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,

The undersigned organizations 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. The undersigned organizations would 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.

The undersigned organizations thank you for your continued leadership and your careful consideration of these recommendations.

Sincerely,

5p- Society
Ago2 Association
AliveAndKickn
Alpha-1 Foundation
Alstrom Syndrome International
American Behcet's Disease Association
Angioma Alliance
Aplastic Anemia & MDS International Foundation
APS Foundation of America, Inc
AXYS
Bobby Jones Chiari & Syringomyelia Foundation
Born a Hero, Research Foundation
BPAN Warriors
Bridge the Gap -Syngap Education and Research Foundation
CADASIL Together We Have Hope
Canavan Foundation
Care4ASH1L

CARES Foundation Inc.
CFC International
CHARGE Syndrome Foundation
Children's Cardiomyopathy Foundation
ClinWiki
CLOVES Syndrome Community
Coalition to Cure CHD2
Congenital Hyperinsulinism International
Costello Syndrome Family Network
CSNK2A1 Foundation
Cure CMD
CURE Epilepsy
Cure HHT
Cure Sanfilippo Foundation
Cure SMA
CURED Nfp
CureSHANK
Cutaneous Lymphoma Foundation
Dravet Syndrome Foundation
Dup15q Alliance
Epilepsy Foundation
Fabry Support & Information Group
Fibromuscular Dysplasia Society of America
FND Hope
FOD Family Support Group
Foundation for Prader-Willi Research
Foundation to Eradicate Duchenne
FOXG1 Research Foundation
Genetic Alliance
Hermansky-Pudlak Syndrome Network
Histiocytosis Association
Hypertrophic Cardiomyopathy Association
International Foundation for CDKL5 Research
International Pemphigus Pemphigoid Foundation
International SCN8A Alliance
Jeffrey Modell Foundation
Lennox-Gastaut Syndrome (LGS) Foundation
Lupus and Allied Diseases Association, Inc.
Lymphangiomatosis & Gorham's Disease Alliance
MEPAN Foundation
Mississippi Metabolics Foundation
MitoAction
MLD Foundation
Mowat-Wilson Syndrome Foundation

Mucolipidosis Type IV Foundation
MYGENEDx Co Pvt Ltd
Myocarditis Foundation
National Fabry Disease Foundation
National Foundation for Ectodermal Dysplasias
National Tay-Sachs & Allied Diseases Association
NBIA Disorders Association
Noah's Hope - Hope4Bridget Foundation
Organic Acidemia Association
Parent Project Muscular Dystrophy
Pathways for Rare and Orphan Studies
Phelan-McDermid Syndrome Foundation
PXE International
Rare Epilepsy Network
Rare New England
RASopathies Network
Ring14 USA
SPAN Parent Advocacy Network
Sudden Arrhythmia Death Syndromes (SADS) Foundation
TBC1D24 Foundation
Team Titin
The Ehlers-Danlos Society
The Global Foundation for Peroxisomal Disorders
The School of Theoretical Modeling
The Sturge-Weber Foundation
Turner Syndrome Foundation
Unique
United Leukodystrophy Foundation
US COPD Coalition
Usher 1F Collaborative
Usher Syndrome Coalition
VHL Alliance
Wilson Disease Association

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