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

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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

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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.

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

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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.

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

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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