What is your diagnosis?

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CLINICAL SUMMARY

A three-year-old female born of non-consanguineous marriage presented with delayed language development skills. She was able to speak only few familiar words. She now had intermittent haematuria since 15 days. Also there was history of two episodes of seizures in infancy. On general examination, she

was afebrile and blood pressure was on the higher side of normal. Hypopigmented patches were seen on the trunk region and macular elevated lesions were seen on the face and neck. Abdominal examination revealed palpable kidneys. CNS and laboratory examinations were within normal limits. Computerised tomography (CT) abdomen and magnetic resonance imaging (MRI) brain was performed (Figures 1 and 2). What is your diagnosis?

Figure 1 Plain axial T2WI magnetic resonance imaging brain.

Figure 2 Axial FLAIR magnetic resonance imaging demonstrates cortical hyperintense tubers.

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Answer to Radiological Quiz - pg. 394

Tuberous sclerosis

Axial T2W brain image (Figure 1) shows cortical lesions which appear heterogeneously hyperintense and subependymal lesions which appear hypointense. On postcontrast study there was heterogeneous mild to moderate enhancement of cortical lesions (Figure 2). Few of subependymal lesions enhanced intensely (Figure 3). The above radiological imaging and clinical findings suggest tuberous sclerosis. Computerised tomography (CT) abdomen demonstrated multiple, different sized cysts in both kidneys causing severe parenchymal thinning (Figure 4). Post-contrast CT shows minimal enhancement of renal parenchyma and delayed excretion of contrast (Figure 5). Bilateral renomegaly was seen. These are typical features of bilateral polycystic kidneys.

Tuberous sclerosis is a rare, multisystem, congenital disorder of autosomal dominant variety that causes benign tumours to grow in the brain.¹ Symptoms may include seizures, developmental delay, behavioural problems, skin abnormalities, lung, and kidney diseases. Brain imaging shows cortical and subcortical tubers due to gyral enlargement, subependymal nodules, and subependymal giant cell astrocytomas. Kidney findings include angiomyolipomas, which can cause hypertension and polycystic kidney disease.² Lung shows multiple cysts causing lymphangioleiomyomatosis like picture. Cardiac findings include rhabdomyomas. Retinal patches, hamartomas, and astrocytomas are common.³ Skin findings include ash leaf-shaped macules, angiofibromas of the face (adenoma sebaceum), congenital shagreen patches and depigmented naevi.⁴

In this case there were brain (cortical tubers and subependymal nodules), kidney (bilateral polycystic kidneys), and skin (ash leaf-shaped macules and adenoma sebaceum) lesions. Clinical details of the patient included delayed development, seizures, early hypertensive findings, and skin lesions.

Figure 3 Postcontrast axial magnetic resonance imaging images demonstrates enhancement of cortical tubers as well as of subependymal tubers.

Figure 4 Plain computerised tomography abdomen shows polycystic kidneys.

Figure 5 Postcontrast computerised tomography abdomen shows polycystic kidneys with thinned out parenchyma.

A combination of imaging findings and detailed clinical evaluation is required in order to establish the diagnosis.

REFERENCES

1. Khan GA, Hassan G, Tak SI, Kundal DC. Depression as the presenting feature of tuberous sclerosis. *JK Practitioner* 2003;10:34–35.
2. O'Callaghan FJK, Noakes M, Osborne JP. Renal angiomyolipomata and learning difficulties in tuberous sclerosis complex. *J Med Genet* 2000;37:156–157.
3. Shields JA, Eagle RC, Shields CL, Marr BP. Aggressive retinal astrocytomas in 4 patients with tuberous sclerosis complex. *Arch Ophthalmol* 2005;123:856–863.
4. Victor M, Ropper AH. Developmental diseases of the nervous system. In: *Adams and Victor's – Principles of Neurology* 7th ed. New York: McGraw Hill, 2001:1050–1105.