

**PLATFORM PRESENTATIONS**

**CACN CHAIR’S SELECT ABSTRACTS**

**A.01**

**Targeted analysis of whole exome sequencing and genotype-phenotype correlation in epileptic encephalopathies**

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**Background:** Epileptic encephalopathy (EE) is a severe condition in which the epileptic activity in the brain itself may contribute to severe cognitive and behavioural impairments above and beyond what might be expected from the underlying pathology alone. Next generation sequencing technologies such as whole exome sequencing (WES) can detect underlying genetic causes of EE. **Methods:** This report describes genotype-phenotype correlation of 29 subjects with unexplained epileptic encephalopathy, in whom WES, targeting a list of 557 epilepsy-associated genes was performed. Epilepsy phenotyping was done according to current ILAE recommendations. **Results:** Median age at seizure onset was 14 months (range 1-48). Electroclinical syndromes were applicable for 16/29, 8/16 had a definite/diagnosis suggesting the association between AR and autoimmunity, focusing on possible precipitants and familial autoimmunity, in comparison with patients with infantile autism (IA). **Methods:** charts of children diagnosed with ASD in 2014 were reviewed, and patients were classified as either AR or IA based on Autism Diagnostic Interview (ADI-R) criteria. **Conclusion:** the association between AR and preceding febrile illness as well as familial autoimmunity, supports the notion of AR as a separate entity within ASD, possibly mediated by autoimmune changes.

**A.02**

**Clinical clues for autoimmunity in the etiology of autistic regression**

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**Background:** Autistic regression (AR) accounts for 20-40% of patients with Autism Spectrum Disorder (ASD).1 Literature demonstrates specific immune changes in AR patients,2 as well as association between AR and autoimmune thyroiditis.3 Our study explores the clinical association between AR and autoimmunity, focusing on possible precipitants and familial autoimmunity, in comparison with patients with infantile autism (IA). **Methods:** charts of children diagnosed with ASD in 2014 were reviewed, and patients were followed over time.

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**A.03**

**Cerebral perfusion and its relationship to post-concussion syndrome in mild traumatic brain injury: a prospective controlled cohort study**

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**Background:** Persistent post-concussive symptoms (PCS) have been linked to increased cortical network activation and decreased cerebrovascular reactivity. Decreased cerebral perfusion could help explain PCS and may be a biomarker to track recovery. **Methods:** Children (ages 8 to 18 years) symptomatic with PCS at one month post-injury were studied. Children who recovered following a mTBI (asymptomatic group) and healthy children acted as controls. **Results:** Global CBF was significantly higher in the AR group (p<0.001). Symptomatic children had increased CBF in the frontal and occipital regions, and asymptomatic children had decreased CBF in the temporal regions compared to healthy controls, and lower in the asymptomatic group (F(2,57) 9.734 p<0.001). Symptomatic children had increased CBF in the frontal and occipital regions, and asymptomatic children had decreased CBF in the temporal regions compared to healthy controls. CBF decreased in symptomatic children over time. CBF was a predictor of cognition (R2=0.235; p=0.001). **Conclusions:** Cerebral perfusion is altered in children with mTBI and is associated with recovery trajectory. Asymptomatic children had decreased CBF suggesting cerebral recovery is ongoing. Further longitudinal studies are required to determine if these perfusion patterns continue to change over time.

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Suppl. 2 – S7