Congresswoman DeGette
US House of Representatives
Rayburn House Office Building, 2111
Washington, DC 20515

Congressman Upton
US House of Representatives
Rayburn House Office Building, 2183
Washington, DC 20515

June 15, 2020

Dear Representatives DeGette and Upton,

On behalf of the Rare Disease Community, we join our voices to thank you for your efforts to build upon the success of the 21st Century Cures Act and engage in a dialogue in shaping Cures 2.0.

While recent innovation has presented new opportunities to diagnose and treat genetic rare diseases, individuals with rare diseases still face vast difficulties in diagnosis, specifically during the COVID-19 pandemic when funding and access is limited. As you know, nearly 80% of all rare diseases have a genetic cause, and half of rare disease cases impact children. The average diagnostic odyssey can last anywhere from five to seven years. You can imagine the anguish of parents watching their children suffer, watching them endure one test after another. The toll this takes on the family, emotionally and financially, is a great travesty in an age where comprehensive genetic screening is available and affordable. Whole Genome Sequencing will alleviate an enormous part of a huge burden these families carry.

To address the current barriers to coverage and patient access of genomic sequencing, we recommend including the H.R. 4144 - Ending the Diagnostic Odyssey Act in the Cures 2.0 legislation. The "Ending the Diagnostic Odyssey Act" would allow states to conduct a pilot program to increase the Federal Medical Assistance Percentage rate (FMAP) to provide Whole Genome Sequencing clinical services for children on Medicaid with a disease that is suspected to have a genetic cause. We are eager to see this bill signed into law so this first-line test can be offered to families, regardless of income.

Knowing the genetic cause of a disease can mean an actionable diagnosis – leading to changes in treatment and management of the condition, preventing additional unnecessary testing, and helping families find a support structure via other families and organizations. This has utility and benefits for the child, the family, and society at large. And even when there is no treatment at the ready, having multiple kids diagnosed early on, with the hope of gathering data on their condition and those of other kids like them, accelerates treatment development.

Thank you for the opportunity to provide comments and feedback on your proposal to develop Cures 2.0 legislation. As all of us know well, and often personally, a diagnosis means a great deal to a family. Just having that information empowers parents to find support, participate in research, and ultimately end the diagnostic odyssey so that they can get the right care.

Thank you again for your leadership on this important legislation.

Sincerely,

22Q Texas
5p- Society
Adult Polyglucosan Body Disease (APBD) Research Foundation
AliveAndKickn
All Things Kabuki
Alstrom Syndrome International
AMENSupport (American Multiple Endocrine Neoplasia Support)
American Lyme Disease Foundation
American Multiple Endocrine Neoplasia Support
American Porphyria Foundation
APS Foundation of America, Inc.
Asthma and Allergy Foundation of America
AXYS
Bale Genetic Consulting LLC
Barth Syndrome Foundation
Batten Disease Support and Research Association
Beyond Celiac
Bridge the Gap – SYNGAP Education and Research Foundation
Canavan Foundation
CARES Foundation
CCARE Lynch Syndrome
Children's Tumor Foundation
Children’s Cardiomyopathy Foundation
Cholangiocarcinoma Foundation
Colorectal Cancer Alliance
Columbia University Irving Medical Center
Congenital Hyperinsulinism International
Cure CMD
Cure HHT
Cure Sanfilippo Foundation
CURED Nfp
CureSHANK
Cutaneous Lymphoma Foundation
Dandy-Walker Alliance, Inc.
Debra of America
Dravet Syndrome Foundation
Dup15q Alliance
Dystrophic Epidermolysis Bullosa Research Association of America
EB Research Partnership
Epilepsy Foundation
Epilepsy Leadership Council
Familial Hypercholesterolemia Foundation
FamilieSCN2A Foundation
Family Voices-NJ
FND Hope
FOD Family Support Group

Foundation for Prader-Willi Research
Foundation for Sarcoidosis Research
Foundation to Eradicate Duchenne
FOXG1 Research Foundation
Genetic Alliance
Global Foundation for Peroxisomal Disorders
Glut1 Deficiency Foundation
Hannah's Hope Fund
Hope for Hypothalamic Hamartomas
Hunter's Hope Foundation
Hydrocephalus Association
Hypertrophic Cardiomyopathy Association
Idaho Parents Unlimited
International FOP Association
International Foundation for CDKL5 Research
International Pemphigus and Pemphigoid Foundation
International WAGR Syndrome Association
Jeffrey Modell Foundation
Kabuki Syndrome Foundation
LGS Foundation (Lennox-Gastaut Syndrome)
Life Raft Group
Lipodystrophy United
LunaPBC
Lupus and Allied Diseases Association, Inc.
Lymphangiomatosis & Gorham's Disease Alliance
MitoAction
MLD Foundation
Myocarditis Foundation
National Blood Clot Alliance
National Eosinophilia Myalgia Syndrome Network
National Fabry Disease Foundation
National Foundation for Ectodermal Dysplasias
National Neutropenia Network
National Psoriasis Foundation (NPF)
National Society of Genetic Counselors
National Urea Cycle Disorders Foundation
NBIA Disorders Association
No Stomach For Cancer
Organic Acidemia Association
Parent Project Muscular Dystrophy
Parents and Researchers Interested in Smith-Magenis Syndrome
Pathways for Rare and Orphan Studies
Phelan-McDermid Syndrome Foundation
PreventionGenetics
PTEN World
PXE International

RARE-X
RASopathies Network
Rett Syndrome Research Trust
Ring14 USA
RUNX1 Research Program
SADS Foundation
SCID Angels For Life Foundation
Sickle Cell Thalassemia Patients Network (SCTPN)
SPAN Parent Advocacy Network (SPAN)
Spastic Paraplegia Foundation
Spina Bifida Resource
Stickler Involved People
Sudden Arrhythmia Death Syndromes (SADS) Foundation
SWAN USA (Syndromes Without A Name)
Team Titin
The FH Foundation
The Global Foundation for Peroxisomal Disorders
The International 22q11.2 Foundation, Inc.
The Life Raft Group
The Mastocytosis Society, Inc
The Transverse Myelitis Association
The XXYY Project
Tuberous Sclerosis Alliance
Turner Syndrome Foundation
Turner Syndrome Society of the United States
UMDF – The United Mitochondrial Disease Foundation
United Leukodystrophy Foundation
United Mitochondrial Disease Foundation
Usher 1F Collaborative
Usher Syndrome Coalition
WAGR Syndrome Association
WE ACT for Environmental Justice
Williams Syndrome Association
Wilson Disease Association
Wishes for Elliott: Advancing SCN8A Research
Wishes for Elliott/DEE-P Connections
X4 Health

For more information, please contact Sharon Terry at Genetic Alliance
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