Beneficial Effects of Everolimus on Autism and Attention-Deficit/Hyperactivity Disorder Symptoms in a Group of Patients with Tuberous Sclerosis Complex.

Kilincaslan A, Kok BE, Tekturk P, Yalcinkaya C, Ozkara C, Yapici Z.


Abstract

OBJECTIVES:
Such neuropsychiatric symptoms as autism spectrum disorders, attention-deficit/hyperactivity disorder (ADHD), intellectual disability, aggression, and epilepsy are very common in patients with tuberous sclerosis complex (TSC). Everolimus, a mammalian target of rapamycin (mTOR) inhibitor, is a recent and effective treatment for TSC patients with giant cell astrocytomas and renal angiomyolipoma, and it has been shown to have a potential to reduce tumor volume. However, there is a paucity of studies on the effects of everolimus on neuropsychiatric symptoms. The aim of the present study is to describe the effects of everolimus on emotional and behavioral symptoms and refractory epilepsy in a group of patients with TSC.

METHODS:
Four boys and two girls (median age 16.5; range 7.5-23 years) were included in the study. Information on the clinical and treatment characteristics of the patients was gathered from the medical records.

RESULTS:
Median everolimus dose was 10 mg/day (range 5-20 mg) and median time for follow-up was 17.5 (range 7-26) months. The drug was well tolerated with mild adverse effects, including stomatitis (three cases), increase in triglycerides and cholesterol (two cases), and constipation (one case). The adverse effects encountered during the course of treatment did not make it necessary to discontinue the drug or decrease its dose. All cases experienced very good to moderate response for controlling epileptic seizures. Besides, improvements in social contact, language, repetitive behavior, inattention, hyperactivity, and depression were observed in some patients.

CONCLUSIONS:
Everolimus was well tolerated without severe adverse effects. It was helpful in controlling seizures and additional improvements were noted in autistic, ADHD, and depressive symptoms.