

Double valvular dysfunction in a 4-year-old girl with tuberous sclerosis

Dear Editor,

Cardiac rhabdomyomas are encountered in about 50 % of patients with tuberous sclerosis (TS) and are related with cardiovascular complications¹. We report the case of a 4-year-old girl diagnosed with TS at the age of four months. Her initial cardiological evaluation at the age of six months revealed multiple cardiac rhabdomyomas in the left ventricle (just below the mitral and aortic valve, and in the apex of the ventricle) and at the point of superior vena cava drainage into the right atrium. At the age of 25 months, premature atrial contractions (PACs) were detected in the electrocardiogram. At the age of four years, after about two years without cardiological evaluation, the girl was admitted to our Department due to a new systolic rough murmur, which prompted further evaluation. Echocardiography revealed severe mitral and mild aortic regurgitation (Figure 1). The regurgitation jet in the mitral valve was laterally (left) deviated. Heart function was within normal ranges. The girl was discharged home with captopril and instructions for a regular follow-up. The mitral regurgitation was improved three months later, and after six months the girl (at five years of age) was in good general condition, receiving antiepileptic treatment due to refractory seizures.

Various cardiovascular complications of rhabdomyomas in children with TS have been reported (arrhythmias, cerebral thromboembolism, hemodynamic compromise)¹. Valvular disorders are rarely reported as isolated cardiac manifestations in these patients. However, mitral and pulmonary valve stenosis/regurgitation have been more frequently identified in children with coexistent cardiac rhabdomyomas^{2,3}. In cases of severe complications surgical interventions have good outcomes².

The electrically active tissue of atrial rhabdomyomas in the reported patient explains the presence of PACs. Rhabdomyomas adjacent to mitral and aortic valve constitute an apparent cause of their regurgitation through a potential continuous mechanical effect, leading to valvular cusps degradation. Additional potential causes of valvular regurgitation to be considered in this patient include valve endocarditis by *Mycobacterium tuberculosis* and rheumatic fever. However, Mantoux skin test was negative, while no other Jones criteria (apart from carditis) were met at the time of evaluation.

In conclusion, electrocardiogram and echocardiography should be regularly performed in children with TS, even when physical examination is normal.

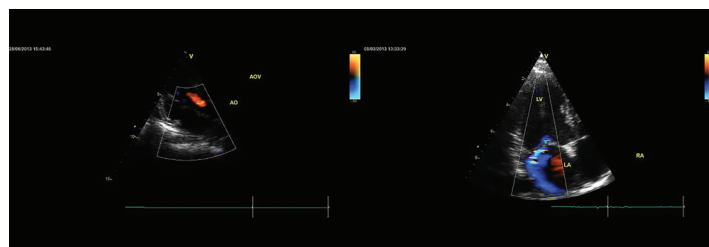


Figure 1: Mitral and aortic valve regurgitation in a 4-year-old girl with tuberous sclerosis.

LA: left atrium, LV: left ventricle, RA: right atrium, AOV: aortic valve.

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Conflict of interest

None.

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Gogou M, Keivanidou A, Giannopoulos A

2nd Department of Pediatrics, AHEPA General Hospital, Medical Faculty, Aristotle University of Thessaloniki, Greece

Corresponding author: Maria Gogou, 44 Dimitriou Nika, 60100, Katerini, Greece, tel: +306978905873, e-mail: mariaangogou@gmail.com