Testimony of Jeff Shaw, M.S.

So we'll begin with Jeff Shaw.

MR. SHAW: Thank you. My name is Jeff Shaw, and I am the director and genetic counselor for the Hereditary Cancer Service of Penrose Cancer Center in Colorado Springs, Colorado, elevation 6,300 feet. I don't quite know what to do with this much oxygen.

(Laughter.)

MR. SHAW: I would like to thank you for giving me the opportunity to present information from our program to this committee. The clinicians here feel that you all were a very difficult group to follow, but one thing we've got is numbers.

I provide genetic counseling for individuals and families in every area of medical genetics, from prenatal to adult-onset conditions. For the last seven years I've worked exclusively with patients concerned about hereditary cancer predisposition, much like the women who have presented here today. The purpose of our program is twofold. One is to provide the best estimate of cancer risk so that screening can be appropriately modified so that if a cancer occurs, it can be caught as soon as possible when survival is the highest and treatment is easiest to go through. Two, it's to provide appropriate implementation of medical and lifestyle interventions to drastically reduce the risk of cancer, especially in those with an inherited predisposition.

Our program is clinically based and merges the gap between research-based programs and the implementation of genetic testing into general medical practice. Although we are clinically based, at the outset of our program we created a large database to collect information we felt would be helpful to the provision of our service. The data presented today covers seven years of clinical service to over 900 individuals for hereditary cancer predispositions.

I'll start with the fear of genetic discrimination. Genetic counseling for presymptomatic cancer predispositions is complex and time consuming. It involves, of course, a detailed family history that we try to confirm with medical records, education regarding the differences between sporadic, familial and inherited cancer predispositions, and also the psychosocial issues and family issues that are involved with this type of testing, because, as you've heard, if you test one individual, that information is going to be applicable to their entire extended family.

If you look at the entire number of people that we see, 61 percent of patients have a family history strong enough to indicate the possibility of genetic testing. If an individual is offered the possibility of genetic testing, we go through a rather lengthy informed consent process discussing the risk, benefits, and limitations of that testing and how it would apply to their medical care. Unfortunately, when I'm supposed to be talking about medical care and medical decisions, the bulk of this discussion I feel I need to be a lawyer, because it is strongly centered around the concerns our patients have regarding genetic discrimination, not only for themselves but for their siblings and, I find most importantly, for their children.

In our program's experience, 20 percent of those individuals who are eligible for genetic testing for presymptomatic cancer decisions declined having the test. Of those individuals, 22 percent did so because of a fear of discrimination. You must also realize that the people who see me are already motivated to learn more about their family history.

Of interest, the patients that did decline based on a fear of discrimination, 90 percent of those had a very significant increased chance for testing positive for a mutation that would increase their risk for cancer. Therefore, the people who would experience the greatest possible benefit from this testing are the most likely not to pursue it.

I'd like to give a few patient experiences because these numbers take on a more personal tone in the context of real people. I recently saw a woman who had a very strong family history of cancer, much like the women who have talked earlier today. She had just been diagnosed with a Stage 1 breast cancer at the age of 46. Her mother died of ovarian cancer at the age of 52. Two maternal aunts had breast cancer in their early 40s and have passed away, and her maternal grandmother died from breast cancer at the age of 41. Due to this strong family history and her own diagnosis, we determined she had at least a 43 percent chance of carrying an inherited mutation that would increase her risk for second primary cancers.

If she pursued the testing and was determined to carry one of these faulty genes, she would have up to a 60 percent increased risk for a second primary breast cancer, a brand new one, and up to a 44 percent chance of developing a primary ovarian cancer. Prophylactic surgical intervention could reduce her risk for these cancers 90 percent or greater. If she tested positive, each of her four daughters would have a 50 percent chance to inherit this faulty gene that could increase their risk for cancer.

She has declined testing, and this has been very emotional and very difficult for her, but she's done so because she is very concerned as to how this information could affect her children's chances of getting health insurance. She doesn't know what profession they're going to go into, she doesn't know if they're going to be group or self insured, she doesn't know what state they're going to end up living in. Because of that, without the genetic testing, it's unclear how to proceed with her medical care, especially the surgical ones that could reduce her risk for cancer developing again.

However, without documentation of a mutation that she would carry, her insurance company will not pay for any of these surgical interventions. Therefore, she remains in a state of anxiety, using imperfect breast and ovarian cancer screening methods and simply hoping that another cancer does not occur. Due to her current employment situation, she might have to change insurance companies. She's afraid that if she were to change insurance companies, she could be denied, and with her current diagnosis she simply cannot afford to be without health insurance.

Another patient we had had a strong family history of FAP. This is a dominantly inherited colon cancer predisposition characterized by early onset of colon polyps, hundreds to thousands of these that can begin as early as the age of 10. Individuals with this condition basically have up to a 100 percent chance of developing colon cancer sometime in their life. He worked his whole life at a relatively small company with a small self-directed group insurance plan. He has been warned by his doctor not to have genetic testing for FAP as he would lose his job or his health insurance if they were to find out about the condition in the family.

At the age of 42 he had significant rectal bleeding and finally went in for evaluation. He was found to have over 400 polyps in his colon. It was so extensive that he needed to have his entire colon removed, a drastic but life-saving technique for these individuals. Luckily, he did not have an invasive colon cancer. Other family members were not as lucky. Most of those affected with FAP in his family died of colon cancer in their late 20s.

At the age of 46, he came to me for genetic counseling. He has two children, ages 22 and 24. He

had not informed them of the condition prior to this time as he did not want the family history in their medical records due to a fear of genetic discrimination. Unfortunately, this meant that these early 20-year-old children were not having appropriate screening. With several genetic counseling sessions with him, he finally decided that he would do the testing, even with his fear of the discrimination, in order to have appropriate medical care for his kids. He was tested and the genetic mutation in the APC gene that was causing FAP in his family was identified.

His children decided to have testing. One child has tested positive, one child has tested negative. The 22-year-old that tested positive is now having appropriate screening, but also lives in fear that at some point she could lose her medical insurance.

The fear of genetic discrimination in this family could have caused the same early deaths in his immediate family as it did in his extended family.

I just saw a 24-year-old patient whose mother tested positive for a mutation in one of the breast/ovarian cancer suppressor genes. She did not have testing based on a fear of discrimination. She was diagnosed with a Stage 3 breast cancer and died in June. She finally had the testing done, and a mutation was identified. Therefore, we could cheaply test her three children to see if they indeed inherited this mutation or not. All three children decided they did want to be tested because they felt it was important for their care. All three children paid for this test out of pocket to keep this information as confidential as possible.

In fact, when we look at individuals when the cheaper test can be performed, this is about $350 when a mutation has been identified in a family, 74 percent of those individuals will pay out of pocket because of their fear of genetic discrimination. Unfortunately, if you're the first person being tested in the family, the test costs about $3,000, making that really not an option for the bulk of individuals that we see.

Although she has informed her family about this mutation, well over 50 percent have decided not to be tested based on a fear of discrimination. Although anecdotal, my experience with hundreds of families shows me that this is the case for many of these family members.

What about after testing? We conduct one-year follow-up surveys of all patients seen by our program. We're happy that we have a 72 percent response rate to these surveys. Of those patients who tested positive for an inherited cancer predisposition, 70 percent report having continued significant anxiety that they would experience genetic discrimination at some point in the future. Fear of future genetic discrimination remains a real concern for our patients, especially those who have tested positive.

Then we were wondering about those people who don't even make it for genetic counseling, they don't even get to the point of being offered testing. In addition to a fear of discrimination from genetic testing, there's also fear of discrimination simply from participating in a genetic counseling session. In 2001, Geer, et al., studied factors that would influence an individual's decision not to come in for counseling. Of those declining genetic counseling, the biggest reason was a fear of genetic discrimination, accounting for 40 percent of those individuals surveyed.

Our program has had a significant number of physician-referred individuals who did not show up for their scheduled appointments. After seeing the Geer study, we wanted to see informally if this was a concern for the people referred to our program. We conducted an informal six-month survey of those patients not pursuing referral for genetic counseling by phone. In this time frame, we had 60 patients that did not show. Fifteen percent would not return our calls, and of those we

could contact, 49 percent stated that they had changed their minds, that they had heard from their physicians and family that they should not have this documented in their medical chart due to genetic discrimination. I was unable to get the bulk of these people in for an appointment.

This informal survey supports the data seen in the Geer study. It shows us that a fear of genetic discrimination is a barrier for individuals that could benefit greatly from genetic counseling and possibly genetic testing for hereditary cancer predispositions.

When I graduated with my genetic counseling degree in 1994, there were but a handful of genetic tests available for inherited conditions. In 2004, just 10 years later, there are over 1,000 genetic tests available on a clinical or research basis. The number of genetic tests that will become available for single-gene and complex genetic disorders is expected to increase exponentially over the next decade. I fear that without strong federal protection, the appropriate use of these tests will continue to be under-utilized and we will not gain the benefit from the genetics revolution.

None of us are genetically perfect. Learning what genetic imperfections we have inherited and how they affect our risk for disease is difficult, sometimes frightening, and a life-changing experience. The decision to have presymptomatic genetic testing is multifaceted. It encompasses issues regarding one's sense of self, family relationships, anxiety, depression, and very complex decisions regarding future medical care. The citizens of our country need to be assured that when they are deciding whether or not to pursue genetic testing, a fear of genetic discrimination is not a factor.

Thank you for your kind attention, and I look forward to answering any questions.

MS. MASNY: Thank you, Mr. Shaw.