

A Case of Tuberous Sclerosis Diagnosed Following Sepsis and Acute Exacerbation of Chronic Kidney Disease

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Abstract**Case Report**

In this case, a whole-body computed tomographic scan performed to evaluate multi-organ failure including sepsis and an acute exacerbation of chronic kidney disease revealed the presence of angiomyolipomas in both kidneys, leading to a suspicion of tuberous sclerosis. Further evaluation revealed autism spectrum disorder; abnormal calcifications or hemangioma-like findings in the cerebral cortex suggestive of cortical nodules or hamartomas; multiple lingual fibromas; multiple enamel hypoplasia lesions; multiple small cysts in the lungs suggestive of lymphangioleiomyomatosis; a well-defined, hyperechoic lesion in the heart suggestive of cardiac rhabdomyoma; osteosclerosis of the extremities; sporadic cutaneous fibromas, and abnormal ossification around the right ribs. Based on the above findings, although there were no typical cutaneous symptoms, a diagnosis of tuberous sclerosis was performed.

Keywords: tuberous sclerosis; angiomyolipoma; hamartoma; atopic dermatitis; chronic renal failure.

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1. INTRODUCTION

Tuberous sclerosis complex (TSC) is a disorder characterized by the systemic occurrence of hamartomas such as subependymal giant cell astrocytoma (SEGA) in the brain, renal angiomyolipomas, pulmonary lymphangioleiomyomatosis (LAM), and facial hemangiomas throughout the body, due to the loss of inhibition of the downstream mTORC1 pathway caused by dysfunction of the complex formed by the proteins hamartin and tuberin, which are produced by the causative genes TSC1 and TSC2, respectively [1,2].

The symptoms of tuberous sclerosis affect nearly the entire body, and the onset and severity of each symptom vary widely. These include cardiac rhabdomyomas that appear from the fetal period through infancy; skin depigmentation present at birth; epilepsy, autism spectrum disorder, and intellectual disability that emerge during infancy and early childhood; facial angiofibromas; brain tumors that often become problematic from infancy through early childhood; retinoblastomas; renal angiomyolipomas and cysts that become prominent from childhood through adolescence; pulmonary LAM and multifocal micronodular pneumocyte hyperplasia (MMPH), which are particularly problematic in women aged 20 and older; and gastrointestinal tumors and uterine lesions, which

become more common in people in their 40s and older. Other conditions frequently observed include periungual fibromas, Shaglin's patches, enamel pitting in the teeth, osteosclerosis, liver tumors, and ovarian cysts [1,2].

In this report, we present a case in which the patient was not diagnosed with tuberous sclerosis until the age of 30 because there were virtually no cutaneous manifestations of the disease. The patient was transported to our hospital due to altered consciousness resulting from an acute exacerbation of chronic kidney disease triggered by sepsis, and a diagnosis of tuberous sclerosis was made based on the presence of angiomyolipomas in both kidneys, as revealed by a whole-body computed tomographic scan (CT).

2. CASE REPORT

A 30-year-old man, whose cardiac hypertrophy and atopic dermatitis had been largely left untreated and who had recently begun experiencing choking episodes,

contracted enteritis and developed diarrhea. A few days later, he was found collapsed at home and was transported to our hospital by ambulance. It appears his mother had been complaining of unexplained pain in her limbs, but since they were divorced, the details were unclear. His grades in school were poor, and after

graduating from high school, he failed his job interviews and remained unemployed and shut himself away at home. On presentation, the patient's vital signs were as follows: Glasgow Coma Scale score of 11, systolic blood pressure of 130 mmHg, pulse rate of 90 beats per minute, respiratory rate of 50 breaths per minute, SpO₂ of 100% (room air), and body temperature of 35.3 °C. On physical examination, the skin showed lichenification and hyperpigmentation due to generalized atopic dermatitis; the lower legs had a fish-scale-like appearance, and small hemorrhages and crusts resulting from scratching were observed in various areas throughout the body. Venous blood gas analysis revealed severe metabolic acidosis under room air conditions, with a pH of 6.764, pCO₂ of 15.0 mmHg, HCO₃⁻ of 2.2 mmol/L, and base excess of -33.2 mmol/L. A cardiac ultrasound revealed impaired wall motion and inferior vena cava collapse. A 12-lead electrocardiogram revealed tent-shaped T waves and negative T waves in leads V4–V6. A diagnosis of sepsis was made, and a whole-body computed tomographic scan (CT) was performed to identify the source of infection [3]. The scan revealed atrophy of both kidneys and findings suggestive of angiomyolipoma, leading to a suspicion of tuberous sclerosis (**Figure 1**). The results of the complete blood count and biochemical tests were as follows: white blood cell count (WBC) 12,500/μL; hemoglobin 8.1 g/dL; platelet count: 219,000/μL; total protein 6.6 g/dL; albumin 3.2 g/dL; total bilirubin 0.1mg/dL; aspartate aminotransferase 32 U/L; alanine aminotransferase 22 U/L; lactate dehydrogenase 810 U/L; alkaline phosphatase 80 U/L; gamma glutamyl transferase 11 U/L; creatine kinase 1,377 U/L; amylase 466 IU/L; blood urea nitrogen (BUN) 216.2 mg/dL; creatinine (Crea) 31.55 mg/dL; uric acid 22.3 mg/dL; sodium 149 mmol/L; potassium 7.1mmol/L; chloride 110 mmol/L; calcium 5.5 mg/dL; phosphate 23.0 mg/dL; blood glucose 63 mg/dL; ammonia 313 n-μg/dL; b-type natriuretic peptide 543.2 pg/mL; high-sensitivity troponin 273.2 pg/mL; c-reactive protein 1.91 mg/dL; prothrombin time international normalized ratio 1.2; activated partial thromboplastin time 44.8 seconds; fibrinogen 471 mg/dL; fibrin degradation products 11.2 μg/mL; urine protein, 3+, WBC +/- . Based on the above, the clinical diagnosis was atopic dermatitis, dehydration, sepsis, acute exacerbation of chronic kidney disease, hyperkalemia, hypocalcemia, hyperuricemia, anemia, tachypnea due to marked metabolic acidosis, hypoglycemia, septic cardiomyopathy, rhabdomyolysis, hyperammonemia, and suspected tuberous sclerosis due to the presence of bilateral angiomyolipomas [4,5]. Due to impaired consciousness and poor respiratory status, it was decided to administer rapid intravenous fluid therapy, correct hypoglycemia and hypocalcemia, sedate the patient and place them on mechanical ventilation, and perform emergency continuous filtration dialysis.

Initially, because of the patient's impaired consciousness and tachypnea, it was unclear whether the patient had a history of enteritis; therefore, sepsis and multiple organ failure resulting from a skin infection were suspected, and cefazolin was administered along with a blood transfusion. Atopic dermatitis was treated with a steroid ointment. On the second day of hospitalization, hypokalemia had decreased to 4.6 mEq/L, however, there was little improvement in metabolic acidosis (pH, 6.964 and HCO₃⁻, 5.6 mmol/L). Since the patient's respiratory and circulatory status remained stable, renal replacement therapy was switched to conventional dialysis. On the third day of hospitalization, metabolic acidosis improved, so the patient was extubated. On the fourth day of hospitalization, we began oral feeding and gait rehabilitation. Subsequently, vitamin D deficiency was identified as the cause of hypocalcemia; we administered vitamin D and daprodustat to treat renal anemia. Since *Staphylococcus aureus* (MSSA) the same strain found in the skin culture was detected in the blood culture, we continued cefazolin administration for two weeks. In addition, since a definitive diagnosis of tuberous sclerosis had not been made, various tests were conducted, and a swallowing assessment was performed because the patient's choking did not improve. Based on the medical history and various test results, in addition to the diagnosis of bilateral renal angiomyolipomas and cysts using the gradient echo and dual echo methods, findings suggestive of autism spectrum disorder, cortical nodular lesions, abnormal calcifications or hemangioma-like findings in the brain suggestive of hamartomas or cortical nodules on magnetic resonance image (MRI) T2*-weighted images (WI) (**Figure 2**), multiple lingual fibromas, multiple enamel hypoplasia (**Figure 3**), multiple small cysts in the lungs suggestive of lymphangioleiomyomatosis (**Figure 4**), a well-defined, hyperechoic lesion in the heart suggestive of cardiac rhabdomyoma, osteosclerosis of the humeral head and femoral trochanter, scattered skin fibromas, and abnormal ossification around the right ribs (**Figure 5**) [6,7]. Based on the above, this case was clinically diagnosed as tuberous sclerosis [1,2]. In this case, the patient's entire body had developed lichenified lesions due to atopic dermatitis; no facial angiofibromas, typical leukoderma, periungual fibromas, or Shaglin patches skin lesions commonly associated with tuberous sclerosis were observed. Laryngoscopic examination revealed multiple small ulcers in the larynx and dysphagia; however, these findings showed signs of improvement. Since diuresis could not be maintained in this case and the patient was highly likely to require chronic dialysis, the patient was transferred to another hospital on the 16th day of hospitalization for maintenance dialysis.

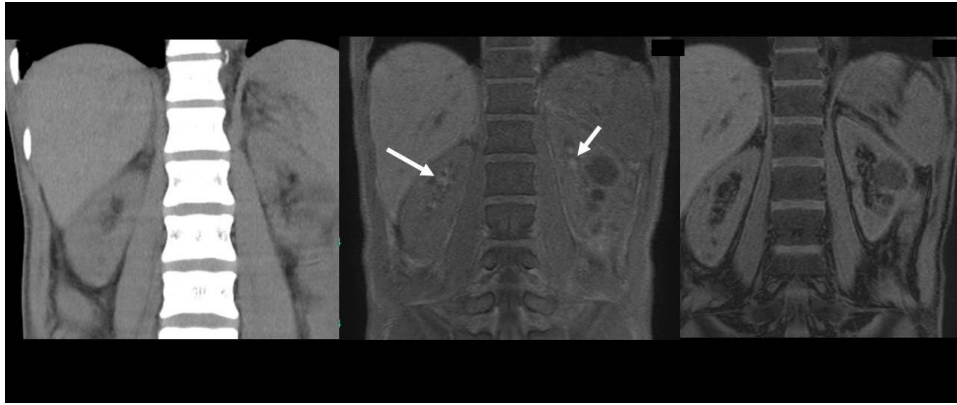


Figure 1: Abdominal computed tomographic (CT) scan (left) and magnetic resonance image (MRI) using the gradient echo and dual echo methods (middle, in-phase; right out-phase)

The CT scan revealed atrophy of both kidneys and findings suggestive of angiomyolipoma, leading to a suspicion of tuberous sclerosis. In the in-phase view, the

high-intensity areas change to a low-intensity areas in the out-phase view, indicating fats.

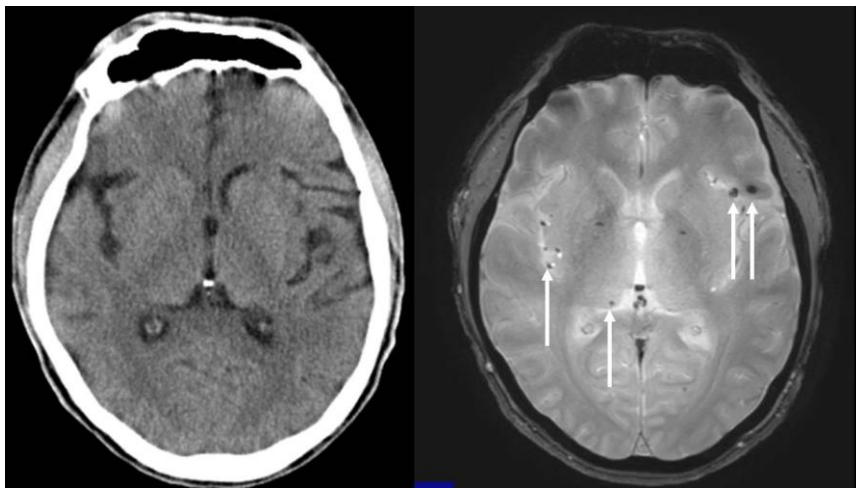


Figure 2: Head computed tomographic (CT) scan (left) and magnetic resonance image (MRI) using the T2*-weighted images (WI) (right)

The MRI shows multiple low-intensity spots in the brain, indicating abnormal calcifications or hemangioma suggestive of hamartomas.



Figure 3: Intraoral findings

Intraoral findings show multiple multiple enamel hypoplasia (white arrows), and lingual fibromas (black arrows)



Figure 4: Chest computed tomographic (CT) scan

The CT shows multiple small cysts in the lung (arrows) suggestive of lymphangioleiomyomatosis

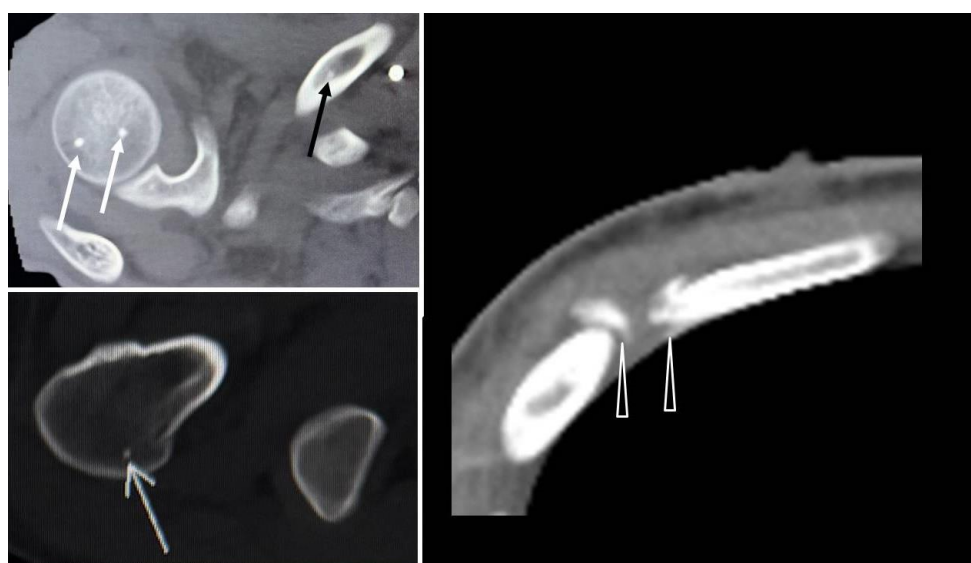


Figure 5: Bony findings

Radiographic studies show osteosclerosis of the humeral head (left upper, arrows) and femoral trochanter (left lower, arrow), and abnormal ossification around the right ribs (right, triangles).

3. DISCUSSION

In this case, the patient was not diagnosed until the age of 30 because he did not exhibit the typical cutaneous manifestations of tuberous sclerosis. The diagnosis of tuberous sclerosis was made following the detection of angiomyolipomas in both kidneys during a whole-body CT scan performed to evaluate multi-organ failure, including sepsis and an acute exacerbation of chronic kidney disease. The gradient echo dual echo technique was useful for distinguishing between cysts and fat, and MRI T2*-WI was useful for identifying images suggestive of cerebral hamartomas that were difficult to detect on head CT [8,9].

One possible reason why this patient was not diagnosed with tuberous sclerosis until the age of 30 is that he did not exhibit the typical skin lesions commonly associated with the condition, and he had widespread atopic dermatitis. Typical skin manifestations of tuberous sclerosis include angiofibromas (approximately 75%), nail fibromas (approximately 20–80% of affected

individuals), fibrous patches on the scalp (approximately 25%), Shagreen-like patches (more than 50%), and localized depigmentation (approximately 90%) [10,11]. In this case, we observed white patches that appeared to be post-inflammatory hypopigmentation; however, the white patches characteristic of tuberous sclerosis are leaf-shaped referred to as “leaf-shaped white patches” and are typically at least 5 mm in diameter, so the findings in this case did not match this description.^{1,2} Since only about 4% of cases present without the characteristic skin manifestations of tuberous sclerosis, it was considered highly likely that the diagnosis in this case was delayed [12]. Furthermore, no reports of cases combining tuberous sclerosis and atopic dermatitis were found in the literature.

This case presented with renal atrophy at the time of admission; combined with abnormally high BUN and Crea levels at that time, we believe this represents an acute exacerbation of chronic kidney disease triggered by sepsis resulting from enteritis. The incidence of renal

failure in tuberous sclerosis is not high: approximately 5% of cases develop chronic renal failure, and only about 1% require dialysis. The primary causes of renal failure are believed to be a decline in renal function resulting from angiomyolipomas or multiple cysts in the kidneys, their enlargement, hemorrhage, embolization procedures to treat bleeding, malignant transformation, or nephrectomy performed to remove a tumor [13-17]. In this case, although there were multiple angiomyolipomas and cysts in the kidneys, they did not occupy a significant portion of the kidneys as seen on MRI images. Typically, dialysis becomes necessary at an average age of 48, and the patient in this case is still young [15]. On the other hand, in this case, atopic dermatitis had been largely left untreated due to the patient's autism spectrum disorder, resulting in lichenification throughout the body; therefore, it was considered possible that chronic inflammation caused by this skin condition may have contributed to the deterioration of renal function [18-19]. Evidence suggests that the decline in renal function associated with tuberous sclerosis can be mitigated by prophylactic treatment with mammalian target of rapamycin inhibitors, highlighting the need for early diagnosis [15].

It appears that the patient had dysphagia prior to admission to our hospital, and this was confirmed by tests conducted during hospitalization. Although there are very few reports of dysphagia in cases of tuberous sclerosis, since there have been a previous case report of pediatric patients developing speech and swallowing disorder, dysphagia caused by tuberous sclerosis is a possible complication [20]. In a report investigating the causes of death in 19 cases of tuberous sclerosis, malignant tumors were the leading cause in 6 patients, followed by seizures and aspiration pneumonia, each accounting for 3 deaths. Thus, dysphagia may be a serious complication that affects the prognosis of tuberous sclerosis [21].

4. CONCLUSION

We report a case of tuberous sclerosis that was incidentally diagnosed during an investigation to identify the cause of sepsis. MRI using the gradient echo dual-echo technique was useful for diagnosing renal angiomyolipomas, and MRI T2*-WI was effective for detecting images suggestive of cerebral hamartomas that were difficult to identify using head CT.

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