Fetuses with Tetralogy of Fallot – important information from prenatal diagnosis

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Objective.
We analyzed prenatal history and perinatal outcome of fetuses with tetralogy of Fallot (ToF). The type of ToF, associated anomalies, karyotype, prenatal evolution and pregnancy outcome were analyzed.

Methods.
Prospective echocardiographic examinations were conducted in the referral center for fetal cardiology since 2002. We performed retrospective review of database and recorded exams for the period 2002–2012.

Results.
There were 99 fetuses (52 males, 45 females, 2 unknown) examined at average 28 weeks (median 28, 36% before 24 weeks). 68 fetuses had classic ToF, 24–ToF with pulmonary atresia (ToF-PA) and 4–ToF with absent pulmonary valve (ToF-APV). In 3 cases a transition from ToF to ToF-PA was observed. Karyotype was checked in 66 cases and was normal in 36 fetuses. Abnormalities included: T21–8 cases, T18–3, T13–3, Klinefelter-1, with 22q21 microdeletion, other–3, 22q21 microdeletion – 12 fetuses. In all DiGeorge cases thymus was small or invisible. This syndrome was detected in 2 out of 4 fetuses with TOF-APV and in 2 out of 3 transitional cases.

Associated anomalies within cardiovascular system included: 4 AVSD (2 in T21 fetuses), 4 I SVC, 13 right aortic arches, 6 aberrant origins of RSA, 19 MAPCAs. We did not find significant correlation between additional cardiovascular lesions and abnormal karyotype. DA was absent in all cases of ToF-APV.

Extracardiac malformations (ECM) were in 32 cases, IUGR – 28. Correlation between IUGR, karyotype and ECM was not significant.

Perinatal outcome was known in 93 cases (94%). There were 81 live births (41-VD, 37-CS), 6 stillbirths and 6 terminations (6.5% of all pregnancies, 20% of diagnosed before 24 week). 14 (17.9%) children were born pre-term. 15 (19.2%) had low body weight. Majority were born in good condition: Apgar score 8-10:56(74%); 5-7: 14(19%); had 1-4:5(7%).

Conclusions.
ToF was diagnosed after 24 weeks in 63% cases. ToF is an indication for detailed USG scan and karyotyping with 22q21 microdeletion exam, especially when thymus is absent. Right aortic arch, aberrant right subclavian artery or I SVC were not markers of abnormal karyotype. Termination rate was low, even in early recognized cases, and occurred more commonly in fetuses with genetic anomalies. Neonatal condition was good in 93% cases.