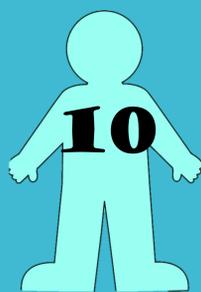


RENAL IMPAIRMENT IN OUR CHILDREN WITH TUBEROUS SCLEROSIS

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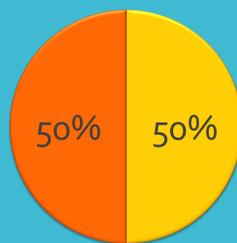
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Tuberous sclerosis is a neurocutaneous syndrome with heterogeneous clinical picture starting in childhood. It often presents with neurologic symptoms and cognitive impairment as well as with renal manifestations. The latter can begin in infancy and increase with age, involving cystic renal disease and angiomyolipomas. The aim of our study was to determine the extent of renal impairment and other clinical characteristics as well as treatment of children with tuberous sclerosis, followed-up in our department.



Median age

13.5 years



Boys Girls

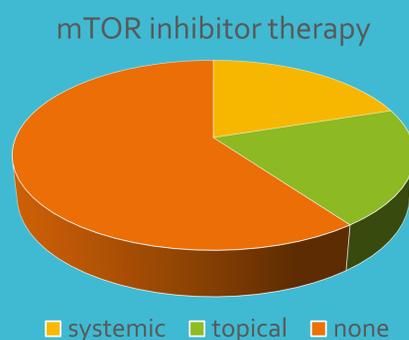
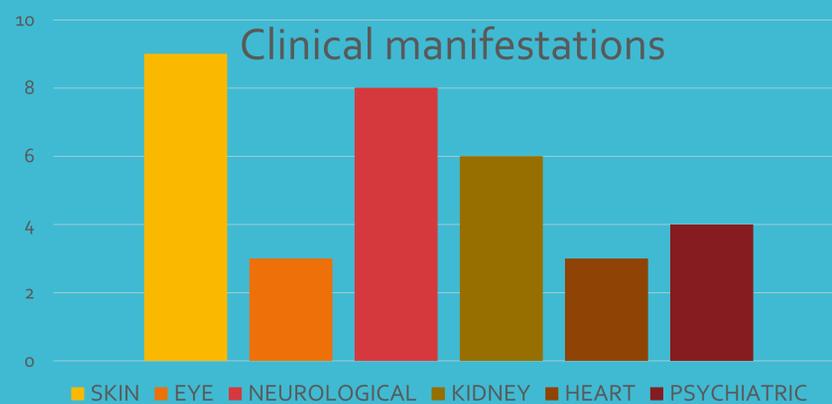
Median age at diagnosis

1 year

RESULTS

Renal manifestations, mostly angiomyolipomas, were noticed in six of our patients. In 2 patients renal impairment was found to be substantial. However, at the moment decreased renal function is present in only one. In two thirds of patients skin pathology was found, both cardiac rhabdomyomas and retinal hamartomas were present in 3 and brain lesions in 7 patients. In 7 patients epileptic seizures and in 3 cognitive disorders were diagnosed, with 4 and 2 of them necessitating antiepileptic or psychiatric therapy, respectively. In 2 patients systemic everolimus was used and in one of them, embolization due to renal haemorrhage was performed. In 2 patients topical everolimus for skin lesions was also prescribed. Until now, genetic analysis was performed in 7 patients and was positive in 5 and negative in 2 of them.

Ten patients with tuberous sclerosis (median age of 13.5 years) were included in our study. Their history, clinical characteristics, including renal manifestations, and treatment were highlighted. In addition, the results of genetic analysis were checked.

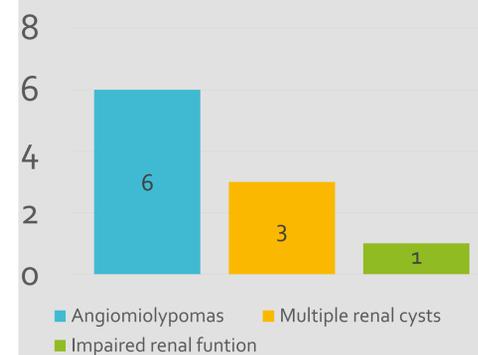


GENETIC TESTING

positive negative not done



Renal manifestations



CONCLUSIONS Our results confirmed that renal impairment in tuberous sclerosis patients is often present. However, in most of the paediatric patients renal function is stable and the treatment is rarely needed.