International Journal of Science and Research (IJSR)
ISSN: 2319-7064
Impact Factor 2025: 7.089

Autopsy Based Case Report of Pentalogy of Fallot with Single Coronary Ostium in an Infant

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Abstract: ***Pentalogy of Fallot is a rare congenital cardiac anomaly characterized by the features of Tetralogy of Fallot with an additional atrial septal defect. This report describes an autopsy-based case of an eight-and-half-month-old male infant who presented with fever, erythematous rash and sudden breathlessness, and was declared dead on arrival at hospital. Medicolegal autopsy revealed a boot shaped heart with a ventricular septal defect, right ventricle hypertrophy, overriding aorta, single atrium and a single coronary ostium. Pulmonary consolidation and inflammatory changes were observed in the lungs. Ancillary investigations were negative for infectious etiologies. The cause of death was determined to be pulmonary consolidation secondary to congenital heart disease. This case highlights the importance of early detection and timely intervention in congenital cardiac anomalies to improve clinical outcomes.***

Keywords: Tetralogy of Fallot, Pentalogy of Fallot, congenital heart disease, autopsy, infant mortality, ventricular septal defect, forensic pathology

Abbreviations: TOF – Tetralogy of Fallot, CHD- congenital heart disease, ASD- atrial septal defect, VSD- ventricular septal defect, RVOT- right ventricular outflow tract

1. Introduction

1.1 Case description

The infant was eight-and-a-half months old, with fever and rashes on the body, developed sudden onset of breathlessness and was brought to the Emergency department, where he was declared dead on arrival. He had history of another bout of fever two months back. On examination, he had erythematous rashes on the back of head, neck, under chin and trunk.

This study aims to present and analyze an autopsy-confirmed case of Pentalogy of Fallot with associated coronary anomaly and discuss its clinical and forensic implications.

1.2 Methodology

The autopsy was conducted at the Department of Forensic Medicine at Government Medical College Thiruvananthapuram, Kerala in July 2019. After external examination, dissection was done using modified Letulle's method. All the organs were weighed. The heart was weighed and dissected using Virchow's inflow - outflow technique. The heart was then fixed in 10% formalin solution for further examination. Ancillary investigations comprising serological tests and histopathological analysis were conducted at the Departments of Microbiology and Pathology at Government Medical College, Thiruvananthapuram, Kerala. Tissue samples were fixed in 10% formalin for histopathological examination, followed by sectioning and staining with Hematoxylin-Eosin staining and examination under microscope. Chemical analysis was done at the Government Chief Chemical Examiner's laboratory, Thiruvananthapuram, Kerala.

1.3 Autopsy Findings

On examination, the child measured 68 cm in length and weighed 5.9 kg. Erythematous rashes on under chin, neck, back of head and back of trunk. His finger nails were pale. Rigor mortis was fully established and there was no sign of decomposition.

On dissection, pericardial sac contained 20ml of straw-colored fluid. Heart (79g) was boot shaped (couer en sabot), with single atrium having two appendages, two ventricles with a ventricular septal defect in upper part of interventricular septum; wall of right ventricle was thicker than that of left ventricle. Root of aorta showed only single coronary ostium and there was a single coronary artery. Aorta overrode the pulmonary trunk and was seen attached to the pulmonary trunk externally. Interventricular septal defect was seen in the upper peri-membranous part of septum and was the cause of VSD. Tricuspid valve leaflets were thickened. Myocardial thickness was 0.2-0.3cm in the single atrium, 1 cm in right ventricle and 0.8cm in left ventricle. Subendocardial hemorrhages were also seen.

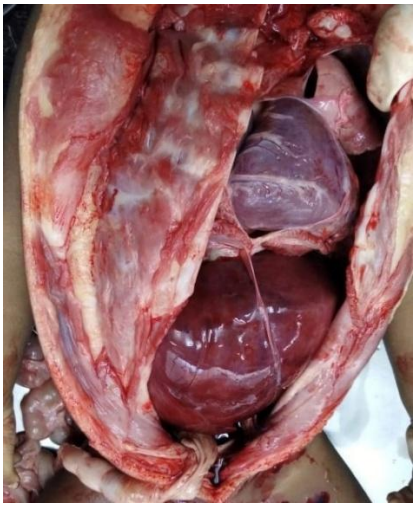

Figure 1: Boot shaped heart seen on dissecting the pericardium

Figure 2: Heart after dissecting and formalin fixation

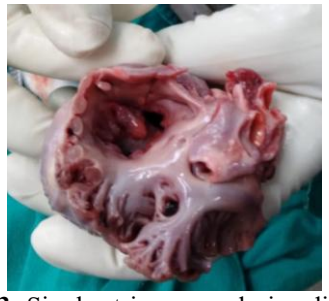

Figure 3: Single atrium seen during dissection

Figure 4: Ventricular septal defect shown in formalin fixed heart

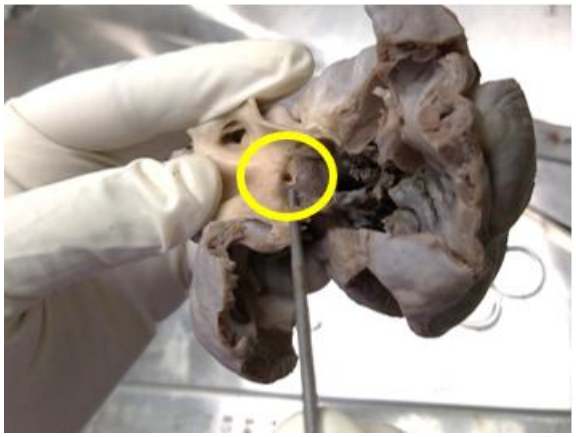

Figure 5: Single coronary ostium in formalin fixed heart

Thymus gland was seen as a fibrous strand. Brain was congested and edematous. Air passages showed reddish discoloration and were obstructed by thick yellow pus. Lungs showed consolidation and petechiae on the surface (right-66g, left-57g). Bronchioles were thickened, containing purulent exudate.

Spleen was congested (24g). Liver was pale (193g). Kidneys were pale with distinct corticomedullary demarcation and cortical hemorrhages and weighed 20g(right), 26g(left). Right adrenal gland was hemorrhagic. Peritoneal cavity contained 10ml of straw-colored fluid. Stomach contained 5ml curd like material having no unusual smell, mucosa was pale.

1.4 Serological tests

Serological tests done included IgM Dengue, IgM Leptospira, IgM Chikun Guniya, all of which were negative.

1.5 Histopathology examination

Histopathology examination showed pulmonary oedema. There were areas of collapse with areas of compensatory emphysema, hemorrhage with lymphoplasmacytic infiltrate and neutrophils. Thymus gland showed hyperplastic areas with neutrophilic infiltrate. Liver, spleen, kidneys and adrenal glands were congested.

2. Discussion

Pentalogy of Fallot is TOF with atrial septal defect.(1) The components of TOF are right ventricular outflow tract obstruction, malalignment type VSD, dextroposition of aorta causing overriding of VSD and right ventricular hypertrophy. The pathophysiology occurs due to severe hypoplasia of the pulmonary valve annulus, causing right ventricular outflow tract obstruction. There is also a large VSD below the aortic valve, causing the aorta to override the VSD, ending in right ventricular hypertrophy.(2)

TOF is a cono-truncal malformation, hence can be associated with(3)

1) DiGeorge syndrome – microdeletion of chromosome 22q11.2
2) Shprintzen velocardiofacial syndrome (CATCH22)
3) Alagille syndrome (JAGGED1 gene mutation)
4) CHARGE syndrome

5) VACTREL syndrome
6) Goldenhar syndrome (oculoauriculovertebral spectrum)
7) Tetralogy of Fallot with an atrioventricular septal defect occurs with Downs syndrome. 22q11 deletion is found in 16% infants.(4)
8) TOF is sporadic and non-familial, but can be associated with chromosomal anomalies (12%- Down's, Patau, Edwards) and monogenic syndromes.(5)
9) Additional anomalies seen in association with TOF+22q11 microdeletion:
   - Right or cervical aortic arch
   - Hypoplasia or absence of infundibular septum
   - Absence of pulmonary valve
   - Discontinuity/ diffuse hypoplasia of pulmonary artery

Severity of symptoms depend on the severity of obstruction to RVOT. The age of first presentation and cyanosis depend on the degree of pulmonary obstruction. There is marked cyanosis in later part of infancy, with clubbing and dyspnea on exertion and squatting. Cyanotic spells may be seen in the first two years of life. There may be recurrent infections which lead to failure to thrive as well as delayed puberty. On examination, right ventricular apex may be palpable, with a left hemithorax bulge. Loud and harsh systolic murmur with intensity inversely proportional to the severity of RVOT obstruction may be heard. In severe cases, the chest becomes silent.(6) Pansystolic murmur may also be heard due to the VSD. If there is ascending aortic aneurysm and aortic regurgitation, it will lead to a high-pitched diastolic murmur.(7)

The work up other than routine blood panel includes X ray, electrocardiography, echocardiography, cardiac catheterization and angiography and MRI. ECG will show right axis deviation and evidence of right ventricular hypertrophy, while echocardiography will establish the diagnosis.(6) Management includes symptomatic treatment as well as surgical repair of the defects, like balloon dilatation, stenting, shunting, valve replacement.(8) All of these surgeries require basic understanding of the pathophysiology and hemodynamics for effective anesthesia and proper outcome.(9) Symptomatic treatment could be given immediately to stabilize the patient. Balloon dilatation of RVOT and stenting of pulmonary arteries may be needed. Symptomatic infants at any age may need balloon dilatation of RVOT and pulmonary arteries with or without stenting. In adults, surgery is recommended and risks are the same as in infants if there are no comorbidities. Palliation is not a permanent treatment, but a stop gap measure, and must be followed by intracardiac repair. Intervention may be needed if residual VSD shunt >1.5:1, severe PR with RV dilatation, exercise intolerance or in case of sustained arrhythmias. Palliation may be done in cases where surgery is not possible. Survival to adult life is rare without surgery and palliation. 25-year survival rate is 94%. Expert cardiology follow up is needed.

Preoperative recognition of single coronary ostium is also very important for planning and execution of surgical repair. First published case of Pentalogy of Fallot with single coronary artery disease is in a 22-year-old female patient, diagnosed via echocardiography and catheterization in 2014. In this case, there was undiagnosed CHD and DiGeorge syndrome.(10) Other procedures like dental surgeries may also be done keeping in mind the possibility of stress induced cyanosis and bacterial endocarditis.(11)

In a study by Veldtman et al., in pregnant women with confirmed TOF, they concluded that there is increased risk of foetal loss and congenital anomalies in their offspring. There is also possibility of adverse maternal events. Intracardiac repair in the mother has permitted survival to childbearing years and excellent quality of life.(12)

3. Conclusion

This autopsy-based case confirms Pentalogy of Fallot with an associated single coronary artery anomaly in an infant, with death attributed to pulmonary consolidation secondary to congenital heart disease. The findings underscore the importance of early diagnosis, neonatal screening and timely surgical intervention in improving survival outcomes. Enhanced awareness and genetic evaluation in high-risk population may further aid in reducing morbidity and mortality associated with complex congenital cardiac anomalies.

4. Recommendations

Genetic counselling especially in high-risk scenarios like recurrence of CHD is important. Detection of genetic abnormalities in antenatal period can change the outcome.(2) Recurrence of CHD is possible, so a high index of suspicion is needed in subsequent pregnancies. Early screening with mandatory neonatal screening especially in high-risk cases. Early surgical intervention before disease progression is also important in improving the prognosis.

References

[1] Partana P et al., Multiple pregnancy in a primigravida with uncorrected Pentalogy of Fallot. BMJ. BMJ Case Reports. Jan 2017. https://pmc.ncbi.nlm.nih.gov/articles/PMC5256472/
[2] Braun-Falco M et al., Pentalogy of Fallot. Encyclopedia of Molecular Mechanisms of Disease.2009;1602–5p. https://link.springer.com/rwe/10.1007/978-3-540-29676-8_1393
[3] Dan L Longo et al., Disorders of the Cardiovascular system, Harrison's principles of Internal Medicine. 22nd ed. Vol 2. 2025. 1926-1927 p.
[4] Neil McIntosh et al., Farfar and Arneil's Textbook of Pediatrics. 7th edition. Elsevier; 2008. 788-789 p.
[5] I B Vijayalakshmi et al., A comprehensive approach to congenital heart disease. Jaypee brothers medical publishers 2013. 547–558 p.
[6] Kleigman et al., Nelson textbook of Pediatrics. 20th edition. Vol. 2. Elsevier; 2015. 2165-2215p.
[7] Mann et al., Braunwald's Heart Disease, A textbook of Cardiovascular Medicine. 10th edition. II. 2015. 1415–1418 p.
[8] K V Krishnadas. Textbook of Medicine. Elsevier; 5th edition. 779–780p.
[9] Anaesthetic management of a patient with pentalogy of Fallot a case report. https://journals.lww.com/ejca/fulltext/2018/12020/anaes

thetic_management_of_a_patient_with_pentalogy.3.aspx

[10] Pentalogy of Fallot with a single coronary artery: A rare case report, Jahangir Rashid Beig, et al., J Tehran Heart Cent. 2014; 9(3): 132-134p. https://pmc.ncbi.nlm.nih.gov/articles/PMC4393836/

[11] Drishti Kaushal et al., Pentalogy of Fallot: A case report and overview dental implications. Spec care dent. Jan 2020 Vol 40 (1). 121-126p. https://onlinelibrary.wiley.com/doi/abs/10.1111/scd.12433

[12] Veldtman GR, Connolly HM, Grogan M, Ammash NM, Warnes CA. Outcomes of pregnancy in women with tetralogy of Fallot. Journal of the American College of Cardiology. 2004 Jul; 44(1):174–80. http://www.onlinejacc.org/content/44/1/174