Testimony of Donald Hadley, M.S.

We'll hear next from Donald Hadley.

MR. HADLEY: Thank you. I'm an associate investigator and a genetic counselor within the National Human Genome Research Institute at the National Institutes of Health. I appreciate the opportunity to present our work to the committee.

My professional experience and work has focused primarily on providing education and counseling to families who are threatened with genetic and inherited diseases. I've had the privilege of working with these families for the last 23 years. In October of 1993, I was invited to join the then newly established National Center for Human Genome Research. Our goal was to develop research that focused on identifying factors that influenced the interest in and uptake of genetic counseling and testing and the associated psychological, social, and behavioral outcomes. Our research agenda was inspired by the identification of a rapidly growing number of genes that predisposed or increased susceptibility to disease.

My research has specifically focused on families who are newly diagnosed with a hereditary cancer syndrome named hereditary non-polyposis colorectal cancer, or HNPCC, and families who specifically have deleterious mutations that have been identified. The identification of families with an HNPCC mutation allowed the opportunity to learn from them before, during and after the provision of genetic counseling and the offer of genetic testing. We felt that identifying the factors influencing decisions and their outcomes was necessary to plan for a future when genetic testing will be more routinely used to predict risk for rare as well as common diseases in the general population.

Within our study, once a family is identified to carry an HNPCC mutation, we sequentially offer participation to first-degree adult relatives who are at 50 percent risk of inheriting the mutation. This offer includes the provision of comprehensive genetic education and counseling about HNPCC and the option of genetic testing. For those electing to pursue genetic testing, the Clinical Center at the NIH pays for genetic studies, removing the issues of cost and insurance coverage from the decisionmaking process. However, prior to the education and counseling sessions, we ask participants to complete a questionnaire that collects demographic information, information about their knowledge of genetics and genetic testing, their perceived cancer risk, and standardized scales assessing mood, coping style, spirituality, their perception of their own control over their health issues, and family relationships.

Additionally, we also ask them to specifically identify what factors motivate them to consider genetic testing, such as the desire to clarify their risk or their children's risk for cancer, or to guide their cancer screening. Likewise, we also ask them to identify factors that concern them about undergoing genetic testing, such as emotional concerns about handling the results for themselves or for other family members, their level of confidence in the prevention strategies that exist, and their concerns about test results affecting insurability. These questions are asked individually so we might obtain a level of significance for each issue.

In addition, we ask the participants to identify the most important reason for them to consider testing, and likewise the greatest concern they have in considering testing. All the questions are validated measures developed and used by the Cancer Genetics Studies Consortium of the NHGRI Ethical, Legal, and Social Implications Program.

As we began to talk individually with each participant, describing the intent of the study and the

process involved, one key issue was consistently identified. That issue was posed in the form of a question: How might participation in this study affect my insurance or that of my family? The question came unsolicited in the beginning of the informed consent process. This question and the associated worry seemed to persist even after we had provided each participant with information that reassured them of the confidential nature of the study, that the study had a certificate of confidentiality issued by the National Institutes of Health; that all participants are issued study ID codes that removes their personal identifiers from the data and the test results; and that the cost of the testing is paid for by the study, so insurers are not involved; and that records are protected by the Clinical Center and only released if written permission is obtained from the participant.

Nevertheless, it was clear that there was an overwhelming concern and in some cases a palpable anxiety about the impact of genetic testing on health insurance. These concerns dominate our informed consent process and recur session after session with an intensity that opened our eyes to the level of concern that the public feels about genetic discrimination. I specifically recall one young woman and her mother, both of whom had experienced uterine and colon cancers at young ages within a family riddled with HNPCC cancers. Even though this young woman had experienced cancer twice and felt there was little residual discriminatory risk to her, she was immobilized by the concerns about the potential of genetic test results branding her family as uninsurable.

She opted to wait on testing but would periodically call to discuss the safeguards our study provided regarding test results and the information obtained. She was admittedly tortured by the concerns about insurance risk, which she felt was keeping her from protecting her family. Finally, after months of considering the implications of testing, she returned to pursue testing, knowing that her results may well prevent others from experiencing what so many in her family had already endured.

Sequencing efforts did identify a deleterious mutation, providing a tool for those within her family to clarify their cancer risk, to focus their cancer screening, and to consider preventive steps such as prophylactic surgery. We anticipated that within the months that followed we'd be hearing from at least a few of her family members, but there were no calls, there were no emails, and there were no letters.

Through follow-up we learned that she had shared the results with her four sisters, and those four sisters expressed that their concerns regarding discrimination were too great to safely allow them to participate in a genetic counseling study with the option of genetic testing. They were worried about being in small companies with limited insurance options and the associated risk that genetic testing posed for them, as well as their children.

In 2003, we published a paper in the Archives of Internal Medicine, which I brought along for your consideration. In the paper we reported on attitudes, intentions, and uptake of genetic testing of individuals within these families. Of particular relevance to this group were findings regarding the level of concern that exists within these families about participating in a genetic counseling and testing research study. The questions that we used to identify their concerns regarding genetic testing are also included in the packet before you.

In looking specifically at what factors influence decisions, we identified and published that 39 percent of participants reported that their most pressing concern was their worry about the potential of a genetic test result affecting their or their family's insurability. I recently looked at our data to see if the level of concern has held true from the earlier analysis, since we've added

approximately 80 additional participants. I found that the current baseline data suggests a number slightly higher than originally published. It's now about 43 percent identifying that their greatest concern regarding genetic testing pertains to their concerns about discrimination.

Furthermore, on follow-up at six and twelve months, a greater proportion of them identified discrimination as the single most worrisome factor. Specifically at six and twelve months, 48 and 54 percent, respectively, identified concerns about genetic discrimination by their insurer as their principal concern. Obviously, this concern is not going away with time and adjustment to the outcome of testing. This seems surprising since research from other studies focused on presymptomatic and susceptibility testing demonstrate that other variables such as anxiety, distress, and mood, seemed to return to pre-test levels by about a year out from testing.

What's different about the concerns regarding insurance? If people have not experienced what they perceive as discrimination, why are there increasing concerns? Participants often ask: "Has anyone in this study ever reported discrimination on the part of their insurance company or employer?" Our answer is always the same: "Not that has ever been reported to us." But apparently, just reassuring them that discrimination in general and within our study is not the norm doesn't help. There is a pervasive mistrust that seemingly worsens with time.

In summary, the prevalence of genetic discrimination by insurance companies does not appear to be the key issue. The real issue is that the public perceives that the potential for genetic discrimination by insurance companies is an overwhelming risk, and in my experience this fear provides a barrier to genetic research and clinical genetics care. This barrier limits our potential for research and basic sciences and social and behavior research. The greatest tragedy, however, is the missed opportunity to prevent cancer or diagnose it early in persons at high risk who are unwilling to risk the potential of discrimination. Providing federal legislation prohibiting genetic discrimination will reassure the public that genetic discrimination is not a risk, provide an increased opportunity for research to address other, more significant issues, and most importantly reduce mortality and morbidity associated with cancers diagnosed at later stages.

Thank you for the opportunity to present our work.

MS. MASNY: Thank you, Mr. Hadley.