B.02

Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy

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Background: *ATP8A2* mutations have only recently been associated with human disease. We present the clinical features from the largest cohort of patients with this disorder reported to date. **Methods:** An observational study of 9 unreported and 2 previously reported patients with biallelic *ATP8A2* mutations was carried out at multiple centres. **Results:** The mean age of the cohort was 9.4 years old (range: 2.5-28 yrs). All patients demonstrated developmental delay, severe hypotonia and movement disorders: chorea/choreoathetosis (100%), dystonia (27%) or facial dyskinesia (18%). Hypotonia was apparent at birth (70%) or before 6 months old (100%). Optic atrophy was observed in 75% of patients who had a funduscopic examination. MRI of the brain was normal for most patients with a small proportion showing mild cortical atrophy (30%), delayed myelination (20%) and/or hypoplastic optic nerves (20%). Epilepsy was seen in two older patients. **Conclusions:** *ATP8A2* gene mutations have emerged as a cause of a novel phenotype characterized by developmental delay, severe hypotonia and hyperkinetic movement disorders. Optic atrophy is common and may only become apparent in the first few years of life, necessitating repeat ophthalmologic evaluation. Early recognition of the cardinal features of this condition will facilitate diagnosis of this disorder.

B.03

Registered EEG technologists can accurately identify ictal and interictal epileptiform patterns on routine EEG

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Background: Registered EEG technologists (RETs) are trained in both the technical aspects of EEG and in preliminary EEG interpretation. However, there is little research evaluating the accuracy of EEG interpretation by RETs. **Methods:** Retrospective study of consecutive routine EEG recordings performed at SickKids Hospital. Preliminary reports by RETs and final reports by neurophysiologists were compared in 5 domains: background activity, focal abnormalities, ictal and inter-ictal epileptiform discharges and summary. **Results:** 500 EEG recordings were analyzed. Sensitivity and specificity of RET reports was high for the assessment of background (85%, 93%), focal slowing (84%, 93%) and inter-ictal epileptiform discharges (92%, 90%). RET reports identified ictal EEG patterns in 32 cases vs. 29 cases identified by neurophysiologists. RET reports were 100% accurate for noting no EEG change for all of 11 cases with non-epileptic events. **Conclusions:** Preliminary EEG reports by RETs were sensitive and specific for all EEG domains analyzed. In the majority of cases, the preliminary interpretation made by the RET was concordant with the final report of the neurophysiologist. Given these findings, RETs may be able to participate in the screening of routine EEG recordings in order to enhance the productivity of busy EEG laboratories.

B.04

Insight into the mesial frontal negative motor area: The girl with a very unusual interest in having her back patted

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Background: Currently, there is limited insight in to the function of the mesial frontal negative motor area (NMA) and the anatomic structures implicated in its function. **Methods:** We present a patient with a Rett-like phenotype, refractory frontal lobe epilepsy, and reflexogenic seizures in which backpatting induced atonic seizures with a semiology resembling the patient falling asleep. The patient underwent video EEG monitoring and ictal/interictal SPECT imaging capturing the reflexogenic seizures. Iterative reconstruction was performed, with images co-registered to previously acquired MRI with subtraction Ictal-Interictal imaging co-registered to MRI. **Results:** Interictally, the patient's EEG showed a slow background and right frontal spikes. Ictally, the patient had numerous subclinical frontal seizures. The reflexogenic seizures had an ictal pattern at the vertex (Cz) with the ictal SPECT imaging, showing hyperperfusion in the right mesial frontal region, both paramedian precentral and postcentral gyri, and right basal ganglia. **Conclusions:** Our findings support the hypothesis that the negative motor area may be activated by the primary sensory cortex; moreover, the ictal SPECT now suggests involvement of the basal ganglia in the NMA's function.

B.05

Nusinersen in infants who initiate treatment in a presymptomatic stage of spinal muscular atrophy (SMA): interim results from the Phase 2 NURTURE study

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Background: NURTURE (NCT02386553) is an ongoing open-label single-arm efficacy/safety study of intrathecal nusinersen in infants who initiate treatment in a presymptomatic stage of spinal muscular atrophy (SMA). **Methods:** Enrolled infants were age ≤6 weeks at first dose, clinically presymptomatic, had genetically diagnosed SMA, and 2 or 3 copies of *SMN2*. Primary endpoint is time to death or respiratory intervention (≥6 hours/day continuously for ≥7 days or tracheostomy). **Results:** As of July 5, 2017, 25 infants (2 copies *SMN2*, n=15;3 copies, n=10) were enrolled. All infants were alive. Two infants (both with 2 copies *SMN2*) required respiratory intervention (but not tracheostomy or permanent ventilation) during an acute, reversible viral infection and thus met the primary endpoint. At last