The diverse manifestations of tuberous sclerosis complex: the experience of a provincial TSC clinic

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Background: Recent consensus recommendations for Tuberous Sclerosis Complex (TSC) stress the importance of multidisciplinary follow-up for these patients. The objective of our study was to review the manifestations of TSC seen in our hospital to determine the care needs of this population. Methods: This was a systematic, retrospective chart review of children with TSC treated at our institution. Patients were identified through epilepsy and clinical neurophysiology databases. Results: The study population comprised 81 patients, born between 1987-2014, who were a median 10 (Range 0.2-23.2) years of age at last follow-up. 88% of patients had epilepsy, including 30% with a history of infantile spasms. Developmental delay was reported in 65%, while 40% had intellectual disability. Psychiatric co-morbidities occurred in 49%. The most common psychiatric diagnoses were autism (25%), ADHD (19%), and anxiety (16%). Cardiac rhabdomyomas occurred in 35% of patients and renal angiomyolipomas in 42%, while only 4% had polyostotic kidneys. Subependymal giant cell astrocytomas were observed in 14% of patients. 86% had skin manifestations. Conclusions: This study reaffirms the multi-system manifestations of TSC and the need to provide comprehensive, multidisciplinary care. As many children are still very young, the prevalence of autism and intellectual disability is likely underestimated.

Epilepsy surgery in tuberous sclerosis complex: the BC Children’s Hospital experience

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Background: Epilepsy occurs in up to 90% of patients with Tuberous Sclerosis Complex (TSC) and is often refractory to medications. Our objective was to assess the safety and outcome of epilepsy surgery in children with TSC at our institution. Methods: We performed a systematic, retrospective chart review of children with TSC who underwent epilepsy surgery at our institution. Patients were identified through epilepsy and clinical neurophysiology databases. Results: 19 patients (out of 81 with TSC) underwent surgery between 1995-2014. Median age at surgery was 4.2 (Range 1.1-15.6) years, with patients having failed a median 4 (Range 0-10) anti-seizure medications. Surgery comprised corpus callosotomy in 2 and resection of one or more tubers in 17. 2 patients had a subsequent second resection. Minor neurologic deficits occurred after 14% of surgeries. Median follow-up was 2.4 years (Range 0.3 -13.8 years) following surgery. At last follow-up, 47% were seizure free, including 2 patients off anti-seizure medication. Conclusions: Epilepsy surgery is safe and effective in carefully selected TSC patients, with the majority having a good seizure outcome. Children with epilepsy secondary to TSC should be referred for epilepsy surgery assessment.