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Senator Doug Jones  
330 Hart Senate Office Building  
Washington, DC 20510

Senator Susan Collins  
413 Dirksen Senate Office Building  
Washington, DC 20510

Senator Bob Menendez  
528 Hart Senate Office Building  
Washington, DC 20510

Senator Martha McSally  
404 Russell Senate Office  
Building Washington, DC 20510

Dear Senators Jones, Collins, Menendez, and McSally,

I am writing to thank you for introducing the Senate version of the “Ending the Diagnostic Odyssey Act.” It is wonderful to see growing bipartisan, and now bicameral, support for this effort.

For me, this is very personal, and a long time coming. My daughter and son were diagnosed in 1994 with a rare genetic condition after a long diagnostic odyssey. They were five and seven years old, and I had been trying to discover what was wrong with them for years. There was no option for affordable whole genome sequencing in those days. A diagnosis means a great deal to a family, as I am sure you know. Just having that information empowers us as parents. In my case, it allowed me to establish a patient advocacy foundation, build a research coalition, and eventually become CEO of Genetic Alliance to do the same for all diseases. I have a strong passion to see that all children receive a diagnosis in a timely manner.

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 from 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.

Knowing the genetic cause of a disease can mean an actionable diagnosis – leading to changes in treatment and management of the condition, prevent additional unnecessary testing, and help 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 when there is no treatment at the ready, just 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.

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.

This legislation has the potential to build upon the promises of the “21<sup>st</sup> Century Cures Act,” furthering the emerging field of precision medicine, and lowering health care costs by facilitating better diagnoses, and the consideration of preventive measures.

Thank you again for your leadership on this important legislation.

Sincerely,



Sharon F. Terry  
President and CEO

22Q Texas  
5p- Society  
Adult Polyglucosan Body Disease (APBD) Research Foundation  
AliveAndKickn  
All Things Kabuki  
Alstrom Syndrome International  
AMENSsupport  
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  
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  
Debra of America  
Dravet Syndrome Foundation  
Dup15q Alliance  
EB Research Partnership  
Epilepsy Foundation  
Epilepsy Leadership Council  
Familial Hypercholesterolemia 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  
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  
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 Foundation for Ectodermal Dysplasias  
National Neutropenia Network  
National Psoriasis Foundation (NPF)  
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  
PTEN World  
PXE International  
RARE-X  
RASopathies Network  
Rett Syndrome Research Trust  
Ring14 USA  
RUNX1 Research Program  
SCID Angels For Life Foundation  
Sickle Cell Thalassaemia 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 XYY Project  
Tuberous Sclerosis Alliance  
Turner Syndrome Foundation  
Turner Syndrome Society of the United States  
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  
X4 Health