

Genetic susceptibility testing for Alzheimer disease: Motivation to obtain information and control as precursors to coping with increased risk

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Abstract

Objective: This study investigated appraisals, including motivation, and coping preferences for undergoing Apolipoprotein E (APOE) susceptibility testing for Alzheimer disease (AD).

Methods: Participants were 60 adult children of individuals affected with AD enrolled in a trial investigating use and impact of APOE susceptibility testing. An exploratory qualitative study was undertaken in which participants were interviewed about their testing experience.

Results: Most participants viewed genetic testing as providing valuable information that could help direct future health care decisions and meet their emotional concerns about living at increased risk. Participants related their motivation for genetic testing to their worries about developing AD, preference to seek information about health threats, and need to feel in control of their health.

Conclusion: Even without prevention or treatment options, genetic testing may be a useful coping strategy for some at-risk individuals.

Practice implications: Once testing becomes clinically available, practitioners need to address the value and limitations of testing as well as appraisals and efforts to cope.

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1. Introduction

Alzheimer disease (AD) is a progressive dementia characterized by declining levels of cognitive function, leading ultimately to disability and death. It is estimated to affect up to 1% of Americans by age 65 year and up to 18% of Americans aged 85 year and older [1]. Approximately, four million people are living with AD in the United States today, and AD is expected to cause even greater social and

financial burdens in the years to come [1]. Investigations into genetic contributions to AD have found a common polymorphism in the Apolipoprotein E (APOE) gene, the $\epsilon 4$ allele, to be associated with later-onset AD [2]. In general, compared to individuals with the common $\epsilon 3/\epsilon 3$ genotype, individuals with one copy of the $\epsilon 4$ allele are approximately three times more likely to develop AD, and those with two copies of the $\epsilon 4$ allele are approximately 15 times more likely to develop AD [3]. The risk of developing AD for a given individual with one or two $\epsilon 4$ alleles varies with age, sex and ethnicity [3,4].

The APOE $\epsilon 4$ allele is neither necessary nor sufficient for the development of AD, and other genetic and environmental factors contribute to the disease. The low sensitivity

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and specificity of APOE testing has led the American Geriatrics Society [5], the National Institute on Aging and the Alzheimer’s Association [6], the American College of Medical Genetics [7], and others [8,9] to issue statements cautioning against the current use of APOE genetic testing to assess future risk of AD. Given the low predictive value of the test, the lack of proven cures or prevention for AD, and the concern that individuals may have difficulty accurately interpreting the results and limitations of APOE-based risk assessment, the APOE genetic test is not currently recommended for asymptomatic individuals [5–7].

Despite these cautionary statements, studies investigating the hypothetical uptake of APOE susceptibility testing by members of the general public [10] and people with a first degree relative with AD [11] have found some individuals are interested in susceptibility testing as a way to help “plan for the future.” One survey of 171 community physicians found 15% had had an asymptomatic patient request APOE testing and 5% reported ordering testing [12]. A study (REVEAL see p. 5) of 149 at-risk individuals found APOE ε4-positive test results did not significantly raise personal perceptions of AD risk, and ε4-negative results were not falsely reassuring [13]. These studies suggest there is interest in APOE testing and that accurate assessment of test results is possible. Yet the usefulness of APOE testing remains largely unknown.

A health behavior model that includes motivation for susceptibility testing could help direct patient education and counseling for this type of testing in the future. Coon et al. proposed using the Transactional Model of Stress and Coping (TMSC) to understand how individuals cope with APOE genetic susceptibility testing [14]. The model, originally developed by Lazarus and Folkman in 1984 from empiric studies of human responses to life stressors, has been applied to a variety of health-related stressors [15]. According to the model (Fig. 1), individuals gauge threats to their health based on perceptions of their susceptibility to a given illness, the severity of the illness and its consequences, the causes of the illness, and their ability to control the illness and their emotional responses to it. How individuals then cope with health threats is shaped by personal coping patterns, such as the tendency to seek out information in times of stress, as well as emotional and social resources.

Several researchers have suggested that undergoing genetic testing may be one way individuals at-risk for developing illnesses such as cancer and Huntington disease cope with their risk for disease [16–18]. The body of research on genetic testing for Huntington disease, a debilitating neurodegenerative disorder that is also neither preventable nor curable, has found testing helps some people cope emotionally by reducing their uncertainty and focusing them on planning for the future [19]. While APOE genetic

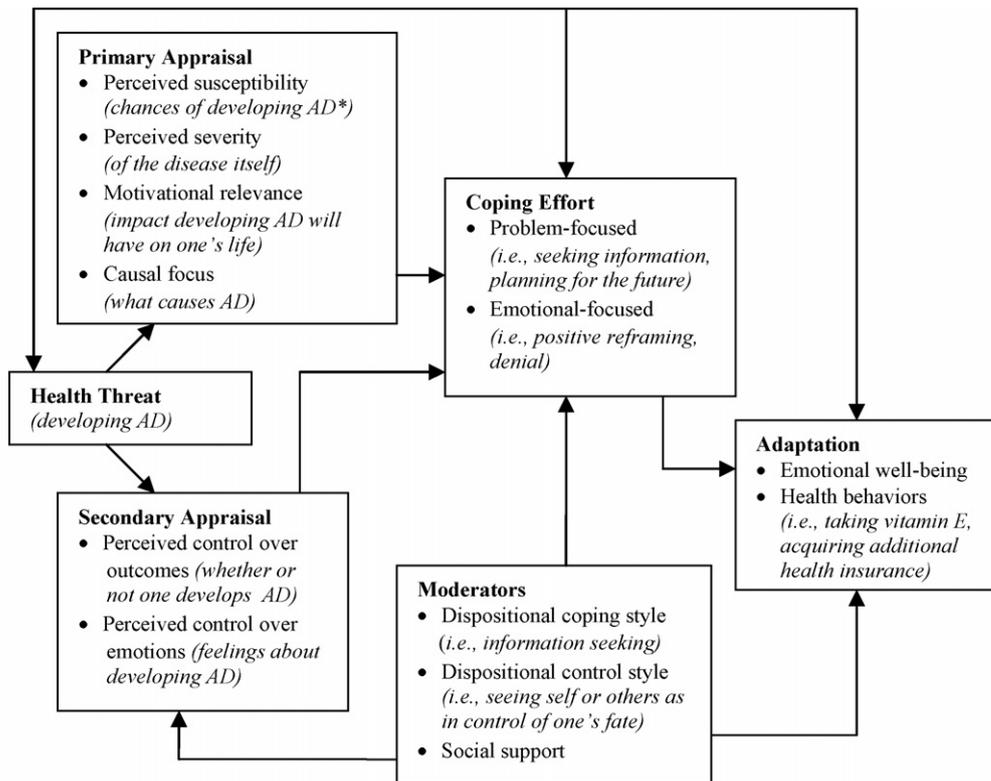


Fig. 1. Transactional Model of Stress and Coping. *AD = Alzheimer disease. Adapted and modified from Lazarus and Folkman [15]. Original model domains are in regular font; applications to those at-risk of developing AD are shown in italics. Arrows indicate the transactional nature of the model, such that appraisals, moderators, coping efforts, and adaptations feedback upon and influence each other.

testing for AD may provide similar benefits, the later onset of AD and the less predictive value of the APOE test create significant differences from genetic testing for Huntington disease. These differences will likely affect how individuals evaluate the usefulness of APOE genetic testing for coping with being at-risk for developing AD.

This descriptive study explores the use of genetic testing as a way to cope with the threat of being at increased risk for AD. We present the experiences of a group of individuals enrolled in a study that included APOE testing as a component of AD risk assessment. A qualitative approach was chosen in order to better understand participants' desire for genetic testing from within the framework of their beliefs about Alzheimer disease and their personal coping styles. Based on the theoretical assertions of the TMSC and prior research on genetic testing and coping [20], we expected individuals to relate their decision to have genetic testing to being worried about their susceptibility to AD; and to describe themselves as preferring to seek out information about threats to their health and as feeling able to cope with learning their results. By studying APOE genetic susceptibility testing, we aimed to characterize individuals' motivation to use genetic susceptibility testing in the future to cope with their risk for developing a variety of adult onset diseases.

2. Methods

2.1. Participants

Participants were adult children of people with AD enrolled in the Risk Evaluation and Education for Alzheimer Disease (REVEAL) study. REVEAL study enrollment, proceedings, and results have been published elsewhere [4,21–23]. REVEAL participants were randomized to receive risk counseling based on their family history and APOE genetic result (intervention group) or family history alone (control group). The 60 individuals who participated in this study, including four people who had declined to complete the REVEAL parent study, participated in the REVEAL Qualitative Research Initiative (REVEAL-QRI), a secondary study which used in-depth interviews to further understand the APOE genetic testing experience.

2.2. Procedures

The REVEAL-QRI team included anthropologists, psychologists, genetic counselors, physicians and nurses affiliated and unaffiliated with the parent REVEAL study. We developed a semi-structured interview guide to address participants' experiences with AD and with the APOE genetic testing process. Participants in the parent study who had received their risk assessment and completed the 12-month follow-up questionnaire were considered eligible for

interview. Genetic counselors from each of the three study sites phoned individuals who they perceived to be willing participants based upon previous interactions during the parent study; no one was excluded from participation in the qualitative study. One hundred and thirty-one individuals were contacted and 56 (43%) agreed to be interviewed. Four additional people who had declined to complete the parent study were also interviewed. Interviews were conducted in-person with participants at each of the three parent study sites (Boston University, Case Western Reserve University, Cornell University) by one of four investigators from the REVEAL-QRI group. Interviews were conducted between December 2002 and January 2004, lasted approximately 1 h, and were audiotaped with the written consent of the participant. Both the parent study and the REVEAL-QRI were approved by the institutional review boards at each study site.

2.3. Data collection

The interview guide was developed from contributions of all REVEAL-QRI team members and contained open-ended questions about personal experiences with AD and participation in REVEAL. Specific domains of the TMSC were addressed with questions regarding participants' perceived likelihood of developing AD; beliefs about the causes of AD; perceived control over AD; motivations for participation in REVEAL; and reactions to receiving results. Because all interviews were conducted many months after participants had received genetic testing and/or family history based risk assessments, participants' comments were all retrospective.

2.4. Data analysis

For this analysis, each of the 60 transcripts was initially read once for content by the principal researcher (HG). Through this reading, it was established that 11 domains from the TMSC were demonstrated in the interviews. A list of specific themes pertaining to each of the 11 general domains was generated through the reading of the transcripts. These themes comprised the original codebook for qualitative analysis. Transcripts were imported as text files into the QSR NVIVO Version 2.0 program (QSR International Pty, Ltd., 1999–2002) for coding and analysis.

Twenty transcripts were then selected at random and coded by the principal researcher (HG) and a second coder (EL). Transcripts were coded inductively; i.e., codes were applied to areas of text based on the interpretation of individual coders. The unit of analysis was thematic; i.e., a code was applied each time a given theme was mentioned by a participant. For the first four transcripts coded, the average interrater reliability for a given code was 60%. The codebook was then revised to reduce ambiguity before a second group of transcripts was coded.

This process was repeated four times until 20 transcripts were coded and interrater reliability reached 76%. All disagreements were reconciled through discussion between the principal researcher and the second coder. The remaining 40 transcripts were then coded by the principal researcher (HG). Data reports were generated using the QSR NVIVO 2.0 software. Data from the disease susceptibility, perceived threat, dispositional coping/control styles, coping effort and emotional adaptation domains are reported.

3. Results

3.1. Participant information

The majority of the 60 participants in the REVEAL-QRI study were Caucasian, college-educated, and of a high socioeconomic status (Table 1). The mean age, ethnicity, educational background, marital status and income of the qualitative interview participants were similar to those of the parent REVEAL study. Seventeen participants had learned they carried the $\epsilon 4$ allele (a “positive result”), 24 had learned they did not (a “negative result”), 15 had received a family history based risk assessment (the “control group”) and 4 had declined to complete the parent study. The qualitative study had slightly more female participants, participants with negative genetic testing

Table 1
Demographic characteristics of participants in the REVEAL study and participants in the REVEAL Qualitative Research Initiative

Demographic characteristic	All REVEAL participants (n = 162)	Qualitative participants ^a (n = 60)
Mean age in years (range)	53 (30–78)	54 (37–76)
Sex, % female (N)	72% (117)	87% (52)
Ethnicity, % Caucasian (N)	94% (152)	95% (57) ^b
Marital status, % married (N)	65% (105)	63% (37)
Number of affected relatives		
One, % (N)	42 (68)	48 (29)
Two or more, % (N)	58 (94)	52 (31)
Mean education in years (range)	17 (10–22)	17 (12–21)
Median income bracket	\$70,000–99,999	\$70,000–99,999
Results		
$\epsilon 4$ positive, % (N)	33 (53)	28 (17)
$\epsilon 4$ negative, % (N)	36 (58)	40 (24)
Control, % (N)	32 (51)	25 (15)
Declined, % (N)	N/A	7 (4) ^a
Site, % at (N)		
Boston University	35 (56)	42 (25)
Cornell University	34 (55)	25 (15)
Case Western University	32 (51)	33 (20)

^a Includes four participants who declined to continue with genetic testing in the parent REVEAL study following the introductory educational session.

^b Two individuals self-identified as African–American and one as Hispanic.

results, and participants from Boston University, than the parent study.

3.2. Perceived susceptibility to AD and desire for genetic testing

Forty-six participants (77%; 15 $\epsilon 4$ negative, 16 $\epsilon 4$ positive, 16 control/declined) discussed feeling at increased risk for AD based on their family history of the disease. Some participants described focusing on their own risk for AD as soon as their parent was diagnosed or became debilitated ($n = 10$). Many individuals with multiple affected family members felt they were at very high risk because of their “family tree” ($n = 12$), while one participant rationalized that she was less likely to get the disease because only her mother had AD. Some ($n = 7$) felt they were at greater risk for the disease because they shared physical or behavior traits with their affected parent, while others ($n = 2$) focused on how different they were from their affected parent in an attempt to distance themselves from the disease. Ten participants directly linked their concerns about their family history of AD to their desire to know their own APOE genetic status. Quotations illustrating participants’ perceived susceptibility to AD are in Box 1.

3.3. Threat of developing AD and desire for genetic testing

Twenty-nine participants (48%; 13 $\epsilon 4$ negative, 9 $\epsilon 4$ positive, 7 control/declined) said that they were frightened of or worried about developing AD, indicating that they felt threatened by their increased risk. According to the TMSC, perceiving oneself as susceptible to a given disease is only part of what makes that disease threatening. Participants also related their fear of being a burden on loved ones to their concerns about AD and desire for genetic testing ($n = 17$), especially if they had had a difficult time caring for their own parent with AD or were still contemplating having children ($n = 2$). Others ($n = 6$) were afraid of having their careers cut short or being unable to enjoy retirement. Fourteen of the 19 individuals (74%) who said learning their genetic risk information was their primary reason for participating in REVEAL indicated that they were worried about their risk. For some ($n = 9$), it was fear of developing AD that motivated them to learn their APOE genetic results, but for others ($n = 2$), this fear was a reason for declining to learn the genetic information. Quotations illustrating participants’ worries about developing AD and how this influenced their genetic testing decisions are in Box 2.

3.4. Dispositional coping styles and motivation for genetic testing

Over half of the participants ($n = 35$, 58%) identified themselves as generally preferring to seek out information when asked what they believed led them to be interested in

Box 1. Quotations illustrating participants' perceived susceptibility to AD**At time of parent's diagnosis**

- "I can't believe it doesn't happen to everybody almost immediately. I immediately thought, 'Oh, my God, this is my destiny. Having Alzheimer's may be what happens to me.' There'd be some person saying, 'Mommy was always so strong, and here I am, I'm taking care of Mommy.'" And I'd be thinking, 'Oh, God, and all I'm thinking about is why aren't they asking the same thing I'm asking? Isn't this going to happen to them, and aren't they scared of that?'" (50 year old F, 3/3)*

As a result of multiple family members having AD

- "And I think any objective person would look at my family tree and say, if I had to place odds, my sisters and I would all be on the wrong side of those odds for the possibility of getting this disease at some point." (37 year old M, 3/4)

Because of similarities with affected relative

- "I've showed you the picture of me and my dad. We look like clones, practically, physically. And nobody's really said—I don't know whether the information is out there because I haven't read it—whether or not that makes a difference, a person's physical appearance. But I have a suspicion that it does." (56 year old F, control)

Relationship to motivation for genetic testing

- "But I got scared to death that I inherited this, and that's why I was anxious to get in the study and see." (73 year old F, 3/3)

*Denotes participant age, sex, and genotype if in intervention group (e.g., 3/3 = genotype APOE3/APOE3), "control" if in control group, or "declined" if declined to proceed through parent study to genetic testing.

genetic testing. Participants described their need for information in a variety of ways. While many spoke of their general desire to acquire information, 11 participants believed it was necessary to collect information about one's health in order to make informed health care decisions. Fifteen of the 19 individuals (79%) who said learning their genetic information was their primary reason for participating in REVEAL mentioned their tendency to seek out information in times of stress. Some ($n = 5$) related it to the information gathering nature of their work (i.e., nursing, journalism) or their general curious nature ($n = 6$), citing examples of how they only read non-fiction or were taught at a young age to ask lots of questions.

For some, information was described as fulfilling psychological rather than practical needs. Four separate participants used the phrase "knowledge is power" when talking about their preference for information and 10 linked

Box 2. Quotations illustrating participants' worries about developing AD**Related to family history of disease**

- "People like me worry about the genetic composition. At least I do. Anybody who has a parent with Alzheimer's always is worried about it." (75 year old F, 3/3)

Related to being a burden on loved ones

- "I was going to make some kind of provisions so that my children didn't have to go through what we're going through [caring for my mom with AD] because it's a very sad, difficult, hard, stressful situation. And you do it because you love that person so very, very, very much, but it can tear a family apart. I don't want that to happen to my children, so I, therefore, first and foremost wanted to know if I was a prime candidate." (58 year old F, 3/3)

Related to making life goals unobtainable

- "And for me, since I've invested so much of my life into becoming what I am [a professional writer], the idea that that would be cut short is really – before I had a chance to do it – is really frightening." (50 year old F, 3/3)

Relationship to desire for genetic testing

- "But I got scared to death that I inherited this, and that's why I was anxious to get in the study and see." (73 year old F, 3/3)
- "I didn't want that knowledge, to say that in 20 years I could get Alzheimer's. That's pretty scary." (55 year old F, declined)

their need for information to their need to feel control over situations. Twenty participants in all (33%) identified themselves as the type of person who needs to feel in control or likes to take charge of their health and well-being; this included 11 of the 19 people (58%) whose primary reason for participating in REVEAL was to learn their APOE genotype. People related their need for control to being raised in a family of proactive people ($n = 3$), to being the eldest and needing to take charge for other siblings' sakes ($n = 2$), and as a reaction to the chaotic nature of their home life ($n = 2$). Quotations illustrating participants' need for information and control are in Box 3.

Even though the development of Alzheimer's disease was seen as uncontrollable by most individuals ($n = 45$), for some the act of acquiring genetic information was seen as a way to confront their risk and therefore exert control. For two participants who self-identified as needing information and control but were assigned to the study's control group, the risk assessment based on their family history was not enough to address their anxieties about developing AD. But for two others, who identified as coping by avoiding information, the idea of confronting their genetic risk for AD

Box 3. Quotations illustrating participants' preferences for information and control

Information seeking

- "And realizing that it was my mother, and my mother's sister, and my mother's mother, and realizing how strong the family history was—then it became more focused on my own probabilities. And I did what I tend to do, which is start seeking information." (53 year old F, 3/4)

Need for control

- "I definitely am of the mind-set of wanting to know, and definitely the spectrum of, you know, passively accepting one's fate or trying to manage one's fate, I'm definitely way out on the scale of, we should try to manage our fates and not accept our fates." (51 year old F, 3/3)

Relationship between information and control

- "Not to coin a phrase, but knowledge is power. I really believe that. I mean, I don't think you can necessarily change your destiny, but certainly to go through life with your eyes only half open doesn't help you at all." (52 year old F, 3/3)
- "The old adage is information is power. And it makes you feel reasonable. Being reasonable makes you feel strong. Actually you could call it doing sort of intellectual triage. That makes you feel competent and good." (50 year old F, 3/3)
- "I mean, just if you grow up with a mother who is always out of control, you want to always be in control. And the way you stay in control is to possess information, which is the driving force of my life." (53 year old F, 3/3)
- "When it comes to genetic testing, I mean, one doesn't really have any control at all. Except that information is control." (53 year old F, 3/4)

was too threatening and so they had declined to complete the study. Quotations illustrating these varied reactions of those not receiving genetic information and how they related to differing needs for information are in [Box 4](#).

3.5. Importance of anticipating emotional reactions to results

The appraisal of potential emotional reactions to APOE results played an important part in many participants' decisions to use genetic testing. Twenty-four participants (40%; 10 ϵ 4 negative, 10 ϵ 4 positive, 4 control/declined) described themselves as having the emotional resources and coping skills necessary to handle their results. Thirteen others (22%; 10 ϵ 4 negative, 2 ϵ 4 positive, 6 control/declined) had worried about their emotional reaction to the news prior to receiving results and for two people this had played a role in their declining to continue with the study. Participants also appreciated that choosing to know genetic

Box 4. Quotations illustrating reactions regarding not receiving genetic information

From individuals desiring their genetic information but assigned to the control group

- "When I was waiting [for the results], I actually woke up screaming in the middle of the night one night, which flipped my husband out. Flipped me out, too. You know, sometimes I need the information to know my feelings. There's a lot of fear involved, a lot of anxiety involved in knowing whether or not you have the gene." (56 year old F, control)
- "The record has to be complete. I can't make informed decisions unless I have all the information. It's something in me. I'm obsessed with having all of the facts that are available. I don't like the loose ends. The loose ends make me really nervous." (54 year old F, control)

From individuals choosing not to receive their genetic information

- "But just to know and then drive yourself crazy is—sometimes a little knowledge is too much." (47 year old F, declined)
- "But like I said, I've almost stopped going to doctors because I don't want any more bad information. This is all I can handle. And I'm healthy, so I'm all set." (55 year old F, declined)

information about one's risk for disease was not for everyone. Half ($n = 30$) the participants said they had encountered siblings or friends who told them they would never consider undergoing genetic susceptibility testing for AD. Many ($n = 15$) felt that people's ability to handle receiving genetic risk information was influenced by personality characteristics and coping abilities. Quotations demonstrating participants' characterization of their own, as well as others', ability to cope with genetic information are found in [Box 5](#).

3.6. Coping with being at increased risk for AD using the APOE genetic test

Participants described using the APOE genetic test both as a problem-focused coping effort and as an emotion-focused coping effort. Twenty-eight participants (47%) saw the APOE genetic test as a way to help plan for the future; most spoke in general terms about "getting things in order" ($n = 17$). Three people planned to use the information to decide about long term care insurance. Others hoped genetic results would allow them to try experimental treatments in the future ($n = 3$) or simply prepare themselves and their families emotionally for the possibility of developing AD ($n = 5$). Seven participants put their desire to use genetic testing to help plan for the future in the context of sparing their children the "burden" of caring for a parent with

Box 5. Quotations illustrating participants' appraisal of their own and others' ability to cope with genetic results

Ability to cope with results themselves

- "I was at peace with myself because I knew that whatever the outcome, if it was a negative, I would deal with it; if it was a positive, hooray for me." (58 year old F, 3/3)
- "I was a little bit nervous. And after I was gung ho, I was going to do this, I'm going to find out. But then before I got the results I was like, oh, my God, what if I really do have it, I'm screwed, you know. So, I mean, some nights I would lay awake thinking what am I going to do." (52 year old F, 3/4)
- "If there was some preventative cure, that would be a different story, but if I found out that I had the marker, and then every time I forgot something or misplaced something, I'd drive myself crazy. I know I would. So I think that was the reason why I chose not to, to get the results." (47 year old F, declined)

Ability of others to cope with results

- "There are certainly people who are looking for more and more ways to remain depressed or be depressed, and there are some people who will take that kind of information and allow it to affect daily functioning. I mean, I think one's coping style is a very important factor." (53 year old F, 3/4)
- "I think it would be useful for those who would find it useful, who would want to know. I think it would be about as useful as a screen door on a submarine for those who don't want to know. It could be risky. You know, it's putting undue psychological burden on somebody who doesn't want to know." (55 year old F, control)

Alzheimer disease, an experience each had personally suffered.

Almost half the participants ($n = 29$, 48%) noted a feeling of relief after receiving their results; this included four of the seventeen participants who learned they had the $\epsilon 4$ allele (24%). They expressed relief that even with the $\epsilon 4$ allele their risk was not as high as they had previously anticipated, demonstrating a positive reframing of their risk assessment. Conversely, six participants, all of whom carried the $\epsilon 4$ allele (35%), expressed greater concern about their risk for AD. These individuals described their genetic results as "depressing," "frightening," and "disappointing," each noting she/he had entered the study hoping for better news. One of these participants felt that knowing her genetic results made her unable to stay in denial about her risk for AD. The remaining 21 participants who received results expressed neither relief nor worry in response to them, including 7 participants who learned they had the $\epsilon 4$ allele. These participants related their lack of emotion to the lack of predictability of the APOE test and the feeling that it only

confirmed what they had already come to accept as their risk for AD.

4. Discussion and conclusion

4.1. Discussion

This study of 60 participants enrolled in a study of risk assessment for AD including genetic susceptibility testing suggests that some people choose genetic testing as a way to cope with living at increased risk for developing AD. As theorized by Lazarus and Folkman (Fig. 1), individuals faced with a threat to their health use coping tactics to face the problems arising from being ill or at increased risk of becoming ill, as well as with the emotions that the threat to their health evokes. *Problem-focused coping* strategies, such as reducing the threat of disease through life-style changes, are more likely to be employed if the stressor is appraised as controllable. *Emotion-focused coping* strategies, such as seeking social support, are more likely to be used when the situation is appraised as uncontrollable [24]. Better psychological adjustment is predicted when the coping strategy used matches the controllability of the stressor [25].

Currently, there are few proven options available for controlling the onset of AD symptoms or their progression, so we would expect those individuals at increased risk for developing AD to focus on coping with their emotional concerns about developing the disease. Searching for more information about a health threat, including using medical tests to clarify risk, is described as a problem-focused coping effort [20]. However, examining the comments of these participants revealed a less explicitly stated emotional need that was being served by the genetic testing process. The degree of uncertainty about their chances of developing AD was distressing to some individuals. For those identifying as preferring to seek information and to be in control in times of stress, pursuing genetic testing appeared to provide an avenue for coping with their emotional concerns about developing AD. This finding is consistent with recent empiric evidence that information-seeking meets both practical and emotional needs [26]. As the reactions from members of the control group who strongly desired their genetic results demonstrate, for some people knowing their genetic information may be a prerequisite for progressing on to other emotion-focused coping strategies.

The most common reason participants mentioned for pursuing genetic testing was to better plan for other problem-focused coping efforts, like financial planning and completing advance directives. This focus on taking concrete actions may help people exert some sense of control over an uncontrollable disease like AD. It would be concerning if individuals who learned they were not at increased genetic risk for AD felt overly reassured and chose not to pursue these planning efforts, which are worthwhile for anyone of advancing age. However, data from the parent

REVEAL study suggests that this has not occurred, as no significant differences in purchasing of health, life, or disability insurance were found amongst the participants with positive or negative testing results [27]. While individuals at increased genetic risk for AD were more likely to purchase long term care insurance, those who learned they were not at increased genetic risk for AD were no less likely than members of the control group to do so [20].

4.2. Conclusion

Even without prevention or treatment options, genetic testing may be a useful coping strategy for some at-risk individuals. Our study suggests that the option of undergoing susceptibility testing may particularly facilitate emotion-focused strategies.

4.2.1. Limitations

Although this study offers preliminary findings about appraisals of APOE testing, including motivation, and coping with results, findings are limited to a very specific demographic. Because participants were mostly from a high socioeconomic group, Caucasian and female, this study fails to represent a large number of people who are at-risk for developing AD. Individuals who differ from this sample may also differ in their level of interest in, comprehension of, and reactions to the APOE genetic test. Yet qualitative research is rarely, if ever, intended to yield generalizable findings. Rather the methods serve to mine descriptive data from previously unstudied phenomena to be used to generate testable hypotheses in broader populations.

Participants were purposefully biased toward those who were sufficiently interested in genetic testing to proceed through a clinical trial and to have reflected on their ability to cope with learning their APOE status. This study is also biased by its focus on individuals who were willing to participate in an interview about their research testing experience. Interviews with four participants who declined testing revealed that similar issues of threat, certainty, and control played a part in their aversion to the genetic test. It is unknown whether similar concerns caused others to decline testing.

Because interviews were conducted months after the disclosure of genetic testing results, participants' impressions of the genetic testing process were likely colored by both the passing of time and their risk assessment. Further, the retrospective design of the study limited the examination of the effect of receiving results on participants' future coping. However, these results may inform future studies aimed at quantitatively investigating relationships amongst personal coping patterns and motivation to undergo susceptibility testing for AD.

4.3. Practice implications

Despite the limitations, the qualitative nature of this work allowed for exploration of motivations for undergoing

APOE susceptibility testing for AD. Our findings have implications for the education and counseling of clients considering genetic susceptibility testing for a variety of adult onset diseases. As scientists continue to research genetic contributions to complex diseases, we are likely to encounter an increasing number of genetic susceptibility tests that are partially predictive of the future development of diseases that, like AD, are not entirely controllable or preventable. Individuals concerned about their risk for these diseases who are motivated to undergo genetic testing should be engaged in a discussion with a health care provider about what they hope to gain from the testing experience. Those who feel they need to know their genetic status in order to proceed with practical concerns, such as financial planning and adopting healthy diet and exercise habits, should be encouraged to value and pursue those strategies regardless of their genetic status. However, practitioners should recognize that genetic susceptibility testing may also serve a useful function by helping some clients cope with their emotional responses to being at increased risk for disease [19]. Before proceeding with genetic susceptibility testing, clients should be encouraged to anticipate how they might cope with either a positive, negative, or inconclusive result.

As postulated in the APOE genetic susceptibility testing consensus statements, the development of preventative or therapeutic agents for those at increased genetic risk for various adult onset diseases may lead to wider implementation of genetic susceptibility testing. However, our findings suggest that even without prevention or treatment options, genetic testing may be a useful coping strategy for some at-risk individuals. While clients' needs should not trump important specifics such as clinical validity, they should be included when decisions about clinical use are under deliberation [28]. When susceptibility testing becomes more widespread, practitioners should counsel individuals not only on the value and limitations of testing, but also on the personal nature of decision-making that addresses client appraisals, including motivation and coping patterns.

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