Methods: We performed ultra-deep sequencing of 13 mTOR pathway genes using a custom HaloPlex target enrichment kit (Agilent Technologies) in 16 resected histologically confirmed FCD specimens.

Results: We identified causal variants in 62.5% (10/16) of patients at an alternate allele frequency of 0.75–33.7%. The spatial mutation frequency correlated with the FCD lesion’s size and severity. Conclusions: Screening FCD tissue using a custom panel results in a high yield, and should be considered clinically given the important potential implications regarding surgical resection, medical management and genetic counselling.

P.049
Quality of life in children with absence epilepsy

M Sidhu (Hamilton)* D Streiner (Hamilton) G Ronen (Hamilton)
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Background: Childhood absence epilepsy is a common generalized epilepsy in pediatric patients. Although this was considered a “benign” syndrome, new data suggests there are associated neurocognitive effects. This is the first study comparing quality of life and social functioning in those with absence epilepsy to those with other types of epilepsy. Methods: This observational study recruited patients from six Canadian academic centers. 106 patients had absence seizures, and 219 had other seizures. Established measures of depression, anxiety, social skills, social support, participation, quality of life, and epilepsy severity were assessed. MANCOVA was used to evaluate differences in social function, quality of life, and epilepsy severity measures, while accounting for age and gender. Results: This yielded a statistically significant result (Wilk's lambda <0.05), with partial eta squared of 0.163. Follow up of between subjects tests revealed lower health related quality of life interpersonal/social subscale and close friend social support scores in those with absence epilepsy, while other measures were not significant. Conclusions: Children with absence epilepsy have similar social function, quality of life and epilepsy severity measures compared to those with other types of epilepsy. This indicates that any dysfunction in these domains is similar to those with other types of epilepsy.

P.050
Epilepsy phenotypes in patients with Sotos syndrome

O Fortin (Montreal)* C Vincelette (Sherbrooke) AQ Khan (Montreal) S Berrahmoune (Montreal) IE Scheffer (Melbourne) J Lu (Boston) K Davis (Saskatoon) KA Myers (Montreal)
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Background: Sotos syndrome is a genetic condition caused by NSD1 alterations, characterized by overgrowth, macrocephaly, dysmorphic features, and learning disability. Approximately half of children with Sotos syndrome develop seizures. We investigated the spectrum of seizure phenotypes in these patients. Methods: Patients were recruited from clinics and referral from support groups. Those with clinical or genetic diagnosis of Sotos syndrome and seizures were included. Phenotyping data was collected via structured clinical interview and medical chart review. Results: 25 patients with typical Sotos syndrome features were included. Of 14 tested patients, 64% (n=9) had NSD1 alterations. Most had developmental impairment (80%, n=20) and neuropsychiatric comorbidities (68%, n=17). Seizure onset was variable (2 months to 12 years). Febrile and absence seizures were the most frequent types (64%, n=16). Afebrile generalized tonic-clonic (40%, n=10) and tonic (24%, n=6) seizures followed. Most patients (60%, n=15) had multiple seizure types. The majority (72%, n=18) was controlled on a single antiepileptic, or none; 4% (n=1) remained refractory to antiepileptics. Conclusions: The seizure phenotype in Sotos syndrome most commonly involves febrile convulsions or absence seizures. Afebrile tonic-clonic or tonic seizures may also occur. Seizures are typically well-controlled with antiepileptics. The rate of developmental impairment and neuropsychiatric comorbidities is high.