Testimony of Mark Brantly, M.D.

The last of our health provider presenters will be Dr. Mark Brantly.

DR. BRANTLY: I'd like to thank the committee for inviting me to come and speak. My name is Mark Brantly. I'm a pulmonary physician and a physician scientist at the University of Florida. I've been involved in alpha-1 antitrypsin deficiency testing since approximately 1983 and have tested about 20,000 individuals, identified about 2,000 alpha-1 individuals over the last 20 years. In recent years I've been testing approximately 5,000 to 6,000 patients per year for alpha-1 antitrypsin deficiency.

I follow approximately 150 alpha-1 antitrypsin deficient individuals in my clinic at the University of Florida and have first-hand experience regarding the impact of this diagnosis on them personally and also their families.

Let me begin by giving a brief expose of alpha-1 antitrypsin deficiency. It's a very easy disease to diagnose. It requires simply an alpha-1 antitrypsin level and a PI type or a genotype. It's one of the more common genetic diseases, with a frequency of 1 in 2,500 to 1 in 4,000 individuals. The phenotype is primarily chronic obstructive pulmonary disease and liver disease. It's oftentimes associated with a rapid decline in lung function punctuated by lung infections. However, it's one of the classic genes in which there's an environment and gene interaction. That is, individuals who have alpha-1 antitrypsin deficiency lose lung function much faster when they smoke cigarettes. Indeed, they die 20 years prior to non-smoking individuals.

Importantly, in my clinic population I have individuals that are 80 years old with profound alpha-1 antitrypsin deficiency who are living active lives. Therefore, prevention of behaviors and interactions is a critical aspect of this disease. It is not all about having expensive therapies. People can live their entire lives with not having disease or disability if they're identified early and we're able to protect them. That, I think, forms the basis of early diagnosis and preventive care being critical if we are to make a significant impact in this disorder.

In the State of Florida only, there are 900,000 individuals with COPD, and 9,000 die per year. Almost 1,000 of these individuals have at-risk alpha-1 antitrypsin deficiency alleles. In the State of Florida we've had a program in which we have done targeted detection. We first began by establishing a consensus among the community with the help of the Alpha-1 Foundation that testing exceeded the risk of testing. We established a high-throughput laboratory, and we provided professional and lay educational materials to deal with some of the educational issues that are associated with alpha-1 antitrypsin deficiency diagnosis. We developed an easy testing system where patients can prick their finger and send it to our central laboratory, yet we still have significant barriers to testing these individuals despite major recommendations from the major thoracic societies recommending a Category A recommendation for testing.

These barriers include genetic discrimination, and particularly fear of genetic discrimination, ignorance regarding the disease among the physician population. We've also established tertiary care referral systems to make sure that when physicians do identify these patients, that they have someplace to go with these patients.

So we have yet still an important job, and that is to be able to -- instead, right now, we have 5,000 individuals that are identified with alpha-1 antitrypsin deficiency, and there are approximately an estimated 95,000 that haven't been identified. If these patients were identified early on, they perhaps could be protected from developing disability.

One of the approaches that we've used is doing a coding testing trial through the Medical College of South Carolina and Charlie Strange. This is funded entirely by the Alpha-1 Foundation, and it's been a longitudinal study looking at the reasons why people do not wish to be tested through their physician. We've tested now more than 3,300 individuals in this testing program and have done some initial longitudinal follow-up. I've provided you with one manuscript that gives you some of the results, but I'd like to focus in on a couple of things most recently that we have done.

The first one is the risk and benefits of genetic testing. Thirty-three percent of individuals said that the reason why they chose the coded testing trial was because of fear for losing their health insurance or higher health insurance costs. The other thing is in the post-test, who would you give your results to? Well, not surprisingly, they would give the results to their children and their spouse, and not surprisingly they wouldn't give it to their ex-spouse.

(Laughter.)

DR. BRANTLY: In addition, they would not provide this information to their health insurance companies or their life insurance companies. Indeed, only about 16 percent would disclose that. Sadly, though, I have to say that only 80 percent of these individuals who were profoundly deficient would even tell their personal physician, and that's problematic as far as I'm concerned.

Finally, one of the things that this study I think brings up in close contrast is that when patients were diagnosed with alpha-1 antitrypsin deficiency, obviously one of the major therapies is to do smoking cessation. While there was a trend towards individuals who had alpha-1 antitrypsin deficiency quitting smoking, this was not significant. In actuality, it was higher for alpha-1 antitrypsin deficient individuals, still there was a large portion, greater than 80 percent, who did not quit smoking.

In my clinic and in many of the physicians' clinics who take care of alpha-1 antitrypsin deficient individuals, I have a 95 percent quit rate for cigarette smoking. The national average is 10 percent. Why is that? That's because I hound these patients to death. I schedule them for appointments to see me every month, I have my nurses hassle them, because I know of all the things that I do for these individuals, getting them to quit smoking is clearly one of the most important things that I can do.

When we have to resort to coded testing and we leave out the physician and the health care provider in helping these individuals cope with and make these changes, we short-change them in a big way. We short-change them because they're afraid, because they can't trust our system to protect them and to give them the correct information. There's only one difference between my patients and me. We all as complex genetic organisms have five to fifteen "lethal mutations" that may be associated with our demise or our disability. The difference between me and my patients is I don't know about mine. My patients know about theirs and they have the ability to do risk prevention.

Thank you very much.

MS. MASNY: Thank you for all your testimony. It continues to clarify that genetic discrimination, and especially the fear of genetic discrimination, is very real.