of neuroimaging. Provincial live births were obtained from statistics Canada. Outcomes were extrapolated to the Pediatric Stroke Outcome Measure (PSOM). *Results:* We identified 74 cases: 49 NHS (26 retrospective, 27 prospective), 4 presumed perinatal HS (PPHS), and 21 hemorrhagic transformation (HT) of ischemic injury. Incidence of NHS was 1:8800 live births (1:5820 for all forms). HT was common (28.4%) including global, arterial venous ischemic lesions. Presumed perinatal hemorrhagic stroke presented with epilepsy. No risk factor was identified in 68% of cases. Outcomes were abnormal (PSOM 1 or more) in 30% and better in the HT group. *Conclusion:* NHS occurs in 1:8800 live births. Imaging classification is essential to define mechanisms.

A.05

Health-related quality of life in children with Duchenne muscular dystrophy: a follow-up study

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Background: Improvement of health-related quality of life (HRQOL) is a major goal in chronic disease management and HRQOL has become an important outcome in clinical trials. Longitudinal data on HRQOL are needed to elucidate change over time and to assess effectiveness of interventions; such research is lacking in the paediatric Duchenne Muscular Dystrophy (DMD) population. *Methods:* We followed up participants from our initial HRQOL study in 2013 a year and a half later. Multidimensional generic and disease-specific measures from the Pediatric Quality of Life Inventory were used to assess HRQOL from child and parent perspectives. Mean changes in HRQOL were calculated. *Results:* Data collection is ongoing and currently, data from 16 families (out of the initial 98) are available. Preliminary results indicated that by both child and parent reports, there were declines in all domains of HRQOL except for social function, in which there was a slight improvement. Mean decline in HRQOL scores ranged from 1.6 to 8.6 for child reports; and 3.3 to 7.7 for parent reports. *Conclusion:* HRQOL of boys with DMD deteriorates over time. Our results may be helpful in interpreting patient reported outcomes in forthcoming clinical trials and determining minimally clinically important changes in this population.

A.06

KCNQ2 mutations: genotype-phenotype association beyond epilepsy

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Background: KCNQ2 abnormalities were described in infants with benign familial neonatal seizures (BFNS) and epileptic encephalopathy (EE). Associated features possibly include abnormal neuroimaging findings such as hypomyelination and/or T2 high signal of basal ganglia. *Methods:* This report describes 4 infants carrying different heterozygous KCNQ2 variants and 2 infants with 20q13.33 deletions encompassing KCNQ2 gene. *Results:* The different KCNQ2 mutations led to EE in 3 patients and included a novel de novo missense variant, p.Arg201Cys/c.601C>T, in an infant with severe EE and global developmental delay, hyperkinetic movement disorder, autonomic dysfunction with chronic hypoventilation, apnea, low GABA levels in CSF, and hypomyelination. She died at age 3 years of respiratory failure. One patient with BFNS and normal MRI has a previously reported c.508delG frame shift mutation in KCNQ2. Of the two de novo 22q13.33 deletions (1.2Mb versus 254.1 Kb) the larger caused a more severe phenotype, including focal epilepsy from infancy until 4 years, moderate developmental delay and diffuse brain volume loss. *Conclusions:* Along with varied epilepsy phenotypes and neuroimaging findings KCNQ abnormalities were associated with severe autonomic dysfunction and reduced CSF GABA levels. This might have further treatment implications, besides that the altered potassium channel function itself presents a therapeutic target.

A.07

Brain stimulation and constraint for hemiparesis after perinatal stroke: The PLASTIC CHAMPS trial

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Background: Perinatal stroke causes hemiparetic cerebral palsy. Constraint therapy (CIMT) improves function in congenital hemiparesis and adult stroke. Repetitive transcranial magnetic stimulation (rTMS) may improve function in adult stroke. The two have not been tested in perinatal stroke. *Methods:* PLASTIC CHAMPS (www.clinicaltrials.gov/NCT01189058) was a controlled factorial trial of rTMS and CIMT in perinatal-stroke hemiparesis. Children 6-18 years participated in a 2 week peer-supported motor learning camp, randomized to daily inhibitory rTMS (1200 stimulations, contralesional M1), CIMT, both or neither. Primary outcomes were Assisting Hand Assessment (AHA) and Canadian Occupational Performance Measure (COPM) at 1, 8, and 24 weeks. Quality-of-life, safety and tolerability were evaluated. Change was assessed across treatment groups over time (linear mixed effects model). *Results:* All forty-five subjects completed the trial (median 11.4yrs). COPM scores increased >100% with maximal gains at 6 months ($p<0.002$). Addition of rTMS and/or CIMT doubled the chances of clinically significant gains. Combined rTMS+CIMT resulted in larger AHA gains at all time points (6 months $p=0.006$). CIMT or rTMS alone had more modest effects. Neither treatment decreased function in either hand. Procedures were well tolerated. *Conclusions:* Children with hemiparesis participating in intensive, psychosocial rehabilitation programs perceive marked increases in function. Non-invasive brain stimulation may enhance motor learning therapy in perinatal stroke hemiparesis.