

Table 1 Patient Characteristics and Diagnostic Studies

Case	Gestational Age (weeks+days)	Prenatal Imaging	Family History	Exam Findings	Postnatal Imaging	EEG	ASMs	Genetic Results
1	40+3	Echo: CR; MRI: CT+SEN	None	Retinal hamartomas	RUS: right RC MRI: CT+SEN	T7/P7 SC seizures	LEV, VGB, PHB, FOS	<i>TSC2</i> DEL ex. 15-38
2	40+1	Echo: CR; MRI: CT+SEN	Epilepsy (paternal cousin)	Systolic ejection murmur, sacral dimple	RUS: RC vs AML; MRI: CT+SEN+SEGA	C3 SC seizures	LEV, VGB, FOS, OXC, Sirolimus	<i>TSC2</i> c.500_509dup10 (p.Gly171Aspfs*21)
3	30+6	Echo: CR; twin pregnancy (normal twin)	None	Respiratory distress at birth	HUS: left grade I subependymal hemorrhage	Prolonged IBI; C3 SC seizures	LEV, VGB	<i>TSC2</i> c.1258G>T (p.Glu420*)
4	40+0	Echo: CR	None	Retinal hamartomas	MRI: CT+SEN	Left posterior-temporal SC seizures	LEV, PHB, FOS	<i>TSC2</i> c.1258-1G>T
5	38+5	Echo: CR	None	Normal	MRI: hypointense T2 lesion in left parietal region	C3 SC seizures	VGB	<i>TSC1</i>
6	39+0	Echo: CR	None	Normal	MRI: CT+SEN	Left central SC seizures	Data unavailable	Data unavailable

ASMs: anti-seizure medications; Echo: echocardiogram; CR: cardiac rhabdomyomas; MRI: magnetic resonance imaging; HUS: head ultrasound; CT: cortical tubers; SEN: subependymal nodules; SEGA: subependymal giant cell astrocytoma; RUS: renal ultrasound; RC: renal cysts; AML: angiomyolipomas; SC: subclinical; IBI: inter-burst interval; LEV: levetiracetam; VGB: vigabatrin; PHB: phenobarbital; FOS: fosphenytoin; OXC: oxcarbazepine; TSC2: Tuberous Sclerosis Complex Type 2 (NM_000548.5)