ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

January 31, 2019

The Honorable Eric Swalwell
U.S. House of Representatives
407 Cannon House Office Building

Sent electronically to: lizzy.fox@mail.house.gov

Re: Advancing Access to Precision Medicine Act

Dear Representative Swalwell:

On behalf of the American College of Medical Genetics and Genomics (ACMG), I want to thank you for taking time to meet with us on January 3, 2019. ACMG appreciates your interest in medical genomics and looks forward to working with your office in the future.

ACMG is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics. Our membership includes over 2,200 biochemical, clinical, cytogenetic, molecular, and medical geneticists, genetic counselors, and other healthcare professionals, nearly 80% of whom are board certified in medical genetics specialties. ACMG engages in coordinated efforts to improve patient care, ensure optimal reimbursement for genetic services, establish standards of care and laboratory policy, educate members about advances important to their practices, and advocate for the responsible application of genetics and genomics in medicine. With the rapid increase in knowledge about the relationships between genetics and disease that has developed over the past couple decades, genetic services have become increasingly more complex. ACMG's members facilitate the delivery of high-quality clinical and laboratory medical genetics services which is a critical component of the precision medicine initiative.

ACMG appreciates your effort to support medical genetics through the Advancing Access to Precision Medicine Act, and we look forward to introduction of this bill in the 116th Congress. We believe that system-wide approaches are necessary to achieve meaningful and lasting enhancements that ultimately improve public health and support the goals of precision medicine. However, even with an appropriate framework in place, our ability to successfully integrate genetics and genomics in medicine will be limited by the availability of quality data. This is especially true for rare diseases and rare genetic variants for which data sharing may be the only way to increase statistical power and support clinically meaningful conclusions. Increasing the breadth and quality of accessible data requires a system to support sharing of data among public and private institutions as well as incentives to share such data. This will be increasingly important as the use of genomics in the private sector continues to rise. Section 2 of the Advancing

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Access to Precision Medicine Act instructs the National Academy of Medicine to study ways in which the Federal Government may be able to help reduce barriers to genetic and genomic testing. Reducing barriers to and creating incentives for sharing of genomic data is another key area in which the Federal Government may be able to help.

This section also mentions improving access to genetic counselors, pathologists, and other relevant professions, including strengthening related workforce education and training efforts. Successful integration of genetics and genomics also requires an appropriate workforce, and medical geneticists (clinical and laboratory) are critical to the foundation of that workforce. Clinical geneticists are licensed physicians (MD or DO) who are also certified by the American Board of Medical Genetics and Genomics (ABMGG), one of the 24 member boards of the American Board of Medical Specialties. Laboratory geneticists (MD, DO, or PhD) are also certified by ABMGG and often serve as directors of clinical laboratories. Through certification, ABMGG ensures high standards of continuous education and minimum proficiency are maintained throughout the geneticist's professional career. In addition to providing patient care and laboratory services, these medical geneticists, as well as genetic counselors, work together through organizations like ACMG to develop educational resources for healthcare professionals and clinical decision support tools to facilitate integration of genetics and genomics into other specialties. The current medical genetics workforce is insufficient, but the specifics of the workforce needs and shortages in the United States are unclear. As a first step in addressing this shortage, the committee report (115-862) that accompanied the 2019 LHHS Appropriations bill included a request for the GAO to perform an assessment of the medical genetics workforce. ACMG hopes to work closely with the GAO to ensure that they are able to obtain the best data currently available.

Section 3 of the Advancing Access to Precision Medicine Act refers to state options to provide whole genome sequencing services for certain children. There is a current effort in the genetics field to shift away from use of the term "whole" when referring to genome sequencing. Genome sequencing means an unbiased sequencing of DNA in an individual's genome, however current technology does not enable coverage of an entire human genome. Although coverage capabilities are improving, there are still sections of the genome that pose significant challenges for current technology. Further, even if we could sequence a person's entire genome, the clinical significance of much of the genome is not well understood at this time. We recommend that "whole" be removed from the text when referring to genome sequencing. In addition, the definition provided in Section 3 for "whole genome sequencing clinical services" includes sequencing of the eligible individual and a biological parent. However, to be informative, genome sequencing is generally needed from both biological parents (typically referred to as trio sequencing). We recommend revising this language to include sequencing of one or both biological parents.

Lastly, the definition of "eligible individual" in Section 3 limits the state option to only children who have already presented with significant disease and required admittance into a pediatric intensive care unit (PICU). For many diseases, early intervention is key to long-term management of the condition and prevention of irreversible damage. Credible suspicion of pediatric-onset genetic disease can be established through outpatient medical specialist evaluations, and genomic testing at that point could lead to early intervention that may preclude the need for an expensive PICU admission. Further, biochemical screenings, such as those performed as part of state newborn screening programs, can identify signs of disease prior to notable symptom onset. In some cases, genomic sequencing may be able to identify the etiologic cause of a biochemical finding, and early intervention can prevent devastating, irreversible damage. It would be interesting to explore the possibility of maximizing the utility of genomic sequencing by increasing its

access to children prior to developing severe disease in the hopes of preventing irreversible damage and costly medical care.

We appreciate the opportunity to introduce our organization and provide comments on the Advancing Access to Precision Medicine Act. If you have any questions, please contact Dr. Michelle McClure, ACMG's public policy director, at mmcclure@acmg.net. We look forward to working with your office in the future and are happy to serve as a resource on the topic of medical genetics and genomics.

Sincerely,

Michael S. Watson, MS, PhD, FACMG
Executive Director