

Successful prenatal management of cardiac rhabdomyoma with mTOR pathway inhibitor

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ABSTRACT

Introduction: Cardiac rhabdomyoma (CR) is a rare benign tumour often associated with tuberous sclerosis complex, with an incidence reported 1 in 6,000 live births. In cases where CR lesions do not lead to severe complications, the pregnancy outcome is generally favourable. Complications associated with CR include arrhythmias, valvular regurgitation, outflow tract obstruction, heart failure, pericardial effusion, fetal oedema, and, rarely, stillbirth. This case report shares the successful prenatal treatment of fetal CR with sirolimus to prevent cardiac failure, which improved neonate outcome. **Case Description:** A case of a 36-year-old woman at 24 weeks in her first pregnancy, an ultrasound scan examination revealed CR in the left and right ventricles of the fetus. Serial scans revealed progression in the size of CR and impending cardiac failure. There was reduced contractility of the right and left ventricles. Ventricular filling was compromised due to obstruction by the tumour. Considering impending cardiac failure, therapy by sirolimus (mammalian target of rapamycin) was administered to the mother at 32 weeks of gestation. Pre-treatment screening of the mother's blood parameters was normal, and no contraindication for starting sirolimus. Mother was monitored for side effects and blood parameters, and she tolerated very well to the treatment. **Discussion:** Successful treatment of life-threatening CR in a fetus by administering sirolimus to the pregnant mother without exceeding 10 ng/ml of blood concentration. Significant reduction of the size rhabdomyoma improved the clinical outcome of the newborn. Sirolimus is a safe option for prenatal treatment in fetuses with CR.