American Journal of Medical Case Reports, 2019, Vol. 7, No. 12, 334-336
Available online at http://pubs.sciepub.com/ajmcr/7/12/8
Published by Science and Education Publishing
DOI:10.12691/ajmcr-7-12-8

Wunderlich's Syndrome in Advanced Tuberous Sclerosis Complex: A Rare Case of Perinephric Hematoma

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Received August 11, 2019; Revised September 16, 2019; Accepted October 08, 2019

Abstract Tuberous sclerosis complex (TSC) is a rare, autosomal dominant disease, associated with mutations in either the TSC gene. The incidence of TSC is estimated as 1 in 5,000-10,000 live births and angiomyolipomas (AML) is one of the characteristic findings of tuberous sclerosis. A 36-year-old Caucasian female admitted to the hospital after two episodes of pre-syncope in March 2016. The patient was tachycardic on examination, and laboratory studies revealed acute anemia with hemoglobin of 8.2 g/dL on admission with a decrease to a nadir of 6.2 g/dL. Imaging studies revealed bilateral fat-rich renal masses with distortion of the renal anatomy consistent with angiomyolipomas (AML). Further as a part of treatment embolization of the right renal artery was performed. However, everolimus is also approved for the treatment of AMLs not requiring surgery. Each patient should have an individualized treatment plan based on the clinical situation as determined by the physician.

Keywords: *tuberous sclerosis complex, sub-ependymal nodules, angiomyolipoma*

Cite This Article: Aisha Siraj, and Negar Salehi, "Wunderlich's Syndrome in Advanced Tuberous Sclerosis Complex: A Rare Case of Perinephric Hematoma." *American Journal of Medical Case Reports*, vol. 7, no. 12 (2019): 334-336. doi: 10.12691/ajmcr-7-12-8.

1. Introduction

1.1. Tuberous Sclerosis

Tuberous sclerosis complex (TSC) is a rare, autosomal dominant disease, associated with mutations in either the TSC1 gene on chromosome 9 or the TSC2 gene on chromosome 16. The incidence of TSC is estimated as 1 in 5,000-10,000 live births [1]. TSC usually presents with neurologic disorders including seizures, mental retardation, autism, brain lesions [cortical tubers, sub-ependymal nodules (SEN)], and sub-ependymal giant-cell astrocytomas. TSC can also present with cardiac, pulmonary, renal, and ophthalmic findings such as angiomyolipomas (AML) of the kidneys and rhabdomyoma of the heart. There are several distinct dermatologic findings, including facial angiofibroma, Shagreen patches, and Hypomelanic macules (Ash-Leaf spots).

Wunderlich syndrome was first defined in 1856 as spontaneous non-traumatic renal hemorrhage into the perinephric space [2].

2. Case Report

We are presenting a case of a 36-year-old Caucasian female admitted to the hospital after two episodes of pre-syncope in March 2016. Characteristic features of TSC including malar angiofibroma, (Figure 1), ash- leaf spots (Figure 2), bilateral fat-rich angiomyolipomas of the kidneys (Figure 3), and sub-ependymal calcifications (Figure 4) were noted. The patient was tachycardic on examination, and laboratory studies revealed acute anemia with hemoglobin of 8.2 g/dL on admission with a decrease to a nadir of 6.2 g/dL.

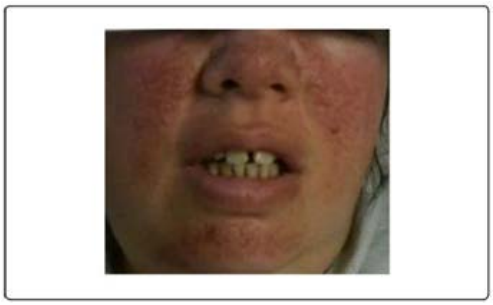

Figure 1. Subcutaneous adenomas Facial Angiofibroma

Figure 2. Ash-leaf appearance on the back

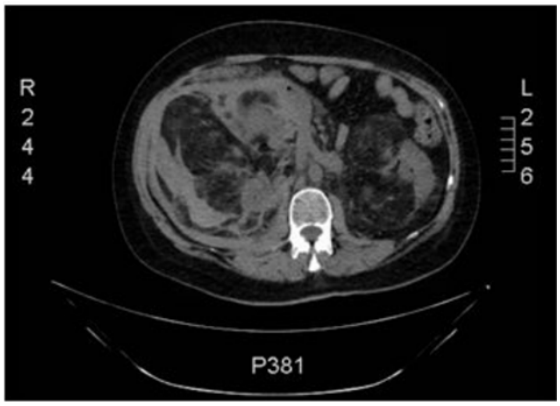

Figure 3. Non-contrast enhanced CT of the abdomen with Bilateral fat-rich Angiomyolipomas of the kidney and extensive right peri-renal fluid consistent with hemorrhage

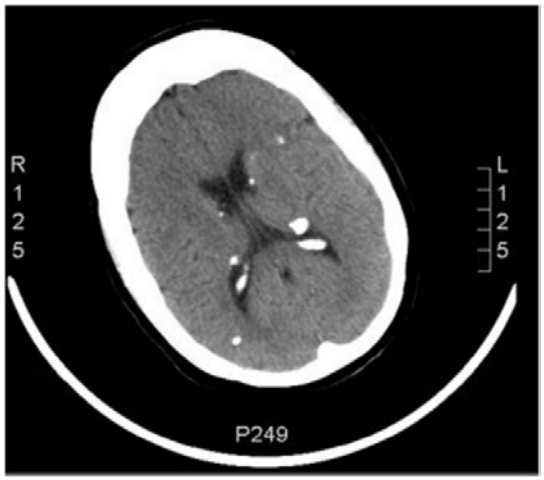

Figure 4: Sub ependymal Nodules as seen in the brain CT scan

Abdominal CT revealed bilateral fat-rich renal masses with distortion of the renal anatomy consistent with angiomyolipomas (AML). Fluid along the lateral aspect of the right kidney, thought to represent hemorrhage into Gerota's fascia, was also evident. The patient was stabilized with fluid resuscitation and red blood cell transfusion and was taken for embolization of the right renal artery with coiling and injection of tris-acryl gelatin microspheres. Post-procedure day 2-3 she had nausea and flank pain that was managed with antiemetics and narcotic respectively. She recovered uneventfully without evidence of further bleeding. Renal function remained within normal limits.

3. Discussion

There are three major renal complications in tuberous sclerosis, AML is the most common with polycystic renal disease (PKD) and renal cell carcinoma (RCC) being rarely described. AML is one of the characteristic findings of tuberous sclerosis. AML is a benign tumor consisting of blood vessels, smooth muscle cells, and fat cells. AML incidence in the average population is about 2% while prevalence in tuberous sclerosis patients is 50%-80% [3].

One of the best approaches for the management of the symptomatic AML is selective arterial embolization (Figure 5). In our patient, the same treatment was done with the combination of three coils and embosphere injection.

Novel therapeutic options for the large AML which cause retroperitoneal bleeding or has the risk of bleeding (Tumors>4 cm) is mTOR pathway inhibitors, which include Sirolimus (also known as rapamycin) and Everolimus. A trial of Everolimus showed a reduction in AML size by 30% in 81.6% of patients and 50% reduction in AML in 64.5% of patients by 96 weeks of treatment, showing added benefit of longer-term Everolimus therapy without additional adverse events [5,6,7].

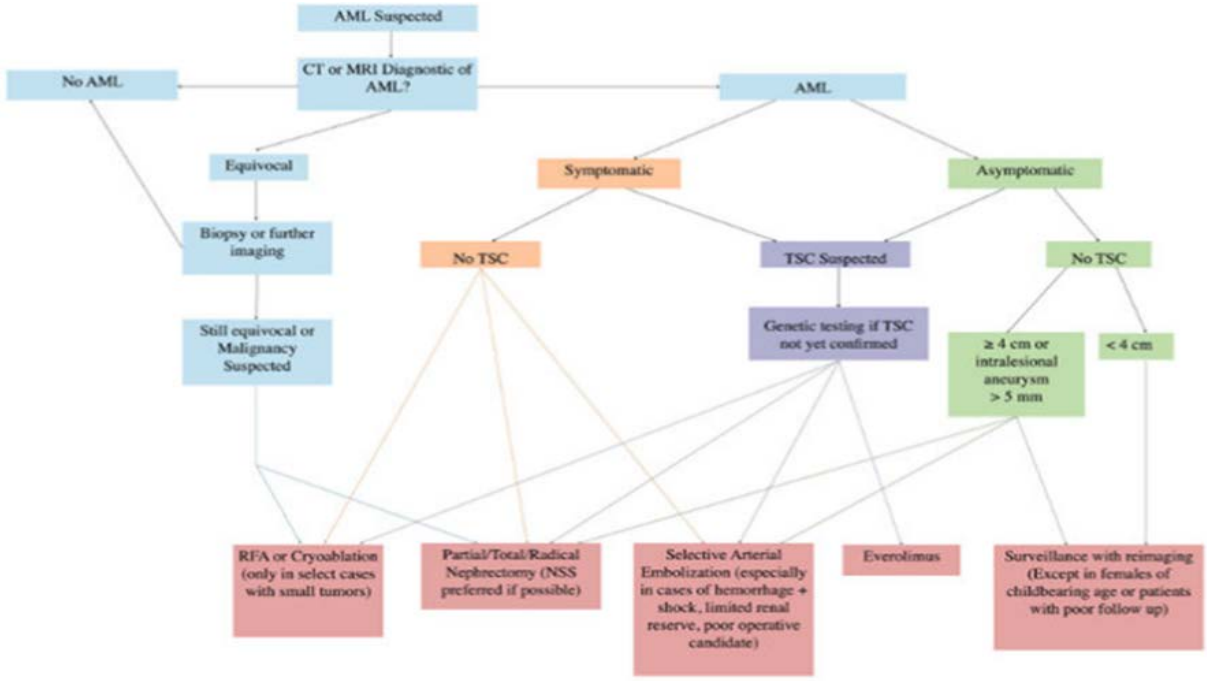

Figure 5. Algorithm for management of Angiomyolipomas [4]. Proposed updated management algorithm for renal AML. While options are suggested, specific treatment decisions should take into consideration individual clinical scenario, and patient and surgeon preference. RFA: Radio Frequency Ablation

4. Conclusion

In large AML tumors in the setting of tuberous sclerosis that have a high risk of spontaneous retroperitoneal bleeding, selective arterial embolization remains a preferred strategy for the treatment. There are other options, like partial or total nephrectomy which have their benefits and risks. Everolimus is also approved for the treatment of AMLs not requiring surgery. Each patient should have an individualized treatment plan based on the clinical situation as determined by the physician.

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