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[Comments as excerpted from the meeting transcript]

I would like to thank the organizers very much for the opportunity for public comment.

I'm an associate professor at University of Miami. I'm speaking for myself from approximately 20 years of life in the trenches in terms of molecular genetic testing from running a diagnostic lab to having to arrange genetic testing for a variety of inherited neurological disorder, as well as running an active translational research lab in gene discovery, as well a member of ASHG and the College.

I want to begin by stating that GeneTest and Gene Review has been an invaluable resource for the genetics community. There's hardly a time in the clinic that we don't access GeneTest or we don't refer a patient to Gene Reviews, and I am very glad to see that Gene Reviews will be included in the new plans for the GTR.

It also has been very helpful in the GeneTest to be able to see labs as a research setting, as well as labs that are throughout the world for rare diseases.

As we go on to develop a Genetic Testing Registry, I certainly applaud NIH in this very important effort and you hear many of the challenges and complexities that have been brought up to date. I want to bring up two additional concerns.

One is that we should have increased usage of laboratories throughout the world. They are sometimes the only source or one of the few sources for rare disease testing, as well as research laboratories. Now this, of course, brings up a problem if we're going to have CLIA testing and CAP certification for all the things to be included in GTR and this is one thing that has to be considered. In many cases—I'll take limb girdle muscular dystrophies, for example—there are only a few labs in the country and other places in the world that are offering the kind of testing that is needed and we have to have a resource for that.

My second concern is that many academic laboratories have grown up being providers of rare diseases. Homes for these kinds of tests, although they often provide also common disorders, they are often either the sole source or the expert in these areas. And when we test for rare diseases as you've heard these concerns brought up before today, they--we don't often have the data about prevalence, incidence and the other--some of the other datasets that are being called for, and so we need to consider this.

In regards to specific questions that we're to answer today, I think if we are to take a phased-in approach it should be taken with the rare--with the common diseases first. Those are also commonly tested by many laboratories.

I fully agree with Dr. Korf's comments that were at the beginning of the session in regards to testing for rare disease and rare variants.

Again, more importantly, due to our niche, to consider what to do with new state of the art approaches. Things like NextGen sequencing, exome sequencing, CNVs, and how to use this kind of information when the clinical utility is still not know and there is no FDA validation for

this and yet many laboratories are moving to these technologies.

And in reference to Question No. 3 I think that cost should not be included. They are often subjective. They are dependent on contract arrangements, Medicare/Medicaid, and they are going to be changing constantly.

In closing, I would hope that the GTR as this goes forward carefully considers the very careful comments that have been prepared by the ASHG, the ACMG, the NSGC, Genetic Alliance, and AMP in reference to this and encourage them to use members of these communities to move forward in this process.

Thank you.