

Response to Request for Information Regarding Sections 101 through 104 of the Genetic Information Nondiscrimination Act (GINA) of 2008

From: The American Society of Human Genetics

Date: December 18, 2008

The American Society of Human Genetics (ASHG) represents about 7500 professional members, including researchers, laboratory practitioners, clinicians, genetic counselors and trainees in human genetics. We thank you for the opportunity to address issues related to the implementation of the Genetic Information Nondiscrimination Act of 2008, and therefore are responding to the Request for Information in the Federal Register (RIN 0938-AP37). This is a very significant piece of legislation and our comments are focused from our scientific perspective. As an organizational member of the Coalition for Genetic Fairness, ASHG has endorsed the policy comments made by that Coalition. ASHG would like to commend the policy experts of the Genetics and Public Policy Center of Johns Hopkins University and the Georgetown Health Policy Institute for their thoughtful and extensive comments.

ASHG wishes to make specific statements in three areas:

- (1) comments related to definitions that would assure that the regulations are developed to adapt to the rapid scientific and clinical advances being made in genetics;
- (2) assertion of the critical importance that the prohibition of use of genetic information to discriminate is assured henceforth, no matter when that genetic information was actually collected; and
- (3) offering the expertise of The American Society of Human Genetics to policymakers should there be an opportunity to be of assistance at any time.

Definitions

The definitions of “genetic tests” were extremely difficult to articulate in the GINA legislation, and the final wording is acceptable, although potentially problematic if not interpreted with some flexibility. In addition to changes in DNA sequence, for example, recent molecular analyses of DNA structure have demonstrated that DNA structure can change without altering its sequence. One such change, methylation (attachment of a specific molecule to the DNA structure), alters the level of activity of a gene or stretch of DNA. The term “epigenetic” change is used to describe molecular changes in DNA conformation and expression, whether or not the DNA sequence itself is mutated. We would consider analyses that measure epigenetic effects to be genetic tests.

While the definitions in the law are rather concrete and methodologically based, they should be interpreted flexibly enough to include new advances in the future. Another situation is the evaluation of metabolites that may not have a one-to-one correlation with

a specific mutation, but indicate the presence of any one of several genetic abnormalities. For example, newborn screening that tests for extremely high levels of the amino acid phenylalanine indicate the presence of one of several possible genetic defects in phenylalanine metabolism, and not only phenylketonuria (PKU). These metabolite tests are designed so that professionals can further evaluate newborns to determine the specific mutational cause of high phenylalanine in the blood. We believe the definitions appropriately include these tests, and their interpretation must be sufficiently flexible to include variations in such testing as progress in the study of genetic disorders is made.

Previous Testing

ASHG members have expressed concerns that potential acts of discrimination may be perpetrated after GINA goes into effect based on information gathered *prior* to the enactment of GINA. We encourage strong affirmation that GINA prohibits discriminatory acts or misuse of genetic information no matter when that information was garnered or recorded.

Offer of Expertise and Assistance

The American Society of Human Genetics wants to assure policymakers that our organization is willing to participate in any discussions, briefings or consultation in which the expertise or experience of the thousands of geneticists within our membership may be helpful to the further development or implementation of the Genetic Information Nondiscrimination Act.

Thank you for the opportunity to comment on issues related to this very important piece of legislation from our perspective.

Contact: Joann A. Boughman, PhD
Executive Vice President
The American Society of Human Genetics
jboughman@ashg.org
(301) 634-7307