# The Association of Fetal Umbilical–portal–systemic Venous Shunts and Genetic Variants: Another Case Report

**Li Zhen, Dong-Zhi Li***

Prenatal Diagnostic Center, Guangzhou Women and Children's Medical Center Affiliated to Guangzhou Medical University, Guangzhou, Guangdong, China



Dear Editor,

Congenital portosystemic venous shunts are vascular anomalies with communications between the portal venous system and the systemic venous circulation. They have been recognized as an important pediatric condition, but these vascular anomalies have been reclassified as umbilical–portal–systemic venous shunts (UPSVSs) for fetal cases, taking into account the unique anatomy *in utero*.[1] Indeed, the umbilical, portal, and ductus venosus all can produce a shunt with systemic veins. Here, we report a fetal UPSVS case which is associated with a *GATA4* variant.

A 28-year-old woman, G2P0A1, was referred to our center for further evaluation because of abnormal fetal abdominal vascular anomaly at 20 weeks' gestation. She conceived naturally and had a nuchal translucency of 7.2 mm at 12 weeks' gestation. Chorionic villus sampling reported a normal microarray result. Her first pregnancy ended with fetal demise at 20 weeks with no known causes. Both partners were healthy and nonconsanguineous. At this referral, a detailed ultrasound showed an appropriate-for-gestational-age female fetus with normal amniotic fluid volume and placental morphology. However, the umbilical vein-left portal vein-ductus venous complex was not detected in the fetal liver, and the intrahepatic portal vein was unclear on ultrasonography. Instead, the umbilical vein entered into the abdomen and ran on the liver surface and directly connected with the azygos vein, and through the superior vena cava to the right atrium [Figure 1]. Fetal echocardiogram showed a normal heart with persistent left superior vena cava. An umbilical-systemic shunt was the prospective diagnosis. Trio exome sequencing was required, and this test detected a *de novo* likely pathogenic *GATA4* (NM_001308093.3) variant c.884C>A (p.Ala295Asp) in heterozygosity. Considering the clinical and genetic findings of the fetus, the parents decided pregnancy termination.

Previous studies indicated that prenatal cases with ductus venosus-systemic shunts or intrahepatic portal-systemic shunts have a high risk of aneuploidy, mainly trisomy 21 and 18. However, common trisomies would seldom be identified in fetuses with UPSVS in late pregnancy in the modern era with perfect first-trimester aneuploidy screening approaches. Variants in the *GATA4* gene have been associated with congenital heart malformations and neonatal- or childhood-onset diabetes.[2] We first report the association of UPSVS with a *GATA4* variant. Although rare, these vascular shunts can be a part of presentations in some genetic syndromes. For example, they have been reported in cardiofaciocutaneous syndrome, Noonan syndrome, and Costello syndrome, together belonging to RASopathies.[3] Other syndromic associations include neurofibromatosis, Cornelia de Lange syndrome, Holt–Oram syndrome, Adams–Oliver syndrome, and Rendu–Osler syndrome.[4,5] In terms of postnatal course, the umbilical-systemic shunt had a worse prognosis when characterized by an absence of or abnormal intrahepatic portal venous system, or the presence of heart enlargement, heart failure, or even fetal hydrops due to increased right atrial volume load in the earlier stage of gestation. In most cases, however, UPSVS is an isolated feature which is treatable and has a favorable outcome if affected fetuses manifest no heart enlargement. Our study further indicates that parents should be alerted to the possibility of a genetic condition in the cases of prenatal UPSVS, in addition to focusing on the shunting consequences.

## Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent form. In the form the patient has given her consent for her images and other clinical information to be

**Address for correspondence:** Dr. Dong-Zhi Li, Guangzhou Women and Children's Medical Center, Guangzhou, China.
E-mail: drlidongzhi2014@sina.com

**Received:** 09-10-2023 **Accepted:** 23-10-2023 **Available Online:** 15-02-2024

Access this article online

Quick Response Code:

Website: https://journals.lww.com/jmut

DOI: 10.4103/jmu.jmu_126_23



**How to cite this article:** Zhen L, Li DZ. The association of fetal umbilical–portal–systemic venous shunts and genetic variants: Another case report. J Med Ultrasound 2024;32:186-7.

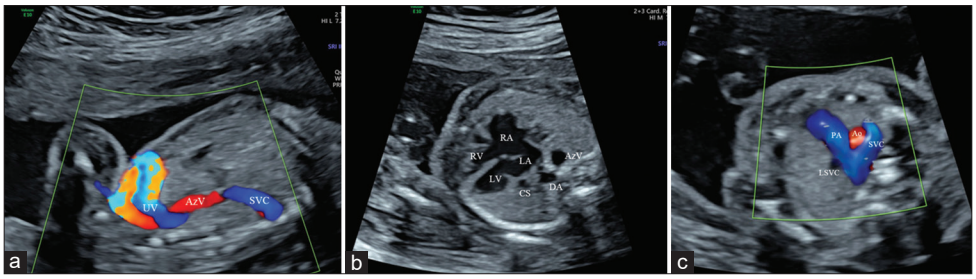


**Figure 1:** A prenatal case of umbilical-systemic shunt. (a) An umbilical–systemic shunt with direct connection between UV and AzV; (b) Four-chamber view with dilated CS. (c) Three-vessel view with persistent left superior vena cava. Ao: Aorta; DA: Descending aorta; LA: Left atrium; LV: Left ventricle; PA: Pulmonary artery; RA: Right atrium; RV: Right ventricle; SVC: Superior vena cava; UV: Umbilical vein; AzV: azygos vein; CS: Coronary sinus; LSVC: Left SVC

reported in the journal. The patient understands that her name and initials will not be published and due efforts will be made to conceal the identity, but anonymity cannot be guaranteed.

## Financial support and sponsorship

Nil.

## Conflicts of interest

There are no conflicts of interest.

## References

1. Achiron R, Kivilevitch Z. Fetal umbilical-portal-systemic venous shunt: *In-utero* classification and clinical significance. Ultrasound Obstet Gynecol 2016;47:739-47.
2. Shaw-Smith C, De Franco E, Lango Allen H, Batlle M, Flanagan SE, Borowiec M, *et al. GATA4* mutations are a cause of neonatal and childhood-onset diabetes. Diabetes 2014;63:2888-94.
3. Zhen L, Han J, Li DZ. Congenital extrahepatic portosystemic shunt: An unusual feature in cardio-facio-cutaneous syndrome. Eur J Obstet Gynecol Reprod Biol 2022;273:107-8.
4. Digilio MC, Capolino R, Marino B, Sarkozy A, Dallapiccola B. Congenital intrahepatic portosystemic venous shunt: An unusual feature in LEOPARD syndrome and in neurofibromatosis type 1. Am J Med Genet A 2005;134:457-8.
5. Pérez-García C, Martín YR, Del Hoyo AA, Rodríguez CM, Domínguez MC. Adams-Oliver syndrome with unusual central nervous system findings and an extrahepatic portosystemic shunt. Pediatr Rep 2017;9:7211.