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3-year Clinical Lessons Learned from the Alberta Spinal Muscular Atrophy Newborn Screening (SMA-NBS)

JK Mah (Calgary) TR Price (Calgary) M Crone (Calgary) H Kolski (Edmonton) F Niri (Edmonton)*

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Background: Spinal muscular atrophy (SMA) is caused by biallelic mutations in the *SMN1* gene. Early diagnosis through newborn screening (NBS) and presymptomatic treatment optimize health outcomes. Methods: SMA-NBS began in Alberta on 28February2022. A multiplex quantitative PCR assay detected homozygous deletions of exon 7 in dried blood spot samples. Screen-positive infants underwent genetic confirmation by multiplex ligation-dependent probe amplification to determine *SMN1/SMN2* copy numbers. We report clinical outcomes of SMA diagnoses through Alberta NBS over 3 years. Results: From 28February2022-31December2024, twelve infants were confirmed SMA positive, including two with 2 *SMN2* copies and six with 3 *SMN2* copies. Median age at initial positive screen was 6 days (range=3-9), and at diagnosis, 15 days (range=11-27). Seven infants (median age=29 days, range=18-142) received onasemnogene abeparvovec-xioi. Two received nusinersen (Day 22) or risdiplam (Day 72), followed by onasemnogene abeparvovec-xioi (Day 48 and 111, respectively). Two infants received risdiplam after 3 months of age. One infant was symptomatic at treatment initiation. Post-treatment evaluations showed ongoing motor milestone achievements. Conclusions: SMA incidence in Alberta during 2022-2024 was 8.2 (95%CI: 3.5-12.8) cases per 100,000 live births. Efforts continue to shorten age at treatment initiation, especially for those with two *SMN2* copies, and to promote uniform coverage for 4-copy cases.

Other Child Neurology (CACN)

P.079

Body image in youth and adolescents with CP living in Ontario

B Ahmadi (Hamilton) R Mesterman (Hamilton)*

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Background: Body image research in young people with physical disabilities like cerebral palsy (CP) has received very little attention. The goal of this pilot study is to ask youth with CP (of all levels of disability) directly about body image to learn their perspective. Methods: Our study includes quantitative data of quality-of-life measures, along with qualitative interview data summarized via thematic analysis. Our data is augmented with input from siblings (without CP) of our primary participants to represent a control group in the same family unit. Results: Twelve youths with CP (7 male, 5 female) participated in the study. Withe the higher score representing more positive the body image, scores averaged 17.93/25 (SD 4.73) for those with CP, 18.62/25 (SD 5.45) for those without CP. There were higher scores for males and those ≤13yo compared to 14-18yo. Interview thematic analysis uncovers themes of functional capability, the wish to reduce burden on family, pride in the CP identity, and mixed desirability of media representation. Conclusions: There is greater difference between age groups and genders than there is between those with CP and not. Interviews with participants revealed the important recurring theme of functional capacity connected to positive self-image, which may be considered justification for interventions.

P.080

Head circumference values among Inuit children in Nunavut, Canada: a retrospective cohort study

KM Joyal (Winnipeg) S Collins (Victoria) A Miners (Iqaluit) N Barrowman (Ottawa) E Sucha (Ottawa) J Allen (Iqaluit) S Edmunds (Iqaluit) A Caughey (Iqaluit) M Doucette (Iqaluit) S Khatun (Iqaluit) G Healey Akearok (Iqaluit) L Arbour (Victoria) S Venkateswaran (Ottawa)*

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Background: Inuit children have been observed to have high rates of macrocephaly, which leads to burdensome travel for medical evaluation, often with no pathology identified. Given reports that WHO growth charts may not reflect all populations, we compared head circumference (HC) measurements in a cohort of Inuit children with the WHO charts. Methods: We extracted HC data from a retrospective cohort study where, with Inuit partnership, we reviewed medical records of Inuit children, born between 2010-2013, and residing in Nunavut. We excluded children with preterm birth, documented neurologic/genetic disease, and most congenital anomalies. We compared HC values with the 2007 WHO charts. Results: We analyzed records of 1960 Inuit children (8866 data points). Most data were from ages 0-36 months. At all age points, the cohort had statistically significantly larger HC than WHO medians. At age 12 months, median HC were 1.3 cm and 1.5 cm larger for male and female Inuit children. Using WHO growth curves, macrocephaly was overdiagnosed and microcephaly underdiagnosed. Conclusions: Our results support the observation that Inuit children from Nunavut have larger HCs, and use of the WHO charts may lead to overdiagnosis of macrocephaly and underdiagnosis of microcephaly. Population specific growth curves for Inuit children should be considered.

P.081

Real-world benefits and tolerability of trofinetide for the treatment of Rett Syndrom: interim analysis of the LOTUS study

S Bond (Toronto) H Mayman (San Diego) J Downs (Perth) L Cosand (San Diego)*

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Background: Trofinetide is approved for the treatment of Rett syndrome (RTT) in patients aged ≥2 years. Here, we present the benefits and tolerability of trofinetide in the treatment of RTT with the 12-month follow-up of LOTUS. Methods: Caregivers of patients who are prescribed trofinetide under routine clinical care

are eligible to participate. Assessments include the Behavioral Improvement Questionnaire (BIQ), the Quality-of-Life Inventory-Disability (QI-Disability) Questionnaire, and the Gastrointestinal Health Questionnaire. Due to ongoing enrollment, data are reported to 9 months since the initiation of trofinetide. Results: In total, 192 patients were included. The median dose reported at week 1 was 45.0% of the target weight-banded label dose; by week 9 onwards, the median dose was at least 80.0% of the target weight-banded label dose. Behavioral improvements reported with the BIQ were nonverbal communication (49–62%), alertness (43–62%), and social interaction/connectedness (32–52%). The QI-Disability Questionnaire median total scores indicated overall improvement in quality of life (QoL) with trofinetide. Caregivers reported that patients were most likely to void normal stools over the follow-up; most reports of diarrhea were contained inside the patient's diaper. Conclusions: Caregivers of patients with RTT in LOTUS reported behavioral improvements of RTT symptoms and improvement in patients' QoL.

P.082

Real-world benefits and tolerability of trofinetide for the treatment of pediatric and adult patients with Rett Syndrom: the LOTUS study

S Bond (Toronto) H Mayman (San Diego, California) J Downs (Perth) L Cosand (San Diego)*

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Background: Trofinetide is approved for the treatment of Rett syndrome (RTT) in patients aged ≥2 years. Here, we present the benefits and tolerability of trofinetide in pediatric and adult patients with RTT from the LOTUS study. Methods: Caregivers of patients who are prescribed trofinetide under routine clinical care are eligible to participate. This subgroup analysis of the 12-month follow-up of LOTUS focused on pediatric (0–17 years of age) and adult (≥18 years of age) patient populations. Due to ongoing enrollment, data are reported to 9 months since the initiation of trofinetide. Results: In total, 117 pediatric and 74 adult patients were included. The median dose reported at week 1 was 45.0% and 41.0% of the target weight-banded label dose for pediatric and adult patients, respectively; by week 8, the median dose was at least 86.0% and 70.0% of target, respectively. Behavioral improvements included nonverbal communication (pediatric: 53–64%; adult: 41–58%), alertness (pediatric: 50–69%; adult: 33–65%), and social interaction/connectedness (pediatric: 36–58%; adult: 26–46%). Most reports of diarrhea were contained inside the patients' diapers. Conclusions: Caregivers of pediatric and adult patients with RTT in LOTUS reported improvements consistent with the general population of the study.

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A novel mutation in YARS2 gene in a patient with MLASA

A Eisenkoelbl (Ottawa) M Carter (Ottawa) H McMillan (Ottawa)*

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Background: MLASA (myopathy, lactate acidosis and sideroblastic anemia) is a rare autosomal recessive mitochondrial disorder, which affects oxidative phosphorylation and iron metabolism in skeletal muscle and bone marrow. Three genes have been identified so far, *PUS1* is the most common, followed by *YARS2* and *MT-ATP6*. We present a patient with a novel variant in *YARS2* and a literature review. Methods: We report a 20-months-old girl with ptosis and low birth weight. She presented with delayed motor milestones and bulbar weakness with feeding difficulties. She had mild anemia and elevated lactate, echocardiogram revealed a mild to moderate left ventricular hypertrophy without LVOT obstruction. Results: Genetic testing showed two heterozygous variants in *YARS2*. The maternal one (c.948G>T, p.Arg316Ser) has been reported previously in a compound heterozygous state, while the paternal one (c.917T>C, p.Phe306Ser) has not been previously described. Genetic findings were supported by enzyme activities, which showed reduced complex I +III and complex IV activities and reduced cytochrome oxidase (COX). Conclusions: In this case report we describe a 20-months-old girl with clinical features of MLASA. A novel variant in the *YARS2* gene was found, pathogenicity could be proven with clinical phenotype and enzyme activity testing.

P.084

Epidemiology and burden of illness in patients with Rett Syndrome in Ontario, Canada

S Bond (British Columbia) J Murray (Mississauga) A Datta (British Columbia) MF Rafay (Winnipeg) L McAdam (Toronto) C Neish (Mississauga)*

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Background: Rett Syndrome (RTT) is an X-linked neurodevelopmental disorder, characterized by gradual loss of motor, verbal and social skills. This study describes the epidemiology and healthcare resource utilization (HCRU) of RTT in Ontario, Canada. Methods: RTT patients (≥ one ICD-10-CA code F84.2) were identified using data held at the Institute for Clinical Evaluative Sciences (ICES), between September 2018-August 2023. Incidence and prevalence rates from Ontario were extrapolated nationally using the Stats Can population estimates. Results: A total of 246 patients were indexed; 95% female, median age 21 years and 40% from central Ontario. There were 57 incident and 257 prevalent RTT cases identified in Ontario. National extrapolations estimated 175 incident and 613 prevalent RTT cases. Common comorbidities included developmental disability (85.4%) and epilepsy (49.6%). Patients frequently had outpatient visits (primary care 96.7%, specialists 86.6%), emergency department visits (76.8%) and inpatient hospitalizations (54.5%). Most patients (95.1%) had at least one public claim for all-cause medication. Disease-specific medication claims were for anti-infectives (69.1%) and anti-seizure medications associated with mood effects (65.0%). Conclusions: This study provides population-based estimates of RTT in Canada. Findings highlight the high burden of illness and HCRU of RTT and the opportunities to improve healthcare outcomes in this population.