How Do We Diagnose and Treat Epilepsy with Myoclonic-Atonic Seizures (Doose Syndrome)? Results of the Pediatric Epilepsy Research Consortium Survey

Abstract

Objective: To obtain and assess opinions on EMAS diagnostic criteria, recommended investigations, and therapeutic options, from a large group of physicians who care for children with EMAS.

Methods: The EMAS focus group of PERC created a survey to assess the opinions of pediatric neurologists who care for children with EMAS regarding diagnosis and treatment of this condition, which was sent to members of PERC, AES, and CNS. A Likert scale was used to assess the respondents’ opinions on the importance of diagnostic and exclusion criteria (five point scale), investigations (four point scale), and treatment (six point scale) of EMAS. Inclusion/exclusion criteria were then classified as critical, strong, or modest. Investigations were classified as essential, recommended, or possible. Therapies were classified as first line, beneficial, indeterminate benefit, or contraindicated.

Results: Survey results from the 76 participants determined the following:

EMAS inclusion criteria: history suggestive of MAS (critical), recorded or home video suggestive of MAS, generalized discharges on inter-ictal EEG, normal neuroimaging, normal development prior to seizure onset (strong).

EMAS exclusionary criteria: epileptic spasms, abnormal neuroimaging, focal abnormal exam, seizure onset <six months or >six years (strong).

Recommended investigations: EEG and MRI (essential), amino acids, organic acids, fatty acid/acylcarnitine profile, microarray, genetic panel, lactate/pyruvate, CSF and serum glucose/lactate (strong).

Recommended treatments: valproic acid (first line), topiramate, zonisamide, levetiracetam, benzodiazepines, and dietary therapies (beneficial).

Significance: To date, no similar surveys have been published, even though early syndrome identification and initiation of effective treatment have been associated with improved outcome in EMAS. Medications that exacerbate seizures in EMAS have also been identified. This survey identified critical and preferred diagnostic electro clinical features, investigations, and treatments for EMAS. It will guide future research and is a crucial first step in defining specific diagnostic criteria, recommended evaluation, and most effective therapies for EMAS.

Key Words: EMAS, Doose, epileptic encephalopathy, evaluation, treatment