

Agenesis of ductus venosus

Delgado Martínez A, Gonzales Gabarra R, Álvaro Navidad M, Gutiérrez Martínez M, García-Puente Mesa L
Hospital Universitario Fundación Jiménez Díaz, Madrid, Spain

Objective

To analyse ductus venosus anomalies and its association with chromosomal disorders, malformations, perinatal and postnatal outcome.

Methods

A descriptive retrospective study was carried out sampling 17 patients diagnosed with agenesis of ductus venosus by prenatal ultrasound during 2015-2019. Two patients have been excluded from the analysis: ongoing pregnancy patient diagnosed with ADV with intrahepatic drainage and right umbilical vein persistence, and a patient diagnosed with ADV with extrahepatic drainage in 12th week (demostrating normalcy in 20th week).

Results

In selected 15 patients diagnosed with ADV, 5/15 (33.3%) had extrahepatic drainage, 8/15 (53.4%) had intrahepatic drainage and in 2/15 patients (13.3%) kind of drainage could not be defined. In cases in which type of drainage could not be determined, Noonan syndrome was diagnosed. Both of them associated multiple malformations (hydrops, ventricular septal defect, clubfoot), and a legal termination of pregnancy was carried out. Agenesis of ductus venosus, extrahepatic drainage: 2/5 cases were associated with right umbilical vein persistence (40%), 1/5 cases with restricted intrauterine growth (20%), 1/5 patient (20%) with other cardiac malformations (myocardial hypertrophy and cardiomegaly), and 3/5 patients (60%) presented with multiple extracardiac malformations (single umbilical artery, inferior vena cava agenesis, retrognathia, hypertelorism, hepatic and intestinal calcifications). 1/5 case was diagnosed with Temple syndrome (20%). In 3/5 (60%) cases, a legal termination of pregnancy was carried out, and 2/5 patients (40%) completed pregnancy with livebirth: 1/5 with postnatal diagnostic of non-obstructive byventricular hypertrophy and 1/5 with cardiac malformations that spontaneously resolved (interatrial septal defect, tricuspid incompetence). Agenesis of ductus venosus, intrahepatic drainage: 1/8 case (12.5%) was found to have right umbilical vein persistence, 2/8 cases (25%) were small-for-gestational-age, and 2/8 (25%) cases had generalized hydrops. 1/8 case (12.5%) had Noonan syndrome and 1/8 (12.5%) associated duplication of maternal origin (2q24.3-q31.1) and another de novo mutation in gen PTPN11, which is associated with Noonan syndrome. 5/8 (62.5%) pregnancies were concluded with livebirth, 2/8 (25%) with legal termination of pregnancy and 1/8 (12.5%) with antepartum fetal death at 27th week. 2/8 (25%) cases were postnatally diagnosed with mild cardiac malformations that spontaneously solved (patent ductus arteriosus, interatrial septal defect).

Conclusion

ADV has better fetal prognosis if the drainage is intrahepatic and if it is not associated with other malformations or chromosomal disorders.

	Prenatal diagnosis	Karyotype	Associated anomalies	End of pregnancy	Postnatal diagnosis
1	Agenesis of DV	Noonan Syndrome	Hidropic fetus	Legal abortion	
2	Agenesis of DV	Normal	Hydrops, muscular VSD, clubfoot	Legal abortion	
3	Agenesis of DV, EH	Normal	-	False positive	
4	Agenesis of DV, EH	Temple Syndrome	-	Legal abortion	
5	Agenesis of DV, EH	Normal	IURG, pericardial effusion y intestinal and hepatic calcifications	Preterm cesarean section	Non-obstructive byventricular hypertrophy and hepatosplenomegaly
6	Agenesis of DV, EH	Normal	RUVV, UUA, agenesis ICV, retrognathia, hypertelorism	Legal abortion	
7	Agenesis of DV, EH	Normal	Cleft palate, absent corpus callosum, short "long bones"	Legal abortion	
8	Agenesis of DV, EH	Normal	Myocardial hypertrophy, cardiomegaly	Term cesarean section	Interatrial septal defect and tricuspid incompetence
9	Agenesis of DV, IH	Normal	-	Preterm birth	Interatrial septal defect and patent ductus arteriosus
10	Agenesis of DV, IH	Normal	Generalized hydrops	Fetal death	
11	Agenesis of DV, IH	Noonan Syndrome	Hypoplastic nasal bone, increased NT, hygroma	Legal abortion	
12	Agenesis of DV, IH	Normal	-	Term birth	Normal transthoracic echocardiogram
13	Agenesis of DV, IH	-	RUVV, small for GA	Term birth	
14	Agenesis of DV, IH	Normal	Small for GA	Term cesarean section	Interatrial septal defect and dysplastic pulmonary valve
15	Agenesis of DV, IH	Normal	-	Term birth	Normal transthoracic echocardiogram
16	Agenesis of DV, IH	46XX, Dup 2q24.3-q31.1 and PTPN11 de novo mut.	Increased NT, hygroma	Legal abortion	
17	Agenesis of DV, IH	-	RUVV	Ongoing pregnancy	