Long term outcome in 32 fetuses with cardiac rhabdomyomas

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Background: Rhabdomyomas (R) are more common cardiac tumors detectable in fetuses. Prenatal
counselling regards a possibility of association to tuberous sclerosis (TS). The aim of this study was
to analyse retrospectively the long term outcome of our cases.

Material and methods: Between 1987 and Dec. 2015 32 fetuses (0.63% of 5010 studied by
echocardiography) showed cardiac masses, suggestive of R, at 21-36 w.g.; they were followed-up in
utero and postnatally for a median period of 7 years (6m - 27 yrs).

Results: The diagnosis (dg) was made before 24 w.g. in 9 fetuses, 6 of them opted for the termination
of pregnancy (TP) having a postmortem histological dg. of R. Two fetuses diagnosed at 27 and 28 w.
with multiple large masses opted for TP (in another country), after positive MRI findings for TS.
The remaining fetuses were diagnosed at 29-36 w.g., had 2 or more larger masses with partial inflow
and/or outflow obstruction in half of them. Two fetuses had mothers with TS - one had a large
intrapericardial mass. One fetus had polycystic kidneys of adult type.

Growth in utero: Cardiac masses grew usually till 35-36 w.g. Diagnosis of TS was made in 18 cases
by positive CNS MRI findings of supendymal/cortical tuberi after birth- in 16 of 24 (67%) continuing
pregnancy and in 2 after the late TP; total percentage of 69%. Genetic analysis was performed up to
now in a third of cases.

Outcome: none of the live born cases needed cardiac surgery and the cardiac masses progressively
regressed over time, 8 cases have extrasystolic arrhythmia and one had a short episode of
supraventricular tachycardia. All TS cases have neurological signs- seizures, 3 were operated for
astrogliomas. One case is on dialysis, others have minor renal, dermatological and ocular signs.

Conclusions: Cardiac R show variable characteristics, postnatal regression of cardiac masses but a
relevant association with TS that conditions the clinical state.