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Case Report

Incidental finding of renal pseudoaneurysm in child with tuberous sclerosis presenting with atypical clinical symptoms

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ABSTRACT

Tuberous sclerosis (TS) is a complex medical disorder with multisystemic clinical manifestations. Although the renal manifestations of this disease are well researched, the complexities of clinical diagnosis are raised significantly in patients with cognitive impairments, particularly in the pediatric population. We present a case of a 12-year-old male with intellectual disabilities and renal angiomyolipomas associated with his TS complex presenting with subtle cognitive and behavioral changes leading to the eventual diagnosis of a renal pseudoaneurysm. The purpose of this case report is to highlight the subtleties of diagnosis and management of patients with TS and cognitive impairments and maintaining a high clinical index of suspicion for life threatening complications when presenting symptoms are nonspecific. This case also demonstrates the importance of obtaining a thorough clinical history from parents and caregivers of these children and educating them on significance of recognizing changing behavior patterns. The healthcare responsibility for diagnosis and management must be shared by all levels of personnel that participate in the care to allow for improved morbidity, mortality, and quality of lives for these patients.

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Clinical history

We present a case of a 12-year-old male with an incidental finding of a large pseudoaneurysm discovered incidentally on routine ultrasound examination. The patient's history was significant for Tuberous sclerosis (TS) complex with multiple renal angiomyolipomas (AML) and autosomal dominant polycystic kidney disease associated TSC2 mutation. The renal AML were monitored with routine renal ultrasounds and MR abdomen. The patient presented to the emergency room with low grade fever and a sore throat. At this time, the parents also reported subtle behavioral changes, asymmetric left side back swelling, and inability to lay on his right side. Physical exam demonstrated left lumbar area fullness, firmness on palpation without fluctuance. A back and pelvis ultrasound was done for the left flank and back swelling.

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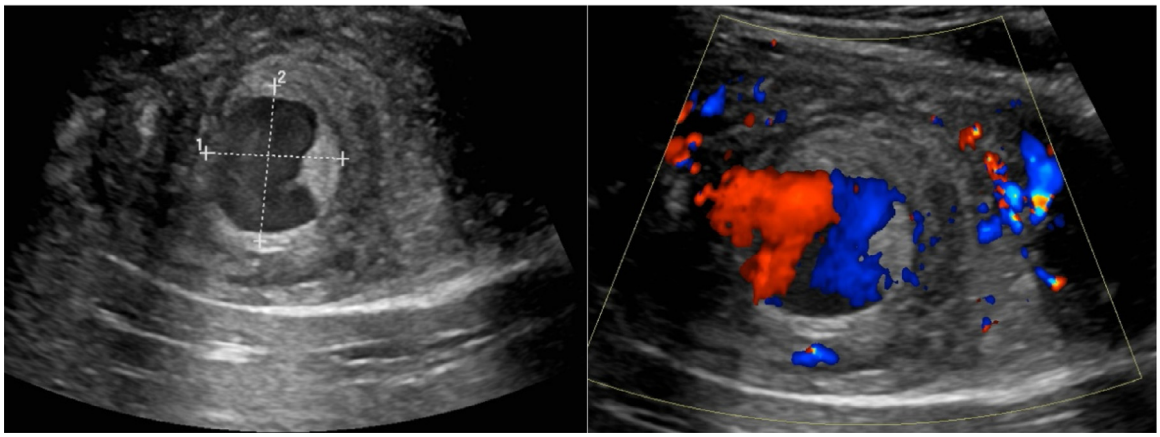

Fig. 1 – Right kidney pseudoaneurysm with the classic "yin-yang" appearance.

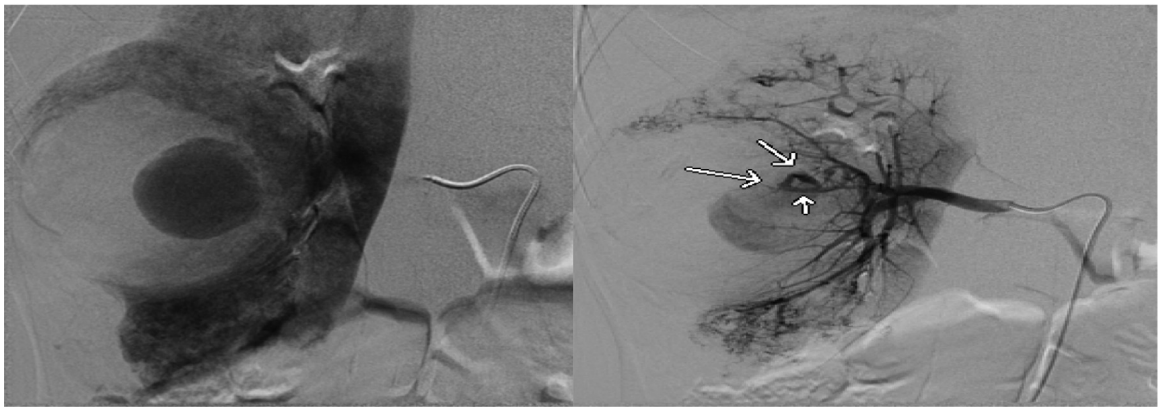

Fig. 2 – Selective right renal artery embolization pre and post 300-500 um embosphere particles.

Imaging findings

The ultrasound exam demonstrated asymmetry in the soft tissues on the left, without a focal collection. In the setting of known renal AML's, an additional scan of the kidneys was also performed by the ultrasound technologist. Incidentally found was a large 3.2 cm pseudoaneurysm in the mid-pole of right kidney which demonstrated the classic "yin-yang" appearance on color Doppler evaluation (Fig. 1).

The patient was sent to the Pediatric Emergency Department for admission due to the concerning imaging findings. Interventional Radiology was consulted and performed a renal arteriogram followed by an abdominal aortogram which confirmed a large pseudoaneurysm in the mid-pole of the right kidney (Fig. 2).

Selective right renal artery embolization (Fig. 2) was performed with 300-500 um embosphere particles. After sluggish flow was obtained, a combination of Tornado and Nester coils were used to embolize the feeding arterial branch distal to, at, and proximal to the pseudoaneurysm neck. Postembolization and coiling selective arteriogram showed no further filling (Fig. 3).

Discussion

AML are known renal manifestations of TS and the most common renal lesion associated with this complex [1,2]. Clinically they present with hemorrhage, hematuria, or mass effect [2–4]. These lesions can develop microaneurysms or macroaneurysms which can spontaneously rupture and have life-threatening complications. This necessitates maintaining a high clinical index of suspicion for diagnosis, particularly in pediatric patients with cognitive impairments. Although the multiorgan presentations of TS are well known in the literature, many patients are asymptomatic or report minimal symptoms of their renal disease. Patients with the TS complex frequently have concurrent behavioral or cerebral impairments hindering their ability to report their symptoms, complicating the diagnosis of acute clinical manifestations.

Cognitive impairments are complications of the TS complex that affects 44%-65% of patients [5,6]. The effect on cognition varies widely within the pediatric population with impacts on behavior and intelligence quotient [7]. Autism and autistic behaviors are common in children with TS complex with prevalence ranging from 40% to 90%. These factors limit

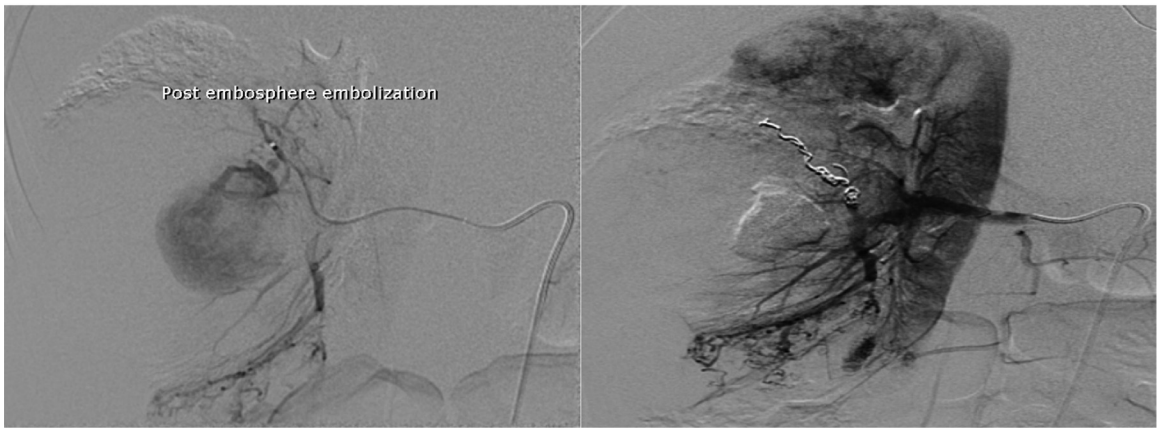

Fig. 3 – Postembolization and coiling selective arteriogram.

the ability of children to raise a clinical concern in the event of an acute change. Our patient has known cognitive impairments limiting his ability to communicate his symptoms requiring a thorough clinical history be obtained from the parents. The child reportedly had significantly reduced activity levels for days prior to presenting to the hospital, started to curl onto his left side when sleeping and did not sleep on his right side. Although he had a fever and concerns of an infection at the time of presentation, the history of asymmetric back swelling was also simultaneously worked up with the ultrasound which then led to the incidental diagnosis of a large renal pseudoaneurysm. This highlights the importance of addressing all clinical concerns in children with cognitive impairments as potentially life-threatening complications may present as subtle behavioral changes or be masked by other symptoms and may be overlooked clinically.

Another key feature to highlight is the reliance on parents and caretakers to obtain pertinent clinical information and the importance of a thorough diagnostic workup. Parents and caretakers play a critical role in the management of such patients and must be thoroughly educated by the clinical team in the recognition of behavior patterns and cognitive changes that warrant immediate clinical attention. Clinicians rely on this critical information to formulate treatment plans and guide management. Thorough knowledge of the clinical history and focused physical exams are key in every case, however situations like these mandates an even closer focus and a level of responsibility to be shared by all levels of healthcare. In our case, the diligence of an ultrasound technologist to scan the patient's kidneys knowing the underlying history of AML was imperative in uncovering this diagnosis even though the requisition was for superficial back swelling.

In cases like these, it is important to maintain a high index of suspicion for the presence of acute potentially serious complications. In an age of rising emergency room visits, hospital admissions, and reliance on medical imaging, it is imperative to perform detailed history taking and clinical exams. These challenges are magnified with managing a pediatric population with cognitive impairments with complex underlying medical histories. The detailed knowledge of the child's medical history, prior imaging studies and interventions, and access to all participating provider clinical notes is a responsibility that must be shared by all healthcare personnel participating in the care. This level of meticulousness on a presumably routine case of a viral upper respiratory infection (URI) and associated behavioral changes led to the diagnosis of a pseudoaneurysm requiring emergent intervention with a very successful outcome.

Conclusion

TS is a complex multiorgan disease with clinical manifestations that can be life-threatening in some cases. The diagnosis of these clinical presentations can be furthermore complicated by the presence of concurrent cognitive disabilities that hinder a child's ability to report any acute changes to their health. AML are the most common renal manifestation of this complex and can give rise to pseudoaneurysms that can grow in size and rupture leading to life-threatening hemorrhage. Patients with cognitive and mental impairments are unable to vocalize their symptoms but rather may demonstrate changing activity and behavior levels, fluctuations in mood and appetite, and physical symptoms such as side preference while sleeping or refusal to weight bear. These symptoms are usually nonspecific and can be masked by other concurrent medical issues that would regularly be treated in a nonacute fashion. This requires medical personnel to maintain a high clinical index of suspicion when such patients present for care. The clinical responsibility must be relayed to all levels of healthcare that participate in the care of these patients. It is also critical to incorporate the parents, care takers, or other healthcare proxies in the management of these children. This concerted all rounded effort may aid the diagnoses of an underlying life-threatening complication and allow for the necessary management and intervention, thereby preserving the quality of life and reducing the overall morbidity and mortality.

REFERENCES

[1] Rakowski SK, Winterkorn EB, Paul E, Steele DJ, Halpern EF, Thiele EA. Renal manifestations of tuberous sclerosis complex: incidence, prognosis, and predictive factors. Kidney Int 2006;70:1777.

[2] Ewalt DH, Sheffield E, Sparagana SP, et al. Renal lesion growth in children with tuberous sclerosis complex. J Urol 1998;160:141.

[3] Nelson CP, Sanda MG. Contemporary diagnosis and management of renal angiomyolipoma. J Urol 2002;168:1315.

[4] Seyam RM, Bissada NK, Kattan SA, Mokhtar AA, Aslam M, Fahmy WE, et al. Changing trends in presentation, diagnosis and management of renal angiomyolipoma: comparison of sporadic and tuberous sclerosis complex-associated forms. Urology 2008;72:1077.

[5] Yates JR, Maclean C, Higgins JN, Humphrey A, le Maréchal K, Clifford M, et al. The tuberous sclerosis 2000 study: presentation, initial assessments and implications for diagnosis and management. Arch Dis Child 2011;96:1020.

[6] Joinson C, O'Callaghan FJ, Osborne JP, Martyn C, Harris T, Bolton PF. Learning disability and epilepsy in an epidemiological sample of individuals with tuberous sclerosis complex. Psychol Med 2003;33:335.

[7] Kopp CMC, Muzykewicz DA, Staley BA, Thiele E, Pulsifer B. Behavior problems in children with tuberous sclerosis complex and parental stress. Epilepsy Behav 2008;13:505.