ADDENDUM



ADDENDUM: Genetic counseling and testing for Alzheimer disease: joint practice guidelines of the American College of Medical Genetics and the National Society of Genetic Counselors

Jill S. Goldman, MS, MPhil^{1,2}, Susan E. Hahn, MS³, Jennifer Williamson Catania, MS, MPH^{1,2}, Susan LaRusse-Eckert, MS², Melissa Barber Butson, ScM⁴, Malia Rumbaugh, MS⁵, Michelle N. Strecker, MS^{6,7}, J. Scott Roberts, PhD⁸, Wylie Burke, MD, PhD⁹, Richard Mayeux, MD, MSc^{1,2} and Thomas Bird, MD¹⁰

Genetics in Medicine (2019) 21:2404; https://doi.org/10.1038/s41436-019-0559-1

Addendum to: Genetics in Medicine 13: 597–605 (2011); https://doi.org/10.1097/GIM.0b013e31821d69b8, published online 16 May 2011

This document was reaffirmed by the ACMG Board of Directors as of 25 June 2018 with the following addendum:

We suggest the following considerations in reaffirming this document:

- 1. To use the phrase "pathogenic variant" rather than the word "mutation" in discussing pathogenic variants related to autosomal dominant early-onset Alzheimer disease. This would be consistent with current ACMG/AMP Guidelines for Variant Interpretation and Reporting¹.
- Because this document no longer meets the criteria for an evidence-based practice guideline by either the American College of Medical Genetics and Genomics (ACMG) or National Society of Genetic Counselors (NSGC), NSGC reclassified this document as a Practice Resource in 2016, and ACMG is also classifying it as a Practice Resource as of this reaffirmation.

Correspondence: acmg@acmg.net

REFERENCE

Genetics in Medicine

1. Richards S, Aziz N, Bale, S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015;17:405–423. https://www.nature.com/articles/ gim201530. Accessed 14 June 2019.

Published online: 19 June 2019

¹Department of Nephrology, Sergievsky Center and Taub Institute for Research on Alzheimer Disease and the Aging Brain, Columbia University, New York City, NY, USA; ²Sergievsky Center and Taub Institute for Research on Alzheimer Disease and the Aging Brain, Columbia University, New York City, NY, USA; ³John P. Hussman Institute for Human Genomics, University of Miami, Miller School of Medicine, Miami, FL, USA; ⁴Department of Bioethics, Case Western Reserve University, Cleveland, OH, USA; ⁵Department of Neurology, University of Washington, Seattle Institute of Biomedical and Clinical Research, Seattle, WA, USA; ⁶CombiMATRIX Diagnostics, Irvine, CA, USA; ⁷Division of Medical Genetics, University of California San Francisco, San Francisco, CA, USA; ⁸Department of Health Behavior & Health Education, University of Michigan School of Public Health, Ann Arbor, MI, USA; ⁹Department of Biomethics and Humanities, University of Washington, Seattle, WA, USA; ¹⁰Division of Neurogenetics, University of Washington, Seattle, WA, USA. Correspondence: Jill S. Goldman (jg2673@columbia.edu)