Research Square

Preprints are preliminary reports that have not undergone peer review.
They should not be considered conclusive, used to inform clinical practice,
or referenced by the media as validated information.

A complex tetralogy of Fallot case with an unusual cluster of malformations diagnosed by multiple prenatal ultrasound views

Yun Wu (861945636@qq.com)
Chengdu University of Technology https://orcid.org/0000-0002-6343-3036
Liu-ying Zhou
chengdu women's and children's hospital

Case report

Keywords: Complex tetralogy of Fallot, Right aortic arch with mirror image branching, Left ductal arch, Left innominate artery, Pulmonary artery atresia

Posted Date: July 16th, 2020

DOI: https://doi.org/10.21203/rs.3.rs-41676/v1

License: This work is licensed under a Creative Commons Attribution 4.0 International License.
Read Full License

Abstract

Background Tetralogy of Fallot (TOF) is a complex congenital anomaly with a wide variety of prenatal presentations beyond the core malformations, creating difficulty in prenatal diagnosis, prognosis, and perinatal management. Indeed, TOF should be considered the ‘complex’ of Fallot.

Case presentation Through a series of precise prenatal ultrasound views, we describe a rare case of complex TOF with multiple malformations including main pulmonary artery atresia, left ductus arteriosus connecting subpulmonary arteries to the left innominate artery, and a right aortic arch with mirror image branching. The prenatal diagnosis and all ultrasound findings were confirmed by autopsy.

Conclusions Tetralogy of Fallot (TOF) is a complex congenital anomaly with various branches of blood vessels. A set of standard views plus additional views (the upper chest coronal view) are required for prenatal diagnosis of complex TOF.

Introduction

Complex tetralogy of Fallot (TOF) is classified into three main types, TOF with pulmonary artery atresia (PA), TOF with absence of pulmonary valve, and TOF with atrioventricular septal defect [1–3]. However, complex TOF is associated with many additional vascular malformations aside from these core features[3]. Some of these associated malformations are extremely rare, such as abnormal connections to the ductus arteriosus. We describe a rare case of complex TOF (pulmonary atresia type) with PA, left ductus arteriosus connecting subpulmonary arteries to the left innominate artery, and a right aortic arch with mirror image branching, revealed by a precise sequence of prenatal ultrasound views.

Case Report

General information

A 41-year-old pregnant female (26^{+1} weeks gestational age) who was in good health and had no personal or family history of birth malformations or genetic disorders received an X-ray examination for a cold during early pregnancy. Anatomical structural screening of the fetus revealed cardiac structural abnormalities but no extracardiac abnormalities. Detailed fetal echocardiography was then performed with a GE 10 Ultrasound (Tiefenbach, Austria) equipped with a 2 – 7 MHz convex probe (GE).

Ultrasonographic findings

Four-chamber views revealed a 0.59-cm discontinuity of the interventricular septum and color Doppler flow imaging (CDFI) demonstrated a bidirectional shunt at this ventricular septum defect.

A view of left ventricular outflow showed that the ventricular septal defect was of the perimembranous type. The dilated aorta overrode the ventricular septal defect by 50% (Fig. 1). A right ventricular outflow view was not acquired.

Three-vessel and three-vessel tracheal views could not visualize the main pulmonary artery but did reveal a confluence of dilated right and left pulmonary arteries. The ductus arteriosus also was not found at the normal anatomical location. The aortic arch was positioned to the right of the trachea. The first branch of the aortic arch was the dilated left innominate artery running left and anterior to the trachea. The second branch was the right common carotid artery, following by the right subclavian artery. On CDFI, reverse bidirectional blood flow was detected in the subpulmonary arteries (Fig. 2).

The upper chest coronal view showed a dilated vessel to the left and anterior to the trachea originating from the subpulmonary arteries and further connected to the dilated left innominate artery. Pulse wave Doppler (PW) demonstrated that this dilated artery was the ductus arteriosus and CDFI revealed reversed blood flow in the ductal arch (Fig. 3).

The final ultrasound diagnosis was complex TOF with main pulmonary atresia, right aortic arch with mirror image branching, and left ductus arteriosus between the subpulmonary arteries and left innominate artery.

All ultrasound findings were confirmed by autopsy (Fig. 4).

Discussion

Asymmetric fusion of the artery bulbar and truncal ridges resulting in anterosuperior displacement of the outlet septum generates tetralogy of Fallot [2]. The anterosuperior deviation of the outlet septum relative to the rest of the ventricular septum causes an anterior misalignment type of ventricular septal defect, pulmonary obstruction, overriding or dextroposition of the aorta, and right ventricular hypertrophy [7]. In addition to the main features, TOF is associated with other structural abnormalities [3], such as a right aortic arch [5], a deviated cardiac axis, an aberrant subclavian artery, and an absent ductus arteriosus [7].

Pulmonary atresia (PA) type accounts for 30% of TOF cases. In pulmonary atresia, the subpulmonary artery is reperfused by the ductal arch or by the somatopulmonary collateral from the descending aorta. This case of TOF with PA featured a right aortic arch with left ductus arteriosus connected between the dilated left innominate artery and subpulmonary arteries. A recent study reported prenatal TOF detection rates of only 30–60% [1], and differentiation of TOF subtypes with unusual associated malformations is even more challenging [1,7]. Ignorance of this potential connection between dilated left innominate artery and subpulmonary arteries via left ductus arteriosus can lead to misdiagnosed as absent ductus arteriosus.

In the current case, we suspected that the ductus arteriosus may exist but with abnormal connectivity based on detection of dilated subpulmonary arteries with reversed bidirectional blood flow. Furthermore, a right aortic arch with mirror image branching is usually associated with left ductus arteriosus connected to the descending aorta or the left innominate artery. Subsequently, we found that the dilated left innominate artery originated at the aorta arch, providing a rationale to scan the upper chest in the coronal

plane to look for the linkage between dilated left innominate artery and subpulmonary arteries. Finally, the ductal arch connected between the dilated left innominate artery and subpulmonary arteries was detected, thus avoiding prenatal misdiagnosis as absent ductus arteriosus, and the pulse wave Doppler (PW) demonstrated the expected flow pattern. The correct structural assessment of the aortic arch, the branches of the aortic arch, ductus arteriosus, and pulmonary artery by prenatal ultrasound could aid in fetal prognosis of complex TOF [4].

According to the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) guidelines, a set of standard views plus additional views (the upper chest coronal view) are required for diagnosis of complex TOF. Complete acquisition of these standard views combined with careful analysis could reduce the risk of prenatal misdiagnosis and be useful for pregnancy counseling.

Declarations

Ethical Approval and Consent to participate

Prenatal ultrasound screening was conducted according to the Maternal and Infant Health Care Law of China. Written informed consent was obtained from all pregnant women before prenatal ultrasound screening. The study was approved by the Ethics Committee of Chengdu Women's and Children's Central Hospital.

Consent for publication

Above all consents could be published.

Availability of data and materials

All data and materials were available.

Competing interests: None.

Funding: This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Authors' contributions

Yun Wu is the first author who wrote the case report and analyzed the case.

Liuying Zhou is the co-first author provided case pictures and analyzed the case.

Acknowledgements

Thanks to the Affiliated Hospital, School of Medicine, UESTC Chengdu Women's and Children's Central Hospital l for supports.

References

1. Reeves C. Obstetric ultrasound in tetralogy of Fallot. Sonography. 2019;6:56–64.
2. Dabbagh A, Conte H, Lubin L. Congenital heart disease in pediatric and adult patients: Anesthetic and perioperative management. Switzerland: Springer Nture; 2017.
3. Guerra FA, Quiroz L, Rodriguez JG, Enriquez G, Villalobos R, Leon A, Zuniga L. Complex of Fallot is a more appropriate name for tetralogy of Fallot ? Ultrasound in Obstet Gynecol. 2009;34(Suppl. 1):62–176.
4. Tuo G, Volpe P, Buffi D, De Robertis V, Marasini M. Assessment of the ductus arteriosus in fetuses with tetralogy of Fallot and the implication for postnatal management. Congenit Heart Dis. 2014;9:382–90.
5. Poon LC, Huggon IC, Zidere V, Allan LD. Tetralogy of Fallot in the fetus in the current era. Ultrasound Obstet Gynecol. 2007;29:625–7.
6. Yoo S-J, Lee Y-H, Kim ES, Ryu HM, Kim MY, Yang JH, Chun YK, Hong SR. Tetralogy of Fallot in the fetus: findings at targeted sonography. Ultrasound Obstet Gynecol. 1999;14:29–37.
7. Zhao Y, Abuhamad A, Fleenor J, Guo Y, Zhang W, Cao D, Zeng S, Sinkovskaya E, Zhou Q. Prenatal and postnatal survival of fetal tetralogy of Fallot. Journal Ultrasound M. 2016;35:905–15.

Figures

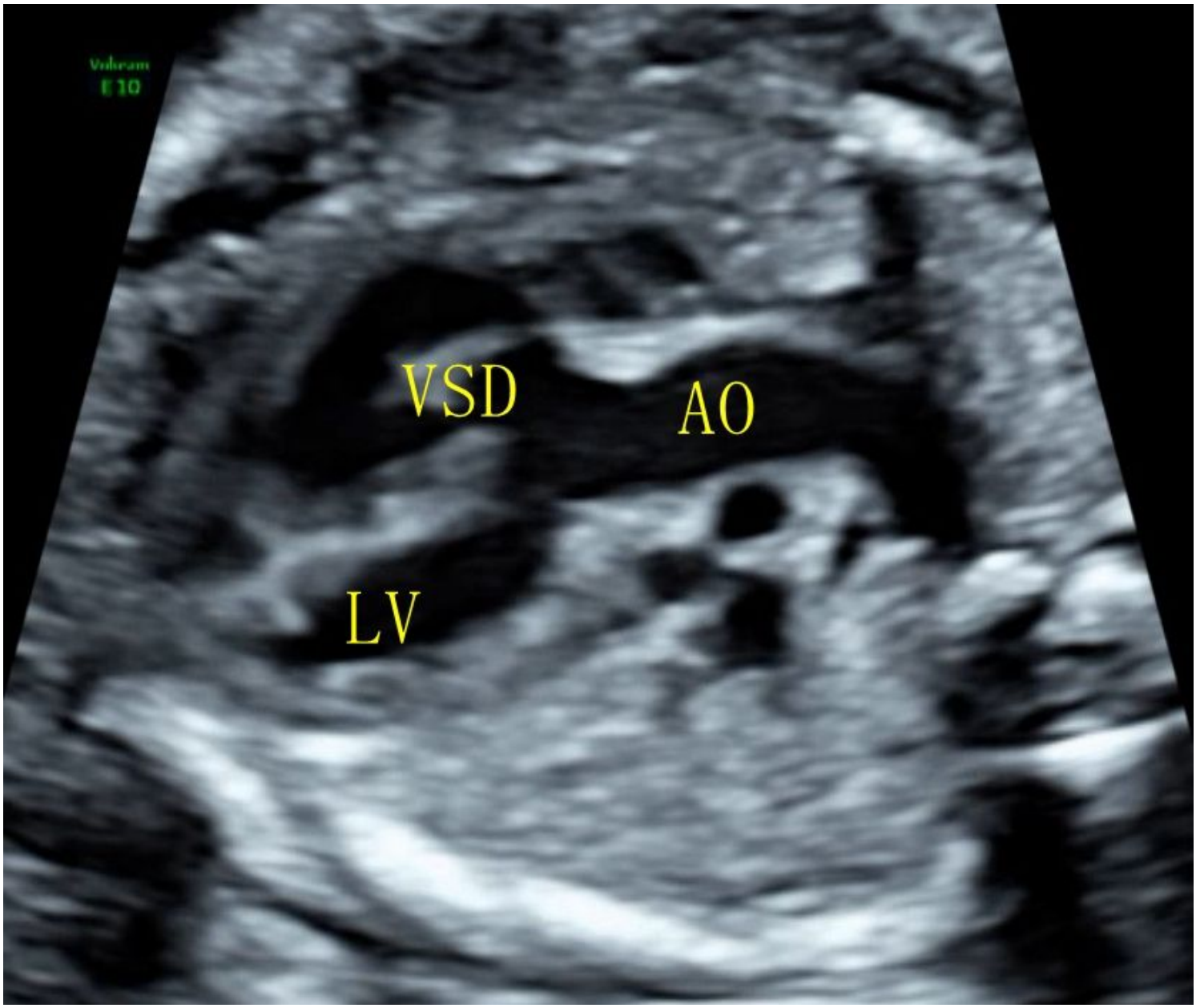

Figure 1

The dilated aorta (AO) overrode the ventricular septal defect (VSD), Left ventricular (LV).

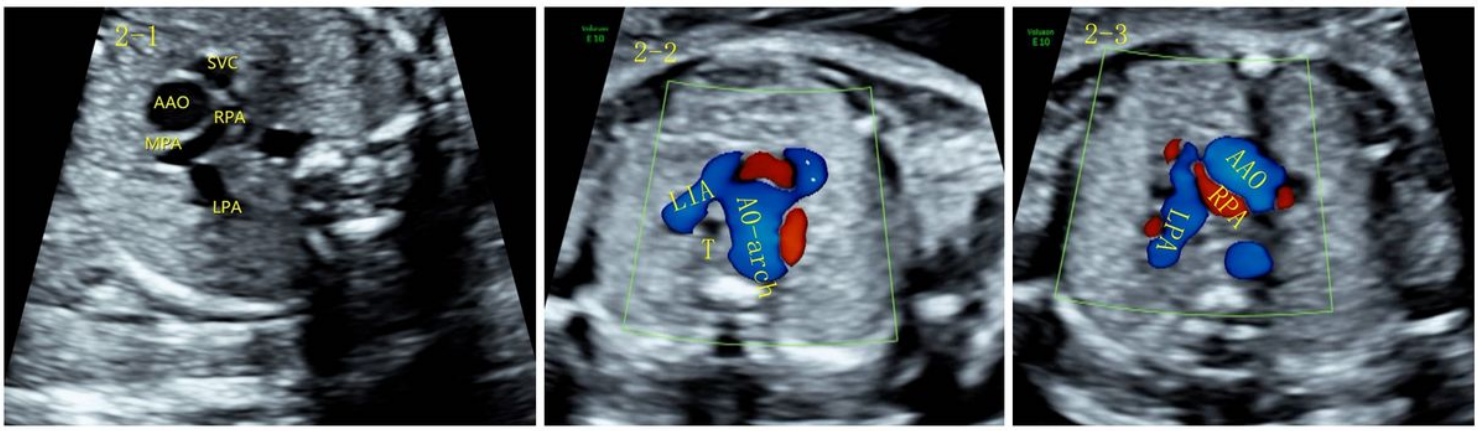

Figure 2

Three-vessel and three-vessel tracheal views: main pulmonary artery (MPA), right pulmonary artery (RPA), left pulmonary artery (LPA), ascending aorta (AAO), the aortic arch (AO-arch), left innominate artery (LIA), trachea (T).

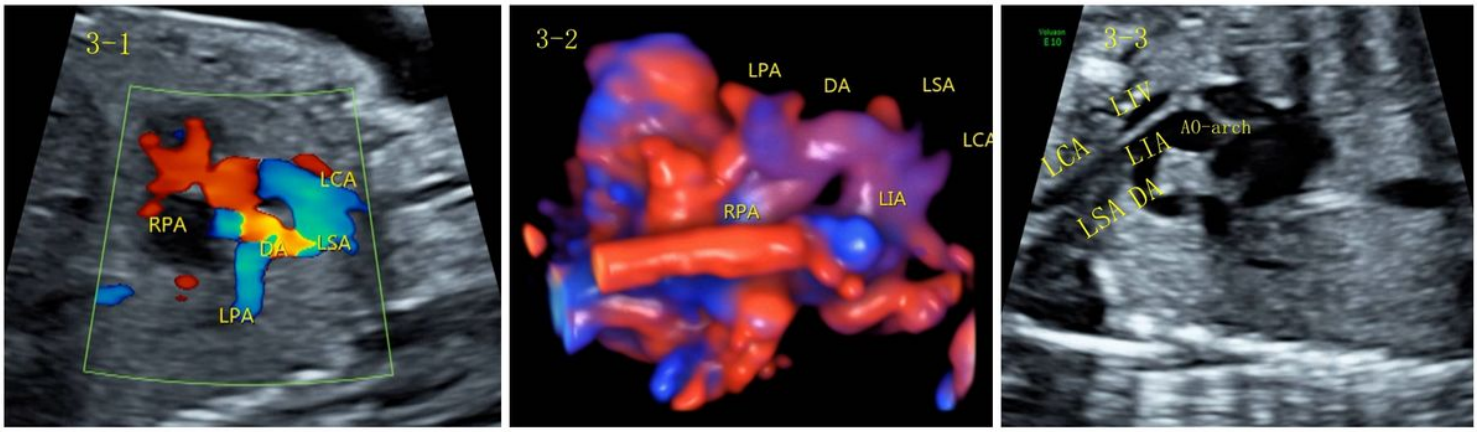

Figure 3

The upper chest coronal view: ductus arteriosus (DA), left common carotid artery (LCA), left subclavian artery (LSA), left pulmonary artery (LPA), right pulmonary artery (RPA), left innominate vein (LIV), left innominate artery (LIA).

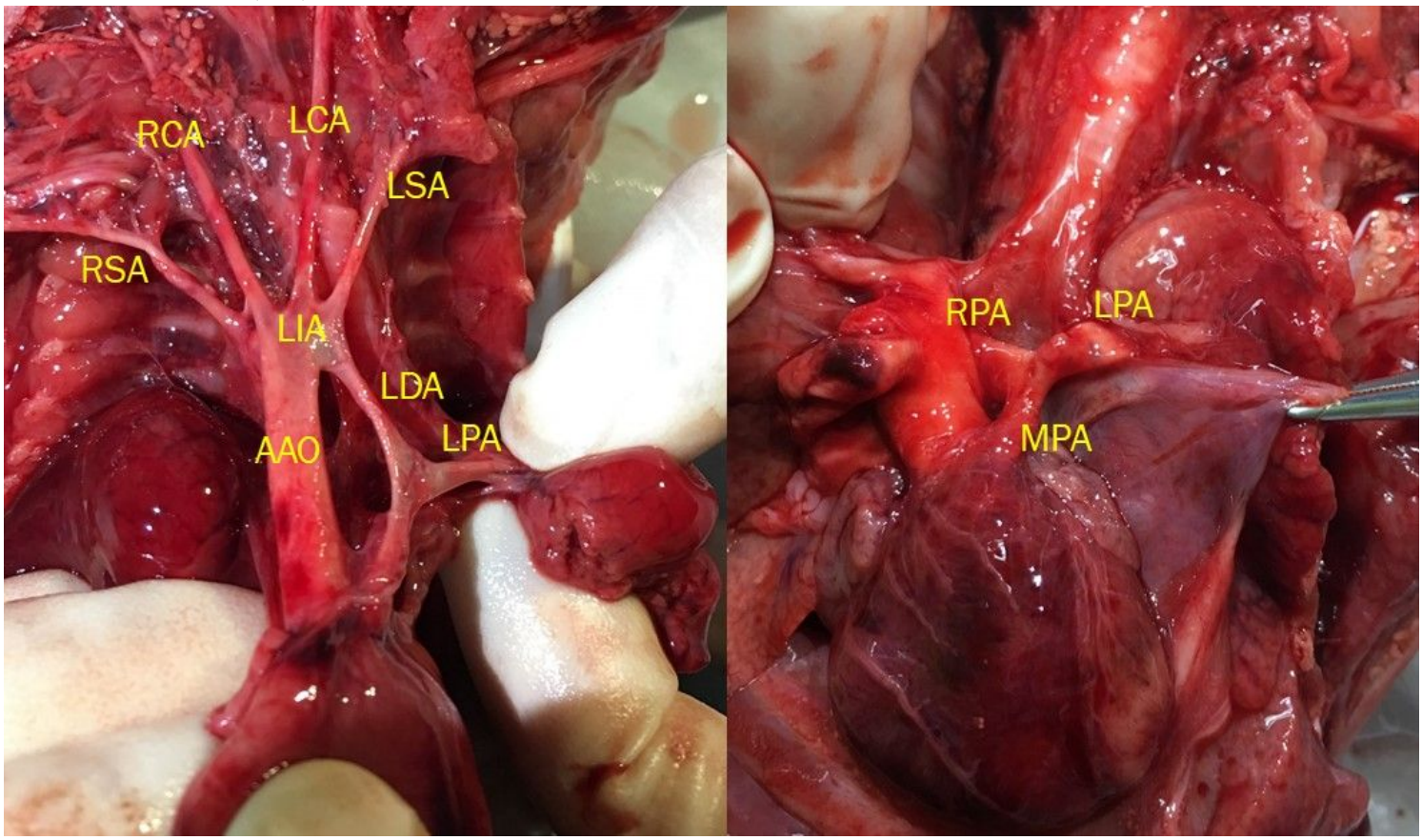

Figure 4

The autopsy: left ductus arteriosus (LDA), left common carotid artery (LCA), left subclavian artery (LSA), right common carotid artery (RCA), right subclavian artery (RSA), left pulmonary artery (LPA), right pulmonary artery (RPA), left innominate artery (LIA), ascending aorta (AAO) , main pulmonary artery (MPA).