

National Institute on Aging Alzheimer's Family-Based Study genetic result sharing protocol: insights from the initial stage of implementation

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BACKGROUND

- The National Institute on Aging Alzheimer's disease Family Based study (NIA AD FBS) started in 2003 and has recruited over 1,400 families.
- Study participants did not receive genetic testing results in past iterations of the protocol
- A new genetic results sharing protocol was developed to inform participants of results associated with autosomal dominant dementia

METHODS

- Eligible participants were offered CLIA-certified genetic results in genes associated with autosomal dominant dementia (*APP*, *PSEN1*, *PSEN2*, *MAPT*, *GRN*, *C9orf72*)
- Participants (or their proxy) received pre- and post-test genetic counseling with a certified genetic counselor
- Demographics and pre- and post- genetic test surveys were given to the subset of participants who received genetic testing results and had the cognitive capacity to complete questionnaires
- Data analysis utilized descriptive statistics

RESULTS

- Most participants' results were not diagnostic (negative and uncertain) (89%, n=25)
- Of participants who completed the questionnaires (n=8), the majority understood what results meant for themselves (75%, n=6)
- 75% (n=6) were not at all or only a little uncertain regarding implication of results for family members
- While most participants did not receive a genetic explanation for their symptoms, the majority (89%, n=7) indicated results were somewhat to a great deal helpful for future planning and were not frustrated about recommendations for care based on genetic results (75%, n=6).

Table 1. Participant demographics

Category	Participants (n=28)	
Average Age of Onset (years)	59	
Sex	Female	19 (68%)
	Male	9 (32%)
Race/Ethnicity	White	24 (86%)
	Asian or Pacific Islander	1 (4%)
	Other ethnicity not listed	3 (11%)
First degree relative with dementia	Yes	10 (36%)
	No	18 (64%)

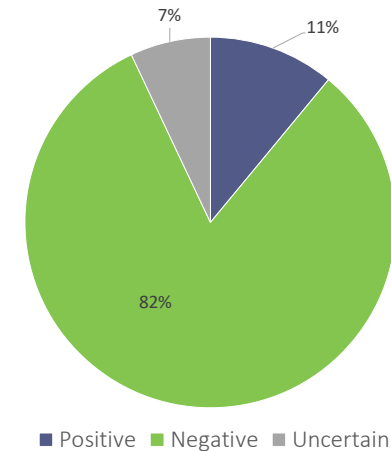


Figure 1. Genetic testing results (n=28)

How uncertain did you feel about what your genetic test result means for you?

How uncertain did you feel about what your genetic test result means for your child(ren) and/or family's risk of disease?

How helpful was the information you received from your genetic test result in planning for the future?

How frustrated did you feel about recommendations for your care based on the genetic test result?

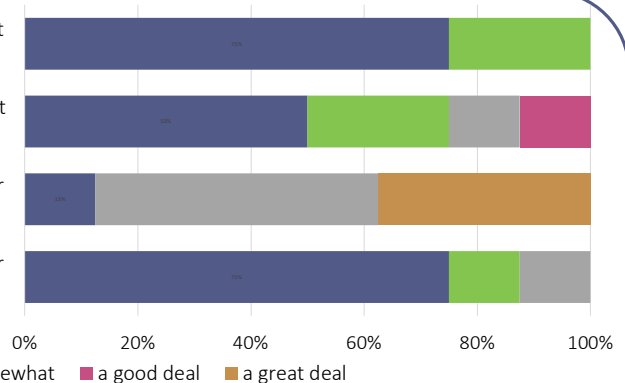


Figure 2. Select questions from Feelings About genomic Testing Results questionnaire (n=8)

CONCLUSION

Our project demonstrates the feasibility of implementation and attitudes regarding receiving genetic results among participants with Alzheimer's disease or mild cognitive impairment. Participants demonstrated that non-diagnostic genetic results were still informative for future planning and there was limited frustration for care implications of results.