

Prenatal diagnosis of 51 fetuses with Aortic Arch Anomalies :Prevalence, associated findings and perinatal outcome in a single institution.

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Objective: To evaluate the incidence and associations of AAA in a population of fetuses referred for fetal echocardiography in our third level center (AORN dei colli – AO Monaldi, Second university of Naples). To report the pre and postnatal outcome of affected cases.

Methods and results: Since January 1995 to March 2015 we practiced 9048 fetal heart scans. Among these, we diagnosed 1281 (14%) consecutive fetuses with CHD. An AAA was diagnosed in 51/9048 (0.6%) fetuses (51/1281 CHD; 4%). Mean gestational age at diagnosis was 24.4 +/- 2.4 weeks. The detection rate of a AAA increased over the study period: 41/51 (80%) cases were diagnosed from 2007 to 2015; this finding is in accord with literature.

There were 51 (42 %) AAA, 47 (92%) of which were RAA (right aortic arc) , 2 (4%) of which were ARSA (right subclavian artery aberrant) and 2 (4%) were DAA (Double aortic arch).

There were 18 (38 %) isolated RAA. A RAA was found in association with ct-CHD in 29 cases (62 %): 20 Tetralogy of Fallot, 5 Pulmonary atresia with ventricular septal defect , 3 double outlet right ventricle with pulmonary stenosis and 1 ventricular septal defect .

Karyotype was performed during fetal life in 28/51 AAA (55%), 10/28 (55%) were pathological.

Of the chromosomal abnormalities : 7 /10 (70%) were 22q11 deletions ,in 2 of the 7 cases (29%) RAA was isolated and 5/7(71%) RAA was associated with CHD .2/10 (20%) were trisomy 21, of which one was isolated ARSA and one TOF with RAA, 1/10 (10%) was trisomy 18.

There were 9 (18%) pregnancy interruptions, 1 (2%) intrauterine death, 33 (65%) live births, 4 (8%) neonatal deaths. In the remaining 4 (8%) pregnancies is still continuing.

Conclusions AAA are rare, but during the last years, we observed an increasing number of AAA because of the increased skill of fetal echocardiographers.

An AAA can be associated to ct-CHD and /or anomalies of the Karyotype in a significant proportion. This is the reason why, when diagnosed an AAA, the couple must be offered a genetic study