

Repeated rapid regression of two cardiac rhabdomyoma in a newborn with Everolimus

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INTRODUCTION

Cardiac rhabdomyoma (CRM) occur in 50% of patients with tuberous sclerosis complex (TSC). CRM are frequently asymptomatic and tend to regress spontaneously. However, some CRM cause arrhythmias, obstructions or valve movements disorders. Current treatment of choice in these patients is surgical resection. Recently encouraging experiences with mTOR-inhibitors such as Everolimus have been reported.

CASE DESCRIPTION

A giant left ventricle tumor was detected antenatally with ultrasound in a fetus. The tumor was considered to be a rhabdomyoma, the most common heart tumor in infants. The child was born on term and showed good primary postnatal adaptation. Two Ash-Leaf spots on the right forearm indicated the presence of TSC, which made the diagnosis of CRM even more likely. Prostaglandin E1 was administered from the first hour of life. Echocardiography after birth showed a large subaortal CRM (10x6x8mm) with subtotal obstruction of the LVOT (supravalvular gradient 70mmHg) and a second septal right ventricular CRM (22x13x10mm). Surgical therapy was rejected due to the high risk of aortic valve destruction. Thus, oral therapy with Everolimus with 0.03mg/kg/d and a target serum trough level of 5-8ug/l was initiated on the third day of life.

Echocardiography showed a volume regression of more than 80% in both CRMs within six weeks. There was also no longer any supravalvular gradient. Everolimus was therefore ceased 3 months after birth to evaluate whether further regression will occur spontaneously.

Four months later follow-up echocardiography revealed regrowth of both CRMs and increasing LVOT obstruction without clinical symptoms. Immediate restart of Everolimus resulted once again in rapid regression of both CRMs and LVOT obstruction was no longer measurable after four weeks. Regular follow-up echocardiographies confirmed continuing shrinkage of both CRMs even though they are still present at the age of 19 months. Due to TSC-associated intractable seizures Everolimus is still administered.

CONCLUSION

Beside established indications Everolimus may be a promising alternative for high-risk surgical cases with hemodynamically significant CRM. As shown in our patient, CRM can respond repeatedly which offers the opportunity of early treatment cessation awaiting spontaneous regression as well as avoiding therapy-associated side effects.