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Agenesis of the ductus venosus: Prenatal diagnosis and perinatal outcomes

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Objective

The aim of this study was to describe the prenatal diagnosis and perinatal outcomes in fetuses with agenesis of the ductus venosus (ADV) including shunting type, morphological and chromosome-associated pathology, and perinatal outcome.

Methods

Retrospective descriptive study of all cases with prenatal diagnosis of ADV, occurring between 2011 and 2018 in three different tertiary care hospitals. We reviewed patient data of all ADV cases. Prenatal diagnosis, intrahepatic or extrahepatic shunting type, aneuploidy association and perinatal outcomes are described. Continuous variables with abnormal distribution data are shown as mean and ranges. Categorical variables are shown as absolute value and proportion.

Results

Thirty-one cases of ADV were diagnosed during the study period. Mean maternal age was 27 years (range 16-40 years) and mean gestational age at diagnosis was 21 weeks (range 11-31 weeks). ADV was diagnosed at first trimester screening in fourteen patients (45%), ten of whom presented with increased nuchal translucency. Seventeen were diagnosed at the second trimester ultrasound, fourteen of whom had an associated major abnormality. Shunt was intrahepatic in 15 patients, extrahepatic in 14, while 2 were non-determined. In the extra hepatic group, eleven communicated directly to the inferior vena cava, two to the right atrium, and one to the iliac vein. Chromosome analysis was performed in 18 of 31 cases; 11 of the fetuses were aneuploid (9 had Turner syndrome and 2 had Down syndrome). Regarding perinatal results, of 31 patients 9 died, 8 were born without complications, 7 are living with a major defect, and 1 had severe fetal growth restriction. 3 are still pregnant and 3 were lost to follow-up.

Conclusion

ADV is a rare condition and it is associated with poor perinatal outcomes. It is highly associated with aneuploidies, especially with Turner syndrome. In our series the disease-free survival rate was only 30%.