

EVEROLIMUS IN TUBEROUS SCLEROSIS COMPLEX TREATMENT

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OBJECTIVES

Tuberous Sclerosis Complex (TSC) is an autosomal dominant disease with variable expressivity and multisystem involvement. Everolimus, mTOR inhibitor, is indicated for the treatment of kidney angiomyolipoma and subependymal giant cell astrocytoma (SEGA) associated with TSC.

The objectives of the study are to evaluate the effectiveness and safety of the treatment in TSC

STUDY DESIGN

Retrospective observational study of patients treated with everolimus from July 2013 to April 2014.

PATIENTS	Sex, age, affected organs, dose, duration and reason for treatment
EFFECTIVENESS	Reduction in size of SEGA equal to or greater than 30%, reduction in size of the kidney angiomyolipomas in at least 25%, improvement of dyspnoea and/or absence of lung acute episodes
SAFETY	Adverse reactions

RESULTS

- ✓ 4 patients were included
- ✓ Follow up (median, range): 17(12-27) months
- ✓ All patients continue with the treatment

	Patient 1	Patient 2	Patient 3	Patient 4
Sex, age (years)	Female, 32	Female, 38	Male, 21	Female, 15
Affected organs	Skin, brain	Brain, skin, bones, heart, lungs	Skin, eyes and brain	Skin, heart, kidneys and brain
Dose	7,5 mg QD	7,5 mg QD	7,5 mg QD	10 mg QD
Reason for treatment	SEGA	Pulmonary lymphangiomyomatosis	SEGA	Kidney angiomyolipomas and SEGA
Effectiveness	No	Yes	Yes	No
Adverse reactions	Stomatitis grade 1	Non-infectious pneumonitis grade 2	Stomatitis grade 1 and mild microalbuminuria	No
Dose reduction	No	Yes → 5 mg QD	No	No

CONCLUSIONS

Everolimus is the only well-tolerated treatment for TSC, but its effectiveness is variable. In the cases where no response was observed, the lesions were stabilised. The number of patients is limited due to the low prevalence of this disease and to the restrictive criteria for initiating the everolimus treatment. More studies are needed to determine the optimal dose and duration of the treatment

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