Persistent Right Umbilical Vein in Trisomy 18: Sonographic Observation

Luc De Catte, MD, Kaan Osmanagaoglu, MD, Inge De Schrijver, MS

Persistent right umbilical vein is a rare entity, with a prevalence of about 0.22%.¹ The intrahepatic variant is observed most frequently, and 72% of the fetuses are normal.^{2,3} In a very limited number of cases, the persistent right umbilical vein terminates directly into the right atrium, without passage into the liver. The majority of fetuses with this variety have multiple congenital malformations.²

We report a case of persistent right umbilical vein draining directly into the right atrium in association with trisomy 18. Although particular sonographic markers related to Edwards syndrome were present in our case, we would recommend fetal karyotyping in all cases of persistent right umbilical vein.

CASE REPORT

A 35 year old Turkish woman had had three uncomplicated pregnancies and deliveries. A fourth pregnancy ended in an unexplained fetal death at 6 months' gestation. The parents refused postmortem investigation. The woman's medical history was unremarkable. A routine second trimester ultrasonographic examination at 22 weeks of gestation showed a normal fetal biometry for gestational age, mild polyhydramnios, and the presence of a single umbilical artery. In addition, a choroid plexus cyst measuring 7×15 mm was observed in the left ventricle, and both ears were low set. The fetus had rocker bottom feet, and both hands were clenched. The lower lobes of the right lung were echogenic. The gallbladder was enlarged and had an abnormal transverse position close to the anterior abdominal wall (Fig. 1). The umbilical vein did not connect with portal circulation in the liver but coursed over the anterior abdominal wall through the anterior portion of the diaphragm directly into the right atrium (Figs. 2, 3). The ventricular outflow tracts were normal, and venous return in the right atrium by the inferior and superior venae cavae was intact. The diagnosis of persistent right umbilical vein draining directly into the right atrium in a fetus showing characteristics of trisomy 18 was made. Fetal blood sampling for fast karyotyping confirmed the presence of a female trisomy 18 fetus in all examined metaphases. Termination of pregnancy was performed.

Gross pathologic examination clearly revealed the presence of the clenched hands and the rocker bottom feet. The fetal abdomen was slightly distended; right and left liver lobes were of equal size. The enlarged gallbladder was

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Address correspondence and reprint requests to Luc De Catte, MD, Department of Feto-Maternal Medicine, University Hospital Brussels, Laarbeeklaan 101, 1090 Brussels, Belgium.

Figure 1 Transverse section at the level of the fetal liver shows the transversely oriented and enlarged gallbladder.

Figure 2 Longitudinal scan of the fetal abdomen reveals the course of the right umbilical vein (arrow) from the umbilicus to the heart, anterior to the liver. C, Fetal caudal end; H, fetal cranial end.

draped over the inferior surface of the liver from left to right. A small craniocaudal groove on the anterior surface of the liver guided the umbilical vein from its entrance at the umbilicus directly into the right atrium. No other vascular abnormalities were observed. Intestinal malrotation also was present.

DISCUSSION

Obliteration and disappearance of the right umbilical vein starts in the fourth week of embryonic development.^{2,4} The initial connection of both umbilical veins with the sinus venosus is lost when the umbilical veins anastomose with the hepatic sinusoids. Failure of this process results in the direct connection of both umbilical veins to the right atrium. The growth of the fetal liver then causes kinking of the right umbilical vein, subsequent occlusion, and regression. However, on rare occasions and for yet unknown reasons the left umbilical vein becomes obliterated. Since normal flow in the liver was already established, the ductus venosus does form, but the blood flow in the liver is abnormal.

The prevalence of persistent right umbilical vein, estimated through targeted prenatal fetal sonographic examinations, is now at 2.2 per 1000 births; this is not as rare as previously believed.^{1,3,4} Persistent right umbilical vein is diagnosed sonographically on the transverse fetal section obtained to measure the abdominal circumference.⁴ Normally, the left umbilical vein passes at the left side of the gallbladder and connects with the portal vein. It then curves right, away from the stomach. In the intrahepatic form of persistent right umbilical vein, the

umbilical vein passes lateral and to the right side of the gallbladder, fuses with the left portal vein, and then bends toward the stomach. Whenever the right umbilical vein does not connect with the portal system but enters the right atrium directly, as in our case, or enters the superior vena cava, the vein runs anteriorly over the liver. This form of persistent right umbilical vein has rarely been diagnosed prenatally. Color Doppler sonography facilitates the identification of the abnormal vessels and their course in cases of poor image resolution.⁴⁻⁶

The spectrum of sonographic, clinical, and autopsy findings in trisomy 18 syndrome has been reported extensively.^{7,8} In the present case, typical sonographic markers for trisomy 18 were noted: mild polyhydramnios, choroid plexus cyst, clenched

Figure 3 Transverse section of the fetal thorax at the level of the four-chamber view shows the entrance of the right umbilical vein (u) directly into the right atrium (a).

hands, and rocker bottom feet. Although at least 30 different cardiovascular malformations have been described in trisomy 18, a persistent right umbilical vein draining directly into the right atrium had not been observed previously.

Persistent right umbilical vein has been associated with a large variety of anomalies related to the intestinal, cardiovascular, urogenital, and skeletal systems, as reported by Shen and coworkers.¹ In the absence of malformations, isolated persistent right umbilical vein probably is of little significance to pregnancy outcome.^{1,3} Of the 74 fetuses (Table 1) with intrahepatic persistent right umbilical vein reported in the literature, 53 (72%) were normal. In

eight of the 74 fetuses more than one organ system was involved; two of these were diagnosed as representing Noonan syndrome. Severe central nervous system lesions have been reported in four cases, and cardiovascular malformations were found in six fetuses.^{1–6,9–13} Although Jeanty has reported the initial survival rate as low as 33%,⁴ recent compilations of cases show a better prognosis.^{2,6}

All but one of the published cases (n = 8) of persistent right umbilical vein connecting to the superior vena cava or the right atrium manifested at least one malformation commonly encountered in the Edwards syndrome^{2,10,12,14–16} (Table 2). The absence of one umbilical artery was the most consistent finding

Table 1: Intrahepatic Persistent Right Umbilical Vein and Associated Malformations

Organ System	Number	Congenital Malformations	Reference (Number)
Central Nervous System	4	Hydrocephaly	9
		Anencephaly	6
		Dandy-Walker, IUGR	2
		Meningocele, hydromelia, ventriculomegaly, Chiari II	2
Cardiovascular	6	Mitral atresia	3
		Mitral atresia, DORV, coarctation of the aorta	4
		Asplenia syndrome	6
		ASD	2
		VSD (glandular hypospadias)	2
		Dextrocardia	1
Urogenital	2	Kidney dysplasia, dysmorphic kidney	6
		Hypospadias	3
Multiple malformations	8	Noonan, SUA	3
		TAPVR, unilateral kidney agenesis, phocomelia, unicornuate uterus	10
		SUA, goiter, atrial septum aneurysm	4
		Nuchal fold, ascites, hypospadias, allantoic duct remnant	4
		Hypertrophic cardiomyopathy, hydrops, Noonan syndrome	3
		Multicystic kidney, VSD, hemivertebrae	6
		Caudal regression syndrome, absent fibula, clubfeet, SUA, aortic stenosis	6
		Truncus arteriosus, bilateral cleft lip, clubfeet, SUA, IUGR	6
			6
IUGR	1	—	4
Normal	53		11(1)
			4(2)
			6(27)
			3(8)
			5(2)
			12(1)
			13(1)
<i>Total</i>	<i>n</i> = 74		2(4)
			1(7)

IUGR, Intrauterine growth restriction; DORV, double outlet right ventricle; ASD, atrial septal defect; VSD, ventricular septal defect; SUA, single umbilical artery; TAPVR, Total anomalous pulmonary venous return.

Table 2: Persistent Right Umbilical Vein Draining into the Right Atrium: Associated Malformations and Outcome

Reference	Associated Malformations	Outcome	Prenatal Diagnosis
14	SUA	Stillbirth	-
10	SUA, esophageal atresia, tracheoesophageal fistula; annular pancreas; duodenal atresia; imperforate anus; accessory spleens	Full term pregnancy	-
10	SUA; situs inversus of the viscera; bilateral hydronephrosis; sirenomelia	Preterm delivery at 35 weeks	-
10	SUA; exstrophy of the heart, liver, spleen, stomach; tetralogy of Fallot	Preterm delivery 8 months	-
15	SUA; right-sided cardiac apex; intestinal malrotation; left-sided liver	Full term pregnancy	-
12	None	Full term pregnancy	+
16	SUA; agenesis of the right kidney; interruption of the intrahepatic inferior vena cava with hemiazygos continuation; bifid right thumb; hemivertebra	Delivery at 39 weeks	+
2	SUA; right hydronephrosis; hypertelorism; syndactyly; hemivertebra	Not available	+

SUA, Single umbilical artery.

in this group (seven of eight), and this finding was also present in our case. Although gastrointestinal, kidney, and skeletal malformations were present in several cases, no uniform pattern was recognized. The structural defects in our case reflected two different pathologic entities: those associated with the persistence and abnormal course of the right umbilical vein (enlarged symmetric liver, transverse position of the gallbladder) and those recognized as part of a trisomy 18 syndrome (choroid plexus cyst, clenched hands, rocker bottom feet, and polyhydramnios). Because in the past fetal or neonatal karyotyping in relation to persistent right umbilical vein had not been performed systematically, chromosomal abnormalities might have been underestimated.⁶

We report a case of persistent right umbilical vein with a chromosomal abnormality. The finding of an abnormal vascular connection of the umbilical vein to the fetal venous circulation in association with structural defects in multiple organ systems merits chromosomal analysis. Further study is needed to clarify any relationship of persistent right umbilical vein and chromosomal defects or to deem this observation purely incidental.

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