In utero treatment of a large symptomatic rhabdomyoma with sirolimus

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Objective

- To describe the use of sirolimus for the treatment of a large, rapidly growing, fetal rhabdomyoma, and consequently ventricular dysfunction at 31 weeks gestation.

Case Report

- Healthy 27-year-old nulliparous woman
- Multiple fetal rhabdomyomas diagnosed and cerebral tubers on MRI
- Progressive growth of rhabdomyomas was noted on serial echocardiography (graph) resulting in poor cardiac function at 31 weeks gestation: poor biventricular contractility, severe tricuspid & mitral regurgitation and pericardial effusion
- Given the poor prognosis, the option of experimental treatment with sirolimus, an mTOR inhibitor in utero, was offered following multi-disciplinary counselling and informed consent.

Antenatal & Postnatal Course

- Transplacental treatment with sirolimus initiated at 31.6 weeks of gestation via maternal administration, aiming for maternal serum trough levels between 10-15ng/ml
- Weekly surveillance with fetal ultrasound and maternal bloodwork, to monitor immune, renal and hepatic function
- Significant reduction in size of largest mass within 4 weeks of treatment (graph), with improvement in ventricular function and resolution of tricuspid regurgitation
- Sirolimus discontinued at 36.1 weeks gestation to allow recovery of maternal immune system prior to delivery
- Male infant weighing 4,300g delivered at 39 weeks
- first postnatal ECHO: mildly reduced biventricular systolic function and a trivial pericardial effusion. Hemodynamically stable.
- Cytogenetics confirmed TSC2

Conclusion

- Sirolimus is a therapeutic option for treatment of symptomatic cardiac rhabdomyomas in utero
- Given limited safety data, this option should be reserved for cases with poor prognosis.

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