

P43: Congenital heart defects in isolated and syndromic context

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Introduction: Congenital heart defects (CHD) is defined as a coarse structural abnormality of the heart or intrathoracic large vessels and which has functional significance.

Objectives: The objectives of this study were to determine the percentage of CHD in fetal autopsies and its frequency in isolated and syndromic context evaluate the most frequent types of CHD and determine the impact of histological study in the diagnosis of cardiac structural abnormalities.

Materials and Methods: Retrospective study of 367 fetal autopsies performed on Centro de Genética Clínica. Evaluation of variables: maternal age; maternal pathology; spontaneous fetal death (SFD) or medical termination of pregnancy (MTP); fetal sex; gestational age (GA); presence of CHD isolated or associated syndrome and classification of the type of CHD.

In the statistical analysis was used the statistical function in Microsoft Office Excel 2013 and the chi-square test.

Results and Discussion: After the study it appeared that 85(23.2%) autopsies were diagnosed with the presence CHD, from these 8.45% occur in isolated and 14.71% in context syndromic. The most common CHD was ventricular septal defect (VSD), with 56 (33.0%) cases, and 12 (7.0%) were histologically diagnosed. MTP occurred in 65 (78.3%) cases and SFD in 18 (21.7%). It was verified that 48(56.5%) are female and 37 (43.5%) are male. In syndromic CHD the mean maternal age was 36 years and the GA was 17 weeks. The mother of the fetus exhibiting CHD, 2(0.5%) had, at least, one maternal pathology. The percentage of CHD obtained is similar to other studies, which refer to a percentage of 20%.The most common type has CHD was VSD with a percentage close to 30% reported by other authors.

Conclusion: The CHD appeared in 23% of fetal autopsies and is related to the presence of syndrome, the most frequently CHD was VSD and histological analysis was diagnosed with CHD in 7%.

References

1. Rocha IJG, Nogueira RMAN, Carriço ALC. Non-compacted myocardium and foetal left isomerism as a hydrops' aetiology. *Cardiology in the young* 2010 Mar;20(2):223-5.
2. Barness EG, Spicer DD. *Embryo and Fetal Pathology – Color Atlas With Ultrasound Correlation*. 1st ed. Cambridge: Cambridge University Press; 2004.
3. Hoffman JIE. Incidence of Congenital Heart Disease: II. Prenatal Incidence. *Pediatric Cardiology* 1995;16:155-65.