Don’t order a duplicate genetic test for an inherited condition unless there is uncertainty about the validity of the existing test result.

Prior to ordering a genetic test for an inherited condition, the health care provider should ask a patient about prior genetic testing and review the medical record for previously performed genetic tests. Repeating a genetic test should be considered if the existing result is inconsistent with the individual’s clinical presentation or if the test methodology has changed and may yield a different result from the original report that could impact patient management.

Don’t order APOE genetic testing as a predictive test for Alzheimer disease.

APOE is a susceptibility gene for later-onset Alzheimer disease (AD), the most common cause of dementia. The presence of an ε4 allele is neither necessary nor sufficient to cause AD. The relative risk conferred by the ε4 allele is confounded by the presence of other risk alleles, gender, environment and possibly ethnicity. APOE genotyping for AD risk prediction has limited clinical utility and poor predictive value.

Don’t order MTHFR genetic testing for the risk assessment of hereditary thrombophilia.

The common MTHFR gene variants, 677C>T and 1298A>G, are prevalent in the general population. Recent meta-analyses have disproven an association between the presence of these variants and venous thromboembolism.

Don’t order HFE genetic testing for a patient without iron overload or a family history of HFE-associated hereditary hemochromatosis.

The majority of hereditary hemochromatosis is due to inheritance of HFE gene mutations. HFE gene mutations are common among individuals of European ancestry; however, only a small proportion of individuals with these mutations develop clinical disease. Other genetic and non-genetic factors contribute to disease expression. HFE genotyping should only be performed among individuals with iron overload (e.g., elevated fasting transferrin saturation >45%) or a known family history of HFE-associated hereditary hemochromatosis. In the setting of genome or exome sequencing, it is now recommended that patients who are homozygous for the pathogenic variant C282Y in HFE should receive this result and consider evaluation.

Don’t order exome or genome sequencing before obtaining informed consent that includes the possibility of secondary findings.

The informed consent discussion for exome and genome sequencing should include the possibility of secondary findings unrelated to the indication for testing. In addition, before ordering an exome or genome sequencing test, review with the patient the potential benefits (e.g., confirming a suspected genetic diagnosis), potential harms (e.g., psychosocial concerns), limitations of testing (e.g., a mutation may be missed), implications of the test results for family members, and alternatives to exome or genome sequencing.
How This List Was Created

The American College of Medical Genetics and Genomics (ACMG) list relies on input from a number of committees in developing clinical practice guidelines and laboratory technical standards and guidelines. For the Choosing Wisely® campaign, input from the Laboratory Quality Assurance Committee, Professional Practice and Guidelines Committee and Therapeutics Committee was solicited. A list of 18 items was reviewed by the ACMG Board of Directors and the five items currently thought to most likely improve quality and reduce waste related to genetic testing were selected. The recommended list was approved by the ACMG Board of Directors, March 24, 2015.

For the ACMG’s disclosure and conflict of interest policy, please visit www.acmg.net.

Sources


About the ABIM Foundation

The mission of the ABIM Foundation is to advance medical professionalism to improve the health care system. We achieve this by collaborating with physicians and physician leaders, medical trainees, health care delivery systems, payers, policymakers, consumer organizations and patients to foster a shared understanding of professionalism and how they can adopt the tenets of professionalism in practice.

To learn more about the ABIM Foundation, visit www.abimfoundation.org.

About the American College of Medical Genetics and Genomics

The American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics and genomics. ACMG has more than 1,750 members, nearly 80% of whom are board certified clinical and laboratory geneticists and genetic counselors. The College’s mission includes the following major goals: 1) to define and promote excellence in the practice of medical genetics and genomics and to facilitate the integration of new research discoveries into medical practice; 2) to provide medical genetics and genomics education to fellow professionals, other health care providers, and the public; 3) to improve access to medical genetics and genomics services and to promote their integration into all of medicine; and 4) to serve as advocates for providers of medical genetics and genomics services and their patients.

For more information, visit www.acmg.net.

For more information or to see other lists of Things Providers and Patients Should Question, visit www.choosingwisely.org.