Successful Live Related Renal Transplant in a case of Tuberous Sclerosis

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Abstract

Tuberous sclerosis (TS) is an autosomal dominant multisystemic disease involving primarily the skin, the brain and the kidneys. Inspite of the kidney being involved in 40-80% of patients with this disease, the incidence of end stage renal disease is only about 1%. There are only 34 reported cases of successful renal transplantation in tuberous sclerosis patients with end stage renal disease. We report a case of successful renal transplantation in a patient of tuberous sclerosis with bilateral polycystic kidneys presenting with renal failure who also underwent bilateral native nephrectomies on follow up.

INTRODUCTION

Tuberous Sclerosis (TS) is a rare familial disease with the prevalence of about 1 in 10-14 thousand. It is characterized mainly by neurological (epilepsy, mental retardation) and dermatological (adenoma sebaceum) signs. The kidney is involved in about 40-80% of patients with this disease. The renal lesions comprise of angiomyolipomas, renal cysts and rarely renal carcinoma. However, the incidence of end stage renal disease is about 1%. There are only 34 reported cases of successful renal transplantation in tuberous sclerosis patients. Previous reports have shown good results with transplant in this group of patients. We report here a case of successful renal transplantation in a patient of tuberous sclerosis with bilateral polycystic kidneys presenting with renal failure.

CASE REPORT

A 24-year-old male was referred to us for renal transplantation with a diagnosis of tuberous sclerosis with chronic renal failure and end stage renal disease. He was dialysis dependent for the last six months. He had history of repeated episodes of generalized tonic-clonic seizures since the age of three years and was given anticonvulsants for five years. A computed tomogram (CT) of the brain done at around 8 years of age was normal. The patient was completely normal till the age of twelve when he developed features of chronic renal failure. There was no family history of similar illnesses.

On general physical examination, he had adenoma sebaceum on the face (Fig. 1) and ash leaf spots on thigh and lumbar region of the back. His pretransplant serum creatinine was 10.6 mg%. Patient’s ultrasound abdomen had shown presence of bilateral polycystic kidneys with the right kidney measuring 17 cm and the left kidney 19 cm. He underwent a living related renal transplantation and was kept on triple drug immunosuppression with cyclosporine (8mg/kg), mycophenolate mofetil (2gm/day) and prednisolone (0.5mg/kg). He had an uneventful immediate postoperative course. At 1 year posttransplant, CECT of the abdomen was done to follow the renal lesions which revealed presence of haemorrhage into a cyst in the right kidney (Fig 2). Bilateral nephrectomy was done in view of suspicion of malignancy. Biopsy of the kidneys showed grossly enlarged kidneys with bosselated surface and the cut...
section showed numerous cysts and solid areas with haemorrhage. Microscopic findings were of angiomylipoma and renal cysts. There was no evidence of malignancy. On last follow-up, at 21 months posttransplant has a serum creatinine of 1.0 mg%.

**DISCUSSION**

Tuberous sclerosis (TS) or Bourneville’s disease is a familial syndrome transmitted through an autosomal dominant gene with incomplete penetrance. It is classically characterized by a triad of adenoma sebaceum, epilepsy and mental retardation. Other manifestations of the disease include cutaneous lesions i.e. ash leaf hypomelanotic macules, lumbosacral shagreen patch, retinal phakomas and hamartomas of many organs including kidneys, lungs, pleura and bones. The earliest and most frequent complaint is seizures in cases with TS and careful investigation for hypomelanotic macules or other skin manifestations typical for TS in cases presenting with convulsion makes early diagnosis possible and obviates unnecessary investigations. Renal involvement is usually bilateral and inspite of renal involvement being present in 40-80%, the incidence of end stage renal disease in TS is about 1%. There are only 34 case reports of successful renal transplants in cases of TS with chronic renal failure.

The second renal lesion found in TS is the renal cyst. Renal cysts in TS have a unique epithelial lining. The etiology of the cystic changes in tuberous sclerosis is unknown. Treatment of patients with renal cysts is rarely indicated unless the cysts become large and cause pain, obstruction or haemorrhage. Decompression of renal cysts has been reported to protect adjacent renal parenchyma from atrophy, but only in selected cases.

Surgery is reserved for life threatening bleeding from angiomylipoma and suspected malignancy. Since nephrectomy is an important cause of renal failure, bleeding should be controlled by embolization or by conservative surgery.

Ultrasound and CT scans are the imaging studies of choice in diagnosing the renal lesions in patients with TS. In 95% of cases, a CT scan can differentiate an angiomylipoma from malignancy.

TS with renal failure is characterized by late diagnosis (about 29 years) and by a predominance of females. In approximately 50-75% patients in this group, renal failure is the first manifestation of TS. Renal impairment is the second commonest cause of death in TS after neurological involvement. Renal failure in TS is uncommon possibly owing to early death from neurological complications of the disease. Renal failure in TS is because of nephron reduction due either to replacement by cysts or angiomylipomas or to nephrectomy. Nephrectomies done before the onset of dialysis are an important cause of renal deterioration, so angiomylipomas in these patients should be treated as conservatively as possible. Rarely patient may develop chronic renal failure due to bilateral kidney rupture. The cause of renal failure in our patient was presence of multiple cysts.

The prevalence of malignancy in TS is 4% which is higher than general population. Some studies have reported a higher prevalence of 13.8%. Because of this risk of hidden malignancies, which might flare up on
immunosuppression, some authors have recommended bilateral nephrectomy before or at the time of renal transplantation.\textsuperscript{1,9} If the native kidneys are not removed at the time of renal transplantation, they should be checked yearly by CT scans and nephrectomy should be done at the slightest suspicion of malignancy.\textsuperscript{1,2} Because of increased risk of malignancy developing under immunosuppression, our patient underwent bilateral native nephrectomy on follow-up.

**REFERENCES**