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**Public Comments**

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DR. TUCKSON: We are reconvened.

One of our critical functions is to serve as -- I knew there'd be trouble over there with the caffeine -- a public forum for deliberations on the broad range of health and societal issues raised by the development and use of genetic technologies. So we do greatly value the input we receive from the public. We set aside time each day of our meetings to hear from members of the public, and we welcome and appreciate the views they share with us. In the interest of our full schedule, I'd ask the commentators to keep their remarks to five minutes. We have copies of your full statements which will be made a part of the meeting record.

Today we will hear from Dr. Mittman, who works for the Maryland Department of Health but is speaking on her own today.

Dr. Mittman?

DR. MITTMAN: Good afternoon and thank you for letting me be here and make my comments. Understanding the genetic basis of life has an overarching impact on the ideological frames within which people view themselves, their actions, and others in society. Whereas in the past scientists targeted ethnically-defined groups for the study of mostly rare or recessively inherited conditions, a new paradigm shift is driving studies to target more common afflictions such as diabetes, hypertension and asthma, conditions with a lifetime risk of at least 60 percent in the general population. In order to facilitate these studies, scientists are once again turning to genetically simplified population isolates such as the old order Amish, Finnish, Icelanders, and Ashkenazi Jews.

The "Jewish" case in genomic research is particularly troubling. In my research, I identified that we can learn a lot by focusing on this population that is quite experienced in population-based studies. The Ashkenazi Jewish population has received disproportionate attention with respect to genetic mapping as genomic researchers have capitalized for decades on the ease of founder mutations, the relative simplicity of gene patterns, and the perceived willingness of Jewish individuals and families to partake in genetic testing.

The disproportionate targeting of Ashkenazi Jews for genomic studies and the obvious lack of sensitive social studies on the impact of such research on this population was very costly for this group. Unlike other populations targeted for genomic studies, Jews carry the scars of the Holocaust, calculated genocide rooted in the "scientific" notion that Jews are members of an inferior race. Jewish community leaders indeed are concerned that the focus on the genetics of Jews will revive the fallacy of the inferiority of the Jewish people and lead to stigmatization and discrimination of this group.

Although American Jews constitute a distinct cultural and religious minority group, they receive little attention in the minority health literature, as do other religious minority groups like Hindus and Moslems. In spite of the racial, ethnic, cultural and economic diversity of Jews, they are often viewed as a monolithic group and stereotypically portrayed as a model minority of affluent, well-connected, and well-educated people. Even when social studies are conducted on perceptions of Jewish community members related to genetic testing, no distinction is made as to the various subgroups that make up the Jewish community, and conclusions tend to be generalized to the entire Jewish population. As an example, a recent study published this month in the American Journal of Public Health portrays Ashkenazi Jews as an advantaged group

receiving preferential treatment in genetics and enjoying easy access to BLCA testing at the expense of other less advantaged groups.

There are many issues pertaining to the U.S. Ashkenazi Jewish population that are at times overlooked. Conservative estimates suggest that more than 600,000 Jews are recent immigrants to the U.S., and thus may face linguistic, economic and cultural barriers to health care. American Judaism is also diverse with respect to religious sub-denominations. The majority of U.S. Jews define themselves in religious terms. Forty percent are conservative, 30 percent are reform, and about 11 percent are orthodox. The various branches of Jewish religious denominations differ in their interpretation of the level of adherence to the written and oral laws of the Jewish religion. While conservative and reform Jews may be fully Americanized and share the values of surrounding communities, orthodox Jews strictly adhere to ancient rules dictated by the Torah and maintain very distinct cultural and religious beliefs.

Importantly, the Jewish population shows wide socioeconomic disparities. While the average Jewish household income is well over the national average, one-fifth of Jewish households fall in the lowest income category.

It has been well documented the (inaudible) identified in the Ashkenazi Jewish population have inadvertently led to discrimination and stigmatization of this group. Moreover my dissertation, which focused on the impact of genetic screening on the orthodox Jewish community, identified serious issues related to within-group discrimination, especially related to marriage prospects in this population which largely utilizes arranged marriages. It appears that an alarmingly growing segment of the population, especially women, are finding it hard to secure a match in part because of perceived undesirable genetic endowment. In this community, inability to marry and procreate may doom a person to a meaningless existence. These issues seem to emerge from misconceptions propagating within the Jewish community with respect to its genetic endowment. This may be the result of linking Jews with common disorders. For example, some of the studies already under way to identify genes for common disorders which target Ashkenazi Jews include inherited mental disorders such as schizophrenia and bipolar disease, autism and Alzheimer's disease.

Targeting socially identifiable communities for genetic studies possesses a serious risk that socially constructed prejudices will be defined in biological terms. Linkage of ethnically and socially defined groups with diseases and particular therapy regimens, as we spoke about earlier in the case of BiDiI, stands to deemphasize a host of important social and environmental construct in disease causation and treatment and encourages stereotypes leading to unequal access to medical intervention, and as a whole could lead to widening health disparities.

In summary, I recommend that when studying public perceptions related to large population-based genetic studies, we should explain the definition of vulnerable populations. They should include all stakeholders, especially those groups already experienced with population studies in order to truly make every voice count. Second, as exigent genetic technology impacts health and health care disparities, it might be beneficial to have federal officers charged with eliminating health disparities take part in these important deliberations.

Thank you.

DR. TUCKSON: Thank you very much. Thank you for taking the time to do that. Let me just, before you run away, as if there are any questions. I think you were very specific about how you

want us to consider your comments, especially in the context of the large population study analysis, but I wonder if people have any other comments.

DR. McGRATH: It's not a question but a great thank-you for bringing to our attention more specific examples of phrases we throw around easily here, genetic discrimination and stigma. After a while we sort of forget what those phrases mean, but thanks for doing that.

DR. TUCKSON: Thank you very much.

Dr. Debra Leonard is presenting on behalf of the Association of Molecular Pathology.

DR. LEONARD: This isn't to make light of my comments. Reed, members of the committee, good afternoon. I'm changing hats from a previous member of the SACGHS --

(Laughter.)

DR. LEONARD: -- to representing the Association for Molecular Pathology, also known as AMP. So I want to make clear that I am representing AMP here. I'll take the hat off so you don't think that this is totally humorous.

The purpose of these comments is to provide AMP's perspective on four issues relevant to the charge of the SACGHS. First are two draft guidances from the FDA on ASRs and IVDMIAs. AMP is finalizing comments to the FDA on two draft guidances which raise concern for the membership of AMP.

The first draft guidance, entitled "Commercially Distributed Analyte-Specific Reagents (ASRs): Frequently Asked Questions," defines a much narrower interpretation of the ASR rule than is currently in practice. AMP is concerned that these more stringent interpretations will limit the availability of ASRs, which provide high-quality reagents for the validation of laboratory developed tests by clinical laboratories under CLIA regulations. If ASRs become more limited in availability, either laboratories will find other sources for these reagents that are of poorer quality, such as research reagents, or laboratories will stop performing many tests that are currently standard of care. This could lead to decreased patient access to molecular testing services.

The second draft guidance, entitled "In Vitro Diagnostic Multivariate Index Assays (IVDMIAs)," defines the FDA's regulatory approach to complex multivariable tests. While AMP agrees that complex tests with interpretive algorithms that are not transparent and are able to be manipulated by the laboratory warrant FDA regulation, the use of an interpretive algorithm is routine in medical practice and should not in and of itself raise specific concerns with the FDA.

The purpose of raising these issues with SACGHS today is to assure that this advisory committee is aware of these draft guidances, and I was not aware at the time that these were being presented right after the public comment period, and their potential impact on genetic testing services. Once AMP finalizes its comments in response to the two draft guidances, the letter can be provided to SACGHS, if that would be of interest.

My second point of concern is the tabling of CLIA genetics specialty regulations. AMP is very concerned by the recent decision not to incorporate a genetic specialty into CLIA regulations. Frankly, we are mystified by this action, which follows years of largely favorable comment from the clinical laboratory and genetics communities in response to the initial CDC's Notice of Proposed Rulemaking. The current CLIA regulations, which have not changed in almost 20 years

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relative to genetics, define genetic testing in terms of classical cytogenetics only. Defining genetic and molecular diagnostic testing explicitly would allow for appropriate regulation and oversight of these tests by the agency best suited and legally mandated to regulate laboratories performing these clinical tests. Defining a genetics specialty within the CLIA regulations would promote expansion of proficiency testing programs, provide better oversight of genetic tests, and reassure the public and members of Congress about the quality of genetic testing performed in CLIA-certified laboratories. The members of AMP strongly urge SACGHS to bring this concern to CMS and CDC and encourage the incorporation of a genetic specialty within the CLIA regulations.

The third issue is assessment of coverage and reimbursement for genetic testing services. AMP members perform genetic tests and other types of molecular tests for the management of patient care. We remain concerned that the CPT codes and reimbursement levels set for these codes are less than the cost to perform these tests. While the SACGHS report on coverage and reimbursement issues made recommendations that CMS develop a plan to address this issue, AMP is not aware of any action taken to date on this issue. AMP applauds SACGHS for its recommendations and asks SACGHS to follow up to determine if action will be taken to correct the inadequate payment levels for these CPT codes.

Finally, the gene patent issue. AMP asks that SACGHS give full consideration to the negative impact of exclusive licensing and enforcement practices for gene patents on the future of genetic testing. We understand that SACGHS set this as a high-priority issue and is now formulating an approach to investigate the impact of gene patents on patient access to molecular tests. AMP wants to assure SACGHS that gene patent enforcement continues to limit the tests clinical molecular laboratories can perform for their patients. We urge SACGHS to develop a plan of recommendations to the Secretary of Health and Human Services to address the clinical impact of these practices.

AMP remains, as always, available to the SACGHS to assist with or provide additional information for your thoughtful deliberations and important work. On behalf of AMP, I thank you for the opportunity to speak to you today.

DR. TUCKSON: All right. With that hat still on, are there any questions to follow up?

(No response.)

DR. TUCKSON: Terrific. So you'll be available? Since we're about to turn to one of the key recommendations you made, which is around the oversight issues, you'll be able to --

DR. LEONARD: You're also going to hear about the FDA draft guidances as well as you'll hear from Judy Yost, I believe, about the CLIA decision not to have a genetic specialty.

DR. TUCKSON: Okay, that's what I'm getting at. We're getting ready to get into the oversight stuff, the CLIA and --

DR. LEONARD: And I didn't know that you were hearing about those otherwise.

DR. TUCKSON: Great. This is perfect. So we'll have a chance to follow up specifically in just a second. Thank you very much. Very well done.

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Please note that there are written public comments also from the International Society of Nurses in Genetics, ISONG, in your table folders.

If there are others in the audience who would like to make comments, we have another public comment period scheduled tomorrow. Please sign up at the registration desk if you would like to speak at that session.