General Pediatric Neurology

P.002

Neurodevelopment at 2 years in asphyxiated newborns treated with hypothermia can be predicted by early neuroimaging markers

FJ Al Amrani (Montreal) P Wintermark (Montreal)*

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Background: Brain imaging in asphyxiated newborns treated with hypothermia has shown that brain injury can be identified as early as day 2 of life and continue to evolve over the first month of life. *Methods:* Asphyxiated newborns treated with hypothermia were enrolled prospectively. Apparent diffusion coefficient (ADC) and fractional anisotropy (FA) were measured on magnetic resonance imaging (MRI) performed over the first month of life. Neurodevelopment was evaluated around 2 years of age. *Results:* Twenty-six asphyxiated newborns treated with hypothermia were enrolled. In asphyxiated newborns treated with hypothermia, who developed cerebral palsy, ADC values were significantly decreased on day 1 of life, on day 2-3 and around day 10 of life in the thalamus. In the same newborns, the FA values were significantly decreased on day 10 of life. *Conclusions:* Early MRI measurements permitted to identify the newborns developing cerebral palsy as early as on day 1 of life and could thus be used in the future to predict the long-term neurodevelopmental outcome asphyxiated newborns treated with hypothermia.

P.003

Health-related quality of life (HRQOL) for genetically determined leukoencephalopathy patients and their families

A Mirchi (Montréal) F Pelletier (Montréal) LT Tran (Montréal) A Pizzino (Washington) M Dilenge (Montréal) N Braverman (Montréal) A Vanderver (Washington) F Roncarolo (Montréal) G Bernard (Montréal)*

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Background: Genetic leukoencephalopathies are a group of neurodegenerative diseases imposing a great burden on patients and families. There is no previous systematic study looking at the impacts of these diseases. *Methods:* HRQOL was assessed using the Pediatric Quality of Life Inventory (PedsQL) model. A total of 24 patients with genetically determined leukoencephalopathies and their family members completed the PedsQL questionnaires. Detailed clinical assessments were performed at the time the questionnaires were filled. HRQOL results were correlated with the severity of the clinical features and the presence vs. absence of a definitive molecular diagnosis. *Results:* Preliminary results show lower PedsQL total scores for patients without compared to with a molecular diagnosis. Emotional and physical functioning scores were significantly impaired in patients without a molecular diagnosis. Lower total scores were obtained for patients who presented more severe clinical features such as lost ambulatory functions and dysphagia. *Conclusions:* Overall, our preliminary results indicate that patients without a molecular diagnosis have an impaired HRQOL and that more severely affected patients have a poorer HRQOL. Further analyses and studies on a larger population of patients in a prospective fashion are required to assess the burden of these diseases and identify potential modifiable factors.

P.004

Endocrine and growth abnormalities in 4H leukodystrophy patients with a molecular diagnosis

F Pelletier (Montréal) A Mirchi (Montréal) FK Cayami (Amsterdam) LT Tran (Montréal) N Ulrick (Washington) C Polychronakos (Montréal) A Vanderver (Washington) NI Wolf (Amsterdam) G Bernard (Montréal)*

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Background: 4H or POLR3-related leukodystrophy is an autosomal recessive disorder characterized by hypomyelination, hypodontia and hypogonadotropic hypogonadism caused by mutations in *POLR3A*, *POLR3B* and *POLR1C*. The endocrine abnormalities have never been systematically studied. *Methods:* A cross sectional international multicenter study was performed and the following variables were assessed: weight, height, head circumference, pubertal history, hormone levels and neurological and non-neurological features. Data was analyzed to determine whether there was a correlation between the presence of endocrine abnormalities and mutations in a specific gene and/or the presence of specific symptoms such as other non-neurological symptoms. *Results:* Data was collected on 156 patients. Endocrine data were available for 144 patients. The most common endocrine abnormalities seen in this cohort were short stature (54/90 patients (60%)) and delayed puberty (53/70 patients (76%)). 13 of the 58 patients tested (22%) had abnormal thyroid function. Patients with *POLR3A* mutations were more likely to have endocrine abnormalities. *Conclusions:* Our results confirm that the most common endocrine features in 4H leukodystrophy are short stature and pubertal abnormalities. However, the other potential endocrine abnormalities are typically under-investigated in this patient population. A prospective study is required to investigate the extent and severity of the endocrine abnormalities in 4H leukodystrophy.

P.006

Keeping neurosarcoidosis on the differential

E Spinelli (Edmonton) G Blevins (Edmonton) J Mailo (Edmonton) L Atilano (Edmonton) H Leonard (Edmonton) EA Yeh (Toronto) H Kolski (Edmonton)*

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Background: Sarcoidosis is a multiorgan autoimmune disease characterized by the presence of non-caseating granulomas. The diagnosis can be difficult, particularly with central nervous system (CNS) involvement, and pathology outside of the CNS has to be carefully evaluated. Early and correct diagnosis is crucial for appropriate management particularly in children where sarcoidosis and neurosarcoidosis are rare. *Methods:* We describe a 16 year old previously healthy boy who presented with progressive pyramidal neurological signs and symptoms localizable primarily to the brain stem. *Results:* Initial imaging revealed striking brainstem, as well as cerebral, cerebellar and spinal cord perivascular enhancement. Lung involvement was subclinical with a miliary pattern on chest imaging and needle biopsy revealed an interstitial lymphocytic infiltration. Extensive serum