



Practice Parameter:

Genetic testing alert

Statement of the Practice Committee Genetics Testing Task Force of the American Academy of Neurology

In clinical practice, neurologists may order tests that yield genetic data about patients or members of their families. Tests can, for example, identify a disease-specific gene or disclose a biochemical abnormality thought to be genetically determined or influenced. Many of these tests are generally accepted and widely performed (e.g., tests of phenylketonuria or sickle cell disease, measuring of cholesterol or lipoprotein levels) and engender little controversy. Other tests may identify genes that predict severe and untreatable neurologic disease (e.g., Huntington's disease) or that suggest vulnerability to such a disease (e.g., Alzheimer's disease). Tests in this latter category have profound implications for patients and their families and merit careful consideration before ordering.

Neurologists are now receiving advertising and other materials that encourage "genetic testing" of their patients. Among other things, these materials may assert that new molecular techniques enable reliable identification of carriers of genes for particular neurologic diseases or of genotypes that predict susceptibility to such diseases. Although molecular diagnostic technologies are undeniably powerful and increasingly accurate, the information they reveal can generate various problems. For example, testing a patient may be of little or no predictive value if family members are not tested as well. Also, unless a variant genotype absolutely predicts clinical disease (as in Huntington's disease), identifying that genotype may have little utility. Indeed physicians or genetic counselors may find that attempting to explain an "abnormal" genotype of uncertain predictive value may only confuse or upset patients or their families. Moreover, once a result of a genetic test that predicts disease or significant susceptibility is entered into a person's medical record, an evident risk is that health insurers, employers, or others will gain access to it and use it in ways that adversely affect the person's welfare. Although medical records are legally protected as confidential documents, patients sometimes, wittingly or unwittingly, allow oth-

ers access to their records to obtain reimbursement for medical expenses or to claim other benefits.

For these reasons, among others, the Genetics Task Force of the American Academy of Neurology's Practice Committee recommends to neurologists that they consider the following points in weighing a decision whether to order genetic testing for patients or family members suspected of harboring genes that cause or increase susceptibility to major neurological disorders.

The following points should be considered:

1. **Predictive power of the test.** Neurologists should understand what a positive or negative genetic test implies with respect to probabilities that a patient, family member, or potential offspring has or will develop a particular neurologic disease. In this regard, a neurologist should appreciate the predictive power of a particular genotype, distinguishing among genotypes that foretell disease with a high degree of certainty (Huntington's disease), those that suggest a possibly heightened susceptibility to a disease (apoE4), and their positive predictive power.
2. **Counseling.** Sensitive and informed counseling provides patients and families a foundation for decisions about testing for serious neurologic disorders. Therefore, before ordering a genetic test in such a setting, neurologists should either possess the training and experience to provide competent counseling about the test and its implications or take steps to ensure access to a qualified genetic counselor. In most situations, genetic testing for major neurologic disorders should not be performed until adequate counseling has been afforded. In the case of predictive testing for Huntington's disease, for example, psychological counseling by appropriately trained persons is essential before testing and after results of testing are disclosed.
3. **Informed consent.** As to diagnostic testing, the doctrine of informed consent asserts that physicians must disclose to patients information that is

See page 1344 for committee members.

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material to a decision about testing and that consent to testing must be informed and voluntary. Therefore, before ordering a genetic test for a major neurologic disorder, neurologists should ordinarily establish that a patient or lawful surrogate is capable of comprehending relevant disclosures and capable of exercising informed choice. If these conditions exist, the neurologist or collaborating genetic counselor should disclose why the test is recommended, the predictive weight of the test, the potentially adverse consequences of a positive test (e.g., extreme emotional distress, stigmatization, loss of health insurance or employment), the benefits of enhanced knowledge about genotype (whether test is positive or negative), and any negative consequences of not testing (e.g., transmission of a detectable disease-associated genotype to offspring). Once there has been adequate counseling, documented disclosure of material information, and voluntary agreement to testing by a competent patient or lawful surrogate, neurologists are ethically justified in ordering a genetic test that may diagnose, predict, suggest vulnerability to, or exclude a major neurologic disorder. Neurologists should not order genetic tests of this sort at the request of members of patients' families or other third parties (e.g., insurers, employers) without the express written consent of a patient or lawful surrogate.

4. Confidentiality. Test data confirming that patients or family members carry genes that indi-

cate or predict susceptibility to major neurologic disorders are highly sensitive. Accordingly, neurologists who obtain such data should implement rigorous measures to ensure their confidentiality and should never disclose such data to third parties without explicit written authorization from tested personal or their lawful surrogates.

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Note. This statement is provided as an educational service of the American Academy of Neurology. It is not intended to include all possible proper methods of care for a particular neurological problem or to exclude any specific alternative therapies. The AAN recognizes that specific patient care decisions are the prerogative of the patient and the physician caring for the patient, based on all of the circumstances involved.

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