

Adult with uncorrected Tetralogy of Fallot, anomalous coronary artery origin, left ventricular non-compaction, atrial septal defect and recurrent right ventricular outflow tract stenosis

Dorośla z Tetralogią Fallota, anomalią odejścia tętnicy wieńcowej, niescaleniem lewej komory, ubytkiem w przegrodzie międzyprzedsionkowej i nawracającym zwężeniem w drodze odpływu prawej komory

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Abstract

Advances in therapeutical possibilities for patients with complex congenital heart defects are unquestionable. Nonetheless, it is still probable to encounter unique challenges.

The study presents a case of a symptomatic 25-year-old female patient with uncorrected Tetralogy of Fallot who has never been qualified for surgery due to an unusual constellation of cardiac congenital comorbidities: anomalous coronary artery origin and non-compaction of the left ventricle.

Key words: Tetralogy of Fallot, anomalous coronary artery origin, non-compaction, uncorrected

Folia Cardiologica 2023; 18, 1: 38–41

Introduction

Tetralogy of Fallot (ToF) is the most common complex cyanotic congenital heart defect observed in humans [1]. It contains ventricular septal defect (VSD), pulmonary stenosis, overriding aorta and right ventricular (RV) hypertrophy. Typically, it is diagnosed prenatally or soon after birth due to

the quick presentation of desaturation and cyanosis. Proper diagnosis is usually followed by anatomical correction – the open-heart cardiac surgery as the natural history is linked with a bad prognosis. Untreated ToF results in progressive hypertrophy of the RV, increasing desaturation due to right-to-left shunt through the VSD and development of congestive heart failure (HF) [2].

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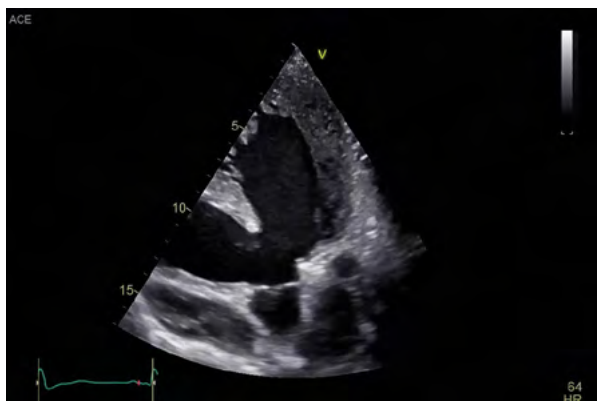


Figure 1. Modified apical view – visible features of Tetralogy of Fallot: large ventricular septal defect with the overriding aorta and visible features of left ventricle non-compaction

Case report

The 25-year-old female patient with uncorrected ToF with a history of serial pulmonary balloon valvuloplasties providing temporary reduction of the RV overload was admitted to the study department due to exacerbation of the right-sided HF – up to New York Heart Association (NYHA) III class, central cyanosis and irregular heartbeat. According to previous data anomalous single coronary artery origin and non-compaction of the left ventricle (LV) were the main contraindications for repair surgery in extracorporeal circulation in early life. At the time of admission, the patient was cyanotic, with maximum oxygen saturation (SatO₂) of 86% at rest while breathing room air, with blood pressure of 110/60 mm Hg and heart rate of 60 bpm. A loud systolic murmur was heard over the pulmonary valve and Erb's point.

During the 6-minute walk test (6MWT) the patient reached a distance of 500.5 m but with ongoing dyspnoea and severe desaturation (SatO₂ 74%). Lab tests revealed an increase of plasma N-terminal prohormone of brain natriuretic peptide (NT-proBNP) level – 2320 pg/mL. In Holter-ECG monitoring ventricular multifocal arrhythmia with several episodes of non-sustained ventricular tachycardia were observed. Echocardiographic examination revealed enlarged and hypertrophic LV with prominent features of non-compaction with preserved systolic function with LV ejection fraction (EF) of 65% and reduced global longitudinal strain (GLS: -15.7%), normal parameters reflecting RV function: tricuspid annular plane systolic excursion 28 mm, peak systolic tricuspid annular velocity (RV S') 11 cm/s, hypertrophic perpendicular RV with narrowed outflow tract, significant complex pathology of pulmonary valve (PV) after 3 balloon valvuloplasties (last one in 2015) – recurrent high grade stenosis with mild pulmonary regurgitation (PV max velocity: 5.38 m/s, peak gradient: 116 mm Hg, mean

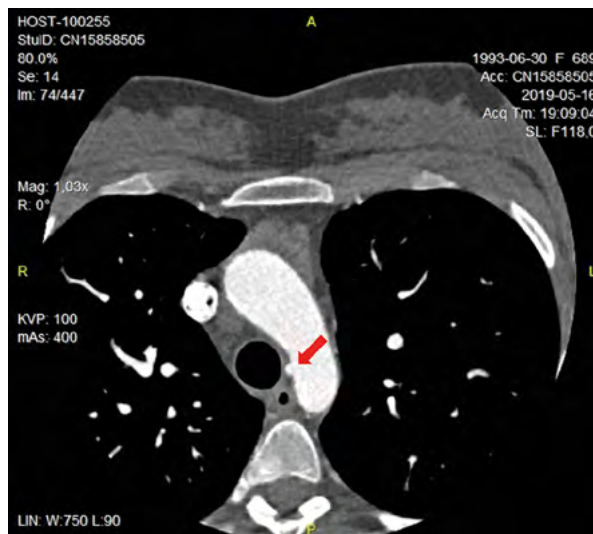


Figure 2. Computed tomography angiography scan – the visible anomalous origin of a single coronary artery in the aortic arch (red arrow)

gradient 57 mm Hg, PHT [pressure half-time] 200 ms), mild tricuspid regurgitation, non-collapsible inferior vena cava (estimated RV systolic pressure 40 mm Hg), overriding aorta without significant pressure gradients, large ventricular septal defect (26 mm) with bidirectional flow, small atrial septal defect, small additional vessel of unknown origin between aortic arch and pulmonary trunk (Figure 1).

Further therapeutic decisions were postponed after the completion of the scheduled multimodality approach. The hybrid-computed tomography angiography and magnetic resonance imaging (angio-CT/MRI) scans of the heart, great vessels and coronary arteries were obtained. MRI revealed disproportion of pulmonary and aortic inflow volume (168 and 57 mL respectively), severe subvalvular pulmonary stenosis with increased velocity (480 cm/s), hypoplastic RV with reduced function (EF 41%, SV 28 mL) and enlarged left chamber and atrium with preserved EF (EF 64%, SV 207 mL). Angio-CT scan revealed VSD with the size of 37 × 34 mm, tri-commissural bicuspid pulmonary valve, dilated pulmonary trunk (38 × 33 mm), proximally stenotic left pulmonary artery (diameters: 14 × 10 mm) with post-stenotic dilation (diameters: 36 × 32 mm), dilated right pulmonary artery (36 × 30 mm), optimal dimensions of ascending aorta, small patent ductus arteriosus with a pinpoint lumen. What is more, a single coronary vessel (diam. 3.0–3.5 mm) with the origin on the right side of the aortic arch was found (Figure 2). In its proximal section, it was laying between the curvature of the aortic arch and the superior wall of the left pulmonary artery (at this level 70% area reduction was observed due to compression by great vessels). In the distal part, it was coming between the pulmonary trunk and ascending aorta

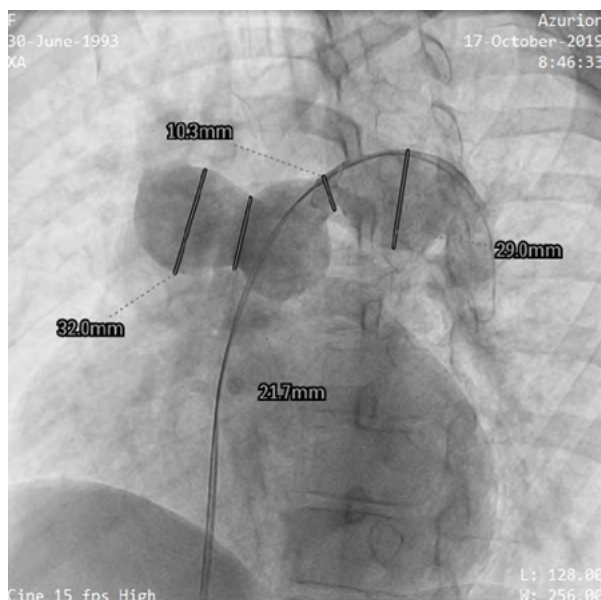


Figure 3. Cardiac angiography – visible pulmonary artery bifurcation with visible left branch artery stenosis

without compression reaching the anterior interventricular groove. In its later part, it was expanding into a wide vascular sinus (10 × 5 mm) connected with the lumen of the RV and heading towards the apex, splitting in the end into 3 vessels supplying the RV myocardium and the apical segments of the LV. The heart catheterization procedure was scheduled and performed with general anaesthesia. The examination revealed left pulmonary branch artery stenosis and supravalvular stenosis of the pulmonary valve. After additional heart team assessment, the left pulmonary branch artery angioplasty procedure was performed with simultaneous CP 34 mm stent implantation with a good final result (Figure 3 and 4). During the periprocedural period, the patient reported a short episode of chest pain, but no troponin elevation, nor electrocardiographic changes were observed.

The echocardiography was performed just after the procedure revealed an immediate reduction of pressure gradients in the RV outflow tract (PV max velocity: 4.14 m/s, peak gradient 68 mm Hg, mean gradient 43 mm Hg). The plasma level of NT-proBNP dropped to 1102 pg/mL. At discharge, the patient’s symptoms were of II NYHA class. However, the second Holter-ECG performed a few days after the catheterization revealed an increased number of premature ventricular beats (from 1198/day up to 9198/day). It could be due to the left pulmonary artery stent and anomalous coronary artery proximity, but no further ischemic symptoms were observed. Nonetheless, another heart catheterization was scheduled in 6 months to prepare the patient for a plausible heart transplant as future exacerbation of HF was inevitable. However, after a few

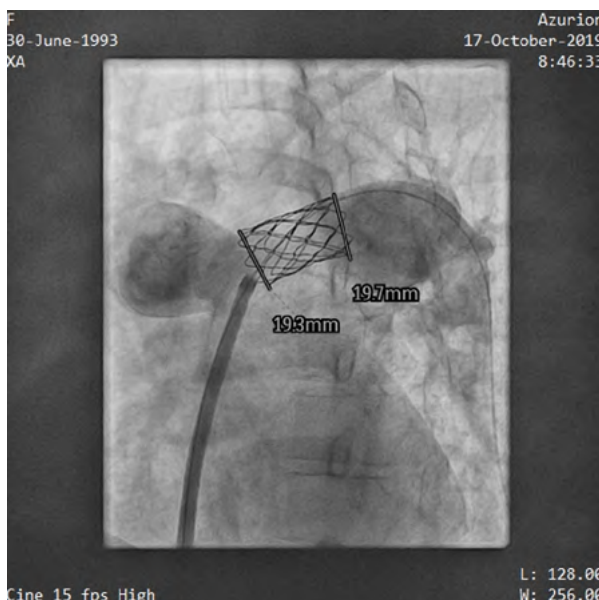


Figure 4. Cardiac angiography – visible stent in left pulmonary branch artery after successful pulmonary balloon angioplasty

months of follow-up, the patient was still in II NYHA class, feeling much better than before the described procedure.

Discussion

Patients with uncorrected complex congenital heart defects who reach adulthood are out of any guidelines as they are not common due to unfavourable natural history [3, 4]. What is more, most of the survivors do not belong to any homogenous group. Each case is a genuine challenge for physicians and the healthcare system as usually a late correction is not a viable option. When such a patient is encountered, it is necessary to perform a full examination to determine the patient’s condition. If any instability is observed, all accessible diagnostic modalities must be taken into consideration as any deterioration may be reversible if the direct cause is detected and treated properly. For the presented patient, the culprit was pulmonary branch artery stenosis affecting the haemodynamics between the ventricles, which was depicted as significant only after a combination of myocardial CT/MRI scan and cardiac angiography.

Pulmonary branch artery stenosis is a common finding in patients with congenital heart defects [5]. It may be diagnosed as a congenital comorbidity but also as an acquired disease complicating the initial therapeutic approach. It is a life-threatening condition leading to cardiopulmonary ventilation/perfusion mismatch and increased overload of the pulmonary circulation and RV predisposing to congestive HF [6]. Narrowing of the pulmonary branch artery may also alter existing shunts – in the case of VSD, the flow is prone

to be bidirectional or right-to-left, especially during exercise. Proper management requires optimal visualisation of the stenotic vessel and surrounding structures to choose adequate and safe interventional therapy. The method of choice is balloon angioplasty of the stenotic pulmonary branch artery with simultaneous implantation of stent selected in advance [7]. The outcome is usually good as ventilation/perfusion mismatch is reduced although restenoses may be observed during long-term follow-up thus such patients should be considered candidates for life-long specialistic grown-up congenital heart care [8].

On the other hand, patients unfit for full correction of the defect may also develop severe HF without any curable causes. Optimal management is based on life-long care and regular check-ups providing continuous observation of the severity of the symptoms. Patients in the II NYHA class and those without any significant ventricular tachyarrhythmias may persist in such an approach. However, any unexplained deterioration or new dangerous arrhythmias may be the landmark for considering intracardiac devices and heart transplantation qualification [9]. Cardiac transplantation in patients with congenital heart defects is associated with higher risk than in the general population, but

eligible candidates may benefit from longer life with preserved quality [10]. It is a great challenge to determine the perfect moment for such intervention, especially without typical criteria of ventricular insufficiency and with altered morphology of the native heart.

Conclusion

Patients with complex uncorrected congenital defects who develop progressive symptoms of HF require a full multi-modality approach as repairable structural lesions such as pulmonary arteries stenoses may be the culprits. Even patients with such anatomical cardiac odds as presented may be responders for specific interventions. Lifelong care in specialistic grown-up congenital heart centres should be provided to every patient with uncorrected defects. Proper monitoring at follow-up and cooperation with the patients being aware of their condition are key factors in terms of upcoming diagnostic and therapeutic decisions.

Conflict of interest

None declared.

Streszczenie

Postępy w możliwościach terapeutycznych dla pacjentów ze złożonymi wrodzonymi wadami serca są nie do zakwestionowania. Jednakże wciąż istnieje prawdopodobieństwo napotkania wyjątkowych wyzwań. W niniejszej pracy zaprezentowano przypadek objawowej 25-letniej pacjentki z nieskorygowaną Tetralogią Fallota, która nigdy nie była zakwalifikowana do operacji z powodu unikatowej konstelacji wrodzonych schorzeń serca: anomalii odejścia tętnicy wieńcowej i kardiomiopatii z niescalenia lewej komory.

Słowa kluczowe: Tetralogia Fallota, anomalia odejścia tętnicy wieńcowej, niescalenie, nieskorygowana

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References

1. Apitz C, Webb GD, Redington AN. Tetralogy of Fallot. *Lancet*. 2009; 374(9699): 1462–1471, doi: [10.1016/S0140-6736\(09\)60657-7](https://doi.org/10.1016/S0140-6736(09)60657-7), indexed in Pubmed: [19683809](https://pubmed.ncbi.nlm.nih.gov/19683809/).
2. Bourland BJ, McNamara DG. Tetralogy of Fallot: natural course, indications for surgery, and results of surgical treatment. *Cardiovasc Clin*. 1970; 2(1): 195–209, indexed in Pubmed: [4106588](https://pubmed.ncbi.nlm.nih.gov/4106588/).
3. Stout KK, Daniels CJ, Aboulhosn JA, et al. 2018 AHA/ACC guideline for the management of adults with congenital heart disease: executive summary: a report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. *J Am Coll Cardiol*. 2019; 73(12): 1494–1563, doi: [30121240](https://doi.org/10.1016/j.jacc.2018.08.005), indexed in Pubmed: [30121240](https://pubmed.ncbi.nlm.nih.gov/30121240/).
4. Baumgartner H, De Backer J, Babu-Narayan SV, et al. 2020 ESC Guidelines for the management of adult congenital heart disease. *Eur Heart J*. 2021; 42(6): 563–645, doi: [10.1093/eurheartj/ehaa554](https://doi.org/10.1093/eurheartj/ehaa554), indexed in Pubmed: [32860028](https://pubmed.ncbi.nlm.nih.gov/32860028/).
5. Meot M, Lefort B, El Arid JM, et al. Intraoperative stenting of pulmonary artery stenosis in children with congenital heart disease. *Ann Thorac Surg*. 2017; 104(1): 190–196, doi: [10.1016/j.athoracsur.2016.12.012](https://doi.org/10.1016/j.athoracsur.2016.12.012), indexed in Pubmed: [28274523](https://pubmed.ncbi.nlm.nih.gov/28274523/).
6. Vida VL, Rito MLo, Zucchetta F, et al. Pulmonary artery branch stenosis in patients with congenital heart disease. *J Card Surg*. 2013; 28(4): 439–445, doi: [10.1111/jocs.12121](https://doi.org/10.1111/jocs.12121), indexed in Pubmed: [23718834](https://pubmed.ncbi.nlm.nih.gov/23718834/).
7. Zablah JE, Morgan GJ. Pulmonary artery stenting. *Interv Cardiol Clin*. 2019; 8(1): 33–46, doi: [10.1016/j.iccl.2018.08.005](https://doi.org/10.1016/j.iccl.2018.08.005), indexed in Pubmed: [30449420](https://pubmed.ncbi.nlm.nih.gov/30449420/).
8. Hiremath G, Qureshi AM, Prieto LR, et al. Balloon angioplasty and stenting for unilateral branch pulmonary artery stenosis improve exertional performance. *JACC Cardiovasc Interv*. 2019; 12(3): 289–297, doi: [10.1016/j.jcin.2018.11.042](https://doi.org/10.1016/j.jcin.2018.11.042), indexed in Pubmed: [30732734](https://pubmed.ncbi.nlm.nih.gov/30732734/).
9. Burchill LJ. Heart transplantation in adult congenital heart disease. *Heart*. 2016; 102(23): 1871–1877, doi: [10.1136/heartjnl-2015-309074](https://doi.org/10.1136/heartjnl-2015-309074), indexed in Pubmed: [27836946](https://pubmed.ncbi.nlm.nih.gov/27836946/).
10. Houyel L, To-Dumortier NT, Lepers Y, et al. Heart transplantation in adults with congenital heart disease. *Arch Cardiovasc Dis*. 2017; 110(5): 346–353, doi: [10.1016/j.acvd.2017.01.002](https://doi.org/10.1016/j.acvd.2017.01.002), indexed in Pubmed: [28237697](https://pubmed.ncbi.nlm.nih.gov/28237697/).