

RettEd Q&A: Epilepsy Incidence, Treatments and Research

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Recording link: <https://attendee.gotowebinar.com/recording/2303318923907771398>

ATTENDEE QUESTIONS	RESPONSE	REFERENCES
AED Questions: Treatments and Triggers		
I want to know what about relation between pain, digestive pain, apnea and seizure discharge.	Stressors are anecdotally reported to lower the seizure threshold- these would all be stressful features that could lower seizure threshold and theoretically increase seizure frequency.	
Any opinions on fycopma?	Very limited experience so far to make a clear judgment. I have tried it in 2 Rett patients without great success, both in patients with very severe seizures.	
Is there a way to tell the difference between a seizure and a “Rett spell” other than EEG?	There is a slide in the talk that goes over some features, such as retained awareness, repetitive nature of the events, that can give some hints as to the nature of the event, but only EEG can definitively discern the difference.	
My daughter has epilepsy and always has seizures. She takes medication Onfi and Keppra and still have seizures. What should I do? I need help and ideas thanks.	Without knowing all of her clinical details, it is hard for me to give recommendations. Depending on seizure types, EEG, and previous medications tried, I would suggest other meds such as Rufinamide, valproic acid or trying the ketogenic diet.	
Do you see seizures connected to puberty or periods?	You can look at Jane Lane’s RettEd about puberty . Seizures are hormonally responsive and having increase in seizures around the period is a well described issue that can be treated with hormonal therapies (oral contraceptives).	
I am interested in learning about triggers (full	As stated above, people have reported weather,	

<p>moon and ovulation times are 2 of them and primary to my daughter) so I can watch and working on prevention with medicine increase, hormonal support (not interested in birth control). Thank you.</p>	<p>environmental triggers for seizures. Clearly, perimenstrual seizures are well described in the literature. Hormonal therapies can be used these are termed oral contraceptives, but are the appropriate hormonal therapies. Increasing a baseline medication or using a benzodiazepine to bridge during known triggers can be considered.</p>	
<p>My granddaughter is 9 and has severe breathing holding spells. It starts at wake up time and almost immediately ends up with a seizure. Often a cluster. She is not on any epilepsy medication as her parents fear the devastating side effects</p>	<p>The family has to first determine if these are definitely seizures. If they are, then to decide to treat or not is a conversation with the neurologist.</p>	
<p>If she stopped having seizures for 4 years, is it possible to have them again? Sometimes she has symptoms similar to what she had after the seizures.</p>	<p>Yes- seizures can come and go in Rett. It is worth trying to take her off medication if she has been seizure free for a long time.</p>	
<p>My daughter is on Kepra, Onfi, Lamictal plus VNS and still continues to have seizures. Is there something else we can try?</p>	<p>Yes- there are a number of other medications, the ketogenic diet and if they also have an electroclinical diagnosis of Lennox-Gastaut Syndrome, the soon to be available CBD could be tried.</p>	
<p>My daughter has been on anti-seizure medication since she was 3. She is 33 today. As of March 2018 her meds are administer via her g-tube. She has gained some weight and wondering if the meds need to be changed as the seizures are breaking</p>	<p>Yes- this would suggest that she may need a medication adjustment. This should be weighed with current side effects and how the seizures affect her. Need to balance seizures and side effects.</p>	

through again.		
Our daughter is currently on epilim, clobazam and tegretol (max doses) and still having daily seizures. We are desperate for advice on where to go next. We realize we may not be able to achieve seizure free periods however her epilepsy is so uncontrolled.	Without further information, it is difficult to give specific recommendations. There are a number of other medications, VNS, and the ketogenic diet to try. You also should discuss with the neurologist the balance between side effects and seizures as if the seizures are brief and not causing any injury, then tolerating more with fewer meds could be considered.	
Rescue medications – what are they, how are they administered, when do we use them and how well do they work short and long term?	Rescue meds are typically benzodiazepines. This is used as they are fast onset, very effective to stop seizures, and can be administered in different ways (by mucosa (rectally, cheek, nose), by stomach, or by IV or intramuscular). Benzos can be used as daily treatment but due to sedation, getting used to them and difficulty to stop, they are primarily used as rescue, except for Onfi which is a unique type of benzo.	
Genetic tests/predictors		
What about these gene tests to tell which anti-seizure drugs might be more effective, and which may not be helping much at all in an individual. What do you think? Are they useful and/or reliable?	As of now, none of the genetic tests that are said to predict responsiveness of seizures to medications very well and are better at predicting side effects. None of these have even been thought about for kids with Rett or other severe epilepsy syndromes.	
Is there any reason to screen for the p. Val66Met polymorphism in the BDNF gene? She has intractable epilepsy. Has had multiple EEGs, admissions, tried many	No- there is no clinical reason to do this at this time. There is some data (see two references at right) that this variant may predict who will develop seizures earlier or has more severe disease but this information will not change treatment at this	1. Neurology . 2008 May 27; 70 (22 Pt 2): 2145-51. 2. Neurology . 2009 Apr 7; 72(14): 1242–1247. PMID: 19349604

<p>AEDs, keto diet, VNS, CBD, and is not usually responsive to rescue medications. If she has this polymorphism, could it point us in a new direction for care? Please explain why or if we should pursue this test.</p>	<p>time.</p>	
<p>Males with Rett, MECP2 mutations/deletions, or CDKL5</p>		
<p>My son has Cdkl5 Deficiency Disorder and I am interested in this information as it may apply to us too. Thank you!</p>	<p>The general seizure information holds for CDD kids as well. The Rett specific does not, but one slide in the talk showed seizures in CDD which spasms are very common where they are not in Rett.</p>	
<p>Anxious to hear ANY info on boys w/ early diagnosis (1yr old)</p>	<p>There will be a paper coming out soon about the natural history study experience with MECP2 mutation boys. There is variability in the severity and symptoms in this group. More than people have thought.</p>	
<p>Was hoping to hear more about boys w/ Rett. My patient was diagnosed at 1 yr old, told had 1-2 yrs to live. Trach/ vent dependent Once started on felbamate after Onfi, seizures cut by 50 q 2 days to 6 in 2 days. Has been way more alert, eyes open 30 min. instead of closed, better neck control. Getting stander to wk up to ambulation. Does Dr Marsh feel as hopeful</p>	<p>With good seizure control I am not very surprised that your patient has some improvements in function. I am always concerned that if seizures worsen some of the gains you see may dissipate. The Natural History Study group is thinking about these boys and including them in research which is coming in the future.</p>	

<p>w/ these outcomes as we are? Can they expect further drop in seizures, can they plateau, or like w/ Onfi will they drop/ then return to previous levels? I know stats/docs are rare in boys w/ Rett. Family recently went to Atlanta, to Center for Rare Neurological Diseases who's doing prelims for research. Does Dr. Marsh know of any more options of studies? My patient is 17mo. Had lost all purposeful movements exc seizures. Now is blowing kisses, reaching for things. Had eyes open on flight to Atlanta!! Parents very knowledgeable! Ck out FB for WARRIOR WILLIAM, mom's blog. Any info, mom can be reached there.</p>		
<p>CBD and Medical Marijuana Questions</p>		
<p>I'm interested in hearing opinion on medical cannabis for treatment of seizures and/or "Rett episodes".</p>	<p>FDA cleared CBD from Greenwich Pharma will be an option in Rett syndrome patients with Lennox-Gastaut epilepsy. I would avoid over the counter CBD as the preparations are not very trustworthy. As for using CBD for treatment of Rett episodes, we don't have good data on that.</p>	
<p>Can CBD replace the other AEDs? My daughter is on CBD and AED combination and her spasms are refractory. Currently she takes vigabatrin, sodium valporate, rivotril,</p>	<p>CBD should be added to existing meds. If seizures improve on the combination, then you can wean other meds. For this case, I would try to first simplify her regimen to figure out if anything is working before making other changes.</p>	

meloset, hemp oil plus ketogenic diet. Please suggest what should be done.		
What studies have been done specifically on Rett syndrome and CBD oil? If none have been done when will there be a study done?	None- GW/Greenwich pharma is considering a study for CBD in Rett syndrome.	
Various types of marijuana use and their names?	THC is the component that gets people 'high'. CBD does not give a 'high' but works for seizures. There are over 80 compounds in marijuana that may have biological activity. THC and CBD are the most abundant in the plant making them easiest to study.	
Is the new CBD-based drug sustained release?	No- it is twice a day dosing. The half life of CBD is about 8-12 hours so it can't be dosed only once daily.	
My daughter seems to be developing tolerance to CBD which requires increasing the dosages. Is that common and what is an upper limit of CBD before it causes significant side effects?	This is uncommon in my experience where patients can stay on the same dose of CBD for years. No one really knows the upper limit, a few doctors who were part of the CBD national expanded access program pushed CBD to as high as 50mg/kg/day- no additional side effects were present but also no additional improvement in seizures.	Devinsky et al, 2016, Lancet Neurology
Can CBD full spectrum oil help with seizures and or rigidity?	CBD has shown efficacy in epilepsy and spasticity. In spasticity, some THC may be needed.	