Diffuse Cystic Renal Involvement in Tuberous Sclerosis

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Dear Sir,

We have read with interest the article 'Chronic renal failure in patients with tuberous sclerosis. Association with renal cysts', in Nephron 30: 85–88, 1982, and we would like to comment on a case recently studied by us. We would like to call attention to the diffuse cystic involvement of the kidney as a sign suggestive of the diagnosis of tuberous sclerosis in infancy, even prior to the appearance of the major signs which characterize this disease.

The patient was a 13-month-old boy admitted to hospital because of lower limb hypotonia, spasmodic crisis and psychomotor retardation. Physical examination revealed bilateral abdominal masses. There was no family history of kidney disease and the father had sebaceous adenomas on the face and thorax. Renal and hepatic functions were normal. Intravenous pyelogram (IVP), abdominal echo-gram and renal arteriography were all consistent with the diagnosis of polycystic kidneys. The microscopic examination of the liver was normal and the microscopic examination of the kidney was performed from a subcapsular specimen obtained by lumbotomy renal biopsy from a macroscopically normal cortical zone located among macroscopic cysts. There were no hamartomatous tumors and only some tubular cyst was observed. The CAT scan showed periventricular calcification (phacomias) that were diagnostic of tuberous sclerosis (fig. 1).

Fig.1. CAT scan: periventricular calcifications (arrows) diagnostic of tuberous sclerosis.

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Fig. 2. Abdominal echogram consistent with the diagnosis of poly-cystic kidneys.

Since the review of tuberous sclerosis done by Lagos and Gomez [1], the incomplete forms are universally accepted. When the diagnosis is made in infancy, the clinical manifestations are usually incomplete. The infantile spasms are the epileptic equivalent, in 69–80% of the cases diagnosed before 2 years of age [2]. The sebaceous adenomas appear usually late and the hypochromic lesions are normally present earlier. The key to a diagnosis of tuberous sclerosis is the periventricular calcifications present in the CAT scan as were found in our patient. These calcifications

along with the spasms and/or hypochromic lesions are diagnostic of tuberous sclerosis in infancy.

The kidney is the most frequently affected viscus in tuberous sclerosis. The diagnosis is easily made if there are multiple and bilateral hamartomatous tumors. When these tumors are not demonstrated, the kidneys can be mistaken for polycystic disease of the adult type, even in infancy [3], with potential evolution to renal failure produced by compression on the normal tissue (fig. 2). Therefore, large cystic kidneys in infancy with normal hepatic anatomy and function and without family history of polycystic kidney disease should lead us to the possibility of tuberous sclerosis. This diagnosis should be confirmed with a CAT scan before the skin lesions and typical neurologic symptoms appear.

References

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