

## Down Syndrome with Unbalanced Complete Atrioventricular Septal Defect and Truncus Arteriosus

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### Abstract

Truncus arteriosus (TA) is a rare cardiac anomaly constituting less than 1% of all congenital heart defects. The association of complete atrioventricular septal defect (AVSD) and truncus arteriosus is uncommon. The management of this uncommon combination of lesions has not been well characterized and there is limited surgical options for treatment of these patients. We present our case report of a late presentation of Down syndrome with unbalanced complete AVSD and type 1 truncus arteriosus in a tertiary hospital in South Africa.

**Keywords:** Persistent truncus arteriosus; Down syndrome; Atrioventricular septal defect; Management

### Introduction

Truncus arteriosus (TA), also known as common arterial trunk, is a cyanotic congenital heart defect. It is a rare cardiac anomaly constituting less than 1% of all congenital heart defects. Embryology of truncus arteriosus-during the early stages of a normally developing heart, the great arteries originate from a single truncal root. A wall forms within this root (truncocoanal septum) that divides the root into a fully developed main pulmonary artery and ascending aorta by the end of the fifth week of gestation. Over the next two weeks of development, there is closure of conal septum as the truncocoanal septum fuses with the endocardial cushions and the interventricular septum, which separates the newly formed right and left ventricle. Disturbance in the development of truncocoanal septum results in conotruncal abnormalities including truncus arteriosus [1]. Atrioventricular septal defect (AVSD) is a congenital heart defect with different subgroups that vary in severity and complexity. The defect is a result of developmental abnormalities in the atrioventricular canal, which leads to varying degrees of left-to right shunting between the atria, ventricle and AV valve incompetence [2]. It occurs in about 7% of all congenital heart disease and in 3, 5 per 10,000 live births [3]. It is well known that AVSD is associated with genetic and chromosomal abnormalities, and the most frequently seen in Down syndrome.

The association of complete atrioventricular septal defect (AVSD) and truncus arteriosus (TA) is uncommon [4]. A combination of a TA and AVSD is probably a result of failure of the embryologic process of septation at both the atrioventricular and ventriculoarterial junctions.

In patients with truncus arteriosus and an unbalanced AVSD, the immediate problem in the neonatal period is difficulty in maintaining a balance between the systemic and pulmonary circulations. As the pulmonary vascular resistance (PVR) falls after birth, pulmonary over-circulation results further complicate the already tenuous hemodynamic status [5].

Most reported cases of AVSD and TA were diagnosed postnatally. There has been a case report of truncus arteriosus and AVSD diagnosed prenatally by fetal echocardiography [4]. Fetal diagnosis of AVSD and TA may be difficult, but careful screening of the four chamber view and outflow tracts should help identify both components of this complex congenital heart disease. Fetal diagnosis of AVSD is quite straightforward and includes an abnormal four chamber view with a defect at the crux of the heart that is bridged by a common atrioventricular valve. The initial clue to the diagnosis of TA is finding a single semilunar valve and great artery that appear large and overrides the ventricular septal defect [4].

The surgical management of truncus arteriosus and AVSD as individual lesions has steadily improved over the past two decades, resulting in improved survival and long term outcomes [6]. The management of this uncommon combination of lesions has not been well characterized and there are limited surgical options for treatment of these patients. There has been reports of patients operated for TA and balanced AVSD, and these patients underwent two ventricle repair [7,8]. Neonates with a combination of TA and unbalanced AVSD with single ventricle physiology and pulmonary over circulation can be successfully offered palliative surgical options.

Pulmonary over circulation in these neonates can be controlled either by bilateral pulmonary artery banding or by excision of the pulmonary artery, patch reconstruction of the truncal defect, and aortopulmonary shunt. The first stage will be followed by bidirectional Glenn shunting and Fontan procedure as the last operation [5].

We present our case report of a Down syndrome patient who presented at about 1 month of age with unbalanced complete AVSD and type 1 truncus arteriosus in a tertiary hospital in South Africa.

### Case Report

A 24 day old infant born by normal spontaneous vaginal delivery at 40 weeks gestation. Patient had normal Apgar scores at birth, with a birth weight of 2, 6 kg. Pre-natal ultra sound scan was not done, and therefore the diagnosis of TA and AVSD was made postnatally. The

patient presented for the first time to cardiology unit at 3 weeks of age. She was referred from a peripheral hospital, with complaints of, poor feeding, shortness of breath and oedema.

### On physical exams

She had dysmorphic features in keeping with Down syndrome, and features of congestive heart failure.

### Chest radiography

Revealed cardiomegaly and dilated main pulmonary artery.

### Electrocardiogram

Showed dilated right atrium and right ventricle. Superior axis deviation.

Echocardiography on admission: showed type 1 truncus arteriosus (Figure 1A), there was no truncal valve regurgitation. Unbalanced atrioventricular septal defect (Figure 1B), dilated right ventricle, hypoplastic left ventricle, straddling single atrioventricular valve, inlet ventricular septal defect and premium atrial septal defect with left to right shunting on colour flow Doppler and mild atrioventricular valve regurgitation (Figure 1C).

### Blood test

Trisomy 21 confirmed on cytogenetic testing. FISH for Di-George syndrome was negative. Management on admission: patient was started on Lasix and spironolactone for congestive cardiac failure. In view of the limited surgical options and the poor prognosis, the patient is for comfort care management.



**Figure 1A:** Transthoracic two-dimensional echocardiogram in short axis view image showing type 1 truncus arteriosus, truncal valve arising from the right ventricle. TV=Truncus valve; RPA=Right pulmonary artery; LPA=Left pulmonary artery.



**Figure 1B:** Transthoracic 2 dimensional echocardiogram in apical 4 chamber view image showing a complete unbalanced atrioventricular septal defect with hypoplastic left ventricle. RV=Right ventricle; LV=Left ventricle; RA=Right atrium; LA=Left atrium.



**Figure 1C:** Transthoracic 2 dimensional echocardiogram in apical 4 chamber view image showing unbalanced complete atrioventricular septal defect with hypoplastic left ventricle, dilated right ventricle, common atrioventricular valve, inlet ventricular septal defect and premium atrial septal defect. RV=Right ventricle; LV=Left ventricle; VSD=Ventricular septal defect; AVV=Atrioventricular valve; ASD=Atrial septal defect; RA=Right atrium; LA=Left atrium.

## Discussion

A combination of TA and AVSD is probably a result of failure of the embryologic process of septation at both the atrioventricular and Ventriculo-arterial junctions [9]. Truncus arteriosus is a congenital heart defect in which a common arterial trunk gives rise to the aorta, pulmonary arteries and the coronary arteries. It constitutes less than 1 % of all congenital heart defects among live born infants. It is usually associated with 22q11 microdeletion syndromes.

Atrioventricular septal defects is characterised by a deficiency of the atrioventricular septum of the heart. They account for about 5 % of all congenital heart disease, and are most common in infants with Down syndrome. Forty five percent of children with Down syndrome have congenital heart disease, of these, 35 to 40% have AVSD [10].

The association of TA and AVSD is exceedingly rare [4]. The majority of cases that have been reported were postnatal diagnosis including post mortem reports [11-13]. The majority of cases of TA and AVSD are diagnosed with neonatal echocardiography immediately after birth. Our patient was diagnosed 3 weeks after birth when he presented for the first time in heart failure.

Majority of the level 1 hospitals in South Africa do not have ultra sound scan facilities and medical staff with expertise of doing cardiac ultrasound during pregnancy, and therefore this patient was not diagnosed during prenatal period. Also with the shortage of skilled medical practitioners at the local hospitals, these cases are sometimes missed at birth and are referred late to cardiology unit for diagnosis and management. This is a typical case of a patient that was missed at birth and therefore only referred to cardiology due to congestive cardiac failure symptoms.

The surgical management of TA and AVSD as individual lesions has steadily improved over the past two decades, resulting in improved operative mortality and long term outcome [3].

The management of this uncommon combination of lesions has not been well characterised, and therefore there is limited surgical options and poor prognosis [4]. Some patients with TA and AVSD are offered comfort care due to the limited surgical options and poor prognosis.

There has been reports of patients operated for TA and balanced AVSD, and these patients underwent two ventricle repair [7,8]. Neonates with a combination of TA and unbalanced AVSD with single ventricle physiology and pulmonary over-circulation can be successfully offered palliative surgical options. Early diagnosis and referral of these patients with AVSD and TA to a tertiary center that offers surgery for congenital heart disease leads to early intervention and improved outcomes. In South Africa we need to improve early

diagnosis, with prenatal and/early postnatal echocardiography and early referral of these patients to a tertiary hospital so as to improve their outcomes and also prevent development of pulmonary hypertension.

## Conclusion

Truncus arteriosus and atrioventricular septal defect is a rare combination. Neonates with balanced AVSD and TA can be fully repaired (2 ventricle repair) surgically, and patients with an unbalanced AVSD and TA with single ventricle physiology can be successfully offered palliative surgical options.

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