an inverse relationship to AATD participants' perceptions of behavioral control and a direct relationship to their beliefs about genes' influence on disease severity. This suggests a pattern of

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Human Studies and Informed Consent All procedures performed involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.
Informed consent was obtained from all individual participants included in the study.

Animal Studies This article does not contain any studies with animals performed by any of the authors.

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incongruence in which AATD participants have low levels of perceived control over genes' influence on health and high levels of perceived genetic influence on disease severity compared to spouses. With public health communication efforts lagging behind the science of genomics, insights regarding the congruence or incongruence associated with married couples' beliefs about genes' influence on disease afford pathways to guide clinical and public health communication about genomics.

Keywords

Communication; Perceived control; Couples; AATD; Genetic determinism; Beliefs

Genomics makes possible the isolation of multiple genes as co-factors that increase, but do not determine, risk for many adult-onset medical conditions, including cancer and heart disease, as well as less recognized conditions such as alpha-1 antitrypsin deficiency (AATD) (Topol 2008). The complex interplay of genes in disease is not well understood and scientific uncertainty is a counter-point to beliefs among the lay public that often frame genes as essentialist blueprints for the human experience (Parrott and Smith 2013). Despite the call for efforts to disseminate information to communities (Beskow, Khoury, Baker, and Thrasher 2001), public health communication efforts have lagged behind the science of genomics, contributing to lay audience misunderstanding. This situation exists despite the reality that genetic tests are being included with traditional tests when adults present with particular symptoms. AATD, for example, is a primarily adult-onset medical condition that may present liver or respiratory symptoms and jaundice, leading to testing for an infectious disease (e.g., hepatitis) and AATD. AATD is an autosomal codominant disorder caused by a mutation in the *SERPINA1* gene that leads to low levels of, or no, alpha-1 antitrypsin (AAT) in the blood, as well as lung disease; abnormal AAT can also accumulate in and damage the liver (Laurell and Eriksson 1963). The most common version (allele) of the *SERPINA1* gene, called M, produces normal levels of AAT, while other versions lead to reduced levels of AAT; the S allele produces moderately low levels of this protein, and the Z allele produces very little AATD. Individuals with two copies of the Z allele (ZZ) in each cell are likely to have AATD; AATD predisposes affected individuals to adult-onset diseases that include chronic cirrhosis, lung or liver cancer, obstructive pulmonary disease (COPD), and emphysema (Sharp, Bridges, Krivit, and Freier 1969). The potential to prevent chronic conditions may be realized through genetic testing that is highly sensitive and specific in predicting who does or does not have AATD-related mutations (Ljujic et al. 2008). For those with AATD-related mutations, preventive measures, such as avoiding smoking, excessive alcohol consumption, and/or exposure to environmental air pollutants will reduce AATD's effects (Hogarth and Rachelefsky 2008).

Beliefs about illness causation, including their genetic component, have long been viewed as critical contributors to perceptions of control over health (Orbell, Blair, Sherlock, and Conner 2001). A focus on individual beliefs and perceived control over genes, however, ignores the reality that illness frequently occurs within a family context. Adults may be married and living together when the sequence of diagnostic testing occurs, with spouses of affected adults involved in deciding future courses of action, partly based on their own

beliefs about the role of genes in health. The degree of similarity between spouses' beliefs is referred to as congruence, with agreement ranging from congruent to incongruent. With more people getting genetic test results as co-habiting adults in existing marital relationships, insight is needed into the congruence-incongruence of spouses' beliefs regarding the role of genes related to both the likelihood of disease onset and the genetic influence on disease severity of the condition, as well as perceived control over genes relating to both. The congruence-incongruence in couple beliefs may afford a means to understand attitudes, perceptions of personal risk, and behaviors for conditions associated with AATD, including prevention and detection practices related to quality of life in health and relational domains.

Perceptions of Control Related to Genes and Health

As medical science continues to reveal roles for genes in health across a broad spectrum, the genetic counselor's role in communicating the role genes play in health has become increasingly complex. Counseling for genetic testing within a traditional, single-gene disorder setting has long been focused on the family to guide understanding of and decisions related to the presence of genetic mutations. In an era of ever-widening knowledge about genetic contributors to health conditions, genetic testing is assuming a personalized focus in which physicians order diagnostic tests to tailor treatments. Couple beliefs about genetics and health likely affect responses to such clinical testing, diagnosis, and treatment, with co-habiting spouses of affected patients providing a source of possibly conflicting information about the role of genes in health.

The theory of planned behavior explains that our attitudes, beliefs about what we think our friends and family members expect us to do, and perceived control predict our behavior (Ajzen 1991). It has often been found that what we think our friends or family members expect us to do is not what they actually expect us to do, impeding our thoughts and action (Ajzen). Such incongruence between patient and spouse illness perceptions have been found to predict variance in coping strategies, negative affect, and health-related quality of life (Karademas, Zarogiannos, and Karamvakalis 2010; Merz et al. 2011). Congruence relating to perceptions of personal control, on the other hand, has been found to predict better psychological adjustment (Sterba et al. 2008).

Whether AATD patients or their spouses perceive that they have some control in this regard remains unanswered. Little research has considered the psychological effects of a positive AATD genetic test result from the patient's perspective (Fanos and Strange 2004), and no research has considered how the congruence-incongruence of couple beliefs impact psychological effects. Research has shown that 60 % of smokers surveyed anticipated that they would be motivated to quit smoking if they had a gene linked to smoking-related disease, while 40 % say they would feel *de*motivated (Sanderson and Wardle 2005). As mutations in the *SERPINA1* gene are also associated with smoking-related disease, understanding what beliefs may be linked to perceptions of control versus lack of control, and congruence-incongruence for couples' genetic essentialist and threat beliefs, may aid in explaining such conflicted findings.

Genetic Determinism as Essentialist Beliefs

Essentialism promotes our understanding and guides our perceptions by allowing us to sort characteristics into groups and act based on our beliefs about the essence of a category. Essentialist beliefs regarding the role of genes for health have been identified in beliefs that reflect a tendency to explain humans in terms of a molecular entity (Parrott, Kahl, Ndiaye, and Traeder 2012). Genetic essentialist beliefs linked to health or disease infer an absolute life course, with participants in one study that considered heart disease risk, for example, viewing heart disease caused by genetic risk to be less preventable (French, et al. 2000). Genetic essentialism has been found to be directly related to fatalism (Shen, Condit, and Wright 2009), with fatalism contributing to failure to seek health care via perceptions of lack of control over health (Peek, Sayad, and Markwardt 2008).

Genetic essentialism affords one explanation for the research finding that 35 % of 186 smokers surveyed anticipated being less motivated to quit smoking if they had a positive genetic test for smoking-related diseases (Sanderson and Wardle 2005). The latter group, in the absence of communication to guide understanding about the significant role that smoking plays in addition to any genetic contributors, may form essentialist views about the course of their well-being linked to genes and disease. Online direct-to-consumer (DTC) advertisements for DNA testing use language that may reinforce essentialist beliefs (Nordgren and Juengst 2009). In the absence of clinical and public health communication to inform responses, there is much room for essentialist responses to genetics and genomics.

Genetic Determinism as Threat Beliefs

Genetic determinism, when viewed as perceived threat that links one’s genome to disease susceptibility and severity, promotes understanding that genetic expression relates to personal behaviors and medical intervention (Parrott et al. 2012). Threat beliefs that genes make one more susceptible to disease thus may align with a heightened awareness or belief in the importance of personal behavior together with medical intervention. Some smokers who have been informed about a possible genetic predisposition to tobacco dependence, for example, have been found to increase their requests for cessation treatment (Wright, Weinman, and Marteau 2003). This suggests that a sense of perceived control over genes existed in the situation, contributing to belief in the importance of intervention to increase the likelihood of success in quitting smoking.

Awareness of particular genes' roles in disease may motivate individuals and/or their spouses to choose particular behaviors or environments as intentional strategies to decrease the likelihood of traversing a path associated with inherited genes. While couple congruence in perceived genetic contribution to disease susceptibility and severity has not been examined in any context, the significance of congruence in couple beliefs for other outcomes has been observed in other settings. Couple congruence regarding adult onset of diabetes and its severity, for example, has been found to be positively associated with marital satisfaction (Peyrot, McMurry, and Hedges 1988). In that context, the spouse’s awareness of a diabetes diagnosis contributed to marital satisfaction. In the context of rheumatoid arthritis, when spouse views of level of fatigue and/or physical limitations were

incongruent with patient experiences, patients received ineffective support, including spouses' failure to understand feelings or to provide advice when asked (Lehman et al. 2011). At minimum, these scenarios suggest that congruent beliefs about genes' influence may promote perceived control over genes, while incongruent beliefs will impede these perceptions, contributing to ineffective support.

The Present Study

In this study, we explore three research questions. First, what are the levels of four genetic-related beliefs: genetic essentialism, perceived control over genes, genetic threat as beliefs associated with genes and illness susceptibility, and severity among those with mutations associated with AATD? Second, how much congruence appears between the beliefs of people with AATD and their spouses? Third, do differences in beliefs relate to personal protective practices or experiences with AATD therapy?

Methods

Participants and Procedures

The study protocol was reviewed and approved by the Institutional Review Board at the Pennsylvania State University where the researchers reside. The data were drawn from a larger study; data on a larger set of registered members of the Alpha-1 Research Registry (ARR) located at the Medical University of South Carolina (MUSC) (Smith, Wienke, & Baker, 2014a) and different variables on a smaller set of the spousal pairs (Smith, Wienke, & Coffman, 2014b) have been presented. The procedures are described in detail in those studies, and are briefly described here.

Participants were recruited by email through the ARR in the summer of 2012. The registry included 1,788 members. Registered members who indicated willingness to be contacted for research and provided email addresses were told in the recruitment invitation that we were interested in married couples' experiences with AATD test results; a link was provided to access an online questionnaire using REDCap (http://www.project-redcap.org/).

After giving consent, participants were asked whether or not they had a spouse who could also complete the survey. Of the 219 members who started the survey, 40 were not married, and thus not eligible. Of the 179 eligible members who started the survey, 130 completed it; on average, this took 20 minutes. In the previous study of the couples (Smith et al. 2014a), 58 spouses completed the survey, but only 50 pairs could be matched and were analyzed. Since that time, more spouses completed the survey and we were able to complete more matches, resulting in 59 couples available for analysis in this study. One reminder was sent out to registry members. No incentives were provided. Participants completed measures related to genetic beliefs, including perceived genes' influence on disease severity and susceptibility, genetic essentialism, and perceived control over genes related to genes and health. They also provided information about health insurance coverage, current health status and behaviors, and demographic information including self-reported biological sex, self-reported race, employment status, and age.

Power analysis

The analysis included chi-square tests, paired-sample t-tests, and correlations. Using the paired-sample *t*-test, with 59 couples, Cronbach's alpha=.05, and power=.80, and two-tailed tests, we had the power to detect effect sizes (*d*)=0.37 and higher. Cohen (1988) identified medium effect sizes at *d*=.50.

Measures

For all scales used to form measures of genetic essentialism, perceived behavioral control, and beliefs about genetic contribution to disease susceptibility and severity, the response options ranged from "1" = "Strongly Disagree" to "5"="Strongly Agree". Items for each measure were summed and averaged into one score, and assessed via use of Cronbach alpha coefficient to judge internal consistency for reliability, with .70 and greater considered to be adequate (Cortina 1993). Genetic essentialism was assessed via nine Likert-type items (see Table 1), six based on Parrott et al. (2012) and three added based on the context; α=.81; higher scores indicated stronger beliefs that human life and health are solely determined by genes. Perceived behavioral control over the role of genes for health was assessed via seven Likert-type items (see Table 1) based on Parrott et al. (2012); α=.90; higher scores indicated stronger belief in control over the role of genes in health. Seven Likert-type items (see Table 1) based on Parrott et al. (2012) were used to assess beliefs about genes' role in disease susceptibility; α=.69; higher scores indicated stronger beliefs that genes determine susceptibility to disease. Four Likert-type items (see Table 1) based on Parrott et al. (2012) were used to assess beliefs about genes' role on disease severity; α=.70; higher scores indicated stronger beliefs that genes determine the severity of disease.

Participants with AATD and their spouses were also asked about prevention practices, indicating if on a typical day, they: eat five fruits and vegetables, get 30 minutes of exercise, smoke one or more cigarettes, or drink one or more glasses of alcohol. Participants with AATD were asked if they had experienced any of the following in the past year: liver disease, COPD, augmentation therapy, and/or supplemental oxygen therapy.

Data analysis

Having established the reliability of the scales used to operationalize the belief sets, each matched couple's responses were entered as a case using SPSS 21, affording us 59 couple cases to examine. Descriptive item and scale statistics were obtained for participants with AATD and their spouses separately to describe overall patterns in beliefs. Paired sample t-tests and intraclass correlations were calculated to provide comparisons between participants with AATD and their spouses relating to perceived control over genes, and genetic essentialist and threat beliefs. Separate bivariate correlations were also obtained for the belief sets associated with the participants with AATD and their spouses to examine possible relatedness among their belief sets. Absolute value difference scores between the participant's with AATD scale mean response and their spouse's scale mean response were calculated for genetic essentialist, genetic contribution to disease susceptibility, genes' influence on disease severity, and perceived control over genes, with higher numbers indicating greater incongruence between the participant with AATD and spouse scores;

these scores were correlated with the participant's with AATD and their spouse's beliefs. Independent samples t-tests were calculated to evaluate relationships between preventive behaviors and health experiences on participants with AATD and their spouses' beliefs.

Results

The analysis for this study was based on 59 couples matched from the online survey, including 59 participants with AATD (56 ZZ and 3 carriers) and their 59 spouses, with an overall mean age of 58.28 (*SD*=9.45). The average age of the 59 participants with AATD was 58.57 years (*SD*=9.30); ages ranged from one 31-year-old to two 74-year-olds; seven 60 year-olds participated. Table 2 displays additional demographic information for the participants with AATD. Table 3 displays prevention practices among participants with AATD and their spouses; in addition, 32 participants with AATD reported experiencing COPD in the past year, seven experienced liver disease (one received a liver transplant and a double lung transplant), and two were on a transplant wait list to receive a lung. Also, 34 had received augmentation therapy in the past year and 25 received supplemental oxygen therapy.

Perceptions of behavioral control

The overall mean for the perceived behavioral control related to genetics beliefs' scale for participants with AATD was 2.95 on a 5-point scale (*SD*=.95) and, for their spouses, 2.86 (*SD*=.82). The intraclass correlation was insignificant for the couple, r=.00, p=.99, indicating that their perceptions were not related. No significant differences were found between participants with AATD who reported eating the recommended amount of fruits and vegetables each day and those who did not on perceptions of perceived control over genes. On the other hand, for participants with AATD who reported exercising at least 30 minutes on a typical day had higher levels (t[57]=2.20, p<.05) of perceived control over genes (*M*=3.12, *SD*=.94) compared to those who did not (*M*=2.55, *SD*=.87). The absolute value scores calculated for incongruence between participants with AATD and their spouses in perceptions of perceived control over genes were positively related to incongruence regarding the genes' influence on disease severity of a condition (r=.28, p<.05). No other significant relationships were observed with regard to these calculated differences and perceived behavioral control. As summarized in Table 4, participants with AATD and spouses' perceptions of behavioral control showed no relationship to essentialist or threat beliefs for self or other.

Genetic essentialist beliefs

The overall mean for the genetic essentialist beliefs scales for participants with AATD was 2.77 (*SD*=.68) and, for their spouses, 2.85 (*SD*=.60) on a 5-point scale. The intraclass correlation was strongly significant for the couple, r=.46, p<.001, indicating overall, within-couple consistency in these beliefs. A review of the responses in Table 1 reveals some incongruence, however: almost twice as many spouses as compared to participants with AATD agree that "genes are the most important factor in determining a person's health." The absolute value score calculated for incongruence between AATD participants' and

spouses' essentialist beliefs showed no significant relationships to other incongruence scores or genetic beliefs for self or other.

As summarized in Table 4, the genetic essentialist beliefs were strongly related to their own beliefs that genes determine disease susceptibility and severity, as well as their spouses' essentialist beliefs—as already noted by the intraclass correlation, but not their spouses' beliefs that genes determine disease susceptibility and severity. Spouses' essentialist beliefs directly related to their beliefs that genes determine disease susceptibility and severity, as well as participants with AATDs' beliefs that genes determine disease severity. Put differently, spouses' essentialist beliefs showed intrapersonal and interpersonal associations with beliefs that genes determine disease severity. The effects of preventive practices revealed no significant effects for essentialist beliefs associated with self or the spouse. Findings did, however, reveal higher levels (t [57]=2.02, p<.05) of genetic essentialism for participants with AATD who had experienced COPD in the past year (n=32; M=2.93, SD=.68) compared to those who had not (n=27; M=2.58, SD=.66).

Genes' role in disease susceptibility beliefs

The overall mean for beliefs about genes' role in disease susceptibility for participants with AATD was 3.99 (SD=.42) and for their spouses, 3.88 (SD=.43). However, the intraclass correlation was not statistically significant for the couple, r=.06, p=.68, showing their beliefs have no more consistency than if we paired one spouse with a stranger. An interpretive review of the descriptive report in Table 1 may offer some insight into this finding, as the items added for the AATD context showed spouses more likely to agree with the statement "genes make some people more likely to benefit from medicine than others" and participants with AATD more likely to strongly agree with the statement "genes cause each person's body to react differently to things in the environment."

Participants with AATDs' beliefs in genes' contribution to disease susceptibility (see Table 4) directly related to their own essentialist beliefs and belief that genes influence disease severity; spouses' beliefs in genes' role on disease susceptibility were positively related to their own essentialist beliefs. Greater difference between a participant with AATD and their spouse's belief that genes increase disease susceptibility was positively related to greater difference in beliefs that genes increase disease severity (r=.35, p<.01).

No significant differences were found between participants with AATD who reported eating the recommended amount of fruits and vegetables each day, or exercising 30 minutes or more, and those who did not for beliefs about genes' influence on disease susceptibility associated with self or the spouse. Higher levels of perceived genetic contribution to disease susceptibility were observed (t [57]=2.37, p<.05) among the 32 participants with AATD who experienced COPD within the past year (M=4.11, SD=.41) compared to those who had not (M=3.86, SD=.40). Participants with AATD who had augmentation therapy within the past year (n=34) exhibited higher levels of belief in the genetic contribution to disease susceptibility (t [57]=2.61, p=.01; M=4.11, SD=.42) compared to AATD participants who had not (M=3.83, SD=.37). For participants with AATD who had supplemental oxygen therapy within the past year (n=25), their spouses exhibited lower levels (t [57]=2.79, p<.01) of perceived genetic contribution to disease susceptibility (M=3.70, SD=.35) compared to

participants with AATD who had not had supplemental oxygen therapy in the past year (*M*=4.00, *SD*=.45).

Genetic influence on disease severity beliefs

The overall mean for the genetic influence on disease severity beliefs' scales for participants with AATD was 3.84 (*SD*=.48) and, for spouses, 3.80 (*SD*=.53) on a 5-point scale. The intraclass correlation was not statistically significant for the couple, *r*=.19, *p*=.15. As already noted, incongruence between participants with AATD and spouses' beliefs about genes' influence on disease severity was related to incongruence in beliefs about the genetic contribution to disease susceptibility and incongruence in perceived behavioral control. Incongruence in genes' influence on disease severity perceptions was inversely related to participants with AATDs' perceptions of behavioral control (*r*=−.27) and direct relationships to beliefs about genetic contribution to disease susceptibility (*r*=.28), genes' influence on disease severity (*r*=.32), and spouses' genetic essentialist beliefs (*r*=.28).

Participants with AATDs' beliefs about genes' influence on disease severity related to their essentialist and disease susceptibility beliefs (see Table 4), and related to spouses' essentialist beliefs, though not spouses' beliefs about the genetic component of disease susceptibility or severity. Spouses' beliefs regarding genes' influence on disease severity directly related to their beliefs about genes' role in disease susceptibility and genetic essentialist beliefs, but showed no significant relationships to participants with AATD threat and essentialist beliefs.

Discussion

Incongruence between participants with AATD and their spouses in relation to beliefs about perceived control over genes and genetic threat, including beliefs about the genetic contribution to disease susceptibility and severity, emerged in both explicit and implicit ways. First, the intraclass correlations showed that couples evidenced some consistency in their genetic essentialism beliefs, but virtually none in their perceptions of control over the role of genes for health, or beliefs about the genetic contribution to disease susceptibility or severity. Second, mean levels of particular beliefs differed between spouses. Third, while essentialism and genetic threat beliefs showed similar intrapersonal patterns for participants with AATD and their spouses, the intrapersonal patterns differed: specifically, spouses' essentialism beliefs predicted participants with AATD beliefs about how genes influence disease severity beliefs but not their own beliefs about how genes influence disease severity.

Practice Implications

Incongruence in beliefs about genes' influence on disease severity provides the most direct evidence to support the need for intervention in this context. Incongruent beliefs about genes' influence on disease severity directly related to incongruence in perceived behavioral control; divergence between spouses in beliefs about genes' influence on disease severity beliefs widened the gap in their perceptions of perceived control over genes' role for health. The pattern for incongruence aligns with participants with AATD having lower levels of perceived control over genes and higher levels of perceived genes' influence on disease

severity, compared to spouses. This could set in motion learned helplessness on the part of participants with AATD or potential blame from their spouses, and suggests norms in this context, with spouses wanting patients with AATD to do more to control their risk for AATD. It may also offer insights into the existence of caregiver burden as spouses assist with supplemental oxygen therapy and/or attend augmentation therapy sessions.

For both clinical and public health genomic communication endeavors, findings suggest the need for greater clarity regarding the spouse's experiences and possible paths to delay or avert severe effects. Some insight into one such path was revealed by the finding that 20 % of participants with AATD reported drinking at least one glass of alcohol each day, while 25 % of spouses drank; only four couples revealed congruent drinking behaviors. In the incongruent couples, spouses who drank may view AATD as less severe owing to the spouses' habit or blame the spouse with AATD for the severity of their condition; for participants with AATD whose spouses drank, their own disease severity perceptions may have been heightened by a daily reminder of something they may have given up due to their condition. In view of the effects of AATD on liver function and disease, avoiding alcohol as a strategy to prevent disease and limit the genes' influence on disease severity emerges as an important strategic message and immediate area for public health genomic communication research.

Only one participant with AATD, and two unassociated spouses, indicated smoking one or more cigarettes each day, perhaps owing to the reality that breathing impairment may be more easily seen and felt than effects on the liver. Further reinforcing this notion, AATD participants' experiences often predicted their beliefs, as those who had experienced COPD in the past year—more than half the participants—held stronger genetic essentialist and disease susceptibility beliefs. Participants with AATD who had augmentation therapy within the past year also exhibited higher levels of perceived genetic contribution to disease susceptibility. These experiences emphasize a finding based on social cognitive theory that behavior arises from a reciprocal triadic relationship among environmental determinants, personal determinants, and behavioral determinants (Bandura 1986).

Further reinforcing the importance of performance accomplishment linked to the effects of AATD, individuals who exercised regularly reported more perceived control over genes than those who did not. The latter highlights the need to build feelings of self-efficacy, which originates from verbal persuasion, performance accomplishment, vicarious performance—or observing another person successfully accomplishing an activity, and physiological arousal (Bandura 1986). Notably, 24 couples shared daily exercise as a common habit. While verbal persuasion contributing to knowledge may importantly guide knowledge, self-efficacy mediates the relationship between knowledge and behavior (Rimal 2000). Sometimes, however, if individuals are given the knowledge that their condition will improve through some means, then the individual may convince oneself that they are able to cope with their condition (Bandura 1986). This may explain the finding that spouses of the 25 diagnosed participants who had supplemental oxygen therapy within the past year exhibited lower levels of perceived genetic contribution to disease susceptibility; perhaps they diminished genes' role to disease susceptibility with the knowledge that their diagnosed spouses could cope through the administration of oxygen. This result points to the

possibility that strategies for coping with AATD may not be well-understood by spouses. What is the role that the spouse plays in either augmentation therapy situations or supplemental oxygen—are they present? These remain as questions to be addressed toward better understanding the incongruence identified in this research.

Limitations and Research Recommendations

Future research should continue to examine the congruence-incongruence of AATD couples' beliefs, as well as the experiences of couples associated with other adult-onset medical conditions. These results are based on 59 couples, largely middle-aged, recruited from an AATD registry; sample size, age and income level of participants, as well as exclusion of those outside of cohabiting marriage may limit the generaliz-ability of the results (Gibson-Davis, Edin, and McLanahan 2005). It will be useful to build on these data to guide research into communal coping strategic efforts, including survey questions tailored to AATD/AATD-related diseases to broaden practice implications. In designing clinical and public health genomic communication and programs, communal coping should be examined, as it focuses on pooling couples' resources to problem solve together in efforts to overcome stressors and adversity (Lyons, Mickelson, Sullivan, and Coyne 1998). This direction for future research regarding couples experiencing AATD and other couples with adult-onset medical conditions may reveal benefits, and complement clinical advice and health promotion.

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Table 1

Means, Standard Deviations, and Frequencies of Response for Beliefs about Genetics and Health

Item (Responses on a 1 – 5 scale)	% Strongly Disagree		% Disagree		% Neither Agree Nor Disagree		% Agree		% Strongly Agree		Item Mean (Standard Deviation)	
D=Diagnosed spouse (*N*=59) S=Undiagnosed spouse (*N*=59)	D	S	D	S	D	S	D	S	D	S	D	S
Genetic essentialism												
1. Genes are the most important factor in determining a person's health.	5.1	0.0	28.8	23.7	27.1	18.6	27.1	50.8	11.9	6.8	3.12 (1.12)	3.41 (.93)
2. Genes are more important than one's own behavior in determining one's health.	13.6	3.4*	47.5	50.0*	23.7	31.0*	13.6	13.8*	1.7	1.7*	2.42 (.95)	2.60 (.84)*
3. Genetics play a bigger role in whether you will get liver disease than your lifestyle.	5.1	5.1	44.1	42.4	22.0	28.8	25.4	20.3	3.4	3.4	2.78 (1.00)	2.75 (.96)
4. The genes one is born with determine how healthy one will be throughout life.	5.1	3.4	35.6	32.2	32.2	28.8	23.7	32.2	3.4	3.4	2.85 (.96)	3.00 (.97)
5. Genes determine the effects of one's own behavior in determining one's health.	22.0	8.8**	25.4	33.3**	22.0	17.5**	28.8	36.8**	1.7	3.5**	2.63(1.17)	2.93 (1.10) **
6. Genes determine whether medication works.	10.2	3.4*	28.8	37.9*	37.3	31.0*	23.7	25.9*	0	1.7*	2.75 (.94)	2.84 (.91)*
7. Genes determine whether smoking leads to lung problems.	11.9	15.3	32.2	37.3	15.3	15.3	33.9	30.5	6.8	1.7	2.92 (1.19)	2.66 (1.12)
8. Genes determine whether drinking leads to liver problems.	10.5**	8.6*	33.3**	41.4*	19.3	19.0*	35.1**	29.3*	1.8**	1.7*	2.84(1.08)**	2.74 (1.04)*
9. Genes are the most important contributor to human health.	8.6*	0.0	39.7*	47.5	27.6*	32.2	22.4*	18.6	1.7*	1.7	2.69 (.98)*	2.75 (.82)
Perceived control												
10. Good nutrition can keep human genes healthy.	5.1	6.9*	23.7	27.6*	20.3	24.1*	39.0	39.7*	11.9	1.7*	3.29 (1.12)	3.02 (1.02)*
11. Living a healthy lifestyle keeps a person's genes healthy.	6.8	6.8	28.8	30.5	16.9	20.3	42.4	35.6	5.1	6.8	3.10 (1.10)	3.05 (1.11)
12. Your own behaviors can protect your genes.	10.2	5.2*	22.0	25.9*	18.6	22.4*	40.7	43.1*	8.5	3.4*	3.15 (1.17)	3.14 (1.02)*
13. Your own behaviors can change your genes.	27.1	22.0	42.4	59.3	11.9	6.8	16.9	10.2	1.7	1.7	2.24 (1.09)	2.10 (.92)
14. Eating healthy protects your genes.	11.9	6.8	22.0	28.8	22.0	20.3	37.3	40.7	6.8	3.4	3.05 (1.17)	3.05 (1.06)
15. Quitting smoking can make your genes healthier.	13.6	11.9	33.9	37.3	15.3	15.3	27.1	23.7	10.2	11.9	2.86 (1.25)	2.86 (1.25)
16. Drinking alcohol can damage your genes.	13.6	8.5	27.1	42.4	20.3	15.3	30.5	27.1	8.5	6.8	2.93 (1.22)	2.81 (1.14)

Item (Responses on a 1 – 5 scale)	% Strongly Disagree		% Disagree		% Neither Agree Nor Disagree		% Agree		% Strongly Agree		Item Mean (Standard Deviation)	
Genetic susceptibility												
17. My genes make future illness more likely for me.	0.0	0.0	10.2	16.9	6.8	18.6	61.0	50.8	22.0	13.6	3.95 (.84)	3.61 (.93)
18. Genes make some people more likely to become ill from germs.	1.7	0.0	10.2	10.2	8.5	10.2	67.8	69.5	11.9	10.2	3.78 (.85)	3.80 (.76)
19. Genes cause each person's body to react differently to things in the environment.	0.0	0.0	0.0	5.1	3.4	8.5	72.9	78.0	23.7	8.5	4.20 (.48)	3.90 (.61)
20. Genes make some people more likely to get liver disease.	0.0	0.0	1.7	0.0	6.8	5.1	66.1	79.7	25.4	15.3	4.15 (.61)	4.10 (.44)
21. Genes make some adults more likely to get COPD.	0.0	3.4	5.1	3.4	5.1	6.8	55.9	66.1	33.9	20.3	4.19 (.75)	3.97 (.85)
22. Genes make some people more likely to benefit from medicine than others.	0.0	0.0	10.2	10.2	39.0	20.3	42.4	61.0	8.5	8.5	3.49 (.80)	3.68 (.78)
23. Genes make some people more likely to become ill when exposed to toxins in the environment (like second-hand smoke).	0.0	0.0	3.4	1.7	5.1	5.1	61.0	76.3	30.5	16.9	4.19 (.68)	4.08 (.54)
Genetic severity												
24. Genes play a very important role in the health of children.	0.0	0.0	0.0	1.7	8.5	13.6	67.8	71.2	23.7	13.6	4.15 (.55)	3.97 (.59)
25. Genes influence how serious an illness can be.	0.0	0.0	1.7	3.4	13.6	11.9	64.4	71.2	20.3	13.6	4.03 (.64)	3.95 (.63)
26. My genes are the best predictor for how badly a disease will affect me.	1.7	3.4	23.7	16.9	42.4	40.7	27.1	28.8	5.1	10.2	3.10 (.89)	3.25 (.98)
27. Genes make disease more severe for some people.	0.0	0.0	0.0	3.4	10.2	11.9	71.2	71.2	18.6	13.6	4.08 (.54)	3.95 (.63)

Notes:

* 1 participant did not complete this question;

** 2 participants did not complete this question

Table 2

Demographic Characteristics for Participants with AATD

Characteristic	n	%
Sex[a]		
Female	30	50.9
Male	29	49.2
Self-identity		
White	57[b]	96.6
American Indian or Alaskan	1	1.7
No response	1	1.7
Have children		
Yes	52[c]	88.1
No	7	11.9
Employed		
Yes	24	40.7
No	35	59.3
Health insurance		
Employment based	29	49.1
Individual policy	10	17.0
Medicare[d]	20[d]	33.9

[a]The roughly equal split between males and females provided an opportunity to compare the experiences and beliefs of couples based on the biological sex of the AATD spouse using independent samples t-tests; no significant differences were found between groups

[b]Consistent with AATD-related genetic mutations in the U.S. (de Serres, Blanco, and Fernandez-Bustillo 2010)

[c]Thirty-five indicated their children had been tested for AATD

[d]Eight reported that they had Medicare medical

Table 3

Prevention Practices for the Participants with AATD and Spouses

	Participant with AATD	Spouse
Prevention practice	*n* (%)	*n* (%)
Exercise 30 or more minutes per day	41 (69.5)[a]	34 (57.6)
Eat 5 servings of fruits and vegetables per day	37 (62.7)[b]	34 (57.6)
Drink 1or more glasses of alcohol per day	12 (20.3)[c]	15 (25.4)
Smoke 1 or more cigarettes per day	1 (1.7)	2 (3.4)

[a] Twenty-four of these had spouses who exercised ($\chi 2[1, 59]=.05$, $p=.83$)

[b] Twenty-two of these had spouses who ate 5 servings of fruit and vegetables per day ($2[1,59]=.14$, $p=.71$)

[c] Four of these had spouses who indicated drinking 1 or more glasses of alcohol per day ($\chi 2[1, 59]=.50$, $p=.49$)

Table 4

Pearson correlations between measures

Measure	1.	2.	3.	4.	5.	6.	7.	8.
Genetic essentialism	1							
Perceived control	.057	1						
Genetic susceptibility	.426**	−.177	1					
Genetic severity	.529**	.012	.569**	1				
Spouse genetic essentialism	.460**	.061	.255	.346**	1			
Spouse perceived control	.079	−.002	.129	.107	−.113	1		
Spouse genetic susceptibility	.032	−.016	.055	.101	.260*	−.238	1	
Spouse genetic severity	.151	−.008	−.016	.192	.279*	.127	.625**	1

Notes:

* $p<.05$;

** $p<.01$ (2-tailed)