

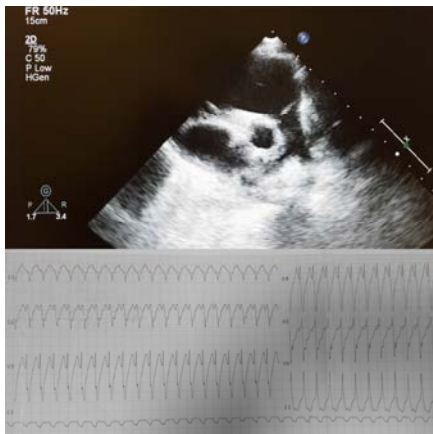
Arrhythmogenic dysplasia, Cutaneous Features and Desmoplakin Mutation; Carvajal Syndrome

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Introduction: Carvajal syndrome is rare autosomal recessive form of ARVD with distinct dermatologic features. We present a case with alopecia, palmoplantar keratosis, sustained VT and biventricular dysfunction.

Case: Four-year-old boy admitted to emergency room with palpitation and vomiting. Previous history of arrhythmia was absent. Parents were non-consanguineous. Mother's aunt has died in sleep in fifties. He was conscious, heart rate was 214/ min, blood pressure was 90/57 mm Hg. Alopecia and palmoplantar keratoderma were present. ECG showed sustained VT with left bundle branch block morphology and superior QRS axis. Tachycardia was unresponsive to adenosine and amiodarone. Lidocain was effective in termination. Resting ECG revealed diminished QRS voltages, epsilon waves in right precordial leads (V1-3), slurring of S waves (>50 msn). Echocardiography revealed dilatation of both ventricles, left ventricular EF was 45%, RVOT was dilated. MRI revealed global hypokinesia of both ventricles, transmural contrast enhancement on apico-basal segments of infero-lateral walls of left ventricle, subendocardial contrast enhancement at right ventricular septum and RVOT. Alopecia and palmoplantar keratosis was present since infancy. Heterozygot c.3564T>A and c.4395T>A mutations were found in exon 23 of desmoplakin coding gene. Mother was carrier for c.3564T>A mutation. Metoprolol, Kaptopril, Furocemide was started; extracardiac defibrillator was implanted for prevention of VT attacks. Pathogenic mutations, sustained VT with left bundle branch block morphology, epsilon waves were compatible with major criteria of ARVD and its syndromic type Carvajal Syndrome.



Conclusion: Cardiocutaneous syndromes with cardiomyopathy and serious rhythm abnormalities should be considered in patients with alopecia and palmoplantar keratosis.