Appendix

EMAS survey questions and possible responses sent to PERC, CNS, and AES.

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| 1. What is your primary profession?   Adult neurologist  Adult epileptologist  Child and adolescent neurologist  Child and adolescent epileptologist | | | |
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| 1. Are you willing to take this survey?   Yes  No, I do not care for patients with Epilepsy with Myoclonic-Atonic seizures (Doose Syndrome) | | | |
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| 1. When making the diagnosis of Epilepsy with Myoclonic-Atonic Seizures (Doose syndrome), how important are the following criteria? 5 point Likert scale: 2. Not all important, its presence or absence provides no helpful information 3. Mildly important 4. Moderately important 5. Very important but not essential to making the diagnosis, providing other factors are supportive 6. Essential to making the diagnosis | | | |
| * Reported myoclonic atonic seizures on EEG (with or without video correlate) * History suggestive of myoclonic atonic seizures * Parent/home video suggestive of myoclonic atonic seizure * Presence of other generalized seizures (absence, tonic, GTC, myoclonic, atonic) | | * Generalized SW discharges on inter-ictal EEG * Normal neuro imaging * Normal development prior to seizure onset * Family history of seizures/epilepsy * Diffuse theta with centroparietal predominance | |
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| 1. When the following “red flag” are present, how likely are you to question whether the diagnosis of Epilepsy with Myoclonic-Atonic Seizures (Doose syndrome) is correct? 5 point Likert scale: 2. Would not question diagnosis at all 3. Argues slightly against diagnosis 4. Argues moderately against diagnosis 5. Argues highly against diagnosis, but does not exclude diagnosis 6. Exclude the diagnosis | | | |
| * Onset before age 6 months or after 6 years * Developmental delay prior to seizure onset * Low CSF glucose * Focal seizures * Tonic seizures | | * Epileptic spasms * Focal epileptiform discharges or background slowing * Abnormal neuroimaging * Febrile status epilepticus * Focal abnormalities on neurologic exam | |
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| 5. In what circumstances would request the following investigations for a patient with possible Epilepsy with Myoclonic-Atonic Seizures (Doose syndrome)? 4 point Likert scale:   1. Always would request this 2. Would request if there are even minor atypical clinical features 3. Would request only if more significant atypical clinical features 4. Would only request if I thought this child did NOT have Epilepsy with Myoclonic Atonic Seizures | | | |
| * Routine EEG (with or without video) * Prolonged video EEG * Microarray * Epilepsy Genetic Panel * Specific gene testing * CSF and serum glucose * Whole Exome Sequencing | | * Serum quantitative amino acids * Urine organic acids * Electrolytes * Fatty acid oxidation profile/acylcarnitine profile * Lactate, pyruvic acid * Additional metabolic testing * MRI brain | |
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| 1. Specify the gene testing you would order. | | | |
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| 1. Specify the additional metabolic testing you would order. | | | |
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| 1. Please rate the following treatment choices for Epilepsy with Myoclonic-Atonic Seizures (Doose syndrome). 6 point Likert scale: 2. I consider this 1st line therapy (1 of my 1st 2 choices) 3. I recommend this early but not 1st line (choice 3 or 4) 4. I recommend this only after several therapies have failed (choice 5 or 6) 5. This could be considered but very late in the course (choice 7 or later) 6. I would not use this in MAE 7. No experience/no data with this treatment in MAE | | | |
| * Acetazolamide * Benzodiazepines (clonazepam, clorazepate, clobazam) * Carbamazepine * Eslicarbazepine * Ethosuximide * Ezogabpine * Felbamate * Gabapentin * Lacosamide | * Lamotrigine * Levetiracetam * Oxcarbazepine * Parampanel * Phenobarbital * Phenytoin * Pregabalin * Primidone * Rufinamide | * Sulthiame * Tiagabine * Topiramate * Valproic acid * Vigabatrin * Zonisamide * Folate/folinic acid * Pyridoxine/pyridoxal 5 phosphate * Ketogenic diet | * Modified Atkins diet * Low glycemic index diet * Medical marijuana (or its derivatives) * IVIG * Corticosteroids (any formulation) * Vagal nerve stimulation (VNS) * Corpus callosotomy * Deep brain stimulation (DBS) |
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| 1. Do you have any other comments, questions, or concerns? | | | |