Ictal SPECT-guided Epilepsy Surgery in a Patient with Forme Fruste Tuberous Sclerosis

Jun-Mo Hwang, M.D., Eun-Ik Son, M.D., Il-Man Kim, M.D., Chang-Young Lee, M.D.
Department of Neurosurgery, Keimyung University School of Medicine, Daegu, Korea

Tuberous sclerosis is an autosomal dominant disease characterised by hamartomas (tubers) in many organ systems and the four major intracranial manifestations including cortical tubers, white matter abnormalities, subependymal nodules and subependymal giant cell astrocytoma. But there is immense variability in the clinical presentation of tuberous sclerosis and many incomplete forms (formes frustes) exist. Almost all patients with tuberous sclerosis have seizures and mental retardation. The authors experienced a 7-year-old boy with medically intractable epilepsy without any skin lesion or mental retardation. In terms of surgical standpoint for determination of extent of resection, corticectomy on the overriding cortex of right premotor and lesionectomy of periventricular calcified lesion were performed according to ictal single photon emission computed tomography (SPECT), which showed hyperperfusion in the subcortical and calcified area. Histopathologic findings showed a few cytologically abnormal neurons with extensive gliosis, containing many Rosenthal fibers, reactive astrocytes and dense calcification, composing of abundant calcospherites which suggested forme fruste tuberous sclerosis. During the follow-up period of eighteen months, seizure was free after surgery.

KEY WORDS: Forme fruste tuberous sclerosis ∙ Epilepsy ∙ Ictal SPECT ∙ Corticectomy ∙ Lesionectomy.
SPECT, which showed hyperperfusion on subcortical and calcified area (Fig. 2). Histopathologic findings showed a few cytologically abnormal neurons with extensive gliosis, containing many Rosenthal fibers, reactive astrocytes and dense calcification, composing of abundant calcospherites which suggested forme fruste tuberous sclerosis (Fig. 3). During the follow-up period of eighteen months, seizure was free after surgery (Fig. 4).

Discussion

Tuberous sclerosis is an autosomal dominant disease characterized by hamartomas (tubers) in many organ systems including the brain, retina, kidney, lung, heart, and skin. Classic triad of tuberous sclerosis are facial nevus (adenoma sebaceum), seizures, mental retardation.

Mutation of TSC1 (9q34) or TSC2 (16p13) is related to tuberous sclerosis. The four major intracranial manifestations of tuberous sclerosis are cortical tubers, white matter abnormalities, subependymal nodules and subependymal giant cell astrocytoma.

The incidence of tuberous sclerosis is 1 in 27,000 overall, with an incidence in children of 1 in 12,000. But there is immense variability in the clinical presentation of tuberous sclerosis and many incomplete forms (forme frustes) exist so the actual incidence may be as high as 1 in 5,800.

Epilepsy is the most common neurological symptom of tuberous sclerosis, occurring in 74 to 98% of patients and often begins during the first year of life. As epilepsy in patients with tuberous sclerosis is often refractory to antiepileptic drugs and also difficult to localize the epileptogenic zone, usually regional or wide area, surgical resection is not so prevalent with unfavorable outcome.

Perot and Weir first reported successful epilepsy surgery in tuberous sclerosis patients in 1966 and some subsequent reports which demonstrate good outcome in approximately 50–60% of drug-resistant patients selected for surgical management.

Unfavourable prognostic factors include onset earlier than 1 year of age, presence of several seizure types (infantile spasms and partial motor or complex partial seizures; drop attacks and atypical absences), multifocal discharges and/or secondary bilateral synchrony, and occurrence of new EEG foci during the evolution. Although Comair already mentioned about the seizure free cases with only PET-guided resective surgery without any other consistent findings to localize the epileptogenic zone, there is no similar report related resective surgery only ictal-SPECT guidance as far as we know. However, selected patients with tuberous sclerosis with medically...

Fig. 2. Ictal single photon emission computed tomography shows abnormally increased perfusion areas on subcortical area above the calcified area of right frontal lobe.

Fig. 3. The histopathologic findings show a few cytologically abnormal neurons with extensive gliosis, containing many Rosenthal fibers, reactive astrocytes and dense calcification, composing of abundant calcospherites which suggest forme fruste tuberous sclerosis (H&E, original magnification ×200). A: partial area of large, bizarre or multinucleated cells with prominent nucleoli and abundant eosinophilic cytoplasm, B: A conspicuous large globular cells with ample pink cytoplasm and eccentric nuclei, C : large areas of numerous calcospherites, D: extensive astrogliosis with Rosenthal fibers.

Fig. 4. In terms of surgical standpoint for determination of extent of resection, corticectomy based on ictal single photon emission computed tomography and ultrasound–guided lesionectomy of periventricular calcified lesion are performed (A, B).
intractable epilepsy verified as localized focal epileptogenic focus especially by ictal SPECT, lesionectomy and/or corticectomy are recommended for better seizure outcome1,3,6).

Conclusion

Although most patients with tuberous sclerosis have intractable seizures, surgical consideration is needed in selected cases based on sophisticated presurgical evaluation especially definitive ictal SPECT.

References


