CASE REPORT

Case Report: A forty year-survivor of Tetralogy of Fallot with pulmonary atresia and chronic pediatric shunt thrombosis; findings from cardiac CT scan [version 1; peer review: 1 approved, 1 approved with reservations]

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Abstract

Here we illustrate a rare case of Tetralogy of Fallot (TOF) with pulmonary atresia in a 40-year-old survivor patient, despite a chronic pediatric shunt thrombosis. The patient became symptomatic at 38-years-old with progressive dyspnea on exertion and short cyanosis spells. The clinical findings were unspecific except for sinus tachycardia. To reassess cardiac abnormalities and associated intrathoracic malformations, a cardiac CT-scan was performed. Here we explain the mechanism for prolonged survival as an expansion of several and huge major aortopulmonary collateral arteries. This case predicts an exceptional late outcome of untreated TOF.

Keywords

Computed tomography, cardiac imaging, Tetralogy of Fallot, Pulmonary atresia, Blalock Taussig shunt.

Open Peer Review

Approval Status

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25 Jun 2020	view	view

1. **Jean-Pierre Laissy** , Assistance Publique - Hôpitaux de Paris, Paris, France

2. **Hanne B Ravn**, Copenhagen University Hospital, Rigshospitalet, Denmark

Any reports and responses or comments on the article can be found at the end of the article.

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Author roles: **Asma A:** Conceptualization, Formal Analysis, Funding Acquisition, Investigation, Methodology, Visualization; **Mnari W:** Supervision, Validation; **Abdelali M:** Visualization; **Zrig A:** Supervision, Validation; **Miladi A:** Visualization; **Ben Messaoud M:** Validation; **Hmida B:** Visualization; **Maatouk M:** Supervision, Validation

Competing interests: No competing interests were disclosed.

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Introduction

Tetralogy of Fallot (TOF) is the most frequent form of cyanotic congenital heart disease. Only a few patients come of age without surgical intervention mainly in the extreme form with pulmonary atresia¹. Here, we relate the case of a patient who survived until the age of 40 years without surgical curative intervention. The patient had an unusual association of chronic Blalock-Taussig shunt (BTS) thrombosis and huge major aortopulmonary collateral arteries (MAPCAs). This case provides the main role that a cardiac CT scan can lay in understanding the late outcome of these untreated cardiac abnormalities.

Case report

A 40-year-old man with “complex” cardiac disease and dyspnea was referred to our department for imaging. Old medical records with conventional angiographic studies inferred the diagnosis of TOF with pulmonary atresia, made at birth, treated by BTS. As the patient was asymptomatic, he had not consulted previously, and had been lost to follow-up. Two years ago, he began to present dyspnea on exertion, and also short cyanosis spells.

Clinical findings on presentation were unspecific except for sinus tachycardia. ECG showed biventricular hypertrophy and incomplete right bundle branch block. Echocardiography confirms the diagnosis of TOF with pulmonary atresia. To reassess cardiac abnormalities, primarily pulmonary trunk morphology and associated intrathoracic malformations, a 128-slice CT scan with ECG synchronization was performed. The scan revealed a large ventricular septal defect with overriding aorta (Figure 1, curved arrow) and right ventricle hypertrophy (Figure 1, asterisks). The pulmonary trunk was atretic, showing characteristic seagull pattern (Figure 2, arrows). The right pulmonary artery had a good diameter, while the left was significantly smaller (Figure 2). Pulmonary blood flow was given by MAPCA connecting blood vessels between the aorta and the pulmonary arteries (Figure 3, arrows). MAPCA was more numerous on the right side supplying the largest pulmonary artery. The BTS was completely thrombosed with a total heterogeneous filling defect and parietal calcifications (Figure 4, arrows); this is compatible with chronic thrombosis.

Figure 1. Four-chamber view from cardiac CT scan with ECG synchronization visualizing ventricular septal defect with overriding aorta and RV hypertrophy. Curved arrow shows the ventricular septal defect; arrows show the overriding Ao; and asterisks show the RV hypertrophy. LA, left atrium; RA, right atrium; LV, left ventricle; RV, right ventricle; Ao, aorta.

Figure 2. Multiplanar reconstruction image from a cardiac CT scan with ECG synchronization showing an atretic pulmonary trunk and hypoplasia of the LPA. Arrows shows the atretic pulmonary trunk. LPA, left pulmonary artery; RPA, right pulmonary artery.

Figure 3. Cardiac CT scan with ECG synchronization visualizing collateral blood supply of the lungs showing coronal-targeted maximum intensity projection (C1) and posterior volume rendering (C2) views of the heart and mediastinal great vessel. Arrows show major aortopulmonary collateral arteries.

Figure 4. Multiplanar reconstruction of CT scan images showing a chronic thrombosis of the Blalock-Taussig shunt from the left subclavian artery to the ipsilateral pulmonary artery. Arrows show a total heterogeneous filling defect of the tube with parietal calcifications.

The mechanism for prolonged survival, despite the old shunt thrombosis, was the expansion of several and huge MAPCA allowing sufficient pulmonary blood flow. Two months after his hospitalization, the patient became asymptomatic, receiving life-long conservative therapy including anticoagulation with a vitamin K antagonist (acenocoumarol 4 mg once a day) and a low dose of diuretic (Furosemide 40 mg once a day).

Discussion

TOF is the most frequent cyanotic congenital heart disease. It includes four characteristic anatomical findings: sub-arterial ventricular septal defect, aorta straddling of the ventricles, a variable obstruction through the pulmonary outflow tract, and right ventricle hypertrophy². The spectrum of severity within this lesion depends upon the anatomy of the pulmonary arteries. The different forms of TOF include TOF with a patent foramen oval/atrial septal defect, TOF with absent pulmonary valve, and TOF with pulmonary atresia, which is the extreme form characterized by absence of flow from the right ventricle to the pulmonary arteries¹. In the latter event, pulmonary vascularization is derived from patent ductus arteriosus and aortopulmonary collaterals with significant variability from one patient to another².

MAPCAs are arteries that grow to irrigate lung circulation when native pulmonary arteries are underdeveloped. MAPCAs often arise from the descending aorta but also from the aortic arch and other systemic arteries like subclavian, the carotid, or rarely the coronary arteries. Pulmonary atresia-ventricular septal defects are classified into 3 types³: type A is associated with presence of the native pulmonary arteries with a pulmonary-aortic duct; type B has MAPCAs and native pulmonary arteries, such as in our case; type C has only MAPCAs, which are seen to provide the pulmonary blood without native pulmonary arteries.

Adulthood clinical presentation, as in our case, is extremely rare. Without surgical intervention, most patients die at a young age with a rate of survival of 66% at one year of age, 11% at 20 years, and 3% at 40 years. In contrast, the survival rate after surgical treatment is over 90% at 40 years old². The surgical option includes palliative forms, such as BTS, which consists in the creation of systemic to pulmonary shunt. Complete repair is the main option and associated with excellent outcome results⁴. In this case, despite a ductus arteriosus closure and a chronic pediatric shunt thrombosis, the patient survived at the age of 40 due to large and developed MAPCAs. The oldest survivor ever reported in the literature is 59 years old⁵.

After a cardiac ultrasound, multidetector angiography CT scan is indicated to the assessment of TOF with pulmonary atresia mainly for associated intrathoracic malformations and to establish the precise MAPCA cartography before curative surgery^{6,7}. Reformatted images allow a comprehensive analysis of pulmonary artery anatomy, measurements of the lumen of ascending aorta, analysis of the origins and course of coronary arteries, study location, the size, and flow in prior shunts⁸. The excellent spatial resolution and fast acquisition of multidetector CT make it a prime imaging tool to demonstrate even distal aortopulmonary collateral anatomy. Radiation should be reduced as low as possible in the pediatric population.

Conclusion

Untreated pulmonary atresia with a ventricular septal defect is uncommon in adults. Most patients die from serious respiratory troubles or congestive cardiac failure very early. An angiography CT scan is the main imaging tool to delineate the pulmonary arterial supply, which is essential for an appropriate surgical approach.

Consent

Written informed consent for publication of their clinical details and/or clinical images was obtained from the patient.

Data availability

All data underlying the results are available as part of the article and no additional source data are required.

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Hanne B Ravn

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Implications of MAPCAS detected this late after the initial event are missing. Data on vital values, including resting oxygen saturations, physical capacity, biventricular function and implications of a long-lasting systemic pressure in the pulmonary circulation are missing. Supposedly, the patient has an Eisenmenger equivalent condition. Apart from the fact that patient is a rarity, there is not much to be appreciated from the case-report. A reference for the survival data in untreated TOF patients is warranted.

Is the background of the case's history and progression described in sufficient detail?

Partly

Are enough details provided of any physical examination and diagnostic tests, treatment given and outcomes?

Partly

Is sufficient discussion included of the importance of the findings and their relevance to future understanding of disease processes, diagnosis or treatment?

Partly

Is the case presented with sufficient detail to be useful for other practitioners?

No

Competing Interests: No competing interests were disclosed.

Reviewer Expertise: Critical care of patients with congenital heart disease

I confirm that I have read this submission and believe that I have an appropriate level of

expertise to confirm that it is of an acceptable scientific standard, however I have significant reservations, as outlined above.

Reviewer Report 13 July 2020

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Jean-Pierre Laissy

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General Comments

This case report describes CTA findings of a 40 year-old patient treated shortly after his birth by a Blalock-Taussig shunt for a tetralogy of Fallot, without use of any exam between the postoperative period and the actual CT. One of the main drawbacks of the BTS is the absence of growth and stretching, so thrombosis is very frequent. Fortunately, the patient developed large systemic to pulmonary arterial anastomoses, classifying the disease as a type B.

The authors state that pulmonary atresia is untreated; this is partially true since BTS was used to compensate pulmonary artery flow from subclavian artery; this is mentioned in the Introduction ("without surgical curative intervention")

Some sentences are unclear and should be rewritten. In particular all the verbs should be in the preterit mode.

Specific comments

Abstract

Last sentence "This case predicts an exceptional late outcome of untreated TOF" is a wrong affirmation since BTS is one of the treatment options as described below.

Case report

Second para. In what way sinus tachycardia is specific?

Line 8. "CT" should be preferred to "The scan"

Line 9. What do the authors mean by "overriding aorta"?

Line 10. "The right pulmonary artery had a good diameter should read "The right pulmonary artery had a good diameter".

Line 12. "MAPCA were more numerous"

Line 16 "the patient becom" should read "the patient became"

Parts of the following sentences should be reformulated such as: "pulmonary flow was given...", "this is compatible with chronic thrombosis " "receiving life-long conservative therapy"...

Discussion

Line 4. "The different forms of TOF include TOF" should read "The different forms include TOF"

Line 9. One should read "other systemic arteries like subclavian, carotid, or rarely coronary arteries. ".

Line 18. The authors mention "In this case, despite a ductus arteriosus closure" Was it

spontaneous or under surgery?

The following sentence is difficult to understand and should be rewritten : "After cardiac ultrasound evaluation, multidetector CT angiography is useful to assess..."

Line 26. What means "study location,"?

References

OK.

Illustrations

OK.

Fig 2 should read "Multiplanar reconstruction image"; the following "from a cardiac CT scan with ECG synchronization" should be deleted.

Is the background of the case's history and progression described in sufficient detail?

Yes

Are enough details provided of any physical examination and diagnostic tests, treatment given and outcomes?

Yes

Is sufficient discussion included of the importance of the findings and their relevance to future understanding of disease processes, diagnosis or treatment?

Yes

Is the case presented with sufficient detail to be useful for other practitioners?

Yes

Competing Interests: No competing interests were disclosed.

Reviewer Expertise: Cardiac imaging

I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard.

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