

An Adolescent Asymptomatic Case with Uhl Anomaly

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ABSTRACT

Uhl anomaly is a very rare disease. Differential diagnosis are arrhythmogenic right ventricular cardiomyopathy and severe tricuspid regurgitation with right ventricular enlargement. Here we present a 18-year male patient without any complaints. The diagnosis was assessed by echocardiography and cardiac MRI imaging.

Keywords: Uhl Anomaly, Arrhythmogenic Right Ventricular Cardiomyopathy, Paper-Thin Wall, No Right Ventricular Aneurysms.

CASE

An adolescent 18-year old male patient presented in the Medical Care Unit for a certification to play professional handball. Chest pain, exertional dyspnoea, and arrhythmias were denied.

His ECG revealed sinus rhythm, low voltage in limb leads, epsilon wave in V1, and T-wave inversions in inferior leads and in lead V1 to V5/V6 without localized QRS prolongation or terminal activation delay.

Echocardiographic function and appearance to the left ventricle appeared to be normal. The right ventricle was grossly dilated with 50 mm in end-diastolic diameter, paper-thin, and right ventricular aneurysms could be excluded. The right ventricle was severely hypokinetic. The tricuspid valve was slightly insufficient (Figure 1).

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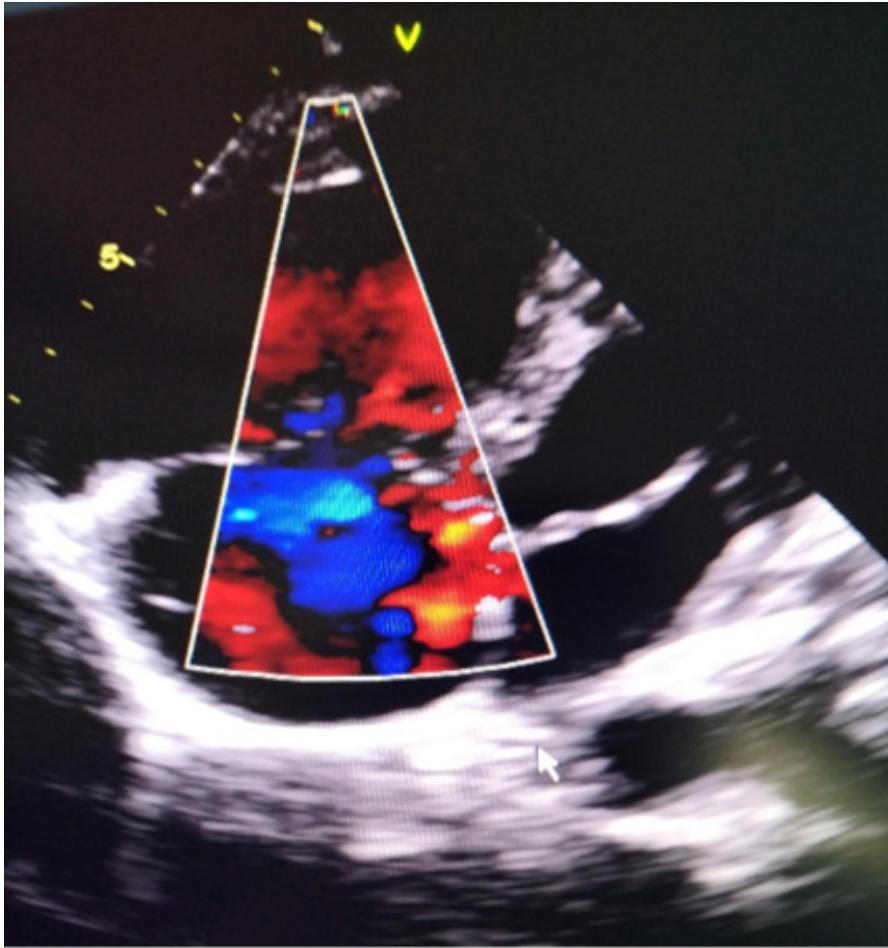


Figure 1. Echocardiography of Uhl anomaly with slight tricuspidal regurgitation.

In MRI of the heart these results could be confirmed with no late enhancement of the left ventricle (Figure 2). Massive fibrosis of the right ventricle and hardly myocardium could

be proven, right ventricular aneuysms and trabecular structure could be excluded. The diagnosis of asymptomatic Uhl anomaly (right ventricle dysplasia) was made.

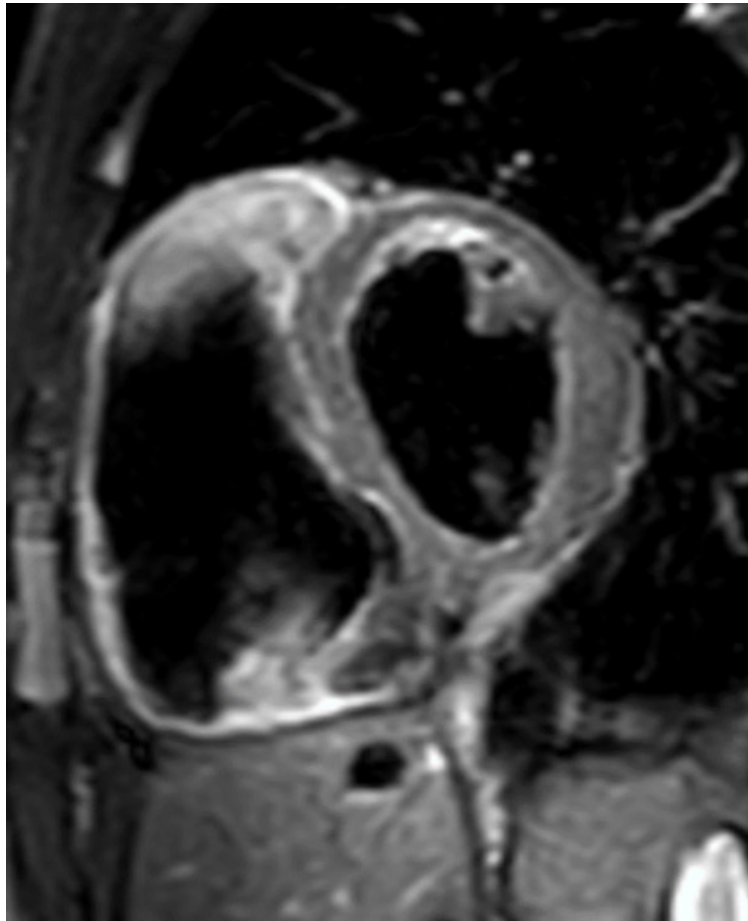


Figure 2a. Cardio MRI with gross dilatation of the right ventricle and normal function of left ventricle.

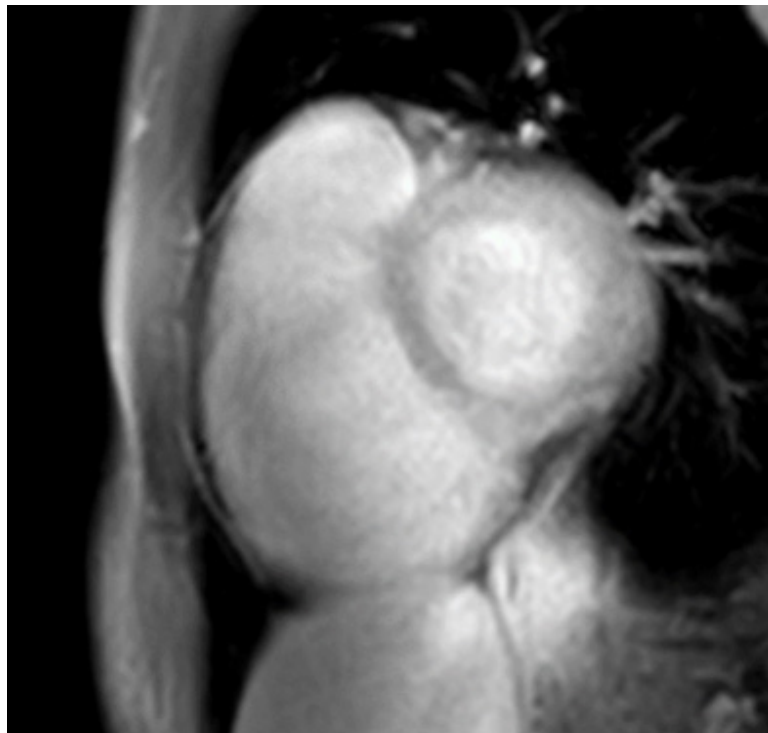


Figure 2b. Cardio MRI with late enhancement of the right ventricle. No late enhancement of the left ventricle.

CONCLUSIONS

The asymptomatic adolescent patient with Uhl anomaly [1] should be observed in a 3 to 6 months follow up. The patient was told not to perform professional handball.

Uhl anomaly is a extremely rare condition, possibly due to interuterine apoptosis of the right ventricle [2]. The dilated right ventricle is hypokinetic, paper-thin, with lacking trabecularisations or aneurysms. Uhl anomaly ends up in progressive heart failure and, to a minor extent, life-threatening arrhythms.

A similar condition is arrhythmogenic cardiomyopathy which includes right ventricular dilatation, right ventricular aneurysms, and highly characteristic ECG changes. Arrhythmogenic cardiomyopathy leads in the majority of cases to arrhythmias and in the minority of cases to heart failure.

The cause of the diseases are completely different with specific gene mutations in arrhythmogenic cardiomyopathy.

In this case of Uhl anomaly the ECG of the patients resembles arrhythmogenic left ventricular or biventricular cardiomyopathy with low voltage in limb leads inferolateral T-wave inversions, but left ventricular function is normal and late enhancement can be excluded.

Epsilon waves can be found in both diseases – arrhythmogenic cardiomyopathy and Uhl anomaly, but also in coronary artery disease [3] and in cardiac sarcoidosis [4]. The ECG in Uhl anomaly is not presented in many case reports, seems to be unspecific.

In this case report extensive T-wave inversions anterolateral are a hint of right ventricular dilatation related to arrhythmogenic cardiomyopathy [5], and possibly plays a role in risk assessment of sudden arrhythmic death.

Another option is right ventricular sarcoidosis. Typical ECG findings are: right precordial T-wave inversions, low voltage in limb leads, epsilon waves, atrioventricular conduction abnormalities, and precordial R' waves [6]. The ECG of the patient reveals three positive criteria, atrioventricular conduction abnormalities and significant precordial R' waves are missing. In cardiac MRI, however, there is no hint of right ventricular sarcoidosis.

Nevertheless, the ECG of the patient suggests that Uhl anomaly seems to be the maximum variant of arrhythmogenic right ventricular cardiomyopathy, although differences (no fatty infiltration, no right ventricular aneurysms or trabecular structure) exist.

ACKNOWLEDGMENTS

None.

CONFLICTS OF INTEREST

The authors declare that there is no conflicts of interest.

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