

Rare association of TOF with multiple anatomic variants: A case report

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Abstract- This case report showcases the various imaging findings in a patient with tetralogy of fallot. TOF is the most common cyanotic congenital disorder. It occurs in three of every 10000 live births and accounts for 7%–10% of all congenital cardiac malformations. The classic tetrad of TOF includes RVOT obstruction, RV hypertrophy, VSD, and overriding of the aorta. There are many anatomic variants, intracardiac and extracardiac anomalies that must be taken into consideration when imaging and planning the surgical procedure is needed. Multi-detector computed tomography (MDCT), with its high spatial and temporal resolution, has an important role in the evaluation of complex anatomical findings in both unrepaired and repaired TOF patients. These could be related to the aortic arch, systemic venous system, pulmonary venous system, coronary arteries, status of outflow tracts and tracheobronchial tree. We herewith report a rarest of rare case of association of TOF with anomalous origin of right pulmonary artery from aorta, right side tracheal bronchus, tracheal diverticulum and azygous lobe which as individual entities are itself rare anomalies.

Keywords: TOF, Tracheal bronchus, Anomalous right pulmonary artery, azygous lobe.

INTRODUCTION

Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease (CHD). Non-invasive cardiac imaging plays a critical role in the initial diagnosis and follow-up of TOF patients. Echocardiography is the initial modality of choice for making the diagnosis and follow-up. However, multi-detector computed tomography (MDCT), with its high spatial and temporal resolution, provides detailed depiction of cardiac anatomy and morphology and thus has a pivotal role in the evaluation of complex anatomical findings in both unrepaired and repaired TOF patients. In addition, performing and interpreting cardiac CT examination in unrepaired TOF is essential to guide the surgical intervention needed for its repair. MDCT also helps in identification of different anatomic variants which may be seen with TOF.

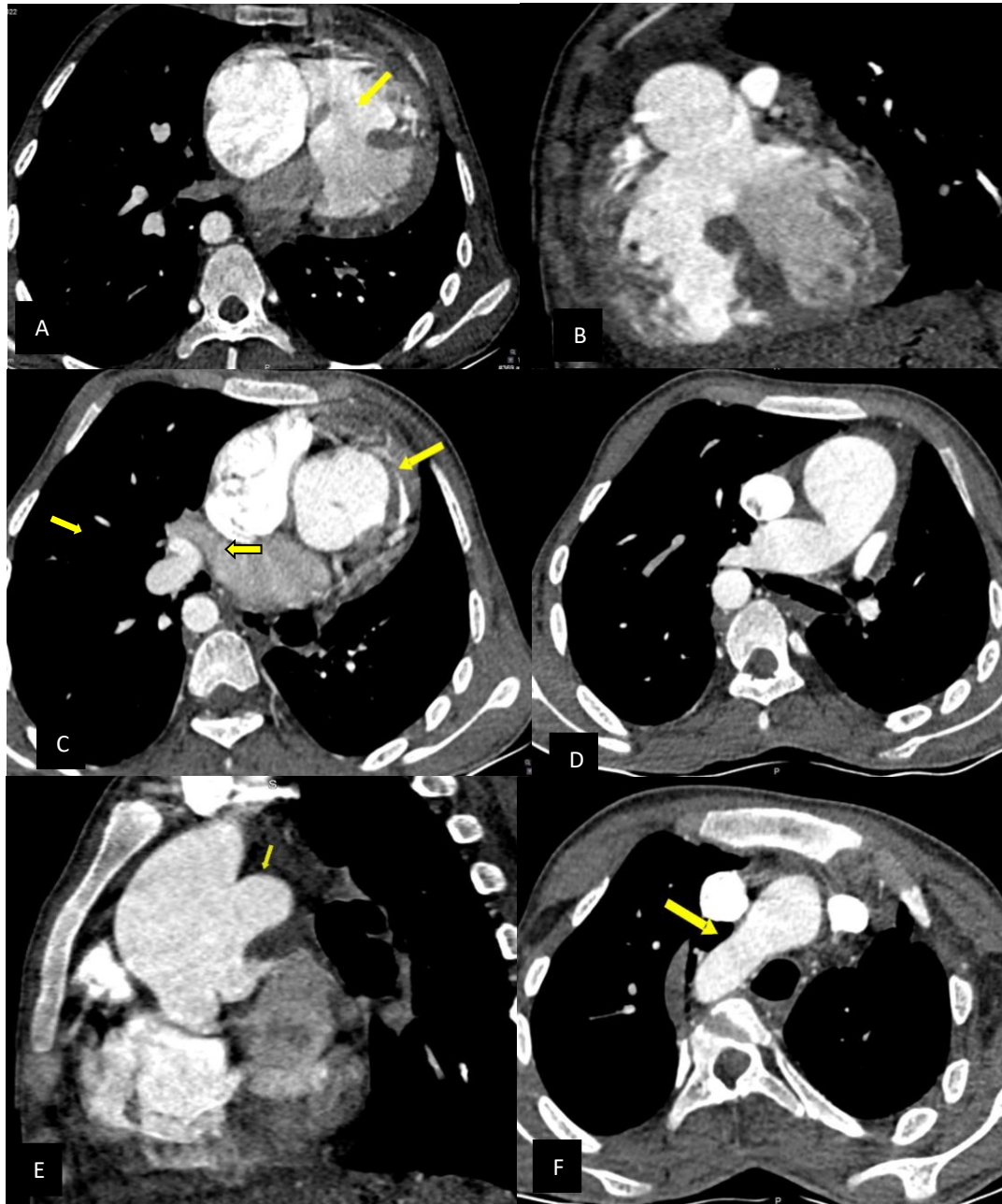
CASE REPORT

A 19 year old male patient presented to our institution with history of cyanotic congenital heart disease and dyspnea on exertion since last 10 years. Patient was diagnosed as a case of Tetralogy of Fallot 10 years before he came to us and he had not undergone any palliative surgery for the same. He currently had mild cough with no history of fever. The patient's laboratory results were found to be within normal limits.

Cardiac color Doppler examination was done and showed features of TOF. MDCT followed by contrast administration was performed as advised by the treating consultant to look for features of TOF and any other associated abnormality and anatomic variants which are required for pre-operative assessment.

Cardiac CT scan showed a large malaligned VSD with overriding of aorta. There was significant stenosis of pulmonary infundibulum which continued distally as left main pulmonary artery. Main pulmonary trunk was not seen and anomalous origin of right pulmonary artery from ascending aorta was noted. Right sided aortic arch was also seen.

Normal anatomic variants were also findings in this patient. Right tracheal bronchus supplying right upper lobe along with tracheal diverticulum arising from left main bronchus was also seen .Right azygous lobe as well was noted.



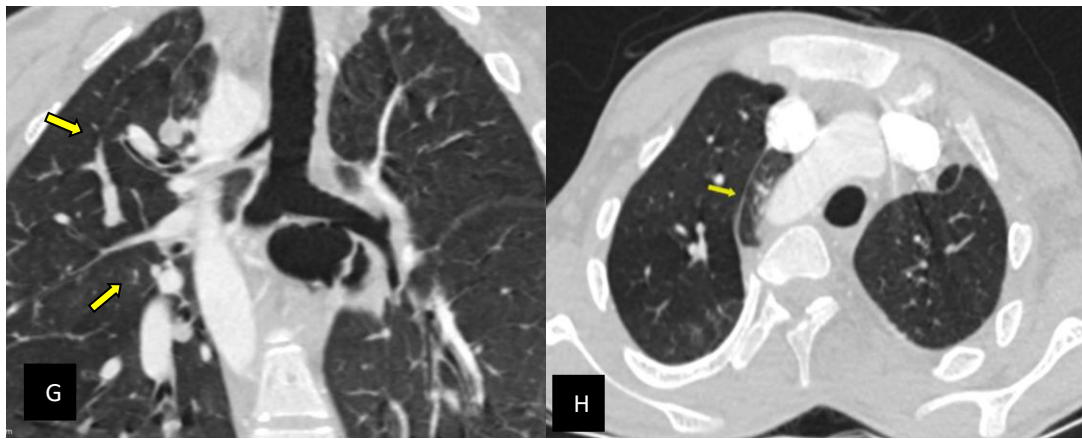


Figure shows MDCT followed by intravenous contrast scan depicting A) Large VSD B) Overriding of aorta along with VSD C) Significant infundibular stenosis which continues as left pulmonary artery D) & E) Anomalous origin of right pulmonary artery from aorta and separate left pulmonary artery F) Right sided aortic arch G) Right tracheal bronchus and tracheal diverticulum H) Right azygous lobe.

DISCUSSION

The classic tetrad of TOF, includes RVOT obstruction, RV hypertrophy, VSD, and overriding of the aorta. There are many anatomic variants, associated intracardiac and extracardiac anomalies that must be taken into consideration when imaging and planning the surgical procedure needed. Multiple anomalies are associated with TOF which calls for a checklist for Preoperative Assessment of TOF. These could be related to aortic arch, pulmonary arteries, system venous drainage, coronary artery anatomy, associated cardiac abnormalities and tracheobronchial anatomy.[1]

Non-invasive cardiac imaging plays a critical role in the initial diagnosis and follow-up of TOF patients which is the most common cyanotic congenital heart disease. Echocardiography is the initial modality of choice for making the diagnosis and follow-up. However, multi-detector computed tomography (MDCT), with its high spatial and temporal resolution, provides detailed depiction of cardiac anatomy and morphology playing an important role in the evaluation of complex anatomical findings in both treated and untreated TOF patients. In addition, performing and interpreting cardiac CT examination in unrepaired TOF is essential to guide the surgical intervention needed for repair as well as depicting any associated anatomic variants. Assessing the anatomy of pulmonary arteries play an important role in order to decide for palliative or correctible surgery.[2]

In TOF, anomalous origin of LPA is more common as compared with that of RPA.[3][4]

Anomalous right or left pulmonary artery arising from the aorta are relatively more rare congenital heart disease entities.[5]

Anomalous origin of the right pulmonary artery from the ascending aorta (hemitruncus arteriosus) is among the very rare congenital anomalies constituting about 0.1% of all cardiac birth defects. A partial or complete developmental failure of the left sixth arch is the underlying etiology of this condition resulting into left to right shunt. The child presents with congestive cardiac failure and onset of early pulmonary hypertension. Early corrective surgery is the definitive treatment of choice to reduce mortality rate.[6]

Right sided aortic arch (RAA) is a rare vascular malformation where the aortic arch crosses over the right bronchus instead of the left. It occurs in 0.1% of pregnancies and accompanies other CHD, most commonly with tetralogy of Fallot. When isolated, RAA does not usually produce symptoms in patients. However, when co-existing with other vascular anomalies, it may result in respiratory symptoms owing to compression of the trachea.[7]

Tracheal bronchus (TB) is a rare congenital tracheal anomaly, which is defined as the presence of an ectopic bronchus arising from the lateral wall of the trachea and supplies the right upper lobe with prevalence between 0.9% and 3%. The majority of TB cases are asymptomatic and diagnosed incidentally by advanced chest imaging or bronchoscopy.

In the setting of tracheo-bronchial branching abnormalities (TBAS), it is worth mentioning that endotracheal intubation merits special consideration. It is often accompanied by other congenital malformations, most commonly laryngomalacia, tracheomalacia, tracheal stenosis, congenital heart disease, and genetic disorders.

Misplacement of the endotracheal tube can occlude the tracheal bronchus (TB) lumen, resulting in atelectasis, hypoxia, or even respiratory insufficiency. Hence, knowledge of tracheal bronchus anatomy is essential perioperatively to take the necessary precautionary measures and avoid complications. Thus association of TB anomalies with congenital heart disease should alert physicians and provide timely management when needed.[8]

An association between tracheobronchial branching abnormalities and TOF has been less reported till date. TBAs are rare congenital anomalies that occur in 0.1% to 2% of the general population. TBAs tended to be more frequent in patients with TOF (27%) than in patients with TOF-PA (pulmonary atresia) (12%). The association between conotruncal heart defects and TBAs possibly involves signaling pathways, such as sonic hedgehog, bone morphogenetic protein 4), and Noggin, which are known to be involved in both conotruncal septation and regulation of bronchial branching morphogenesis in mice.[9]

An azygous lobe is a congenital variation of the lung. It is present in 0.2–1.2% of the population. It can mimic various pathological conditions, which presents challenges during thoracic surgical interventions. In order to prevent misdiagnosis and unnecessary interventions, an understanding of the clinical and imaging features of the azygous lobe is important. Interestingly, we found that some cases with the azygous lobe also had other abnormalities including congenital heart disease, Down syndrome, and other respiratory malformations. Whether formation of azygous lobe is related to genetic variation is a question worthy of further exploration. Preoperative awareness of an azygous lobe is very important for physicians and thoracic surgeons.[10]

CONCLUSION

MDCT is being used with increasing frequency to evaluate patients with TOF due to its wide availability and high spatial and temporal resolution. It helps to assess complex anatomy and associated anomalies in unrepaired TOF patients and guide the surgical approach and type of surgery needed. Besides this it also helps in detecting various anatomic variants which may be incidental finding but are crucial to detect which may be important for surgery.

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