Oral Presentation at the FDA Workshop on Draft Guidance for Proposed Regulation of NGS-based Tests, September 23, 2016

Hello, I'm David Flannery, Medical Director of the American College of Medical Genetics and Genomics, which is the national medical organization dedicated to improving health through the medical practice of genetics and genomics.

We appreciate the opportunity to participate in this public workshop and provide our recommendations regarding possible regulation of next-generation sequencing clinical laboratory tests.

ACMG has already been active in developing the standards by which genomic medicine will be practiced.

As we have commented previously, these NGS tests are not devices, rather they are complex clinical diagnostic procedures performed by specialty trained and experienced clinicians.

We feel that Next generation sequencing based testing is analogous to medical imaging, in which there is a recognized role for FDA. As a result, we recommend that for emerging genome scale testing, FDA should ensure the general analytic performance of manufactured devices used in genetic and genomic testing.

The general capabilities and limitations of different testing platforms for different types of genetic changes should be clear delineated for potential users. With regard to this, ACMG's laboratory quality assurance committee is already at work on ACMG guidelines regarding the best practices for assessment of data quality and best practices for reporting results, including gene variants.

We assert that the use of NGS platforms analytically in clinical laboratories, however, should remain under the oversight of CLIA and as the practice of medicine.

As the use of new genome scale technologies with integrated bioinformatics filtering expands, decisions about which information from the genome anatomy is appropriate for visualization and communication to patients should remain within the practice of medicine.

ACMG guidelines currently assist clinicians in decision-making about running genetic tests appropriately and assist laboratories in accurately and consistently performing the test. We have a joint document with the Association of Molecular Pathology on standards and guidelines for the interpretation of sequence variants.

ACMG is currently updating its policy statement regarding recommendations for reporting of incidental findings on NGS, which should be published soon.

The importance of genomic data sharing has become prominent. ACMG has partnered in the Clinical Genomic Resource, (ClinGen) project, using experts to clinically curate specific genomic variations based on data provided by clinical genetic service providers across the country. This is creating what we call a genomics information commons, which can be used to support the validity claims of associations between specific genes, gene changes and disorders, and also provide a quality improvement resource for clinicians and laboratorians.

ACMG appreciates having the opportunity to participate in this workshop and looks forward to continued engagement with the FDA on these issues. Thank you.