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Epilepsia®

LETTER

Response: Do all individuals with Dravet syndrome have intellectual disability?

Dear Editor,

We are grateful for the opportunity to respond to the letter: *Do all individuals with Dravet syndrome have intellectual disability?* from Reilly and colleagues.[1]

The authors of the International League Against Epilepsy (ILAE) position paper on Epilepsy Syndromes in Neonates & Infants feel that intellectual disability (ID) is an important mandatory criterion differentiating Dravet syndrome from the many other epilepsy phenotypes within the genetic epilepsy with febrile seizures plus (GEFS+) spectrum, which are also associated with loss-of-function pathogenic variants in the *SCN1A* gene.[2] The task force did not define ID based on an IQ score, as there is a variability between individuals with borderline IQ scores in their degree of limitations in intellectual functioning and adaptive behavior. The mandatory features, alerts, and exclusionary criteria tables in the paper refer to the gold standard diagnosis of the established electroclinical syndrome not the syndrome in evolution.[2]

The ILAE developed the position papers because the boundaries of some epilepsy syndromes were not well delineated or previous studies had used broad or differing diagnostic criteria.[3] The syndrome diagnosis criteria, developed by two task forces, peer review and community engagement, provide a consensus to guide clinical practice and research.

The diagnosis of an epilepsy syndrome frequently carries prognostic and treatment implications, and as such it is important that individuals with epilepsies that are self-limited, or that will not be associated with ID, are distinguished from developmental and epileptic encephalopathies. The latest ILAE syndrome definition of Dravet syndrome uses ID to distinguish complex epilepsy phenotypes within the GEFS+ spectrum, which may be therapy resistant but not associated with ID, from Dravet syndrome.[4]

Reilly et al. in their population-based cohorts used diagnostic criteria for Dravet syndrome at onset from 2013 including: (1) normal electroencephalography (EEG) with no pre-existing cerebral lesion in a normal infant; (2) normal development until the first seizure occurring before 1 year of age; (3) refractory clonic or tonic–clonic seizures affecting one or both sides simultaneously or alternatively; and (4) exclusion of any other identified epilepsy syndrome including negative *PCDH19* analysis in *SCN1A*-negative patients.[5,6] This definition is relevant for early recognition of individuals who may develop Dravet syndrome at a stage when the syndrome is in evolution. However, many individuals with these clinical features will not have Dravet syndrome. The development of diagnostic prediction scores comprising age at onset and genetic data will provide greater confidence in diagnosis at an earlier stage in the evolution of the syndrome.[7,8]

CONFLICT OF INTEREST STATEMENT

The authors report no conflicts of interest relevant to the manuscript.

ETHICS STATEMENT

We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

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