

# Atrioventricular Septal Defect with Common Atrioventricular Junction Guarded by a Common Valve Consisting of Left Atrioventricular Trifoliate Valve

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## Case report

### ABSTRACT

**Introduction.** Atrioventricular septal defect with common atrioventricular junction is a rare adult congenital cardiac syndrome. This occurrence with prolonged survival is exceptionally rare. **Case report.** We present the case of a patient who presented with this defect with common atrioventricular junction who survived to the age of 32. We describe a 32-year-old man with atrioventricular septal defect

with common atrioventricular junction guarded by a common valve. His history, clinical course, and anatomic findings are discussed along with the factors which may have contributed to his longevity, which is unique in the medical literature. His management reflected the state of medical knowledge at the time when he presented, and although alternate approaches may have been utilized if the patient presented today. We discuss the findings, frequency, classification, and management

of congenital defects. Development of embryonic structure is altered by interaction between genetics and environmental factors toward a rare associated of congenital cardiac defects-complex congenital heart disease. **Conclusion.** This case demonstrates that patients with very complex congenital cardiac disease may survive to adulthood, presenting challenges in both medical and surgical treatment.

**Key words:** Atrioventricular septal defect, atrioventricular trifoliate valve, echocardiography.

## 1. INTRODUCTION

We present a patient who was born with atrioventricular septal defect with common atrioventricular junction guarded by a common valve with the trifoliate of the left atrioventricular valve. These cases with prolonged survival are exceptionally rare. The medical history, physical examination, laboratory findings, and a review of the individual congenital cardiac defects are discussed.

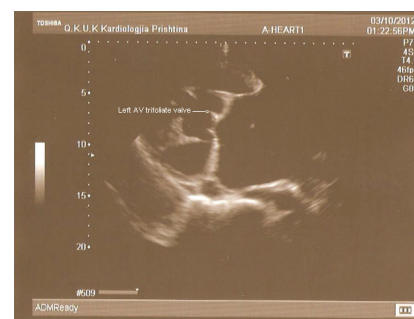
## 2. CASE PRESENTATION

A 32-year-old man presented to our clinic with the following history. His family history was negative for congenital heart defects and the patient had no siblings. Immediately after birth the cardiac status with a systolic murmur along left sternal border leading to a pediatric center where is diagnosed. The growth and mental development were normal, but marked cyanosis, weakness, clubbing, and intolerance of physical activity was present. At the age of nine the patient was referred to a cardio-pediatric center abroad where was diagnosed as complete atrio-

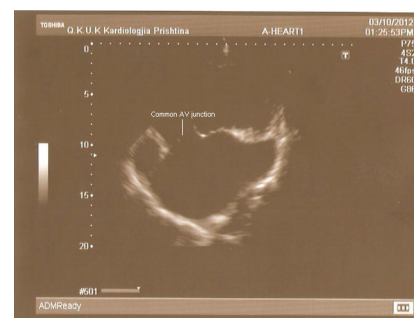
ventricular canal defect, pulmonary atresia, and persistent ductus arteriosus. The patient condition was improved significantly and remained stable for the next eight year. At 17 years of age he was hospitalized again at the same center because of cyanosis, palpitations, and weakness with intolerance to physical activity. The patient had several documented episodes of atrial fibrillation.

At the age of 32, the patient presented to our clinic with severe cyanosis, he complained of shortness of breath with minimal exertion, and chest pain. The physical examination was remarkable for a 4/6 systolic murmur heard at the left sternal border, and cardiac frequency 130 beats per minute. The patient was noted to experience severe shortness of breath with even minimal exertion and was deeply cyanotic.

The electrocardiogram demonstrated atrial fibrillation, with ventricular frequency 100/min, and bifascicular block. Chest radiograph indicated a small pleural effusion on the right and bilateral hilopulmonary stasis. The heart was mod-



**Figure 1.** Parasternal long axis view of atrioventricular septal defect with common atrioventricular junction guarded by a common valve—the left atrioventricular valve is trifoliate.



**Figure 2.** Echocardiography findings showing apical view of the patient presenting with atrioventricular septal defect.

erately enlarged with prominence of the aorta. An echocardiogram was performed (Figure 1, 2).

Patient was diagnosed in his early childhood with a congenital anomaly. In the very early echocardiography findings the presence of rudimentary inter ventricular and inter atrial septum leading to a common atrioventricular canal with one atrioventricular valve was described. This is also in the context of the malformations occurring during the intrauterine development of the embryo. From a perspective of adult congenital heart disease, the enlargement and the slight modifications of the heart cavities through the age, led to echocardiography view of hardly seen the rudimentary part of the interventricular septum, creating a perception of a single ventricular and a single atrium heart. An atrioventricular septal defect with common atrioventricular junction guarded by a common valve was seen. Both atriums were dilated, while pulmonary atresia and patent ductus arteriosus were also diagnosed in the early childhood. The main diagnoses were: atrioventricular septal defect with common atrioventricular junction guarded by a common valve, pulmonary atresia, persistent ductus arteriosus, and congestive heart failure.

### 3. DISCUSSION

Atrioventricular septal defect is an anatomic lesion with common atrioventricular junction as a phenotypic feature (1). These anatomic lesion often describe as atrioventricular canal that may be partial (PAVC)-openings between the left and right atrium and improper formation of mitral valve, and complete (CAVC)-there is a large hole where atria and ventricles meet with one common valve (2). Atrioventricular septal defect is associated with different defects including subaortic stenosis, ventricular hypoplasia, tetralogy of Fallot, atrial isomerism, and rare with pulmonary atresia (3). Pulmonary atresia is a congenital malformation of the pulmonary valve in which the valve orifice fails to develop with the only source of pulmonary blood flow-patent ductus arteriosus. As an uncommon congenital heart disease, this accounts for about 3% of cardiac malformations. A total prevalence

of atrioventricular septal defects is 5.3 per 10,000 births [3]. Both sexes are equally affected, with a slightly higher frequency in female (female/male ratio 1.3/1) and a striking association with Down syndrome is found. In developmental terms atrioventricular septal defect is a deficiency of the tissues that, in the normally structured heart, interpose between the atrial and ventricular chambers. The lesion is produced on the basis of failure of formation of the atrioventricular septum as component of the normal aortic root interpose between the cavities of the left ventricle and the right atrium, and initially a muscular atrioventricular septum, creating an area that is described as a sandwich. The septum and sandwich are absent in almost hearts with atrioventricular septal deficiencies (1). The phenotypic feature of this defect is the commonality of the atrioventricular junction guarded by a common atrioventricular valve. The valve has two leaflets confined to the right ventricle; a mural leaflet positioned on the left ventricle; and two leaflets bridging the ventricular septum (1). In the past, based on the morphology of the superior leaflet three types of complete atrioventricular canal delineated as type A, B or C were described (5).

Congenital Heart disease is usually caused by altered development of embryonic structure, or a failure of the structure to develop beyond an early embryonic or fetal stage. In the etiology of congenital heart disease both genetic and environmental factors participate. From genetics factors atrioventricular septal defect with common atrioventricular junction are often diagnosed in patients with trisomy 21-numeric chromosomal defect (6), than 3p-syndrome, 8p-syndrome-structural chromosomal defects. In the single gene disorders atrioventricular septal defect with common atrioventricular junction is presented in Holt-Oram syndrome, heterotaxy syndrome, and Noonan syndrome. The maternal factors that are present in pregnancies include the use of nonsteroidal antiinflammatory drugs-ibuprofen, or smoking (7). Organic solvents may also be the causes of this entity and other con-

genital heart disease. Pulmonary valve defects are caused from chromosomal defects, and single gene disorders (Holt-Oram syndrome, heterotaxy syndrome, Noonan syndrome, Alagille syndrome). Maternal rubella infection (8), maternal febrile illness (9), maternal medical uses (phenytoin, thalidomide), organic solvents (10) and air pollution with CO (11) may be followed with pulmonary valve defects. Interaction of genetic and environmental factors during pregnancy may cause the unusual complex congenital heart disease unknown till now.

### 4. CONCLUSION

Development of embryonic structure is altered by interaction between genetics and environmental factors toward a rare associated of congenital cardiac defects-complex congenital heart disease.

This case demonstrates that patients with very complex congenital cardiac disease may survive to adulthood, presenting challenges in both medical and surgical treatment.

### REFERENCES

1. Anderson R, Wessels A, Vettukattil J. Morphology and morphogenesis of atrioventricular septal defect with common atrioventricular junction. *WJPCHS*. 2010; 1: 59-67.
2. Mayo Clinic. Atrioventricular Canal Defect. 2012, <http://www.mayoclinic.com/health/atrio-ventricular-canal-defect/DS00745>
3. Craig B. Atrioventricular septal defect: from fetus to adult. *Heart*. 2006; 92(12):1879-1885.
4. Christensen N, Andersen H, Garne E, Wellesley D, Addor MC, Haessler M, Khoshnood B, Mullaney C, Rankin J, Tucker D. Atrioventricular septal defects among infants in Europe: a population-based study of prevalence, associated anomalies, and survival. *Cardiology in the Young*. 2012, <http://dx.doi.org/10.1017/S104795112001400>.
5. Rastelli GC, Kirklin JW, Titus JL: Anatomic observations on complete form of common atrioventricular canal with special reference to atrioventricular valves. *Mayo Clinic Proc*. 1966; 41:296.
6. Paladini D, Tartaglione A, Agangi A, Teodoro A, Forleo F, Borghese A, Martinelli P. The association between congenital heart disease and Down syndrome in prenatal life. *Ultrasound in Obstetrics & Gynecology*. 2000; 15(2): 104-108.
7. Alverson CJ, Strickland MJ, Gilboa SM, Correa A. Maternal smoking and congenital heart defects in the Baltimore-Washington Infant Study. *Pediatrics*. 2011; 127(3): 647-653.
8. De Santis M, Cavaliere AF, Straface G, Caruso A. Rubella infection in pregnancy. *Reproductive Toxicology*. 2006; 21(4): 390-398.
9. Oster ME, Riehle-Colarusso T, Alverson CJ, Correa A. Associations between maternal fever and influenza and congenital heart defects. *The Journal of Pediatrics*. 2011; 158 (6): 990-995.
10. Shaw GM, Nelson V, Iovannisci DM, Finnell RH, Lammer EJ. Maternal occupational chemical exposures and biotransformation genotypes as risk factors for selected congenital anomalies. *American Journal of Epidemiology*. 2003; 157(6): 475-484.
11. Rankin J, Chadwick T, Natarajan M, Howel D, Pearce MS, Pless-Mulloli T. Maternal exposure to ambient air pollutants and risk of congenital anomalies. *Environmental Research*. 2009; 109 (2): 181-187.