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,

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