TUBEROUS SCLEROSIS COMPLEX AND THE KIDNEY: A SINGLE CENTER 17-YEAR PORTRAIT

Rita Leal, Telma Santos, Ana Galvão, Rui Alves
Serviço de Nefrologia, Centro Hospitalar e Universitário de Coimbra, Clínica Universitária de Nefrologia da Faculdade de Medicina da Universidade de Coimbra

54TH ERA-EDTA CONGRESS Madrid, Spain June 3rd-6th 2017 www.era-edta2017.org

INTRODUCTION

- Tuberous sclerosis complex (TSC) is a rare, autosomal dominant disorder caused by mutations in TSC1/TSC2 genes
- It is characterized by tumor lesions in multiple organs, neurological abnormalities and dermatological features
- Renal features → angiomyolipomas and renal cysts → hemorrhage/excessive growth→ impaired renal function

AIMS

Characterize and evaluate the prevalence of renal involvement in patients with TSC at a central hospital over a 17-year period

POPULATION AND METHODS

Retrospective, observational, one-center study – 1st Jan 2000 – 31st Dec 2016

All patients with TSC diagnosis (ICD 9: 759-5)

Clinical features | Genetic tests

Demographic data | Clinical features | Renal involvement | Renal/patient outcome

Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International TSC Consensus Conference

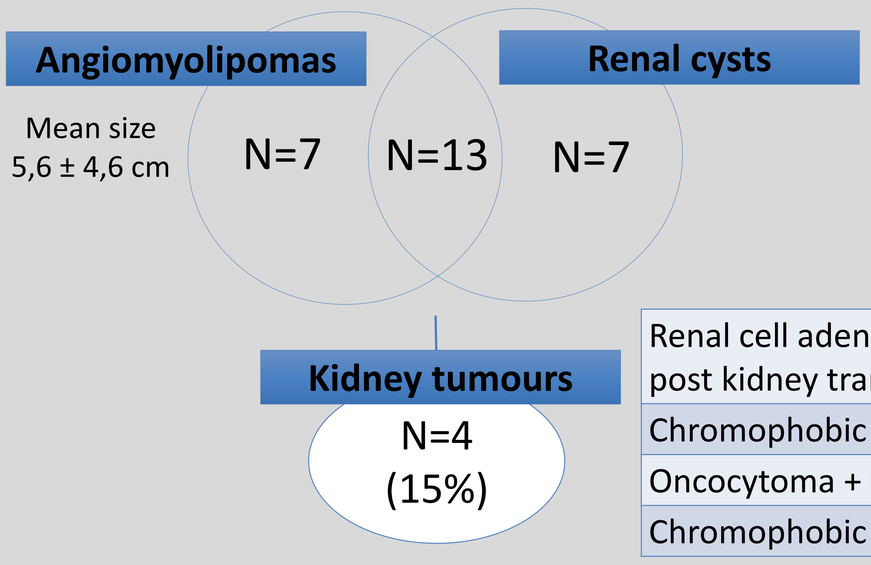
RESULTS

17 years → 37 patients included

DEMOGRAPHICS			
Sex	Female N=19 (51%)	Male N=18 (49%)	
Age (mean± sd [min-max])	39,6 ± 15,9 [16-82] years		
Age at diagnosis	22,7 ± 17,9 [1-62] years		
Genetic testing	10 patients	**TSC2** N=9	Waiting N=1

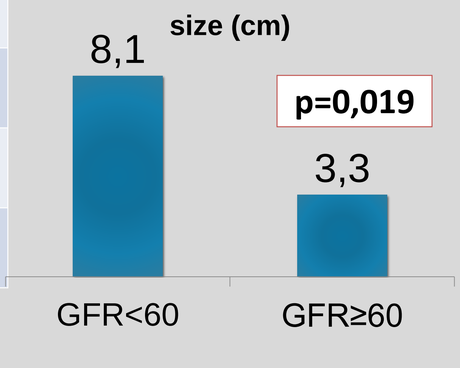
NON- RENAL CLINICAL FEATURES		
Skin lesions	N=35	95%
Neurologic tumors	N=31	84%
Epilepsy	N=19	51%
Behavioral anomalies	N=18	49%
Rhabdmyoma	N=4	11%
Lymphangioleiomyomatosis	N=4	11%
Ophthalmic lesions	N=3	8%
Hepatic cysts	N=2	5%

27 patients with renal involvement – 73%

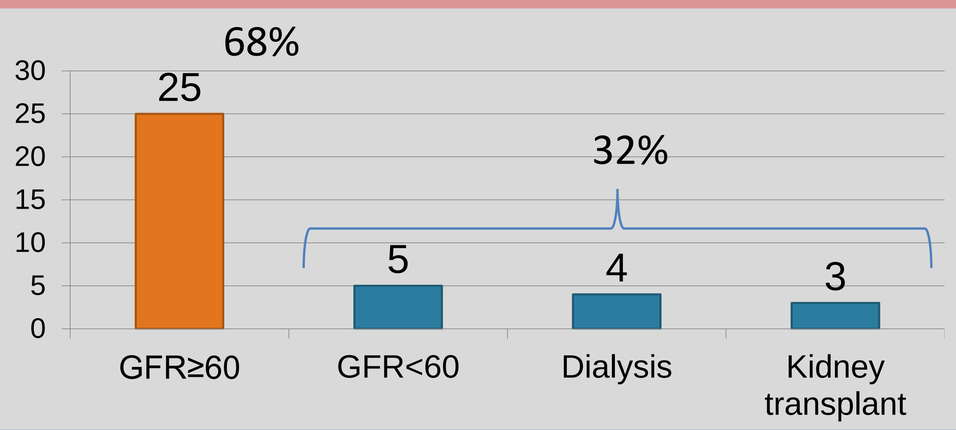


Renal cell adenocarcinoma (RCA) post kidney transplant
Chromophobic carcinoma
Oncocytoma + RCA
Chromophobic carcinoma + RCA

ANGIOMYOLIPOMAS: COMPLICATIONS	
Bleeding	**N=8 (40%)**
Conservative	N=4
Selective angiography	N=2
Nephrectomy	N=2



Patient outcome



- 5 patients on everolimus with staibilization of brain tumour size
- No data on renal outcome

- 3 deaths
  - Hemorrhagic stroke
  - Refractory epilepsy
  - Cardiac arrest - rhabdomyoma

CONCLUSIONS

- Renal involvement in TSC was frequent (73%), of which 44% evolved to renal function impairment
- The severity of the renal disease appears to be associated with the size of tumor lesions
- In our serie, six patients would benefit from mTOR inhibitors, according to TSC 2012 Consensus Conference;

DOI: 10.3252/pso.eu.54ERA.2017
ePosters supported by F. Hoffmann- La Roche Ltd.