26400 Woodfield Road #189
Damascus, MD 20872
202.966.5557
sterry@geneticalliance.org
www.geneticalliance.org

Representative Diana DeGette
US House of Representatives
2111 Rayburn House Office Building
Washington, D.C. 20515-4329

Representative Fred Upton
US House of Representatives
2183 Rayburn House Office Building
Washington, D.C. 20515-4329

July 16, 2021

Dear Representatives DeGette and Upton,

We appreciate your continued support for advancing genomic medicine, as evidenced by key provisions of your "Cures 2.0 Act". In particular, we support your efforts in Section 407 of your June 22 discussion draft to expand access to diagnostic services for children who are suspected of having rare genetic disorders.

We also appreciate the opportunity to provide comments on the discussion draft. We recommend the following two changes:

*** Allow any interested and eligible state to participate in the demonstration program.**

We understand the importance of rigorously evaluating the value of new diagnostic modalities such as whole genome sequencing (WGS), but we also believe that pediatric Medicaid beneficiaries in more than 5 states should be able to access these tests. If more than 5 states can meet the terms of the demonstration program, we should encourage those states to apply. By removing this cap, your bill would generate even more clinical data to demonstrate the value of genomic sequencing, while ensuring that more children have access to the most comprehensive diagnostics available to them.

*** Focus the demonstration program on the newest diagnostic modalities: next-generation sequencing (NGS) clinical services.**

Historically, children suspected of having a rare genetic disease have often endured a diagnostic odyssey that lasts months if not years. For children born into families with known genetic mutations, single-gene testing or genetic panel tests might be appropriate as first-line diagnostic evaluations.

A demonstration program to evaluate and demonstrate the value of these tests is unnecessary: their value is already understood by public and private payers, and these tests are widely covered when clinically appropriate.

However, many children suffer from undiagnosed neonatal-onset or pediatric-onset diseases, the causes of which are unknown. In a world with more than 6,000 known genetic disorders, and new disorders being discovered on a regular basis, access to the newest next-generation sequencing (NGS) diagnostic tests can mean the difference between receiving an answer in days or weeks, or never receiving an answer at all. Unfortunately, access to these new diagnostic modalities is still limited, despite their promise.

To provide the most help to these children, Congress should incentivize states to make the newest and most comprehensive genetic clinical services available where clinically appropriate. Expanding access to next-generation (specifically, whole genome and whole exome) sequencing could move the entire field forward into a more comprehensive testing approach from the moment a clinician suspects a child might be suffering from a rare genetic disease.

Focusing a demonstration project on next-generation sequencing clinical services was the approach taken by California's Project Baby Bear, a small pilot program to assess the value of using whole genome sequencing as a first-line diagnostic tool. In that $2 million pilot, the savings to the health care system made possible by earlier diagnoses (particularly with respect to intensive-care-unit days avoided) nearly paid for the total costs of the program.

We recommend that any similar demonstration programs supported by Congress should focus on next-generation sequencing clinical services. We believe this would build a robust body of evidence on how next-generation sequencing delivers value in our health care system and delivers more timely and comprehensive answers to patients and families in desperate need.

Thank you for your continued leadership and your careful consideration of these recommendations.

Sincerely,

Sharon F. Terry
President and CEO