International Journal Of

Recent Scientific Research

ISSN: 0976-3031

Volume: 7(1) January -2016

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THE OFFICIAL PUBLICATION OF
INTERNATIONAL JOURNAL OF RECENT SCIENTIFIC RESEARCH (IJRSR)
http://www.recentscientific.com/ recentscientific@gmail.com

ISSN: 0976-3031

Available Online at http://www.recentscientific.com

International Journal of Recent Scientific Research
Vol. 7, Issue, 1, pp. 8411-8413, January, 2016

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CASE REPORT

RARE CASE REPORT OF UHL'S ANOMALY VERSUS ARRYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA (ARVD)

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ARTICLE INFO

Article History:

Received 05th October, 2015
Received in revised form 08th November, 2015
Accepted 10th December, 2015
Published online 28st January, 2016

Key words:

Uhl's anomaly, Arrythmogenic right ventricular dysplasia/ cardiomyopathy (ARVD/C), Cardiomegaly.

ABSTRACT

Objective – Uhl's anomaly, a very rare anomaly with unknown aetiology, is characterized by complete or partial absence of the myocardium of the right ventricle. Arrythmogenic right ventricular dysplasia/ cardiomyopathy (ARVD/C) is characterized by local deficiency or fibro-fatty replacement of the right ventricular myocardium. ARVD/C is an inherited cardiomyopathy with right ventricular dysfunction due to fibro-fatty replacement of myocardium, predisposing to ventricular tachycardia and death. ARVD/C and Uhl's anomaly are considered as different manifestations of the same disease. Here we report a rare case of cardiomyopathy encountered at our tertiary hospital, aim is to discuss the clinical findings and the imagining methods.

Case report – A 12-year old girl presented with progressive breathlessness and palpitations. Patient was admitted with right-sided heart failure symptoms. She was treated for cardiac failure. X-ray shows gross cardiomegaly, Echocardiography showed a hugely dilated and diffuse hypokinetic right ventricle

Conclusion – Uhl's anomaly is a rare cause of cardiomegaly, and echocardiography Is beneficial in the diagnostic process of this anomaly.

INTRODUCTION

Uhl's anomaly is characterized by complete or partial absence of the myocardium of the right ventricle. Uhl's anomaly condition was named after Henry Uhl who reported the first case in 1952 (1). William Osler described similar nature of disease called "parchment heart" in 1905(2).

In 1979, Fontaine and his colleagues (3) described arrythmogenic right ventricular dysplasia/ cardiomyopathy (ARVD/C). It is characterized by local deficiency or fibro-fatty replacement of the right ventricular myocardium. ARVD/C is an inherited cardiomyopathy with right ventricular dysfunction due to fibro-fatty replacement of myocardium, predisposing to ventricular tachycardia and death. ARVD/C and Uhl's anomaly are considered as different manifestations of the same disease. Here we report a rare case of Uhl's anomaly encountered at our tertiary hospital in South India.

Case

A 12-year old girl presented with progressive breathlessness and palpitations since 3 months. No history of chest pain, syncope or loss of consciousness was reported. There was no family history of any sudden cardiac death.

On admission, pulse rate was 130 per minutes and regular. Her blood pressure (BP) was 100/60 mmHg and there was bilateral oedema feet. No pallor, icterus, cyanosis, clubbing, lymphadenopathy was observed.

The cardiovascular (CV) examination revealed precordial bulge with grade-I parasternal heave. Early systolic murmur was present in left 2nd intercostal space and pan-systolic murmur was noted in left 5th intercostal space. Basal crepitations were present in both lungs. There was right hypochondriac tenderness. There were no central nervous system abnormalities.

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With these complaints and examination, patient was subjected to investigations. Twelve-lead electrocardiograph revealed atrial flutter with 2:1 atrio-ventricular (AV) block (figure 1). X-ray chest suggested gross cardiomegaly (figure 2).

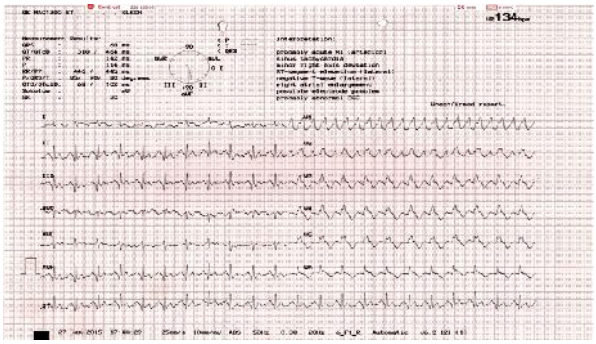


Figure 1 Twelve lead ECG in this 12-year old

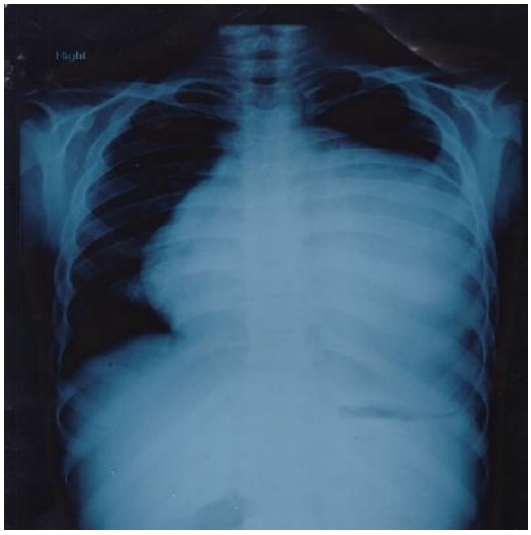
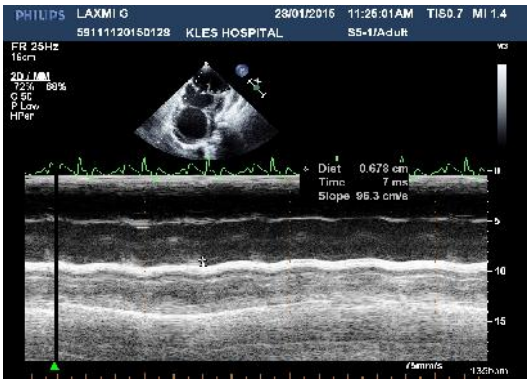
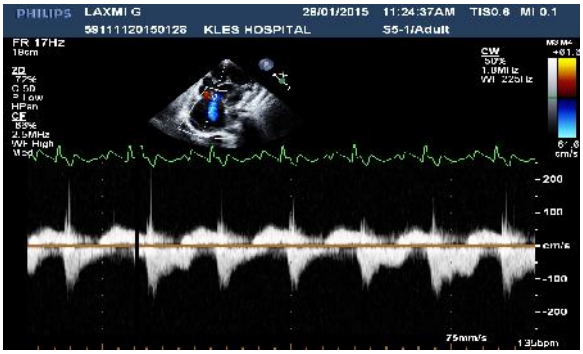


Figure 2 Gross cardiomegaly evident on postero-anteior chest X-ray

Two-dimensional echocardiography (2D-ECHO): revealed situs solitus D loop ventricle, right ventricular cardiomyopathy, gross dilatation of right atrium (RA) and ventricle (RV). RA size measured 6.5 cm transverse diameter, 6.8 cm infero-superiorly. RV size measured 7.2 cm infero-superiorly, 4.3 cm basal diameter, and 4.7 cm at mid ventricular level. RV thickness was 0.7 cm. Aneurismal dilatation and ballooning of RV apex was seen with RV dysfunction (Tricuspid Annular Plane Systolic Excursion [TAPSE] - 0.6 cm, S1 - 0.3 cm/s), interventricular septum paradox positive, inter-atrial septal bulging towards RA, severe TR with peak pressure gradient (PPG) of 9 (low pressure TR). Left aortic arch was normal.





DISCUSSION

Incidence of Uhl's anomaly is very rare and its prevalence is not possible to estimate. In world literature since beginning of the 20th century in a review done in 1993 found only 84 cases . Reported male/female ratio for Uhl's anomaly was 1.27:1, and of ARVD/C was 2.28:1 (4). Prevalence of ARVD/C is approximately 1 in 5,000 (5).

Arrhythmias and conduction abnormalities are not predominant feature of Uhl's anomaly and that is why it differs from ARVD/C where palpitation syncope, ventricular tachycardia, heart block or sudden deaths (often exercise related) are usual mode of presentation.

According to the Task Force Report published by Marcus FI, *et al*. in 2010, this patient comes in to borderline criteria of ARVD (6). Patient was admitted with right-sided heart failure symptoms. She was treated for cardiac failure and was advised to follow up. At 1 month when we enquired for follow-up, we became aware that patient died out of hospital. Hence clinical autopsy was not possible in our case.

Implication to clinical practice

This is rarest rare congenital disease with unusual presentation. These patients are presented with sudden death, as progression of the disease is uncertain and individual for each patient. Understanding its genetic basis, structural and functional characteristics will allow in the future the search for new therapies in the prevention, treatment and monitoring of patients with this rare disease.

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How to cite this article:

Vijay Basayya Metgudmath.*et al*.2016, Rare Case Report of Uhl's Anomaly Versus Arrythmogenic Right Ventricular Dysplasia (ARVD). *Int J Recent Sci Res.* 7(1), pp. 8411-8413.

TSSN 0976-3031