

May 22, 2022

David Berglund, MD, MPH  
Medical Officer / Classification and Public Health Data Standards  
National Center for Health Statistics, Mailstop P08  
Metro IV, 2nd floor, Rm. 2534  
3311 Toledo Rd.  
Hyattsville, MD 20782

Dear Dr. Berglund,

As patient advocacy organizations, we represent thousands of patients, families and caregivers affected by rare, genetic, neurological disorders. Many of these disorders have only been diagnosed and described since the advent of genetic testing. The field of pediatric neurology has evolved rapidly in the last decade due to these advances in genetic testing. These are clinically distinct diseases caused by pathogenic genetic variants. Knowledge of the causal genes behind each disorder explains the mechanism of disease and points to treatments and even cures that were unimaginable twenty years ago.

For example, epilepsy caused by a sodium channel mutation, such as SCN2A Mutation, will need to be treated with a sodium channel medication. Movement issues caused by a citrate transport deficiency, such as SLC13A5 mutations, will need to be treated with precision medications aimed at citrate transport. Developmental delay stemming from the protein misfolding caused by mutations in the STXBP1 gene will likely benefit from a chemical chaperone. Without these specific genetic diagnosis codes in the medical record, it will be difficult to locate, treat, and track the health of these patients.

Please assign a unique ICD-10 code to each of these genetic conditions based on the causal gene.

Most sincerely,

Terry Jo V. Bichell, PhD, MPH  
Founder/Director, COMBINEDBrain  
Vice Chair, Tennessee Rare Disease Advisory Council

Tanya Brown, PhD  
Scientific Director



Terry Jo Bichell, PhD, MPH  
Founder/Director



Nuala Summerfield  
Founder & Chair, The Schinzel-Giedion Syndrome Foundation

Sandra Bedrosian Sermone  
President & CEO, ADNP Kids Research Foundation

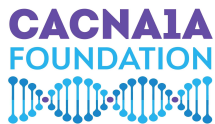


Understanding  
**ADNP SYNDROME**  
Helsmoortel - VanDerAa Syndrome

Nicole Johnson  
Co-founder, Executive Director



Lisa Manaster  
Co-founder and President



Geraldine Bliss  
President, CureSHANK



Amanda Johnson, BS  
Chief Development Officer  
ASXL Rare Research Endowment Foundation



Dominique Lessard, PhD  
Chief Scientific Officer, KIF1A



J. Michael Graglia, MBA MA  
Syngap Research Fund



Jeff D'Angelo  
Founder, President, Research Committee Chair, CHAMP1 Research Foundation



Ashley Fortney Point  
President of the Board, Koolen-de Vries Syndrome Foundation



Charlene Son Rigby  
CEO, RARE-X



Jackie Johnson Steinberg  
Vice President, STXBP1 Foundation



Amber Freed  
CEO & Founder, SLC6A1 Connect



Ben Hutz  
Co-Founder, SHANK2 Foundation



Carrie Howell  
Executive Director, Dup15q Alliance



Liz Marfia-Ash  
President & Founder, GRIN2B Foundation

