Movement Disorders

P.052

Benign spasms of infancy - a mimicker of infantile epileptic disorders

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Background: Benign spasms of infancy (BSI), previously described as benign non-epileptic infantile spasms or benign myoclonus of early infancy, are non-epileptic movements manifesting during the first year of life and spontaneously resolving in the second year of life. BSI are characterized by spasms typically lasting 1-2 seconds, involving to varying degrees the head, neck, trunk, shoulders and upper extremities. Ictal and interictal EEG recordings are normal. BSI are not associated with developmental retardation and do not require treatment. Distinction between BSI and infantile epileptic disorders, such as epileptic spasms or myoclonic epilepsy of infancy, can be challenging given the clinical similarities. Moreover, interictal EEGs can be normal in all conditions. Epileptic spasms and myoclonic epilepsy require timely treatment to improve neurodevelopmental outcomes. **Methods:** We describe a 6-month old infant presenting with spasm-like movements. His paroxysms as well as a positive family history for epileptic spasms were in keeping with a likely diagnosis of West syndrome. **Results:** Surprisingly, ictal video EEG did not reveal epileptiform activity, and suggested a diagnosis of BSI. **Conclusions:** We emphasize that ictal EEG is the gold standard for classification of infantile paroxysms as either epileptic or non-epileptic, thereby avoiding overtreatment of BSI and facilitating timely targeted treatment of infantile epilepsies.

P.053

Whole-genome sequencing identified a frameshift mutation at LMNB1 in a family with early-onset dystonia

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Background: Dystonia is a hyperkinetic condition that produces abnormal movements or postures. Its diagnostic procedure is often challenging and time consuming. Genetic testing provides an effective approach for diagnosis, but currently only very few dystonia genes have been identified. We propose that studying early-onset forms of dystonia with the use of whole-genome sequencing (WGS) will improve the identification of dystonia-relevant genes and mutations. **Methods:** We performed deep WGS using the Illumina HiSeq X technology in a mother-proband pair with dystonia. The mother has generalized dystonia (age of onset: 15) and the proband has myoclonic dystonia (age of onset: 11). **Results:** No pathogenic mutation was identified in any of the known dystonia genes. However, we identified a rare heterozygous frameshift mutation (p.K342fs*7) at *LMNB1* that was shared between the mother and the proband. Duplication of *LMNB1* is known to cause Adult-onset Demyelinating Leukodystrophy. A heterozygous deletion of *LMNB1* has been reported in a patient with microcephaly and global developmental disorder. **Conclusions:** Further characterization of phenotypes in the participants and their family members is needed to confirm the relationship between mutation in *LMNB1* and dystonia. This work provides a proof-of-principle that novel disease-relevant genes can potentially be identified using the proposed approach.

Neurocritical Care

P.054

Electroconvulsive therapy and epilepsy: a case report

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Background: Electroconvulsive therapy (ECT) involves the induction of a generalized seizure with an electrical current and has been used worldwide when treating medically refractory psychiatric illness. Here we describe a patient with no prior history or risk factors for epilepsy who developed temporal lobe epilepsy after chronic treatment of ECT. **Methods:** A 16-year-old right-handed boy with severe refractory depression received ECT treatment every 10 days for 8 months. Six months into his ECT treatment, the patient developed seizures and was admitted to a pediatric epilepsy monitoring unit. **Results:** Initial clinical events included lightheadedness, diaphoresis, and nausea with associated kaleidoscopic vision changes. Seizures progressed to confusion, fear and paranoia by the time the patient was admitted for monitoring. Long-term video EEG captured many focal seizures with impaired awareness, all originating from both temporal lobes. MRI was normal. ECT was terminated and the patient started on carbamazepine. He has been seizure free for the past 2 years on medication **Conclusions:** While rare, we present a case of a patient with no prior risk factors for epilepsy who developed temporal lobe epilepsy after chronic ECT treatment. Although ECT is an indispensable treatment for many medically refractory psychiatric illnesses, we suggest caution in young patient undergoing ECT.

P.055

A clinical pathway of combined EEG monitoring in high-risk critically ill neonates

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Background: overtreatment of neonatal seizures may result in neurological morbidity. aEEG, despite low sensitivity, is widely used, for ease of bedside interpretation. vEEG, is a limited resource needing expert interpretation. We hypothesize that using aEEG combined with vEEG will increase the sensitivity and specificity of seizure detection and reduce anti-convulsants use compared to aEEG alone. **Methods:** Prospective cohort of neonates admitted to CHEO NICU with suspected seizures between April 1st 2018 to present. Seizures (clinical/aEEG) were documented by bedside clinicians and compared to the vEEG. Bedside clinicians could call a neurologist for remote review of the vEEG. Outcomes include concordance of aEEG and vEEG events and number of episodes where management was changed based on both readings **Results:** 27 patients had both

modalities recording simultaneously. No seizure was identified by either modality in 23 recordings. Seizures were identified in 4 vEEG recordings; the aEEG partially identified these seizures.

- aEEG specificity of 0.87, negative predictive value 0.8, sensitivity 0.44 and positive predictive value 0.57
- Bedside clinician contacted a neurologist 9 times; in 2 cases, this prevented unnecessary treatment.

Conclusions: In this small sample, aEEG had good specificity for ruling out seizures, but low sensitivity for detecting them. The new combined pathway may prevent unnecessary treatment.

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Combined conventional and amplitude-integrated EEG monitoring in neonates: a prospective study

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Background: Seizure monitoring via amplitude-integrated EEG (aEEG) is standard of care in many NICUs; however, conventional EEG (cEEG) is the gold standard for seizure detection. We compared the diagnostic yield of aEEG interpreted at the bedside, aEEG interpreted by an expert, and cEEG. **Methods:** Neonates received aEEG and cEEG in parallel. Clinical events and aEEG were interpreted at bedside and subsequently independently analyzed by experienced neonatology and neurology readers. Sensitivity and specificity of bedside aEEG as compared to expert aEEG interpretation and cEEG were evaluated. **Results:** Thirteen neonates were monitored for an average duration of 33 hours (range 15-94). Fourteen seizure-like events were detected by clinical observation, and 12 others by bedside aEEG analysis. None of the bedside aEEG events were confirmed as seizures on cEEG. Expert aEEG interpretation had a sensitivity of 13% with 46% specificity for individual seizure detection (not adjusting for patient differences), and a sensitivity of 50% with 46% specificity for detecting patients with seizures. **Conclusions:** Real-world bedside aEEG monitoring failed to detect seizures evidenced via cEEG, while misclassifying other events as seizures. Even post-hoc expert aEEG interpretation provided limited sensitivity and specificity. Considering the poor sensitivity and specificity of bedside aEEG interpretation, combined monitoring may provide limited clinical benefit.

P.057

Solely neonatal hypoxic ischemic encephalopathy or more? A study examining genetic predisposition towards a clinical picture of HIE

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Background: Neonatal hypoxic ischemic encephalopathy (HIE) is a clinical phenomenon, that often results from pre or perinatal reduced cerebral blood flow and/or hypoxemia. However, in some cases, neonates present with HIE without significant risk factors or have an unusual clinical course. With the advent of advanced genetic testing, we aimed to explore if such infants had genetic risk factors predisposing them to an HIE-phenotype. **Methods:** We reviewed 206 charts of infants meeting local protocol criteria for moderate to severe HIE at Level III NICU's in Calgary, Alberta. Of these, 27 patients had genetic testing such as microarray, whole exome sequencing, or gene panels. **Results:** Six/twenty-seven patients had genetic mutations; two CDKL5 mutations (protein kinase), one CFTR mutation (cystic fibrosis), one PDH deficiency, one CYP21A2 mutation (congenital adrenal hyperplasia), and one ISY1 (VUS; pre-mRNA splicing). Two patients had noted difficult deliveries and four had minor complications, but all were out of keeping with the severity of presumed HIE. **Conclusions:** This preliminary study demonstrates a possible association between genetic co-morbidities and predisposition towards HIE in the context of a relatively uneventful pre/perinatal course. Earlier identification of genetic etiology, recognized by a discrepancy between risk factors and clinical presentation, could aid in treatment decisions and outcome prognostication.

Neuroimaging

P.058

Tuberous sclerosis complex associated intracranial abnormalities identified in utero via antenatal ultrasound

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Background: Tuberous sclerosis complex (TSC) is characterized by growth of benign tumors in the skin, brain, kidneys, lung and heart. Prognosis is mostly determined by the extent of brain involvement as tumors in the brain lead to seizures and cognitive problems. Epilepsy is highly associated with the cognitive abnormalities in TSC and recent evidence suggests anti-epileptic treatment before onset of seizures reduces epilepsy severity and risk of mental retardation. Screening and potential identification of TSC in utero via ultrasound would allow for prophylactic seizure management in these children. The sensitivity of antenatal ultrasound in the identification of brain abnormalities associated with TSC has not yet been published. In this case, we review the antenatal ultrasounds of a child with TSC for evidence of brain abnormalities in utero. **Methods:** Retrospective review **Results:** Retrospective review of antenatal ultrasounds showed some evidence of intracranial abnormalities. Ultrasound at 34 weeks and 4 days gestation revealed an echogenic density in the right ventricle that correlates with SEGA on post-natal MRI brain at 12 days of life. Post-natal brain ultrasound at 37 weeks revealed multiple cranial abnormalities not seen in utero. **Conclusions:** There are limitations to antenatal neurosonography in the detection of intracranial abnormalities associated with TSC.