Clinical Presentation

Please complete the survey below.

Thank you!

In your patients with EMAS-AFTER the initial afebrile seizure WHEN FIRST did you suspect this diagnosis? (Specify percentages at the following time points .Your answers do not have to add up to 100%):

Percentage of patients where you had a strong clinical suspicion for EMAS- within 1 month of first afebrile seizure:	_____
Percentage of patients where you had a strong clinical suspicion for EMAS- 1- 3 months after first afebrile seizure?	_____
Percentage of patients where you had a strong clinical suspicion within 3-6 months after first afebrile seizure?	_____
Percentage of patients where you had a strong clinical suspicion within 6-12 months after first afebrile seizure?	_____
Percentage of patients where you had a strong clinical suspicion within 12-18 months of first afebrile seizure?	_____
What is the youngest age of afebrile seizure onset that you have seen with a definitive evolution of EMAS (in years)?	_____
What is the oldest age of afebrile seizure onset that you have seen with a definitive evolution of EMAS (in years)?	_____

What % of your patients with EMAS have the following development at time of seizure onset:

Percent of patients without clear delay:	_____
Percent of patients with suspected developmental delay	_____
Percent of patients with clear mild delay:	_____
Percent of patients with moderate or greater delay:	_____

Evolution of different seizure types: For the next 2 questions we would like to assess what types and percentage of seizures are seen for the FIRST TIME and whether they are seen at onset versus seen later in the course of EMAS.

For each of the following seizure type(s) indicate the percentage when they are seen for the first time within 6-12 months of onset of first afebrile seizure

	< 25%	25-50%	50-75%	>75%
GTCS- afebrile	○	○	○	○
Myoclonic	○	○	○	○
Myoclonic- atonic	○	○	○	○
Atonic	○	○	○	○
Atypical absence	○	○	○	○
Tonic	○	○	○	○
Nonconvulsive status epilepticus	○	○	○	○

For each of the following seizure type(s) indicate the percentage when they are seen for the first time AFTER 12 months of onset of the first afebrile seizure- please do not carry over patients that continue to have seizure types that had started earlier.

	< 25%	25-50%	50-75%	>75%
GTCS-afebrile	○	○	○	○
Myoclonic	○	○	○	○
Myoclonic-atonic	○	○	○	○
Atonic	○	○	○	○
Atypical absence	○	○	○	○
Tonic	○	○	○	○
Nonconvulsive status epilepticus	○	○	○	○

For each of the following seizure types indicate whether they are mandatory for diagnosis, typically seen but not mandatory, occasionally seen or rarely seen

	Mandatory for diagnosis of EMAS	Usually (>90%) seen but not mandatory for diagnosis	Often seen (50-90%)	Occasionally seen (10-49%)	Rarely seen (< 10%)
GTCS-afebrile	○	○	○	○	○
Myoclonic	○	○	○	○	○
Myoclonic-atonic	○	○	○	○	○
Atonic	○	○	○	○	○
Atypical absence	○	○	○	○	○
Tonic	○	○	○	○	○
Nonconvulsive status epilepticus	○	○	○	○	○

Some clinicians describe a "stormy" course in children with EMAS, or a state of subacute exacerbation of symptoms: where there are typically recurrent bouts of non-convulsive status epilepticus and an increase in other types of generalized seizures including generalized tonic clonic, atonic and tonic seizures, as well as absence and myoclonic seizures.

In what proportion of your patients is this stormy course seen?

- ◯ < 25%
- ◯ 25-50%
- ◯ 50-75%
- ◯ >75%

What % of your patients with EMAS show evidence of a stormy course at the following time points after first afebrile seizure onset

Percentage of patients within 0-3 months after first afebrile seizure onset: _____

Percentage of patients within 3-6 months after afebrile seizure onset: _____

Percentage of patients within 6-12 months after afebrile seizure onset: _____

Percentage of patients within after 12 months of afebrile seizure onset: _____

If you see patients develop a stormy course, how soon after initial seizure onset does this typically occur (give range in months): _____

How long does this stormy course typically last (give range in months): _____

Do you believe developmental regression is also part of the stormy course?

- ◯ Yes
- ◯ No

Do you see a gender difference in the occurrence of EMAS?

- ◯ Yes
- ◯ No

What proportion of your patients with EMAS are boys?

- ◯ < 25%
- ◯ 25-50%
- ◯ 50-75%
- ◯ >75%

What proportion of your patients with EMAS have a prior history of febrile seizures?

- ◯ < 25%
- ◯ 25-50%
- ◯ 50-75%
- ◯ >75%

What proportion of your patients with EMAS have a family history of epilepsy (not febrile seizures) in first degree relatives?

- ◯ < 25%
- ◯ 25-50%
- ◯ 50-75%
- ◯ >75%

What proportion of patients have history of epilepsy in extended family (grandparents, aunts, uncles, cousins etc.)?	◯ < 25% ◯ 25-50% ◯ 50-75% ◯ >75% ◯ Have not asked this routinely
What other features do you typically observe, and when do you see these? You can choose more than one and each choice will have additional clarifying questions	☐ Developmental plateauing ☐ Developmental regression ☐ Hyperactive behavior ☐ Ataxia ☐ Other
% of children with EMAS who develop developmental plateauing:	◯ < 25% ◯ 25-50% ◯ 50-75% ◯ >75%
Developmental plateauing seen even in those without a stormy course?	◯ Yes ◯ No
Developmental plateauing seen mostly in those with a stormy course, or much worse during the stormy course:	◯ Yes ◯ No
% of children with EMAS who develop developmental regression:	◯ < 25% ◯ 25-50% ◯ 50-75% ◯ >75%
Developmental regression seen even in those without a stormy course?	◯ Yes ◯ No
Developmental regression seen mostly in those with a stormy course, or much worse during the stormy course:	◯ Yes ◯ No
% of children with EMAS who develop hyperactive behavior:	◯ < 25% ◯ 25-50% ◯ 50-75% ◯ >75%
Hyperactive behavior seen even in those without a stormy course?	◯ Yes ◯ No
Hyperactive behavior seen mostly in those with a stormy course, or much worse during the stormy course:	◯ Yes ◯ No
% of children with EMAS who develop ataxia:	◯ < 25% ◯ 25-50% ◯ 50-75% ◯ >75%
Ataxia seen even in those without a stormy course?	◯ Yes ◯ No
Ataxia seen mostly in those with a stormy course, or much worse during the stormy course:	◯ Yes ◯ No

Please specify "other":	_____
% of children with EMAS who develop "other":	◯ < 25% ◯ 25-50% ◯ 50-75% ◯ >75%
"Other" seen even in those without a stormy course?	◯ Yes ◯ No
"Other" seen mostly in those with a stormy course, or much worse during the stormy course:	◯ Yes ◯ No
What proportion of your patients with EMAS experience remission of seizures (does not matter if they are still on meds)?	◯ < 25% ◯ 25-50% ◯ 50-75% ◯ >75%
In those with seizure control, how soon after onset does that typically occur? (Given range in months)	_____

In those who experience seizure control, what percentage fall into the following developmental categories

	< 25%	25-50%	50-75%	>75%
Normal development	◯	◯	◯	◯
Mild learning disability but normal IQ	◯	◯	◯	◯
Mild ID	◯	◯	◯	◯
Moderate ID	◯	◯	◯	◯
Severe to profound ID	◯	◯	◯	◯

In those who experience remission of seizures, what percentage are left with symptoms of ADHD?	◯ < 25% ◯ 25-50% ◯ 50-75% ◯ >75%

In those who DO NOT experience remission of seizures, what percentage fall into the following developmental categories?

	< 25%	25-50%	50-75%	>75%
Normal development	◯	◯	◯	◯
Mild learning disability but normal IQ	◯	◯	◯	◯
Mild ID	◯	◯	◯	◯
Moderate ID	◯	◯	◯	◯
Severe to profound ID	◯	◯	◯	◯

Prognostic Factors

Please complete the survey below.

Thank you!

How predictive are the following features on adverse long term outcome (ongoing seizures and more severe cognitive delays).

	Not at all predictive	Mildly predictive of poor outcome	Moderately predictive of poor outcome	Highly predictive of poor outcome
Tonic seizures of any type	◯	◯	◯	◯
Vibratory tonic seizures	◯	◯	◯	◯
Greater numbers of NCSE episodes	◯	◯	◯	◯
Longer duration of NCSE episodes	◯	◯	◯	◯
Interictal EEG outside of stormy period showing very frequent or near continuous irregular generalized spike-wave	◯	◯	◯	◯
Interictal EEG outside of stormy period showing slow spike wave activity	◯	◯	◯	◯
Interictal EEG outside of stormy period showing paroxysmal fast activity	◯	◯	◯	◯
High frequency of drops/ myoclonus at presentation	◯	◯	◯	◯
GTCS in first two years after seizure onset	◯	◯	◯	◯
Family history of epilepsy	◯	◯	◯	◯
Focal spikes	◯	◯	◯	◯
Earlier age at onset (i.e < 2years)	◯	◯	◯	◯
Later age at onset (i.e > 6 years)	◯	◯	◯	◯

Are there other predictive features not mentioned in the above list?

◯ Yes
◯ No

If "yes" please specifiy : you can type in response here

_____

If "yes" to other how predictive is that feature? We are trying to gauge how important you feel this other factor is in predicting outcome.

◯ Not at all predictive
◯ Mildly predictive of poor outcome
◯ Moderately predictive of poor outcome
◯ Highly predictive of poor outcome

Do you ever reclassify the diagnosis of EMAS to ANY alternative (epilepsy syndrome) diagnosis?

◯ Yes
◯ No

How often do you reclassify?

- ◯ Usually
- ◯ Sometimes
- ◯ Frequently

Which of the following factors make you consider reclassification of your diagnosis of EMAS to ANY alternative diagnosis?

	High impact	Low impact	No impact on reclassification
Tonic seizures of any type	◯	◯	◯
Vibratory tonic seizures	◯	◯	◯
Greater numbers of NCSE episodes	◯	◯	◯
Longer duration of NCSE episodes	◯	◯	◯
EEG showing very frequent or near continuous irregular generalized spike wave	◯	◯	◯
EEG showing slow spike wave activity	◯	◯	◯
EEG showing paroxysmal fast activity	◯	◯	◯
High frequency of drops/myoclonus at presentation	◯	◯	◯
GTCS in first two years after seizure onset	◯	◯	◯
Family history of epilepsy	◯	◯	◯
Focal spikes	◯	◯	◯
Earlier age at onset (i.e < 2 years)	◯	◯	◯
Later age at onset (i.e >6 years)	◯	◯	◯
Persistence of seizures >1 year after seizure onset	◯	◯	◯
Persistence of seizures> 2 years after seizure onset	◯	◯	◯
Persistence of seizures> 3 years after seizure onset	◯	◯	◯
Persistence of seizures> 4 years after seizure onset	◯	◯	◯
Persistence of seizures> 5 years after seizure onset	◯	◯	◯
Lack of response to ketogenic diet	◯	◯	◯

If "Tonic seizures of any type" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "vibratory tonic seizures" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "greater numbers of NCSE episodes" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "longer duration of NCSE episodes" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "EEG showing very frequent or near continuous irregular generalized spike-wave" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "EEG showing slow spike wave activity" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "EEG showing paroxysmal fast activity" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "High frequency of drops/myoclonus at presentation" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "GTCS in first two years after seizure onset" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "family history of epilepsy" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "focal spikes" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "Earlier age at onset (i.e < 2 years)" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed): _____

If "Later age at onset (i.e >6 years)" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed):	_____
If "Persistence of seizures> 1 year after seizure onset" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed):	_____
If "Persistence of seizures> 2 years after seizure onset" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed):	_____
If "Persistence of seizures> 3 years after seizure onset" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed):	_____
If "Persistence of seizures> 4 years after seizure onset" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed):	_____
If "Persistence of seizures> 5 years after seizure onset" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed):	_____
If "Lack of response to ketogenic diet" is a factor for reclassification is this factor sufficient in isolation or other factor is needed (elaborate if other factors are needed):	_____

Investigations

Please complete the survey below.

Thank you!

What tests do you perform to establish a diagnosis of EMAS? Note that these tests are not meant to establish etiology of EMAS.

	Always	Usually	Sometimes	Rarely	Never
EEG	◯	◯	◯	◯	◯
Epilepsy panel	◯	◯	◯	◯	◯
Whole exome sequencing	◯	◯	◯	◯	◯
CMA/karyotype	◯	◯	◯	◯	◯
PET scan	◯	◯	◯	◯	◯
MRI	◯	◯	◯	◯	◯
Neurophsychological testing	◯	◯	◯	◯	◯
CSF testing	◯	◯	◯	◯	◯
Metabolic testing	◯	◯	◯	◯	◯

Are there "other" tests performed to establish a diagnosis of EMAS?

◯ Yes
◯ No

If "yes" please specify:

_____

If "yes" to other tests performed how often are they performed to establish a diagnosis of EMAS?

◯ Always
◯ Usually
◯ Sometimes
◯ Rarely
◯ Never

If glucose transporter deficiency is established would you still consider the diagnosis to be EMAS?

◯ Yes
◯ No

What tests do you perform to establish an etiology in EMAS?

	Always	Usually	Sometimes	Rarely	Never
EEG	◯	◯	◯	◯	◯
Epilepsy panel	◯	◯	◯	◯	◯
Whole exome sequencing	◯	◯	◯	◯	◯
CMA/karyotype	◯	◯	◯	◯	◯
PET scan	◯	◯	◯	◯	◯
MRI	◯	◯	◯	◯	◯
Neuropsychological testing	◯	◯	◯	◯	◯
CSF testing	◯	◯	◯	◯	◯
Metabolic testing	◯	◯	◯	◯	◯

Other	◯	◯	◯	◯	◯

What tests do you use for ongoing care of patients diagnosed with EMAS?

	Always	Usually	Sometimes	Rarely	Never
EEG	◯	◯	◯	◯	◯
Epilepsy panel	◯	◯	◯	◯	◯
Whole exome sequencing	◯	◯	◯	◯	◯
CMA/ Karyotype	◯	◯	◯	◯	◯
PET scan	◯	◯	◯	◯	◯
MRI	◯	◯	◯	◯	◯
Neuropsychological testing	◯	◯	◯	◯	◯
CSF testing	◯	◯	◯	◯	◯
Metabolic testing	◯	◯	◯	◯	◯
Other	◯	◯	◯	◯	◯

Do you refer children for a neuropsychological evaluation?

◯ Yes
◯ No

If "yes" please chose an option

◯ < 25%
◯ 25-50%
◯ 50-75%
◯ >75%
◯ Only if not doing well

If "No" what factors lead to a referral?

_____

Treatment

Please complete the survey below.

Thank you!

What therapy should be avoided in treatment of EMAS?

_____

Please list the top two choices of therapy for EMAS

Therapy 1:

_____

Therapy 2:

_____

If the first Therapy has failed list the next 2 choices for therapy

Next therapy 1:

_____

Next therapy 2:

_____

If your first and second line of therapy have failed what would you choose next to treat EMAS?

	Most likely to prescribe	Second best option	Third best option	Will not prescribe
Valproic acid	◯	◯	◯	◯
Levetiracetam	◯	◯	◯	◯
Clobazam	◯	◯	◯	◯
Topiramate	◯	◯	◯	◯
Zonisamide	◯	◯	◯	◯
Lamotrigine	◯	◯	◯	◯
Epidiolex	◯	◯	◯	◯
Artisanal cannabinoids	◯	◯	◯	◯
Ethosuximide	◯	◯	◯	◯
Chlorazepate	◯	◯	◯	◯
Clonazepam	◯	◯	◯	◯
Sulthiame	◯	◯	◯	◯
Phenobarbital	◯	◯	◯	◯
Carbamazepine	◯	◯	◯	◯
Oxcarbazepine	◯	◯	◯	◯
Phenytoin	◯	◯	◯	◯
Vigabatrin	◯	◯	◯	◯

Felbamate	◯	◯	◯	◯
Perampanel	◯	◯	◯	◯
Ketogenic diet	◯	◯	◯	◯
Vagus nerve stimulator	◯	◯	◯	◯
Corpus callosotomy	◯	◯	◯	◯
Responsive neurostimulation	◯	◯	◯	◯
Some other surgical option	◯	◯	◯	◯

Do you ever consider corpus callosotomy in treatment of EMAS?
◯ Yes
◯ No

If "yes" describe clinical scenario (be specific-after how many therapies have failed AND/OR for what seizure types)
_____

Do you ever consider VNS in treatment of EMAS?
◯ Yes
◯ No

If "yes" describe clinical scenario (be specific-after how many therapies have failed AND/OR for what seizure types):
_____

What treatments do you find MOST useful for the stormy phase in EMAS?
_____

What treatments do you find MOST useful for non-convulsive status epilepticus in EMAS?
_____