CASE REPORT

Isolated right ventricular cardiomyopathy with autoimmune hypothyroidism: a rare association in an adolescent

Kavita Yelve,¹ Gajanan Ashok Panandikar,² Amar Pazare,¹ Smrati Bajpai¹

¹Department of Medicine, Seth G S Medical College and KEM Hospital, Mumbai, Maharashtra, India

²Department of General Medicine, Seth G S Medical College and KEM Hospital, Mumbai, Maharashtra, India

Correspondence to

Dr Kavita Yelve,
kavitayelve@yahoo.com

Accepted 1 March 2015

SUMMARY

A 13-year-old girl presented with progressive dyspnoea and palpitation, diagnosed on echocardiography as primary right ventricular cardiomyopathy with atrial fibrillation. Her thyroid profile was positive for antithyroid microsomal antibody, and antithyroid peroxidase antibodies were suggestive of autoimmune hypothyroidism. She was managed with furosemide, digoxin, acenocoumarol and thyroxine following which she showed significant improvement. This is a rare case of isolated right ventricular cardiomyopathy and its association with autoimmune hypothyroidism presenting at the age of 13.

BACKGROUND

Isolated idiopathic right ventricular dilated cardiomyopathy (DCM) is rare and is a diagnosis by exclusion. There is a distinct male predominance. The usual clinical presentations are syncope and right heart failure. ECG may show ventricular tachycardia or left bundle branch block. Diagnosis is usually established based on clinical and laboratory parameters. Confirmation of diagnosis is carried out by studying the pathological features of the heart in an endomyocardial biopsy or at post-mortem.¹ Dilated cardiomyopathy (DCM) is recognised clinically by ventricular dilation associated with low ejection fraction on echocardiography. The left ventricle (LV) is the chamber that is frequently involved due to DCM.² However, there are a few reports of familial DCM affecting the right ventricle (RV) as well.³ This is a report of an uncommon association between isolated right ventricular cardiomyopathy and autoimmune hypothyroidism at an early age of presentation.

CASE PRESENTATION

We present a case of a 13-year-old girl, born of a third-degree consanguineous marriage, with no significant birth history and no history of maternal illness during pregnancy. The child was symptomatic since the age of 1 year with class 2 dyspnoea on exertion and now with progressive dyspnoea for 3 months, cough for 2 months and palpitations over the past 2 days. There was no history of squatting episodes, cyanotic episodes, joint pains, swelling, rash or rheumatic fever in the past. There was no history of sudden death of family members. The girl's pulse was 112 bpm, irregularly irregular, with blood pressure of 90/62 mm Hg in all four limbs. On general examination, she had facial puffiness

with a raised jugular venous pulse with CV complexes and pedal oedema without cyanosis or clubbing. On cardiovascular system examination, she had precordial bulge with pan systolic murmur in the tricuspid area. She also had bilateral basal crepitations and hepatomegaly.

INVESTIGATIONS

On investigating the patient, her baseline biochemical investigations were normal. Chest X-ray (figure 1) was suggestive of massive enlargement of the cardiac silhouette and rounding of left and right heart borders. ECG (figure 2) showed atrial fibrillation with RV enlargement with right bundle branch block (RBBB). Two-dimensional echocardiography showed primary RV cardiomyopathy with severe tricuspid regurgitation, compromised RV function and good LV function, suggestive of isolated RV cardiomyopathy. Cardiac MRI (figure 3) confirmed isolated RV cardiomyopathy with no evidence of arrhythmogenic RV cardiomyopathy (ARVC).

The patient's T3 was 69 ng%, with T4 of 8.2 ng % and thyroid-stimulating hormone (TSH) 11.71, suggestive of hypothyroidism. Antithyroid microsomal antibody titre was 1:3000 with antithyroid peroxidase antibody titre of suggestive of

Figure 1 Chest X-ray showing massive enlargement of cardiac silhouette, and rounding of left and right heart borders.

CrossMark

To cite: Yelve K, Panandikar GA, Pazare A, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2014-206268

Figure 2 ECG showing atrial fibrillation with right ventricular enlargement with right bundle branch block.

autoimmune hypothyroidism. Antinuclear antibody titre was normal and antidouble-stranded DNA antibody was negative. CPK-MB and CPK total were not elevated, ruling out collagen vascular disorder and associated myopathy.

A final diagnosis of isolated right ventricular cardiomyopathy with autoimmune hypothyroidism was made.

DIFFERENTIAL DIAGNOSIS

Considering the isolated involvement of the RV, the most probable differential was ARVC. However, absence of fibro-fatty inflammation affecting both the RV and LV on cardiac MRI

helped to rule out this entity. The patient also failed to satisfy Task Force Criteria for ARVC diagnosis.⁴

TREATMENT

The patient was given intravenous furosemide 40 mg two times a day, along with oral digoxin 0.25 mg and acenocoumarol 2 mg tablets. She was also started on oral thyroxine 50 µg daily.

OUTCOME AND FOLLOW-UP

The patient responded well to conservative management. Cardiac failure improved with controlled ventricular rate. On

Figure 3 Cardiac MRI showing markedly dilated right atrium and right ventricle due to right ventricular cardiomyopathy, and normal left ventricle. No evidence of arrhythmogenic right ventricular dysplasia is seen.

follow-up, the patient's thyroid function tests were normal. The echocardiography findings were unchanged. She was symptomatically better.

DISCUSSION

DCM is the most common form of cardiomyopathy and is characterised by increased myocardial mass and volume. The ventricular wall becomes thin and stretched out, thus compromising the cardiac contractility and resulting in poor ventricular function. The cardiac pathology in isolated RV cardiomyopathy is restricted to the right heart. Although there was thinning of the RV wall, the MRI of the RV wall shows the presence of myocardial fibres and no replacement by adipocyte. These MRI findings help to distinguish it from Uhl's anomaly and arrhythmogenic RV dysplasia/cardiomyopathy (ARVD/C), which involve pathogenic changes in the RV myocardium. ARVD/C and Uhl's anomaly are considered as different manifestations of the same disease.⁵ In Uhl's anomaly, there is virtually complete absence of the myocardium of the parietal wall of the RV, and the parietal wall is composed of the opposing endocardial and epicardial ventricular surfaces, with no fatty tissue interposed between these layers. These changes are congenital and diagnosed by fetal echocardiography. ARVD/C is characterised by patchy and localised replacement of the parietal wall of the RV by fibrofatty tissue. This adipose replacement occurs primarily within the ventricular outflow tract, inlet or apical regions, sometimes progressing to the LV. Isolated idiopathic RV DCM is rare and is a diagnosis by exclusion. The usual clinical presentations are syncope and right heart failure. ECG may show ventricular tachycardia and RBBB.

Our case of isolated RV DCM with its earlier age of presentation and its association with autoimmune hypothyroidism is distinct from other reported cases of isolated RV cardiomyopathy. Autoimmune hypothyroidism usually presents after the fifth decade. The most common reported cardiovascular manifestation is pericardial effusion (30%) secondary to the long-standing autoimmune hypothyroidism. Hypothyroidism can precipitate heart failure and the risk increases with age. This is in contrast to our patient, who had DCM and was symptomatic since childhood. Hypothyroidism may have precipitated cardiac failure in our patient. Although the exact mechanism leading to cardiomyopathy is not clear, thyroid hormone effects on the cardiac myocyte are intimately associated with cardiac function via regulation of the expression of key structural and regulatory genes.⁶ The myosin heavy chain genes encode the two contractile protein isoforms of the thick filament in the cardiac myocyte. The sarcoplasmic reticulum Ca^{2+} -ATPase and its inhibitor, phospholamban, regulate intracellular calcium cycling.

However, isolated RV affection without LV affection cannot be explained by these mechanisms in this case, hence it appears that isolated right cardiomyopathy and autoimmune hypothyroidism may be different entities in the same patient.

To date, very few cases of isolated RV cardiomyopathy have been reported, and none had autoimmune hypothyroidism. The mechanism behind the association is not clear at present.

Learning points

- Isolated idiopathic right ventricular dilated cardiomyopathy should be distinguished from other forms of right-sided cardiomyopathy.
- Association between isolated idiopathic right ventricular dilated cardiomyopathy and autoimmune hypothyroidism is rare. Although causality cannot be ascertained due to the rare nature of the disease, patients of right ventricular cardiomyopathy may be investigated for presence of autoimmune hypothyroidism, as it can precipitate cardiac failure.
- Patients respond well to symptomatic treatment but require regular follow-up due to risk of sudden cardiac death. Other family members should be screened for asymptomatic cardiomyopathy.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Luk A, Ahn E, Soor GS, *et al.* Dilated cardiomyopathy: a review. *J Clin Pathol* 2009;62:219–25.
- 2 Olsen EG. Pathology of primary cardiomyopathies. *Postgrad Med J* 1972;48:732–7.
- 3 Ibsen HH, Baandrup U, Simonsen EE. Familial right ventricular dilated cardiomyopathy. *Br Heart J* 1985;54:156–9.
- 4 McKenna WJ, Thiene G, Nava A, *et al.* Diagnosis of arrhythmogenic right ventricular dysplasia/Cardiomyopathy. Task Force of the Working Group Myocardial and Pericardial Disease of the European Society of Cardiology and of the Scientific Council on Cardiomyopathies of the International Society and Federation of Cardiology. *Br Heart J* 1994;71:215–18.
- 5 Pamuru PR, Dokuparthi MV, Remersu S, *et al.* Comparison of Uhl's anomaly, right ventricular outflow tract ventricular tachycardia (RVOT VT) & arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) with an insight into genetics of ARVD/C. *Indian J Med Res* 2010;131:35–45.
- 6 Klein I, Danzi S. Thyroid disease and the heart. *Circulation* 2007;116:1725–35.

Copyright 2015 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- Submit as many cases as you like
- Enjoy fast sympathetic peer review and rapid publication of accepted articles
- Access all the published articles
- Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow