Tuberous Sclerosis With Hypertension and Abdominal Pain in a Child

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INTRODUCTION
Tuberous sclerosis is an autosomal dominant disease characterized with development of multiple benign hamartomatous tumors in many organs. The classic triad is seizures, mental retardation, and cutaneous angiofibroma. However, the full triad occurs in only 29% of the patients, and 6% lack all three of the characteristics. Other findings in these patients are cortical tubers, subependymal nodules, and cardiac rhabdomyomas. Renal involvement is common in tuberous sclerosis (about 57% of cases), and the most common renal lesions are angiomyolipoma and renal cysts. In 2% to 3% of these patients have bilateral polycystic kidney, because of mutation of two adjacent genes on chromosome 16, which is diagnosed in infancy or early childhood.

This report is on a rather rare association of tuberous sclerosis and bilateral polycystic kidney, and it emphasizes the importance of detailed physical examination in pediatric patients with hypertension to find its underlying cause.

CASE REPORT
A 4-year-old boy presented with abdominal pain. Abdominal ultrasonography showed bilaterally enlarged kidneys with multiple cysts in favor of cystic kidney disease. There was no family history of polycystic kidney or other kidney diseases. He had no history of seizure or abnormal movements suspicious to convulsion, but significant hyperactivity was reported. The parents were not consanguineous and had 3 other children that were apparently healthy.

On physical examination, blood pressure was around 135/85 mm Hg in repeated measurements which was higher than the 95 percentile for the patient’s height and age. He had pinkish papules on face, particularly around the nose, which were treated as allergic eruptions since months before (Figure 1). Skin examination showed multiple

Figure 1. The patient had pinkish papules around the nose.
hypopigmented lesions (Ash leaf spots) over the chest, abdomen, and extremities. Laboratory studies showed blood urea nitrogen, serum creatinine, and electrolytes levels within references ranges. Complete blood count and urinalysis also showed normal results.

With high suspicion of tuberous sclerosis, Brain computed tomography was performed for the patient, which showed calcified lesions in favor of cortical tubers (Figure 2). Ophthalmologic examination showed no retinal involvement. Echocardiography was normal. Psychologic evaluation revealed mild to moderate decrease of intelligence quotient and evidence of hyperactivity disorder.

Treatment of hypertension was started with oral nifedipine, 5 mg, twice per day, which increased to 10 mg, thrice per day. However, hypertension was not completely controlled because of irregular follow-up visits of the patient and low compliance. Two years after the diagnosis, the patient had his first episode of seizure and anticonvulsant treatment was started for him.

DISCUSSION

Tuberous sclerosis is one type of neurocutaneous disease with a prevalence of 1 in 6000 to 1 in 10000 births. Diagnosis of the disease is based on documenting 2 major or 1 major plus 2 minor criteria. The major criteria include hypomelanotic macule, cortical tuber, retinal hamartoma, facial angiofibroma, cardiac rhabdomyoma, and renal angiomylipoma. The minor criteria are pits in dental enamel, bone cysts, multiple renal cysts, and gingival fibroma. Many pediatric patients present with seizure and skin lesions. Epilepsy occurs in more than 70% to 80% of the patients, and almost all subtypes of seizure (simple partial, complex partial, and generalized tonic clonic seizures) have been reported.

There are a few reports of association of tuberous sclerosis with autosomal dominant polycystic kidney disease (ADPKD) in the literature. In most of these reports, there was a positive history of previous seizures in the patients. Sampson and colleagues described 27 patients with tuberous sclerosis and renal cysts, 24 of them, like the presented patient, had a negative family history of polycystic kidney disease. Glazier and colleagues reported 2 infants with ADPKD-like cystic disease and tuberous sclerosis. In the presented case, ADPKD could not be associated with tuberous sclerosis by genetic studies or a positive family history. However, bilateral enlargement of the kidneys and pattern of renal cysts in the presented patient’s ultrasonography urged the radiologist to report “in favor of polycystic kidney disease.” Moreover, a negative family history does not exclude the possibility of ADPKD, as the affected parent may have a clinically silent disease.

Mitnick and coworkers reported a 15-year-old girl with signs of tuberous sclerosis and renal cystic disease. She presented with bilateral flank mass in infancy and skin lesions at the age of 5 years, but diagnosis of tuberous sclerosis was not made until the first seizure episode at 8 years. The patient developed hypertension at the age of 15 years old. Her brother and father were also affected. Michel and associates described 2 half-brothers with polycystic kidney disease with no history of disease in their father. One of them developed adenoma sebaceum at an older age than the age the patient presented here, and the other developed infantile spasms.

The interesting points in this case were: (1) there was no history of seizure and the patient was referred with only a chief complaint of abdominal pain; (2) multiple hypopigmented skin lesions and facial angiofibroma of this patient had not been
adequately noted and followed up; and (3) renal involvement in this case was in the form of bilateral renal cystic disease, presenting with hypertension. This case shows the importance of detailed physical examination and correlating clinical findings of patients even in the absence of the most common presentations of a specific disease.

CONFLICT OF INTEREST
None declared.

REFERENCES

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Received October 2009
Revised March 2010
Accepted May 2010