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 X, 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,

5p- Society
AliveAndKickn
APS Foundation of America, Inc.
Barth Syndrome Foundation
Batten Disease Support and Research Association
Bridge the Gap – SYNGAP Education and Research Foundation
Canavan Foundation
Congenital Hyperinsulinism International
CureSHANK
Cure CMD
Cure HHT
Cure Sanfilippo Foundation
Cutaneous Lymphoma Foundation
debra of America
Dystrophic Epidermolysis Bullosa Research Association of America
Epilepsy Foundation
Epilepsy Leadership Council
FamilieSCN2A Foundation
Foundation for Prader-Willi Research
Genetic Alliance
Global Foundation for Peroxisomal Disorders
Glut1 Deficiency Foundation
Hydrocephalus Association
International Foundation for CDKL5 Research
International Pemphigus Pemphigoid Foundation
Life Raft Group
Lupus and Allied Diseases Association, Inc.
MitoAction
MLD Foundation
National Fabry Disease Foundation
National Neutropenia Network
National Society of Genetic Counselors
NBIA Disorders Association
Organic Acidemia Association
Phelan-McDermid Syndrome Foundation
PreventionGenetics
PXE International
Rett Syndrome Research Trust
SADS Foundation
The RUNX1 Research Program
Tuberous Sclerosis Alliance
UMDF – The United Mitochondrial Disease Foundation
Usher 1F Collaborative
Usher Syndrome Coalition
Wilson Disease Association
Wishes for Elliott/DEE-P Connections

For more information, please contact Sharon Terry at Genetic Alliance
• sterry@geneticalliance.org • 202.966.5557