

score, respectively. Conclusions: All aspects of the SNAP score had negative and steeper slopes prior to neurological decline, whereas only 'voice' in GCS had a negative trend. These findings suggest that the SNAP tool may be useful in earlier identification of acute decline. Ongoing prospective studies are underway.

## NEUROMUSCULAR DISEASE AND EMG

### P.072

#### Alberta Spinal Muscular Atrophy Newborn Screening (SMA-NBS) – 2022 results

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Background: Spinal muscular atrophy (SMA) is a progressive neuromuscular disease caused by biallelic mutations of the *survival motor neuron 1 (SMN1)* gene. Early diagnosis via newborn screening and presymptomatic treatment are essential to optimize health outcomes for affected individuals. Methods: We developed a multiplex real-time polymerase chain reaction assay using dried blood spot samples for the detection of homozygous deletion of exon 7 of the *SMN1* gene. Newborns who were screened positive were seen urgently for clinical evaluations. Copy numbers of *SMN1* and *SMN2* genes were determined by multiplex ligation-dependent probe amplification for confirmatory testing. Results: From February 28, 2022 to December 31, 2022, 42,450 newborns were screened in Alberta. Four infants had abnormal screen results and were subsequently confirmed to have SMA. No false positive newborns were detected. Three infants received adeno-associated virus serotype 9 (AAV9)-mediated *SMN1* gene replacement therapy <31 days of age. One infant received *SMN2*-splicing modulator treatment due to maternally-transferred AAV9 neutralizing antibodies prior to gene therapy at 3 months of age. Conclusions: The estimated incidence of SMA in Alberta is 9.4 (95% CI: 2.5 – 24.1) per 100,000 live-births. During the first year of the SMA-NBS program, 4 asymptomatic infants received treatment and demonstrated excellent developmental progress to date.

### P.073

#### Mapping a national Duchenne muscular dystrophy registry to the International Classification of Functioning, Disability, and Health

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Background: Duchenne muscular dystrophy (DMD) is an X-linked disease that causes progressive muscle wasting. The

Canadian Neuromuscular Disease Registry (CNDR) DMD subset collects data focused on body structure and function. Our objective is to develop a broader dataset including the priorities of those living with DMD in accordance with the International Classification of Functioning, Disability, and Health (ICF) – a framework for describing disease and health functions developed by the World Health Organization. Methods: Clinically relevant ICF categories for DMD were identified and reviewed by two independent committees including two patients and six parent representatives. The Delphi approach was used to narrow ICF categories to a core set representative of DMD, which will be mapped to the CNDR-DMD subset. Results: With full result expected by the conference, the mapping of the ICF to the CNDR-DMD subset will identify data collection priorities in the four domains of functioning and disability: body functions and structures, activities at the level of the individual, participation in all areas of life, and environmental factors. Conclusions: The ICF can be used to identify data collection priorities. Broadening the CNDR-DMD subset will foster future research to include outcome measures important to patients and families affected by DMD.

## NEUROVASCULAR AND NEUROINTERVENTIONAL

### P.074

#### Risk factors for perinatal arterial ischemic stroke (PAIS): A machine learning approach

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Background: Perinatal arterial ischemic stroke (PAIS) is a leading cause of hemiparetic cerebral palsy. Multiple risk factors are associated with PAIS but studies are limited by small sample sizes and complex interactions. Unbiased machine learning applied to larger datasets may enable the development of robust predictive models. We aimed to use machine learning to identify risk factors predictive of PAIS and compare these to the existing literature. Methods: Common data elements of maternal, delivery, and neonatal factors were collected from three perinatal stroke registries and one control sample over a 7-year period. Inclusion criteria were MRI-confirmed PAIS, term birth, and idiopathic etiology. Random forest machine learning in combination with feature selection was used to develop a predictive model of PAIS. Results: Total of 2571 neonates were included (527 cases, 2044 controls). Risk factors uniquely identified through machine learning were infertility, miscarriage, primigravida, and meconium. When compared, factors identified through both literature-based selection and machine learning included maternal age, fetal tobacco exposure, intrapartum fever, and low