Neuroscience Education

P.069

Pediatric neurology subspecialty education development in a resource limited setting

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doi: 10.1017/cjn.2019.169

Background: In 2013, the University of Calgary (UofC) - Mbarara University of Science and Technology (MUST) Pediatric Education Program was established when the Pediatric Department in Mbarara, Uganda identified a need for enhanced education in pediatric subspecialty areas. We report on the experience of developing the pediatric neurology subspecialty curriculum. **Methods:** Pre-visit meetings established mutually agreed upon objectives and learning activities that were implemented over 2-week periods in 2015 and 2018. Pre and post-tests were administered to MUST Pediatric residents. Mean differences in test scores were compared using a Student t-test. Residents provided written feedback following the end of the second visit. **Results:** A pediatric neurologist (AM) visited MUST (2015 and 2018) to deliver the curriculum. The second visit was accompanied by a senior UofC Pediatrics resident (LD). Eight and 14 residents at MUST participated in the curriculum in 2015 and 2018, respectively. Neurology test scores improved in 2015 from a mean of 43% to 61% ($p = 0.011$) and in 2018 from 53% to 84% ($p < 0.00001$). Teaching sessions were well received by MUST residents. **Conclusions:** Collaboration between UofC faculty and MUST established an effective pediatric neurology curriculum that was well-received by residents.

Neurovascular, Stroke and Neurointerventional

P.070

Thrombolysis without large vessel occlusion in a child with acute arterial ischemic stroke

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doi: 10.1017/cjn.2019.170

Background: Stroke is a rare neurological disease in children, with an annual incidence of 2 - 13/100,000 children per year. Pediatric stroke is associated with significant morbidity and mortality lasting many decades. Diagnosis of pediatric stroke is challenging and often delayed, limiting options for acute intervention, and the pharmacological and mechanical recanalization strategies that have revolutionized adult stroke remain undefined in children. Clinicians are left to draw conclusions from other retrospective cohort studies or case reports and extrapolate adult guidelines to the pediatric population. The TIPS trial eligibility criteria are often used in clinical practice, despite not being validated for this purpose. We present here the case of a healthy 14 year old male who was treated with intravenous tissue plasminogen activator (IV tPA) for a presumed arterial ischemic stroke without large vessel occlusion on neuroimaging. **Methods:** Retrospective chart review **Results:** Not applicable **Conclusions:** Following the administration of IV tPA, the patient made a full recovery. While we do not recommend the routine use of IV tPA for treatment of presumed large vessel or small vessel in children, we suggest that the decision to proceed with IV tPA be considered on a case-by-case basis.

Other Child Neurology

P.071

Clinical and demographic predictors of stress in parents of children with genetically determined leukoencephalopathies

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doi: 10.1017/cjn.2019.171

Background: Genetically-determined leukoencephalopathies comprise a rare group of inherited white matter disorders. The vast majority are associated with a progressive disease course and early death. This study seeks to determine the clinical and demographic correlates of stress in parents of leukodystrophy patients, for future clinical guidance. **Methods:** A cross-sectional study including 36 families was performed. Children aged 1 month to 12 years with a diagnosed leukodystrophy or genetically-determined leukoencephalopathy were included. 31 mothers and 24 fathers completed the Parental Stress Index, 4th edition (PSI-4). One demographic questionnaire was completed per family. Clinical data was gathered within 6 months of the questionnaires. Statistical analysis was performed with total stress (TS) scores as the primary outcome. **Results:** Mothers and fathers had comparable TS scores. No clinical or demographic factors predicted the father's TS score. Greater ambulatory impairment, using the GMFCS scale, correlated to lower TS scores in the mother. **Conclusions:** The progressive nature of these conditions makes it such that anticipating a child's inability to walk may cause more stress for mothers than a child's actual inability to ambulate. The inability of all other variables to predict total stress highlights a need for individualized approaches when addressing stress in these families.

P.072

Fetal alcohol spectrum disorder - is this a ciliopathy?

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doi: 10.1017/cjn.2019.172

Background: Fetal alcohol spectrum disorder (FASD) is used to describe the spectrum of birth defects due to prenatal alcohol exposure; these include craniofacial abnormalities and intellectual disabilities. The prevalence of FASD is estimated at 1 in 100. Diagnostic criteria include distinct facial features, neurodevelopmental deficits and confirmation of alcohol use during pregnancy. Unfortunately, often criteria are missed or absent. No biochemical marker is available for screening and diagnosis of FASD that is easy, accurate and cost-effective.

Methods: Five children are being recruited from both the FASD clinic at the Glenrose Rehabilitation Hospital and the Healthy Infants and Children's Clinical Research Program (HICUPP) registry. The levels of exhaled nasal NO will be measured and compared between the two groups. Metabolomics analysis on urine samples targeting metabolites of the NO pathway, along with other urinary metabolites is being performed. Bioinformatic statistical tools will be applied to determine whether measured metabolite profiles can provide distinct signatures between healthy children and children with FASD. **Results:** This project is ongoing. **Conclusions:** We hope to correlate NO levels with FASD, illustrating the relationship between NO, ciliopathies and development of FASD. As well, we hope to determine whether urinary metabolites may yield diagnostic markers of FASD.

P.073

Improving access to urgent neurology care for pediatric patients

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doi: 10.1017/cjn.2019.173

Background: Pediatric neurology referral wait times are increasing, often leading to emergency department (ED) utilization. On average 5% of ED patients present with neurological symptoms and 35% of ED neurological diagnoses are revised after specialist review. A Stollery Rapid Access Neurology (RAN) clinic was created to decrease wait time, and initiate an efficient referral process. **Methods:** The RAN clinic ran weekly from March 2018 until February 2019. This was a prospective study approved by the University of Alberta ethics board. Inclusion criteria were met. Information was collected for diagnosis, along with confidential patient satisfaction surveys. **Results:** Seventy-five patients were referred, 49% from the ED. Wait time averaged 6 weeks. The most frequent referral reason was seizures, with 60% of referring diagnosis being correct. Prior to RAN appointment, 61% of patients presented to the ED, whereas only 0.1% returned in the following 3 months. Neurology follow up was required in 81% of patients. Overall satisfaction was ranked 9.6/10. **Conclusions:** The RAN clinic created an effective urgent triage method. Neurologist review revised 40% of diagnoses. This ongoing study reveals that a RAN clinic can reduce visits to the ED following appointment and initiate appropriate follow up. Future evaluation in cost effectiveness and telehealth appointments are required.

P.074

Infantile idiopathic intracranial hypertension - a case study and review of the literature

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doi: 10.1017/cjn.2019.174

Background: Idiopathic intracranial hypertension (IIH), or pseudotumor cerebri, is an increase in intracranial pressure due to unknown etiology. Presentation in infancy is extremely rare. Little is known about infantile IIH and age-specific treatment guidelines are lacking. **Methods:** Patient data was obtained from medical records at the Children's Hospital of Eastern Ontario. A literature review of infantile IIH was performed. **Results:** A previously healthy 9-month-old boy presented with irritability, decreased appetite, and a bulging fontanelle. CT head imaging and cerebrospinal fluid studies revealed normal results. Symptoms transiently resolved after a lumbar puncture, but 11 days later, his fontanelle bulged again. A second lumbar puncture revealed an elevated opening pressure of 35 cm H_2O and led to a diagnosis of IIH in accordance with the modified Dandy Criteria. Treatment with acetazolamide at a dose of 25 mg/kg/day was initiated and the patient remained symptom-free for 6 weeks, followed by another relapse. His acetazolamide was increased to 38 mg/kg/day, with no further relapses to date. **Conclusions:** A diagnosis of IIH is challenging in infants, since the patients cannot yet verbalize typical IIH-related symptoms, and papilledema is difficult to assess. If undetected and untreated, IIH may result in permanent visual deficits.

P.075

Clinical spectrum of POLR3-related leukodystrophy caused by biallelic POLR1C pathogenic variants

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doi: 10.1017/cjn.2019.175

Background: Biallelic variants in *POLR1C* are associated with POLR3-related leukodystrophy (POLR3-HLD), or 4H leukodystrophy (Hypomyelination, Hypodontia, Hypogonadotropic Hypogonadism), and Treacher Collins syndrome (TCS). The clinical spectrum of POLR3-HLD caused by variants in this gene has not been described. **Methods:** A cross-sectional observational study involving 25 centers worldwide was conducted between 2016 and 2018. The clinical, radiologic and molecular features of 23 unreported and previously reported cases of POLR3-HLD caused by *POLR1C* variants were reviewed. **Results:** Most participants presented between birth and age 6 years with motor difficulties. Neurological deterioration was seen during childhood, suggesting a more severe phenotype than previously described. The dental, ocular and endocrine features often seen in POLR3-HLD were not invariably present. Five patients (22%) had a combination of hypomyelinating leukodystrophy and abnormal craniofacial development, including one individual with clear TCS features. Several cases did not exhibit all the typical radiologic characteristics of POLR3-HLD. A total of 29 different pathogenic variants in *POLR1C* were identified, including 13 new disease-causing variants. **Conclusions:** Based on the largest cohort of patients to date, these results suggest novel characteristics of POLR1C-related disorder, with a spectrum of clinical involvement characterized by hypomyelinating leukodystrophy with or without abnormal craniofacial development reminiscent of TCS.