



# Using Direct-to-Consumer Genetic Testing Results in Alzheimer's Disease Clinical Trial Recruitment



Mary M. Ryan<sup>1,4</sup> (email: marymr@uci.edu), Chelsea G. Cox<sup>1</sup>, Joshua D. Grill<sup>1,2,3</sup>, Daniel L. Gillen<sup>1,4</sup>

<sup>1</sup> Institute for Memory Impairments and Neurological Disorders, University of California, Irvine

<sup>3</sup> Department of Neurobiology and Behavior, University of California, Irvine

<sup>2</sup> Department of Psychiatry and Human Behavior, University of California, Irvine

<sup>4</sup> Department of Statistics, University of California, Irvine

## Introduction

- The apolipoprotein E (APOE) gene is the best described genetic risk factor for Alzheimer's disease (AD).
- Carriage of one or two copies of the  $\epsilon 4$  allele of APOE indicates increased risk for AD.
- APOE genotype can be used as an inclusion criterion or enrichment strategy for preclinical AD trials.
- Direct to consumer (DTC) APOE genetic testing is now available.
- Little is known about potential study participants' willingness to share with researchers their personal genetic information gained through DTC testing.

## Objectives

We sought to:

- Quantify the relationship between participant characteristics and whether a participant has used DTC genetic testing.
- Assess the relationship between participants' use or non-use of DTC genetic services and their willingness to use DTC results to be matched to clinical studies.
- Assess differences in reasons for reluctance to share genetic results, comparing DTC users and non-users.

## Methods

- Links to a survey were emailed to 2,306 members of the University of California, Irvine Consent-to-Contact (C2C) Registry who were at least 50 years old.
- 1,313 valid responses were recorded (56.9% response rate); survey responses were linked to demographic information available in the C2C Registry.
- Participants were asked if they knew their APOE genotype, then how they learned it (through DTC genetic testing, or another genetic testing source).
- Logistic regression models were used to assess the relationship between previous use of DTC testing and race/ethnicity, as well as the relationship between previous use of DTC testing and willingness to use the results of DTC testing for invitation to clinical studies.
- We adjusted for additional potential confounding factors in all models: participant age, years of education, and sex.

## Results

	Know APOE Genotype			Total
	DTC	Other Source	No Testing	
N	91 (6.9%)	14 (1.1%)	1208 (92.0%)	1313
Male	25 (27.5%)	3 (21.4%)	444 (36.8%)	472 (35.9%)
Age	63.54 (8.27)	67.29 (8.05)	66.77 (9.08)	66.55 (9.05)
Years of Education	17.45 (2.77)	16.86 (2.03)	16.53 (2.56)	16.60 (2.57)
<b>Race/Ethnicity</b>				
Non-Hispanic/Latino	74 (92.5%)	11 (84.6%)	1057 (95.1%)	1142 (94.9%)
White	72 (97.3%)	9 (81.8%)	975 (92.2%)	1056 (92.5%)
Black	0	0	12 (1.1%)	12 (1.1%)
Asian	2 (2.7%)	2 (18.2%)	48 (4.5%)	52 (4.6%)
Other	0	0	22 (2.1%)	22 (1.9%)
Hispanic/Latino	6 (7.5%)	2 (15.4%)	54 (4.5%)	62 (5.1%)
<b>APOE4 Status</b>				
Carrier	27 (31.0%)	5 (38.5%)	0	32 (34.8%)
Non-Carrier	60 (69.0%)	8 (61.5%)	1 (0.08%)	69 (75.0%)

Table 1: Characteristics of survey participants.

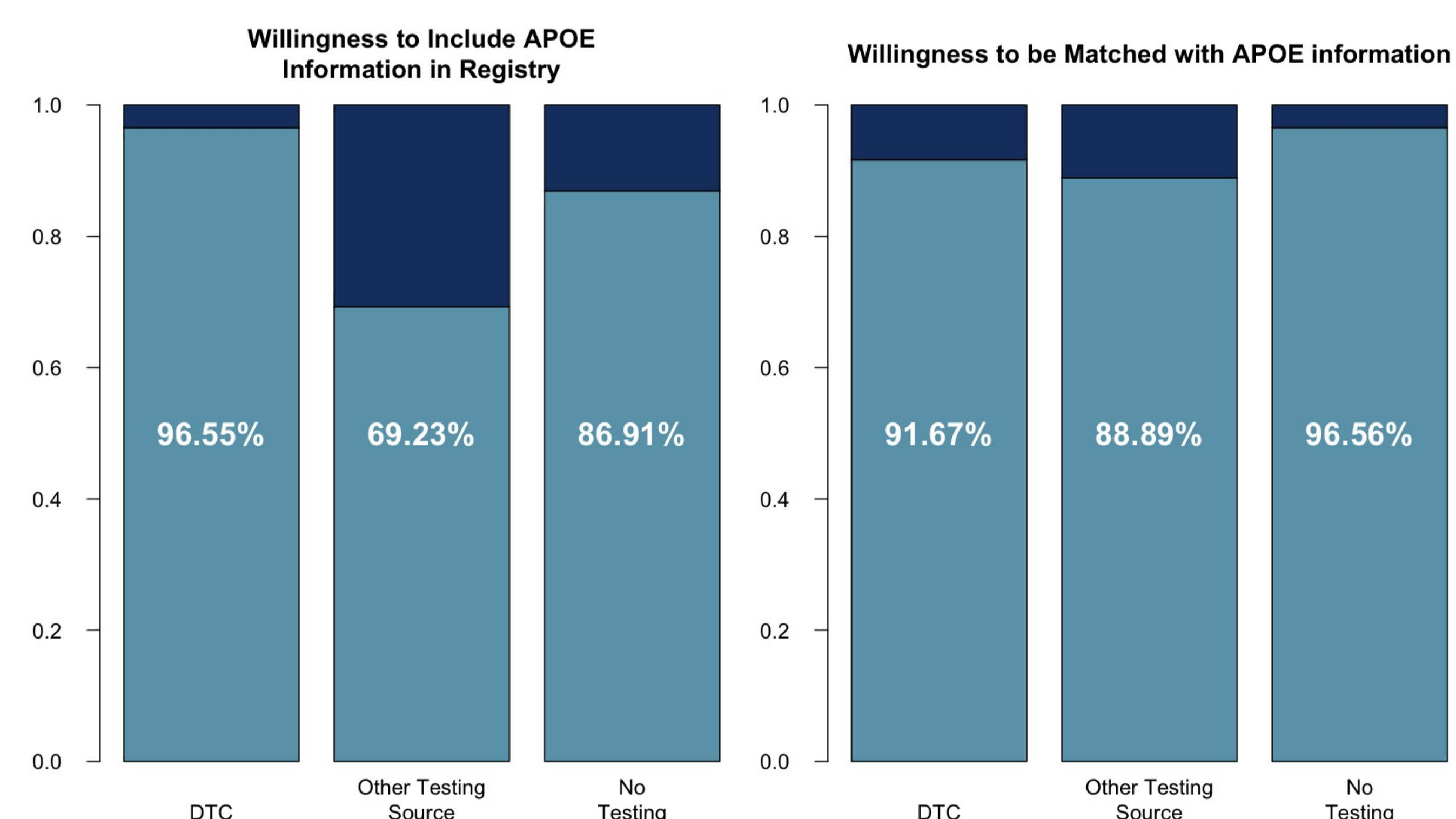


Figure 1: Proportion of survey participants who (left) would be willing to include their APOE information in the C2C Registry, and (right) those who would be willing to be matched to research studies on this information. Proportion of those willing are indicated in light blue, while proportion of those not willing or unsure are indicated in dark blue.

## Reasons for Unwillingness to Include APOE Information in Registry

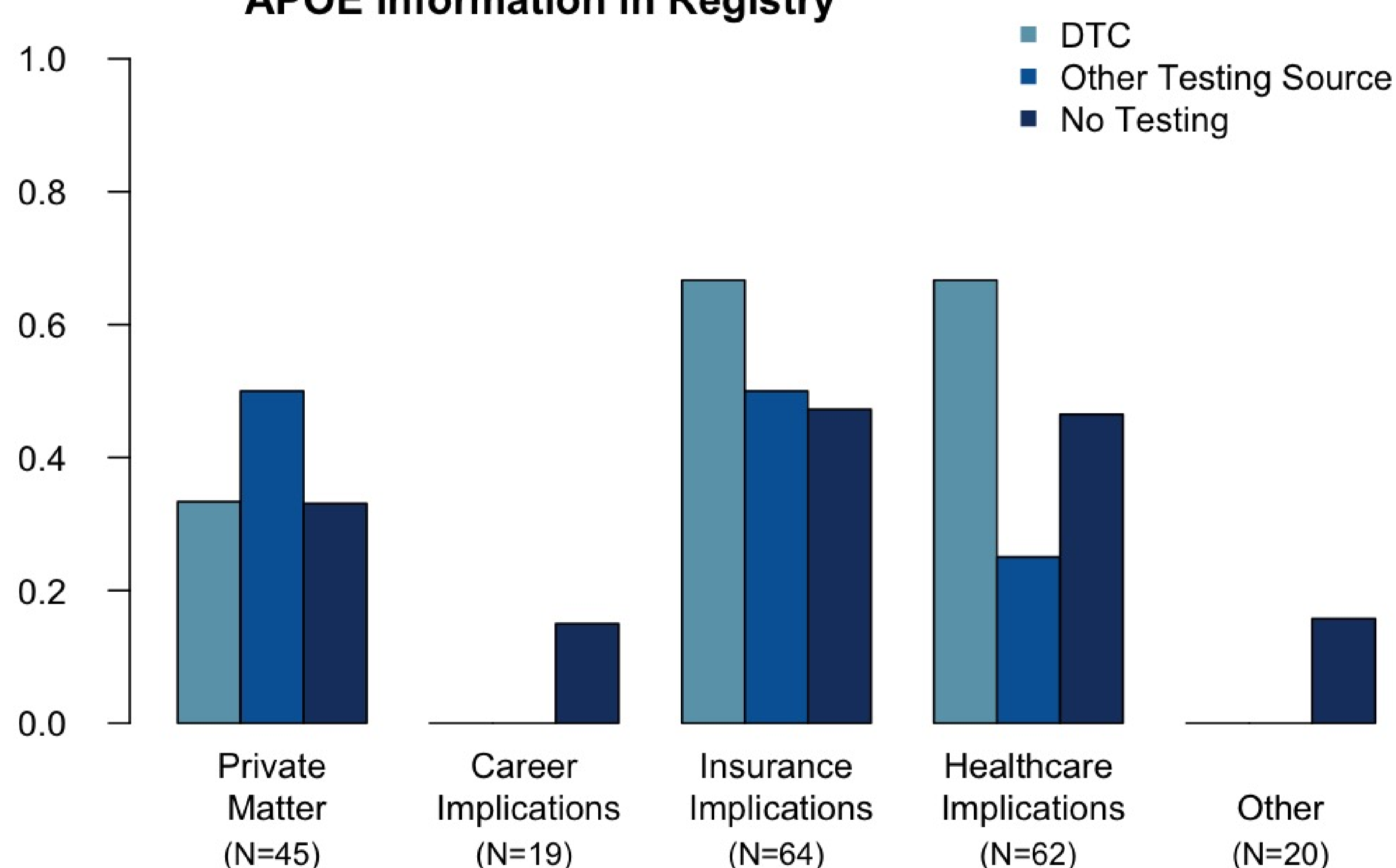


Figure 2: Reasons survey participants gave for why they would be unwilling to include their APOE information in the C2C Registry.

- Most survey participants were aware of DTC genetic testing (N=1,016; 77.4%), while few had used such a test prior to the survey (N=91; 6.9%).
- Most participants who knew their APOE status from DTC tests were willing to share those results in their C2C Registry profile (see Figure 1).
- 86.9% of survey participants who had never undergone any genetic testing were hypothetically willing to include their APOE information in the C2C Registry, and 96.6% were hypothetically willing to be matched to studies (see Figure 1).
- Participants who identified as Asian were less likely to be users of DTC genetic tests, compared to non-Hispanic Whites (OR: 0.08; 95% CI: [0.01, 0.73]).
- Though not significant, participants who had used DTC genetic tests were more likely to be willing to be matched to studies based on their APOE status in their registry profile, compared to non-users (OR: 1.22; 95% CI: [0.11, 14.08]).
- Though not significant, DTC users who were APOE  $\epsilon 4$  carriers were more likely to be willing to be matched to studies based on APOE status than DTC users who were non-carriers (OR: 3.47; 95% CI: [0.37, 32.62]).
- Out of the 134 participants who indicated they would be unwilling to share their APOE genotyping results in the registry, most were concerned about implications to their insurance and implications to their healthcare (see Figure 2).

## Conclusions

- Participants who know their APOE carrier status (regardless of whether they used DTC genetic testing or not) are not less likely to be willing to share that information for clinical trial matching, compared to those who answer hypothetically.
- There is no apparent difference in the results from the regression assessing willingness to use DTC testing results to match to clinical studies. This may stem from the overwhelming majority of survey participants being willing to be matched.
- Concerns about APOE confidentiality do not appear to be a barrier to using genetic test results to enhance AD trial recruitment.