This 45-year old woman with tuberous sclerosis complex (TSC) suffers from mental retardation, seizures, facial and subungal angiomas. She also has bilateral renal angiomyelolipomas which hemorrhaged, requiring right nephrectomy and left endovascular selective renal embolization. The non-contrast head CT (Figure 1) demonstrates multiple calcified subependymal tubers. The contrast enhanced abdominal CT (Figure 2) shows an enormous left angiomyelolipoma (AML) distorting the enhancing left renal parenchyma. Figure 3 is from a selective left renal angiogram, demonstrating an enlarged left kidney with multiple diffuse areas of hypervascularity corresponding to the CT scan.

TSC is an autosomal dominant disease caused by a genetic mutation in the TSC1 or TSC2 gene, which results in the formation of hamartomas in multiple organ systems. The incidence of TSC is about 1 in 6000 to 1 in 12,000. The classic clinical triad in TSC is mental retardation, epilepsy, and adenoma sebaceum (Vogt triad), but up to 50% of TSC patients may have normal intelligence. Most TSC patients demonstrate dermatological, neurological, renal, and/or cardiac manifestations. Skin findings include hypopigmented macules, facial angiofibromas, shagreen patches, and ungular fibromas. Cortical tubers and subependymal tumors are commonly seen. Rarely, subependymal tubers may progress to subependymal giant cell astrocytomas. There is a spectrum of renal abnormalities including AMLs, renal cysts, renal cell carcinoma, and oncocytoma. AMLs are present in 55%-75% of patients and may be complicated by spontaneous hemorrhage, which is a leading cause of mortality. In order to preserve renal function, surgical resection is avoided and endovascular embolization is the treatment of choice. AMLs are embolized for treatment when greater than 3-4 cm in size.
REFERENCES


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