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REVIEW

Isolated hypoplastic right ventricle – a challenge in medical practice

Eliza Elena Cinteză[1,2], Alin Marcel Nicolescu[2], Mihaela Adela Iancu[3], Gabriela Ganea[2], Matei Dumitru[3,4], Gheorghe Gindrovel Dumitra[5]

[1]Department of Pediatrics, Carol Davila University of Medicine and Pharmacy, Bucharest, Romania

[2]Department of Pediatrics, Maria Skłodowska Curie Emergency Clinical Children's Hospital, Bucharest, Romania

[3]Department of Internal Medicine, Family Medicine and Labor Medicine, Carol Davila University of Medicine and Pharmacy, Bucharest, Romania

[4]Department of Pediatrics, Alessandrescu–Rusescu National Institute for Mother and Child Health, Bucharest, Romania

[5]Department of Family Medicine, University of Medicine and Pharmacy of Craiova, Romania

Abstract

Isolated right ventricle hypoplasia (IRVH) is a disease characterized by an underdeveloped right ventricle. It is a congenital heart disease than can associate heterogeneous structural defects and nonspecific clinical features, which can often present a challenging therapeutic management. In this article, there are presented diagnostic methods and treatment options for right ventricle hypoplasia (RVH) according to clinical features, patients age and associated structural heart defects. RVH has a different prognosis in accordance with the severity of the heart defects and the patient's age at which the diagnosis is established. Thus, isolated forms of RVH generally present mild structural and functional defects that can be associated with the onset of symptoms in adolescence or even in adulthood. In these cases, atrial septal defect closure with or without superior cavo-pulmonary anastomosis can be the only procedures needed to correct the hemodynamic abnormalities and relief the symptomatology. Patients with severe form of RVH associated with complex cardiac malformations and onset of the symptoms in the neonatal period require prompt intervention and necessitate palliative procedures. In the long term, these patients could need multiple reinterventions. The family physician should be aware of the cardiac origin of isolated symptoms or clinical signs, such as exertional dyspnea or clubbing fingers, and send the patient for pediatric cardiological evaluation.

Keywords: right ventricle hypoplasia, atrial septal defect, cavo-pulmonary anastomosis, clubbing fingers, cyanosis.

Background

Right ventricle hypoplasia (RVH) is a rare congenital heart disease (CHD). There have been reported isolated cases, therefore the prevalence couldn't be determined yet. Based on scientific publications, this affliction has a prevalence lower than the occurrence of left ventricle (LV) hypoplasia (1:100 000) [1].

Isolated right ventricle hypoplasia (IRVH) is a rare non-cyanotic CHD characterized by the underdevelopment of the trabecular component of the right ventricle (RV) or the entire RV. Underdevelopment of the RV can also appear in pulmonary atresia with intact interventricular septum (PA–IVS). However, the difference between these two variants is that in PA–IVS, the underdeveloped portion of the RV is located at the outlet [2]. This anomaly was described for the first time in 1959 [3], considered initially as an Ebstein's anomaly (on clinic, electrocardiographic and angiographic findings). However, the diagnosis was not confirmed in the operating room. Also, it may be similar to Uhl anomaly, but the presence of myocardium at the RV level does not stand for this diagnosis. Associations with other cardiac structural defects as tricuspid and/or pulmonary valve atresia or stenosis, ventricular septal defect (VSD), coronary arteries development abnormalities can change the clinical presentation from non-cyanotic to cyanotic, however in these cases the RVH is only a component of a particular form of a complex CHD [4]. In general, IRVH is associated with atrial septal defect (ASD) or VSD. The shunt can be bidirectional or with an important right-to-left component.

Etiopathogenesis

The etiology of the RVH is unknown, but there have been reported a series of genetic factors that can be responsible for the etiopathogenesis of this disease [5–8]. In our experience that includes four cases, there is an association with a suspicion of a genetic cardiomyopathy (the father of the boy was diagnosed with cardiomyopathy, non-documented; parents are divorced) and of an association with autoimmune disease including autoimmune thyroiditis and celiac disease. According to reported cases, the means of transmission of this disease seems to be autosomal dominant [9]. Also, isolated hypoplasia of the RV was described in the descendants of people with the same affliction with autosomal dominant pattern of inheritance [2].

Clinical presentation

The clinical features are determined by the severity of the hypoplasia, right-to-left shunt amplitude through the

ASD and the associated structural heart disease. Cases of mild hypoplastic RV disease can be associated with late onset of the cyanosis and can be surgically corrected by ASD closure. Meanwhile, severe RVH associated with valvular abnormalities is characterized by early onset of cyanosis [10]. In two of our cases, considered mild forms, they presented with cyanosis at exertion and dyspnea.

Clinical features and the symptomatology onset are strongly correlated with the severity of the hypoplasia and the associated cardiac structural abnormalities. Patients with mild structural defect of the RV are associated with late onset of the symptoms in adolescence or even in adulthood and the clinical features are characterized by cyanosis and exertional dyspnea. Patients with severe form of RVH and other congenital structural abnormalities, may present congestive heart failure (right), with enlarged liver, dilated turgescent jugular veins and cyanosis in the neonatal period or in infancy [3].

If there is even the slightest degree of obstruction in the pulmonary circulation blood flow, the clinical presentation will later on associate clubbing fingers, central cyanosis and exertional dyspnea [11].

This was the situation of one of our patients that at the age of one-year-old presented with moderate dyspnea and central cyanosis accompanied by clubbing finger (in the presence of hypoplastic RV associated with large apical VSD, severe RV obstruction at the moderator band level, ASD, hypoplastic pulmonary arteries (PAs) and non-compacted LV cardiomyopathy). In case of severe obstruction of pulmonary blood flow in the neonatal period and in the presence of RVH, the patient may manifest respiratory distress syndrome with markedly cyanosis [12].

⊟ Paraclinical and imagistic investigations

To obtain a complete structural and functional evaluation, a wide variety of paraclinical and imagistic investigations are necessary: echocardiography, chest X-ray, computed tomography (CT), magnetic resonance imaging (MRI), electrocardiogram, angiography, and cardiac catheterization.

Ultrasound examination

The echocardiography usually shows a RV with three anatomical regions: inlet, trabecular component, and outlet, but with lower volumes and reduced or absent papillary muscles. RV is considered hypoplastic if the length from the tricuspid annulus to the RV apex is less than half the distance from the mitral valve to the LV apex [13].

RVH may occur from reducing the size of the cavity by a hypertrophic moderator band which divides the ventricular cavity, as in the case of our patient, a 7-year-old boy with hypoplastic RV and ASD (Figure 1a). The reduction in size of the RV cavity may also occur because of the LV apex hypertrophy.

Occasionally, the tricuspid valve may appear dysplastic. From our four patients, only one had a dysplastic tricuspid valve. The lower the volume of the RV is, the more it is required to perform the superior cavo-pulmonary anastomosis procedure. A cardiac defect, usually at atrial level, is frequently associated with bidirectional shunt or right-to-left shunt as in the case of our patient, a 7-year-old boy (Figure 1b).
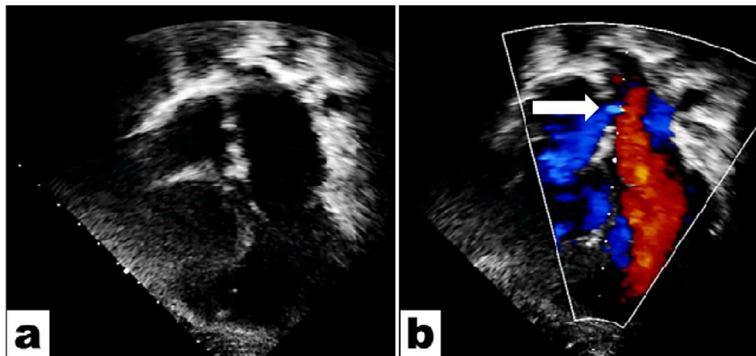

Figure 1 – Ultrasound from apical 4-chambers view. Hypoplastic RV and ASD. (a) Bidimensional evaluation showing a small RV comparing with the LV with increased systolic and diastolic pressure in the RV. (b) Color Doppler evaluation showing a bidirectional shunt at the atrial level (blue yellow color – white arrow). ASD: Atrial septal defect; LV: Left ventricle; RV: Right ventricle.

Chest X-ray

The chest X-ray may not show any obvious pathological change in relation to lower dimensions of the RV due to the overlapping projection of the cardiac silhouette on the diaphragm, as in the case of our patient, a 12-year-old girl, that was diagnosed at birth with ASD, VSD, mild hypoplastic RV, and operated for both defects at the age of four, eight years ago (Figure 2).

Enlargement of the right atrium (RA) may be evident on chest X-ray. The appearance of the pulmonary circulation on the chest X-ray is normal.

CT and MRI

CT and MRI are useful to specify the anatomical details of the RV – tripartite, bipartite, or unipartite RV, tricuspid valve anatomy, dimensions of overlying cavities and non-compacted appearance of the myocardium.

In a 2-year-old patient of ours, which presented with hypoplastic RV associated with ASD, large apical VSD, obstructive moderated band and noncompaction of the LV CT scan showed RV reduced dimension comparing with the LV (Figure 3, a and b).
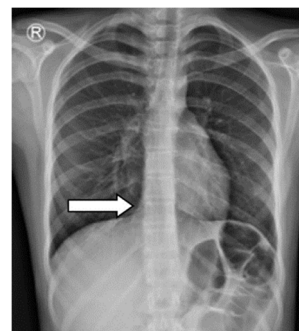

Figure 2 – Chest X-ray. Normal cardiac silhouette, and normal pulmonary vascularity, in a 12-year-old girl, after operation at the age of four (eight years ago) for ASD, VSD, mild hypoplastic RV (white arrow), diagnosed at birth. ASD: Atrial septal defect; RV: Right ventricle; VSD: Ventricular septal defect.

Moreover, MRI can highlight aspects regarding left and RV function. The imaging procedure highlighted, in a case of our patients (a 7-year-old boy) with hypoplastic RV and ASD, the recalibration of the ASD to a diameter of 5 mm after the superior cavo-pulmonary anastomosis (Figure 4, a and b).

CT and MRI can also determine other structural heart defects. Superior imagistic techniques may show also non-compaction of the LV, as in the case imaged in Figure 3b. The three patients in our follow-up that suffered cardiac surgical interventions were evaluated by angioCT and angioMRI before surgery.

Figure 3 – Hypoplastic RV (white arrow) associated with ASD, large apical VSD, obstructive moderated band, noncompaction of the LV. CT scan: (a) Cross-sectional view; (b) Coronal view. ASD: Atrial septal defect; CT: Computed tomography; LV: Left ventricle; RV: Right ventricle; VSD: Ventricular septal defect.
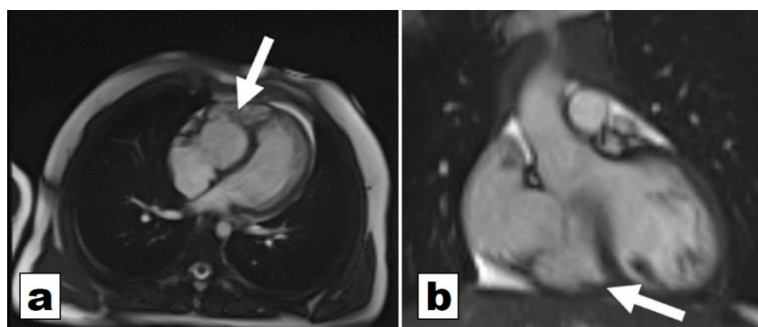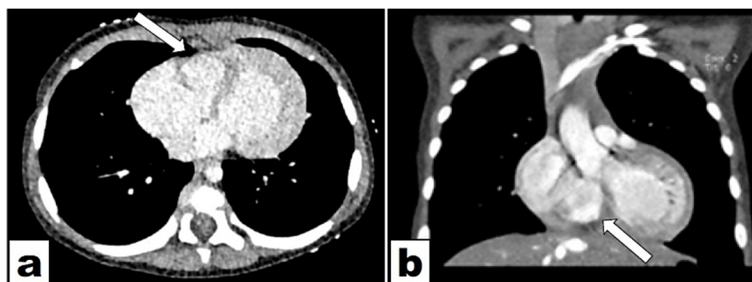

Figure 4 – MRI scan. Hypoplastic RV (white arrow) and ASD after superior cavo-pulmonary anastomosis with recalibration of the ASD. (a) Cross-sectional view; (b) Coronal view. ASD: Atrial septal defect; MRI: Magnetic resonance imaging; RV: Right ventricle.

Electrocardiogram

The electrocardiogram may show dominance of the left leads, with left deviation of the QRS axis. Also, tall and peaked P wave in D1, V1, V2 were described associated as an expression of the elevated pressure in the RA to elements of LV hypertrophy [3, 14].

Angiography

Although the transition from angiography to echocardiography or MRI is noticeable the combining multimodalities to assess isolated RVH hemodynamics and morphology may be important in the diagnosis and therapeutic management of each patient [15].

Cardiac catheterization

Cardiac catheterization is extremely useful. In some cases, this investigation has set the therapeutic management and established the need for surgical correction. Sizing balloon occlusion test is important to evaluate the response of the ventricle after increasing the preload. The risk of contrast agents is not negligible, especially in patients with significant RVH who will undergo cardiac surgery with cardiopulmonary bypass [16]. Pressure evaluation in cardiac chambers may reveal, elevated "a" wave at RA level, due to resistance to flow in the presence of a small RV. ASD closure was the surgical method chosen for patients with appropriate adaptation to increased preload. ASD closure was the only surgical correction needed in patients with RV dimensions at the lower limit of normal with quasi-normal systolic function. In these cases, it was noticed a relief of dyspnea and reversal of cyanosis [3, 9].

RV dimensions were the main criteria for choosing the surgical method. But, sizing balloon occlusion test during cardiac catheterization or surgical correction can provide important information regarding eligible patients for ASD closure [17]. Thus, ASD closure should only be done in patients that present a good adaptation of the right heart at increased preload.

⊡ Surgical methods

Therapeutic management will be established depending on clinical manifestations, associated structural abnormalities, hemodynamic conditions and not the least patient's age. In some author's opinion, if the patient is maintained asymptomatic, no intervention is needed [13].

Surgical correction with the preservation of both ventricles' function remains the ideal goal for maintaining the quality of life of patients with CHD. This outcome is not always possible due to morphological and functional features of the RV which cannot always support the entire preload and pump the blood flow to the pulmonary circulation. Therefore, there are surgical methods which preserve the function of both ventricles ("two ventricle repairs") and surgical methods which preserve the function of one ventricle ("one ventricle repair"). There is also a third category in which the RV presents morphological and/or functional defects and it cannot support the entire preload. Thus, a hypoplastic RV with a potential of use at a lower capacity will be kept at a lower level of function.

The surgical method "one and a half ventricular repair" (also known as partial biventricular repair) consists of performing a direct anastomosis between superior vena cava and right PA (bidirectional Glenn shunt or hemi-Fontan). Initially, there were two indications for this procedure – a small pulmonary ventricle and a dysfunctional, dilated RV. Later, indications were extended to more complex lesions, such as congenitally corrected transposition of the great vessels, Ebstein's anomaly, tricuspid straddling,

etc. [18]. This presents the advantage of maintaining a lower pressure in the RA, together with a pulsating flow in the PA and improves the systemic oxygen saturation level by bypassing the RV and redirecting the blood flow into the pulmonary circulation. This intervention was performed to the first case of IRVH reported in the literature, in 1959 [3, 19]. This procedure reduces the preload and end-diastolic RV pressure. This is an intermediate intervention before performing Fontan procedure where it is needed. Many clinics reported a lowering rate of morbidity and mortality in patients which received bidirectional Glenn shunt [20]. There have been reported cases in which Glenn procedure and ASD closure for maintaining the blood flow, similar to "one and a half ventricular repair", have had positive outcomes in patients with hypoplastic RV in association with an ASD [14]. There has been reported a case which has associated in addition to these two structural abnormalities, moderate pulmonary stenosis and a tricuspid valve dysplasia allowing the maintenance of a pregnancy after two anterior miscarriages [11]. Sometimes, the Glenn anastomosis can be followed by the maintenance of a restrictive ASD that has the role of decompressing the RA of the resistance generated by the small RV [3].

Surgical correction of the congenital cardiac malformation may assume also multiple laborious techniques for specific lesions. In case of the association of hypoplasia of the RV (with all three anatomical segments present) with VSD and pulmonary valve atresia, a pulmonary valvotomy inducing pulmonary regurgitation may be indicated as it was reported in literature [12, 21]. This method supports the development of the RV cavity, and it is an intermediate procedure for future surgical correction leading to a two-ventricle repair.

For patients with hypoplastic RV that associate severe structural abnormalities, total cavo-pulmonary anastomosis, Fontan procedure is the only solution [10]. The Fontan procedure is the final palliative surgical method for patients with congenital cardiac malformations which associate unique anatomical or functional ventricle. Following this procedure, the pulmonary circulation is totally separated from the systemic circulation by performing a total cavo-pulmonary communication using a conduct which connects inferior vena cava to the PA. Thus, the circulation becomes passive and dependent on pressure gradients [22].

In our experience, three of four patients suffered cardiac surgery. One patient had ASD and VSD closure, with a good subsequent evolution. Recently, the 12-year-old girl had an MRI evaluation which showed a mild reduction of ejection fraction of the RV, but at cardiopulmonary testing the result was normal. The 7-year-old boy with bidirectional ASD and hypoplastic RV (Figure 1, a and b; Figure 4, a and b) suffered cavo-pulmonary anastomosis and fenestrated ASD closure with a good evolution at the follow-up remaining asymptomatic after surgery. The 2-year-old girl with complex lesions (large apical VSD, severe RV obstruction at the moderator band level, ASD, hypoplastic PAs and non-compacted LV cardiomyopathy) associated to the hypoplastic RV suffered only cavo-pulmonary anastomosis (bidirectional Glenn shunt) with maintenance of the ASD and VSD. Her evolution initially was good, but, in the presence of the non-compaction LV, the prognosis is reserved. The fourth patient in our experience has a VSD associated to a mild hypoplastic RV. He is followed regularly being asymptomatic.

Occasionally, surgical corrections are contraindicated, especially when cardiac malformations are associated to cardiomyopathies such as LV noncompaction with significant impairment of systolic function of the LV. For such cases, repeated phlebotomies may be the solely therapeutical recommendation [23].

Short- and long-term prognosis is associated with the severity of structural abnormalities. The hemodynamic conditions represent the main factor for decision making process to establish the best therapeutic approach. Cardiac catheterization along with imaging investigations contribute to establish the surgical or interventional approach of isolated RV hypoplasia.

⊟ Conclusions

Patients with RVH can present with a wide spectrum of clinical manifestations from asymptomatic to severe respiratory distress and cyanosis in relation to the severity of the cardiac malformation and the association of other cardiac defects.

The surgical management of these patients is correlated with the hemodynamic response to sizing balloon occlusion test. Besides ASD closure, redirection of the systemic blood flow to the pulmonary circulation may be necessary by creating a superior cavo-pulmonary anastomosis-bidirectional Glenn shunt.

Severe cases of RV, which associate complex structural abnormalities, can be managed by performing the palliative cardiac surgery for unique function ventricle, the total cavo-pulmonary anastomosis, Fontan procedure.

The family physician should be aware of the cardiac pathology which include this rare anomaly of isolated hypoplasia of the RV and be aware of the associated of isolated cardiac symptoms of exertional dyspnea, clubbing fingers, which may appear in mild to moderated forms in adolescents or even adults.

Conflict of interests

The authors declare that they have no conflict of interests.

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Corresponding author
Mihaela Adela Iancu, Associate Professor, MD, PhD, Department of Internal Medicine, Family Medicine and Labor Medicine, Faculty of Medicine, Carol Davila University of Medicine and Pharmacy, 37 Dionisie Lupu Street, Sector 2, 020021 Bucharest, Romania; Phone +40745–031387, e-mail: adela.iancu@umfcd.ro