

**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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**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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**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<sub>2</sub>O 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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**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.