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Experiences with epilepsy treatments: a qualitative content analysis of online patient support group discussions

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doi: 10.1017/cjn.2017.118

Background: To promote patient-centred care in epilepsy, it is essential to understand the issues most important to patients. Literature on patient perceptions of epilepsy treatments is sparse. One source of data is online patient support groups. Patients turn to social media for support from other patients and often express viewpoints not shared with healthcare providers. *Methods:* Using a qualitative content analysis approach, we analyzed major online epilepsy patient support groups. We initially selected a month-long discussion text across these forums, and further threads were sampled with maximum variation until theme saturation was reached. For data coding and analysis, we employed a combination of a priori codes and emergent codes, using NVivo 11 for data analysis. *Results:* In our preliminary analysis, we identified topics and categorized them into themes: 1) differential perceptions and understandings of epilepsy; (2) understanding treatment options; (3) experiences of physiological and psychological treatment side effects; (4) concerns about healthcare providers' knowledge and communication regarding treatments. *Conclusions:* Preliminary results indicate a variety of patient perceptions and understandings of epilepsy and its treatments. Our findings also suggest that patient educational needs should be addressed by incorporating their understanding and concerns. Shared-decision making tools informed by patient perceptions could help healthcare providers better communicate treatment options with patients.

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Focal cortical dysplasia type IIIb associated with oligodendroglioma in a seizure free patient: a case report

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doi: 10.1017/cjn.2017.119

Background: Focal cortical dysplasia (FCD) refers to malformation of cortical development featuring abnormalities of cortical layering, neuronal differentiation and maturation. It is a common cause of medically refractory epilepsy. The coexistence of FCD and low-grade glial neoplasms such as ganglioglioma and dysembryoplastic neuroepithelial tumour is classified by the International League Against Epilepsy as "FCD Type IIIb". We present a case of FCD Type IIIb associated with low grade oligodendroglioma (WHO grade II) in a seizure free patient. *Methods:* A 20-year-old male presented with suspected arteriovenous malformation of the right pinna. Imaging revealed an incidental right frontal lobe mass. Surgical resection was performed. Pathologic analysis revealed FCD Type IIIb associated with low grade oligodendroglioma (WHO grade II). *Results:* The patient recovered uneventfully. Only 4 prior cases of FCD Type IIIb associated with oligodendroglioma have been reported. This is the first reported case of FCD Type IIIb discovered incidentally in a seizure free patient. *Conclusions:* FCD Type IIIb associated with oligodendroglioma is rare. The mechanism(s) by which glioneuronal neoplasms and perilesional cortical tissue jointly contribute to epileptogenicity have not been clearly defined. There may be a reduced risk of seizures with oligodendroglioma rather than tumors with a neuronal component.

P.035

Correlation of thalamic connectivity with the duration of epilepsy in patients with temporal lobe epilepsy

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doi: 10.1017/cjn.2017.120

Background: Morphometry and connectivity studies targeting the thalamus have revealed specific patterns of atrophy and deafferentiation in patients with temporal lobe epilepsy (TLE). We used probabilistic tractography to investigate thalamic connectivity with respect to duration of epilepsy and surgical outcomes in TLE. *Methods:* Patients (N=20) with drug-resistant TLE (10 short duration (<15 years), 10 long duration (>15 years)) were scanned with multi-parametric 3T MRI and compared with 34 healthy controls. The Harvard-Oxford atlas was modified to create 14 target regions in the temporal lobes. Probabilistic tractography (FSL) was used to delineate thalamic subregions most connected to each target. The volume, mean T1, T2, FA and MD of each thalamic sub-region was quantified. Surgical success was quantified using Engel outcome scores. *Results:* Significant decreases in thalamic connected volumes to the hippocampus in patients with longer duration of TLE were revealed. Likewise, when stratified based on surgical success, significant differences in diffusion metrics to the hippocampus, parahippocampal gyrus, and temporal neocortex were found. Significant differences did not withstand false discovery rate (FDR) correction. *Conclusions:* These findings suggest ongoing connectivity changes dependent on epilepsy duration and promote further investigation into the use of thalamic connectivity data as biomarkers for predicting surgical outcomes in TLE patients.

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Targeted molecular therapy with quinidine for seizures in a neonate with KCNT1 mutation leads to poor response

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doi: 10.1017/cjn.2017.121

Background: The KCNT1 gene encodes subunits of the Na+-activated K+ channel, widely expressed in the CNS. Mutations of this gene have been implicated in Malignant Migrating Partial Seizures of Infancy (MMPSI). This early-onset epileptic encephalopathy represents a challenge due to pharmacoresistance. The channel-specific mutation represents the potential for targeted pharmacotherapy. Quinidine is a partial antagonist of the KCNT1 encoded channel; patients with MMPSI have been reported to have responded to doses ranging 34.4/kg/d - 60mg/kg/d. We present a case of MMPSI with a KCNTI mutation (c.G1283A:p.R428Q) trialled on quinidine. *Methods:* Following ineffective trials of 6 anti-seizure medications, this patient was trialled on oral quinidine. This patient was titrated up to a dose of 52mg/kg/d. Twenty-four hour EEG monitoring prior to quinidine therapy, and at target dose were compared. *Results:* Prior

to initiation of quinidine, this patient experienced 22 electrographic seizures over 24 hours. At target dose, this patient experienced greater than 70 seizures over 24 hours. *Conclusions:* Quinidine has previously been reported to be effective in patients with MMPSI with the same and different mutations. We report the second case of a patient with MMPSI and KCNT1 mutation R428Q with poor clinical response to quinidine.

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Role of epilepsy monitoring unit in the investigation of patients with epilepsy and developmental delay

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doi: 10.1017/cjn.2017.122

Background: A significant part of the developmental delay (DD) population has epilepsy (26-70%) and live in an institution. These patients tend to have atypical presentation of epileptic seizures with higher risk of misdiagnosis. Distinguishing their ictal events from paroxysmal behaviors can be challenging.There often is a lack of description of the spells or inadequate history from the caregivers or the patients. These patients often have drug resistant epilepsy requiring polypharmacy with increased risk of morbidity and mortality. The aim of this study was to determine usefulness of Epilepsy Monitoring Unit (EMU) in diagnosis and management of these patients. *Methods:* This is a retrospective observational study of the patients with epilepsy and DD living in institutions that were admitted to the EMU. *Results:* Four patients met the inclusion criteria for this study. The mean age was 45(29-71), 3/4 (N=3) were male and 3/4 had focal epilepsy. All patients had mood disorders and 2 were taking antipsychotic medication. The mean admission-time was 6,25 days (2-15) and there was a correlation with the events and seizures in 2/4 of the patients and the rest had a combination of behavioural-changes and seizures. *Conclusions:* EMU admission can provide an accurate diagnosis of spells in patients with DD and epilepsy, and improve their quality of life.

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Clinical experience with perampanel for refractory pediatric epilepsy in one Canadian center

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doi: 10.1017/cjn.2017.123

Background: Perampanel (PER) is a new anti-seizure medication that inhibits the α-amino-3-hydroxy-5-methyl-4-isoxazolepropionic acid (AMPA) class of glutamate receptors. It is available in Canada for children since 2014. It is important for physicians to be aware of the efficacy and tolerability of drugs in the post-marketing phase. *Methods:* We did a retrospective review of our experience with PER at BC Children's Hospital. Patients on PER were identified. Clinical data, including demographics, efficacy, tolerability, adverse effects (AE) and retention rates were obtained by review of clinical records. *Results:* Of 24 patients pediatric patients prescribed PER, 21 (87%) had focal and three had symptomatic generalized epilepsy. Ten (42%) had greater than 50% reduction in seizures. In fifteen patients, (63%) PER was discontinued due to AE or poor response. Twelve (50%) had behavioral AE and eight (33%) had non-behavioral AE. PER was effective, at lower doses than required for adults. One third experienced serious AE. One patient experienced oculogyric crisis, not previously reported with PER. AE were not associated with high doses and were reversible. Possible risk factors for behavioral AE include behavioral problems with other medications and pre-existing behavioral co-morbidities. *Conclusions:* It is important for clinicians to be aware of and counsel patients about serious AE, particularly behavioral, when prescribing PER.

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mTOR inhibitors as a new therapeutic strategy in treatment resistant epilepsy in hemimegalencephaly: a case report

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doi: 10.1017/cjn.2017.124

Background: Hemimegalencephaly (HME) is a hamartomatous malformation of one cerebral hemisphere, resulting in refractory epilepsy, intellectual disability, and autistic features. Hemispherectomy is the definitive treatment, but there is risk of high morbidity and mortality, especially when done in early infancy. Various preclinical studies have shown that dysregulation of the mTOR pathway has an integral role in the development of various epilepsy syndromes, including tuberous sclerosis complex (TSC), focal cortical dysplasia and HME. Recently, mTOR inhibitors were proven to be effective in treating seizures in TSC. *Methods:* We present a case of a 6 day old female with refractory epilepsy despite the trial of 9 anti-seizure medications and the ketogenic diet. As the patient was awaiting epilepsy surgery, an mTOR inhibitor, rapamycin was initiated. *Results:* After 1 week of the initiation, she had over a 50% reduction in seizures. At two weeks, the parents felt that for the first time, she was making developmental gains. She also appeared brighter and more interactive. Due to her response to treatment, her hemispherectomy was deferred to when she is older, so there will be a decreased risk of complications from the surgery. *Conclusions:* This case exemplifies how mTOR inhibitors should be considered as a treatment option for patients with HME and refractory epilepsy.