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NEWS RELEASE

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New Guidelines on Genetic Counseling and Testing for Alzheimer Disease

Genetics in Medicine Offers Guidance on Which Patients Can Benefit From Testing

PHILADELPHIA, PENNSYLVANIA (July 8, 2011) – When is it appropriate to perform genetic testing for Alzheimer disease (AD), and what information do patients need to understand their risk? The June issue of *Genetics in Medicine*, the official peer-reviewed journal of The American College of Medical Genetics (ACMG) presents a new practice guideline on genetic counseling and testing for AD. The journal is published by Lippincott Williams & Wilkins, a part of Wolters Kluwer Health.

Genetic testing is most appropriate for families with a history of early-onset AD, and should always be accompanied by expert genetic counseling. The new guideline was developed in collaboration between members of the ACMG and the National Society of Genetic Counselors. The lead author was Jill S. Goldman, MS, MPhil, of Columbia University, New York.

Guidelines Highlight Complexity of Genetic Testing for AD

For several reasons including the aging population and the emergence of direct-to-consumer (DTC) genetic tests, more people are interested in genetic testing for AD. The new document seeks to provide health care professionals with guidance on this complex topic.

Tests are available for three known "causative" AD genes which, when mutated, render an individual at extremely high risk for AD. However, even among patients with early-onset AD, no more than five percent of cases are caused by mutations in one of the three genes. Other genes, as well as non-genetic factors likely affect the development of early-onset AD but are yet to be discovered.

A fourth gene, *APOE*, affects susceptibility to AD: people with the epsilon-4 (ε4) form of the gene are at higher risk. However, because the influence of the *APOE* ε4 gene is much weaker than that of truly causative genes, being "neither necessary nor sufficient" for the development of AD, testing provides little useful information in most situations. Even people who test negative for the *APOE* ε4 gene can still be at increased risk, if they have an affected relative.

Genetic Counseling Is Essential Before and After Testing

The new guidelines specify that "Genetic testing for AD should only occur in the context of genetic counseling…and support by someone with expertise in this area." Genetic testing for AD is never recommended in children or before birth. Home or DTC testing for *APOE* is also not recommended due to its poor predictive value and the fact that there exist no proven interventions to mitigate risk.

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In families with early-onset AD potentially caused by a mutation in a strongly determinative gene, testing for causative genes may provide useful information on personal risk. However, those being tested must understand that no current drug or other treatment can reduce the risk of developing AD, regardless of the test results. If no family member with current AD is available for testing, testing of asymptomatic family members is usually unlikely to provide useful information.

In other situations, it is important to assess a person's motivation for pursing genetic testing. Potential conflicts of interest must be addressed—including any disagreement within the family as to whether testing should be performed. Other important considerations include the possible psychological impact of testing, insurance (including life, long-term care and disability) and privacy implications, among others.

The authors hope the guidelines will help to ensure that genetic testing for AD is performed in situations where it will provide useful information, and that patients and family remembers receive accurate information on the meaning of the results. They write, "Genetic testing should be discussed within the context of adapting to familial risk and when clients feel compelled to learn a more refined estimate of their risks to enhance their quality of life."

Note to editors: Interviews with the lead authors available upon request by contacting Kathy Beal, Public Relations Director for the ACMG: phone 301-238-4582 or e-mail kbeal@acmg.net.

About *Genetics in Medicine*
Genetics in Medicine (http://www.geneticsinmedicine.org) is the official peer-reviewed journal of The American College of Medical Genetics. The journal is published by Lippincott Williams & Wilkins, a part of Wolters Kluwer Health, a leading provider of information and business intelligence for students, professionals, and institutions in medicine, nursing, allied health, pharmacy and the pharmaceutical industry.

About the American College of Medical Genetics
Founded in 1991, the ACMG (www.acmg.net) provides education, resources and a voice for the medical genetics profession. To make genetic services available to and improve the health of the public, the ACMG promotes the development and implementation of methods to diagnose, treat and prevent genetic disease. Members include biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors, and other health care professionals committed to the practice of medical genetics. Genetics in Medicine, published monthly, is the official journal of the ACMG.

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