Huge Renal Angiomyolipoma in a Child with Tuberous Sclerosis Complex: A Diagnostic and Therapeutic Dilemma

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Abstract

Tuberous Sclerosis Complex (TSC) is a rare neuro-cutaneous disorder that is associated with the development of benign hamartomas including renal angiomyolipoma (RAML). TSC associated RAML are usually asymptomatic, but it carries a life-threatening bleeding risk. We are sharing a case of a 5-year-old girl who was diagnosed to have TSC with associated subependymal giant cell astrocytoma, cardiac rhabdomyoma and autism. She presented with a history of worsening abdominal distension over 3 weeks duration and clinically noted to be pale with a ballotable left flank mass. Ultrasound and CT scan found to have multiple RAML in both kidneys with a huge mass on the left side. The mass represented a huge RAML (8cm) with aneurysmal formation with suspicion of intratumoral bleeding. The option of conservative management with mammalian target of rapamycin inhibitor followed with partial nephrectomy has been questioned with its life-threatening risk of bleeding and inability to do biopsy to rule out the possibility of renal cell carcinoma. Decision for nephrectomy was then made clearer following a MAG-3 scan which revealed only 11% differential function of the left kidney. She underwent a total left nephrectomy uneventfully and intraoperatively noted to have an enlarging lesion as compared to the previous imaging; 15cm in largest diameter. Histopathological finding was consistent with multifocal angiomyolipoma with intratumoral haematoma. Decision for nephrectomy in TSC-associated RAML need to be justified carefully in view of its risk of losing the contralateral kidney following the disease progression which may end up with life-long renal replacement therapy.

Keywords: Tuberous Sclerosis Complex (TSC), renal angiomyolipoma (RAML), rapamycin inhibitor, partial nephrectomy

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