May 10, 2021

The Honorable Eric Swalwell
U.S. House of Representatives
174 Cannon House Office Building
Washington, DC 20515

The Honorable Scott Peters
U.S. House of Representatives
1201 Longworth House Office Building
Washington, DC 20515

Dear Representatives Swalwell and Peters:

On behalf of the patient and scientific communities, we, the undersigned organizations, are writing to express our support for legislation that would require state Medicaid programs to cover DNA sequencing clinical services to help diagnose individuals - especially children - and give them access to the care they need sooner. Specifically, we believe that the legislative language must empower clinicians to select whatever technology is best suited for their individual patients by providing them with the flexibility to choose from the various diagnostic sequencing technologies.

We want to thank you all for your commitment to improving the lives of patients living with life-threatening, chronic, and rare diseases and for your ongoing efforts to reconcile the differences between your respective bills (the Advancing Access to Precision Medicine Act and the Ending the Diagnostic Odyssey Act). Timely and sustainable access to diagnostic testing that rapidly informs appropriate patient care and treatments is fundamental for rare disease patient care.[i] With robust and ongoing legislative activity happening this year, including Cures 2.0, we hope to see this reform for undiagnosed patients enacted this Congress.

Due to a lack of reliable coverage, children, their families and their clinicians are unable to consistently access genomic sequencing in a timely fashion. Both your bills will improve the ability for all patients and families, not just those covered by Medicaid, to receive timely and accurate diagnoses with access to these new diagnostic technologies, such as whole genomic sequencing (WGS), whole exome sequencing (WES), and gene panels.

A genome comprises all the DNA contained in a cell's nucleus, which totals to about six billion nucleotide bases; this genomic sequence can be captured and examined in whole or in part. Variations in a genome that scientists look for can range from a change in a base pair in one place of the sequence to larger regional changes, deletions, or repeats. Fundamentally, WGS tests look at almost all a patient's genome at a relatively low depth, while WES tests primarily examine the sections that code for proteins at a medium depth; panel tests observe specific genes or other regions of the DNA at the highest depth, with a focus on certain conditions or health issues.

Most rare diseases have a genetic origin, and this disproportionately affects children under 5 years of age. The National Economic Burden of Rare Disease Study in the U.S.[ii] found that the average rare disease diagnosis spans more than 6 years and includes visits to more than 17 healthcare specialists. As a result, many children do not survive long enough to receive an accurate diagnosis or characterization of their condition. Rare genetic mutations are also underlying causes for extremely prevalent developmental disorders, which may have an adverse effect on the care they receive and the ability to develop optimal treatments for many patients living with rare diseases. By identifying genetic causes, patients can begin treatments and interventions earlier, altering the course of a condition, preventing irreversible morbidities, and reducing some negative impacts throughout a patient's life.

In recent years, genomic and genetic sequencing technologies have emerged as a leading diagnostic approach due to the depth of analysis and ability to detect pathogenic abnormalities. These technologies have the potential to improve diagnosis capabilities, reduce the time to diagnosis, and positively impact patient outcomes for many pediatric patients with rare and undiagnosed diseases; however, many patients, including those covered under Medicaid, struggle to access these new technologies.

Congress should ensure that patients, particularly children who rely on federal and state insurance programs, with rare and undiagnosed diseases and who are covered by Medicaid have equitable access to diagnostic tools, including WGS, WES, and gene panels. We support this action because expanding coverage to genomic sequencing through Medicaid would expedite the path to diagnosis and help those most in need. Our hope is that coverage under Medicaid for these diagnostic tests will spur private insurance coverage of WGS and WES – something that remains a barrier to many undiagnosed children and their families.

Furthermore, patients should have access to whatever diagnostic tests their clinicians deem necessary to accurately diagnose and, if possible, treat them. As new technologies develop and current technologies improve, patients should have access to all future diagnostic tools that have been cleared or approved by the Food and Drug Administration (FDA), or others, at the discretion of the Health and Human Services (HHS) Secretary.

We support broader coverage of testing in order to allow patients to receive the best care that meets their specific needs, to avoid giving competitive advantages by favoring certain existing technologies over others, and to not stifle the innovation of other new technologies in the genomic sequencing space. A diagnosis allows patients to receive referrals to critical therapies to manage their disease, even if no FDA approved or licensed therapy exists, like:

- Beginning necessary, non-pharmaceutical therapies (physical, occupational, speech therapy, etc.);
- Building connections with a specific disease community;
- Enrolling in clinical trials, especially for the robust pipeline of over 800 gene therapies in clinical trials for rare diseases; and
- Contributing to the understanding of the natural history of the disease along with the magnitude and diversity of the impacted patient population.

One of the most significant challenges impeding the ability to obtain genomic or genetic sequencing tests is the lack of understanding regarding the coverage under different plans and in different states. We believe that the compromise bill should include language which would either create, generate, or request survey data on current coverage under Medicaid as well as enforce the federal requirements that currently exist.

For example, the Early Periodic Screening, Diagnostic, and Treatment (EPSDT) benefit is a required benefit for Medicaid recipients under age 21 in every state. EPSDT provides comprehensive health coverage by financing an array of pediatric services to ensure that patients receive appropriate and necessary physical, dental, developmental, and mental health services, including preventative measures as well as treatments. However, the inconsistent approach to Medicaid coverage leads some states to classify these genetic and genomic sequencing diagnostic tests as experimental while others consider them “approved”, making standards of coverage under EPSDT unclear.

We believe that this bill should serve as a foundation for increasing the availability of sequencing coverage for all patients, beginning with patients on Medicaid. As such, more data is required to understand the gaps in coverage for these diagnostic treatments and which states choose to offer these services. Though there are numerous approaches that can be taken, we believe legislation must require the U.S. Department of Health and Human Services Center for Medicare and Medicaid Services (CMS) to perform a survey to obtain data for what services states are providing coverage, establish a demonstration or pilot project in a select number of states to generate data, or include a charge to the National Academies of Medicine to study this issue further.

Thank you for your strong leadership to support patients and their families in accessing timely and life-saving diagnoses. We would greatly appreciate the opportunity to meet with your staffs to identify additional areas of collaboration, share valuable insights based on our organizations' extensive databases and patient engagement, and discuss ways to ensure that patients will not have to continue to live with undiagnosed conditions. We appreciate your championship of this critical issue and look forward to working together on patient-centric health reforms that ensure that scientific innovations reach the people who need them the most.

Sincerely,

American Society of Gene & Cell Therapy
CureDuchenne
debra of America
EveryLife Foundation for Rare Diseases
Global Genes
Hemophilia Federation of America
Jett Foundation
Parent Project Muscular Dystrophy
Rare Disease Innovations Institute, Inc.
RARE-X
The Assistance Fund
Tuberous Sclerosis Alliance

Cc:

The Honorable Frank Pallone, Jr.
The Honorable Cathy McMorris Rodgers
The Honorable Diana DeGette
The Honorable Fred Upton
The Honorable Anna Eshoo
The Honorable Brett Guthrie

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[i] Global Genes and Child Neurology Foundation. (2021). Access to Critical Therapies: Guiding Principles of Rare Disease Care and Patient Access. 22 Jan. 2021, https://globalgenes.happyfox.com/kb/article/235-access-to-critical-therapies-guiding-principles-of-rare-disease-care-and-patient-access/

[ii] EveryLife Foundation for Rare Diseases. (2021). The National Economic Burden of Rare Disease Study. 25 Feb. 2021, https://everylifefoundation.org/burden-study/