Case Report:

Truncus Arteriosus Type 1: A Prenatal Diagnosis, Case Report, and Literature Review

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ABSTRACT

Common arterial trunk (persistent truncus arteriosus) is a rare, congenital heart anomaly and characterized by Ventricular Septal Defect (VSD), single truncal valve, and a common ventricular outflow tract. We reported a case of truncus arteriosus type 1 in the fetus of a 28 years-old G2-P1-L1 pregnant female at 24 weeks of gestational age with large sub truncul VSD, truncal overriding, and main pulmonary artery bifurcation to the right and left pulmonary arteries.

1. Context

Congenital Heart Disease (CHD) is a prevalent anomaly in newborns. The prevalence of CHD is estimated to range from 3.7 to 17.5 per 1000 live births globally (1, 2). This rate in Asia is greater than that in Europe (9.3 per 1000 live births vs. 8.2 per 1000 live births) (3). Its variations are due to maternal risk factors, i.e. different from one country to another.

Common arterial trunk (persistent truncus arteriosus) is a rare, congenital heart defect. Its characteristics are a Ventricular Septal Defect (VSD), a single truncal valve, and a common ventricular outflow tract. It is a single arterial vessel formed in the base of the heart through

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a common truncal valve and commonly feeds to the systemic, pulmonary, and coronary arteries (4). Its incidence is 0.01 among 1000 live births (5). In this disease, the systemic venous blood and pulmonary venous blood are mixed at the VSD site, which causes desaturated blood to enter the common trunk. Common trunk usually overrides the VSD, and almost has an equal position to the Right Ventricle (RV) and the Left Ventricle (LV). In some cases, the common trunk can be predominantly originated from one of the ventricles (6, 7).

In Echocardiographic examinations, this defect can be diagnosed by visualizing the pulmonary arteries coming out from a single great artery, i.e. wider than the aorta and overriding on the interventricular septum (5). The main differential diagnosis of this anomaly is pulmonary atresia with the ventricular septal defect. This malformation can be lethal; accordingly, without surgery, death occurs within two weeks to three months after birth and the mortality rate is 85% in the first year of life (8). We presented a case of truncus arteriosus type 1, diagnosed in prenatal fetal echocardiography.

2. Case Presentation

A 28 years-old Gravid 2, Para 1, living child 1, pregnant female referred to our clinic at the 24th week of gestational age. She presented a history of a high degree of nuchal translucency in her fetus in prenatal ultrasonography. In the examinations, systematic diseases were not found. Her family history was normal. She had no history of drug usage prenatally.

In fetal echocardiography, the position and orientation of the heart were normal (levocardia & levy positions). Four-chamber view signified large sub truncal VSD and dilated truncus root (truncul overriding) (Figure 1); outlet view illustrated truncus root and main pulmonary originating from the proximal portion of truncul root (Figure 2); also, the main pulmonary artery bifurcation and division to the right and left pulmonary arteries (Figure 3). No other associated anomaly was observed. Fetal growth was normal and after the routine legal processes, the pregnancy termination was performed.

3. Discussion

Truncus arteriosus is a severe congenital cardiac malfomation. For obstetric physicians and pediatric cardiologists, the prenatal diagnosis of this anomaly is a challenge.

In the study performed by Jaeggi et al., only 15% of heart defects cases could be diagnosed in the prenatal period (9). This rate in the study performed by Tegnander et al. was reported as 26% by imaging 4 chambers in the fetal heart anomaly scanning (9). However, if the major vessels get visualized into the evaluation -in addition to four-chamber imaging-, this rate can surprisingly be increased to 83% (10).

In a study by Marginean et al., of 411 pregnant women, 17 (4.13%) presented truncus arteriosus; they were diagnosed at a gestational age ranging between 15 and 36 weeks. Besides, among which, only 12 had a gestational age lower than 24 weeks. They reported that among them, 9 cases had type 1, 2 cases had type 2, and in 6 cases, they could not establish a well-defined type (11).

This malformation is hard to diagnose with imaging technics. Moreover, as Lee et al. presented, there is confusion between truncus arteriosus diagnosis and aortic or pulmonary valve atresia. According to these scholars, among 17 cases of truncus arteriosus in prenatal diagnosing, by echocardiography or autopsy postnatally, only 12 cases were confirmed to have this anomaly (12).

Differentiating this abnormality with the tetralogy of Fallot and pulmonary atresia with VSD is important. Furthermore, 22q11.2 microdeletion (involving DiGeorge's syndrome & the related forms) presents the most associations with truncus arteriosus. The other genetic syndromes, indexed in the genesis of this cardiac anomaly are as follows: Ivemark syndrome, Fryns syndrome, Carpentier syndrome, Ellis Van Creveld syndrome, Cornelia De Lange syndrome, and Meckel-Gruber syndrome (13). Trisomies 21, 18, or 13 can be observed in 4.5% of the cases (14, 15); even the rare trisomy 8 is also detected in some cases (16, 17).

Patel et al. reported that out of 554 newborns diagnosed with truncus arteriosus, there was a coexistence of noncardiac congenital anatomic abnormalities, genetic abnormalities, and syndromes in 204 (36.8%) cases (18). Marginean C et al. argued that 5 of the 17 (29.41%) cases, have had other abnormalities and one had trisomy 18 (19).

Some maternal factors involved in truncus arteriosus include the following: infections in utero, like maternal rubella; metabolic disorders, like maternal diabetes mellitus type 1 and 2; chronic maternal disease; hypertension, and obesity (20-22).

Some risk factors of gestation are so threatening and can be the cause of malformations, including the following: stress, alcohol use, cigarette smoking, abusing

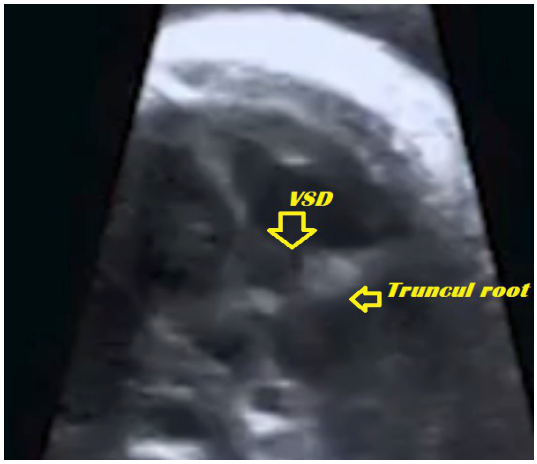

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Figure 1. Four-chamber view showing large sub truncul venticular septal defect and dilated truncul root (truncul overriding)

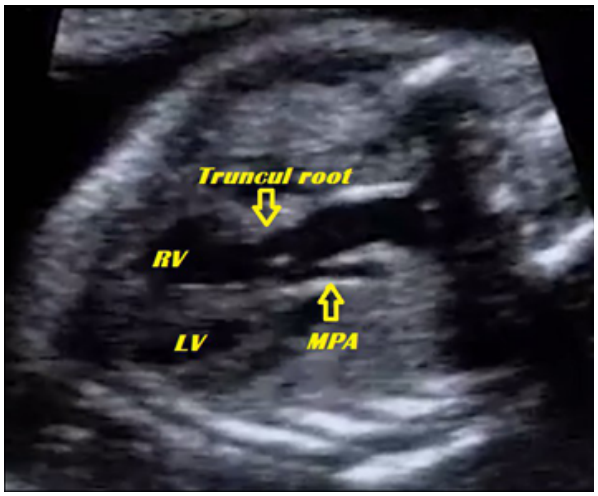

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Figure 2. Outlet view showing truncul root and main pulmonary originated from proximal portion of truncul root
RV: Right Ventricle; MPA: Main Pulmonary Artery.

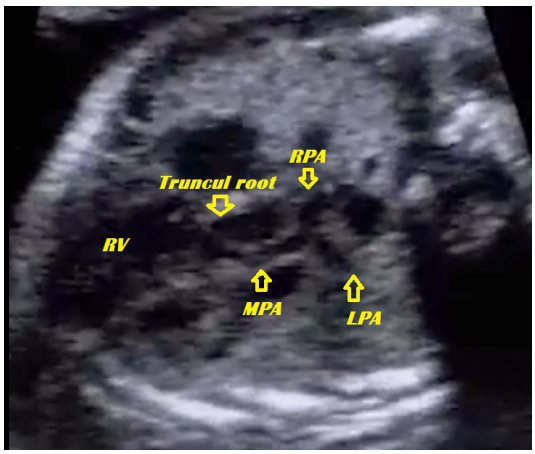

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Figure 3. Main pulmonary artery bifurcation and division to the right and left pulmonary arteries
RV: Right Ventricle; MPA: Main Pulmonary Artery; LPA: Left Pulmonary Artery; RPA: Right Pulmonary Artery.

coffee, retinoic acid, and valproic acid (14). Mother environment is also important and exposure to minerals, hydrocarbons, and oil derivatives before pregnancy or in the early gestation time can lead to heart defects (23). This malformation can be lethal and without surgery, death occurs between two weeks and three months after birth; the relevant mortality rate is 85% during the first year of life (8).

According to a review, post-surgical survival chance equaled 28 out of 48 cases of alive newborns with truncus arteriosus survived. Prenatal diagnosis and postnatal repair surgery can provide a 50% survival chance (24). Naimo et al. stated that in 30 years, the survival rate will reach 73.6% (25). In a study by Jacobs et al., the overall surgical mortality was reported as approximately 9.4% (26). Hussain et al. reported that infants who do not undergo surgical correction encounter a 70% first-year mortality rate and 30% of the infants die within 3 months (27). Surgical repair should be performed within the first 6 months of life to prevent severe pulmonary vasculature obstructive disease (27).

4. Conclusion

Truncus arteriosus is a rare, serious congenital heart abnormality. The good fetal echocardiography in the first trimester of gestation (from 12 weeks of embryonic development) helps an early diagnosis. This is because some types can surgically be repaired in the early stages after delivery. However, this malformation is hard to detect and the accurate diagnosis within this gestational period represents a challenge (28). Differential diagnosis is also essential, as it can be accompanied by other malformations, e.g., right side aortic arch, ASD (atril-septal defect), coarctation of the aorta, Atrio-Ventricular Septal Defect (AVSD), mitral atresia, and the anomalies of the cardiac situs (6).

As mentioned above, this defect can have genetic causes, genetic counseling in recurrent cases, like parental consanguinity, is helpful for better prevention. Postmortem fetopathological examination will provide the most proper diagnosis in fetal death cases, i.e. not diagnosed as an antenatal congenital cardiac disease.

Ethical Considerations

Compliance with ethical guidelines

The patient's information and details have been kept confidential.

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Authors' contributions

Conceptualization: Ali Reza Golbabaei, Azade Rastgar, Mohamad Taghi Majnoon; Methodology: Mohamad Taghi Majnoon, Rana Karimi; Investigation: Ali Reza Golbabaei, Mohamad Taghi Majnoon, Mahsa Naemi; Writing: Behdad Gharib, Sara Memarian; Resources: Ali Reza Golbabaei, Behdad Gharib; Supervision: Ali Reza Golbabaei, Mohamad Taghi Majnoon.

Conflicts of interest

The authors declared no conflicts of interest.

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