

## Naxos disease: an unusual cause of cardiomyopathy

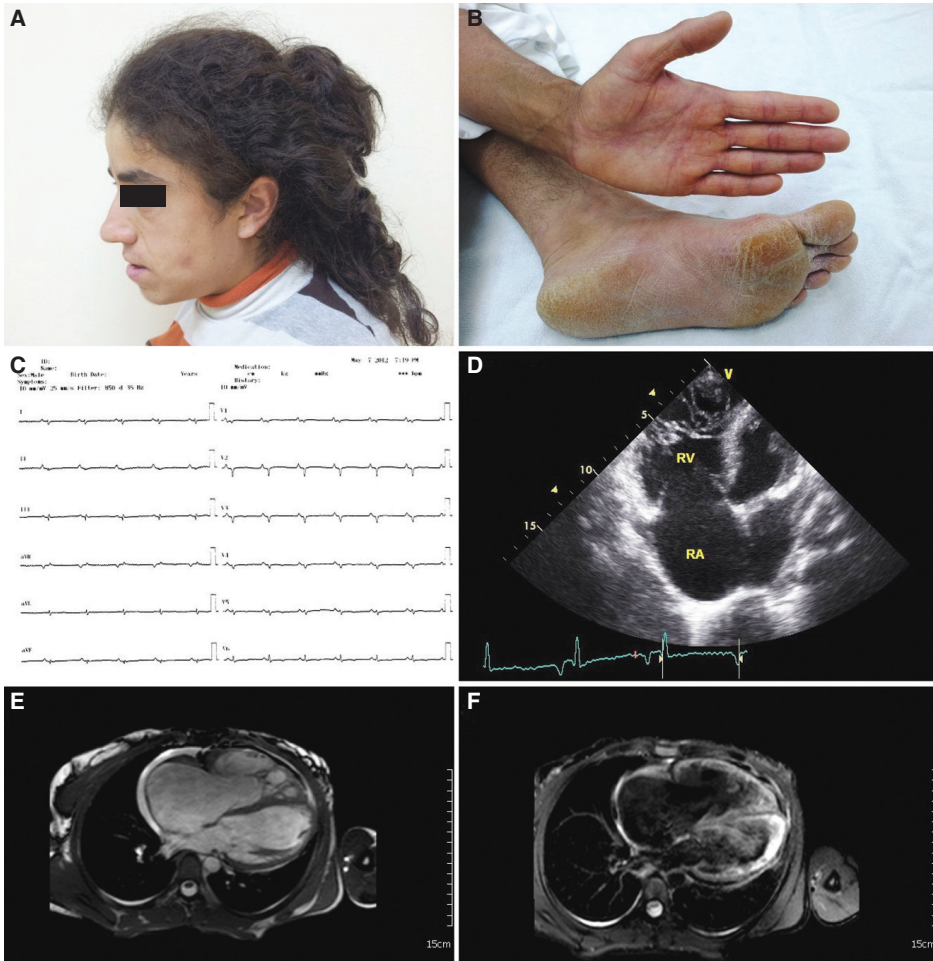
### Kardiyomiyopatinin sıradışı bir nedeni: Naksos hastalığı

Hasan Kaya  
Mustafa Oylumlu  
Faruk Ertaş  
Mehmet Guli Çetinçakmak#  
Departments of Cardiology,  
#Radiology, Dicle University  
Faculty of Medicine, Diyarbakir

A 23-year-old female presented with progressive dyspnea. On physical examination, her blood pressure was 110/70 mmHg and pulse rate was 75/min. On auscultation, a gallop rhythm and basilar rales were detected.

She had woolly hair and palmoplantar hyperkeratosis which had been present since childhood (Fig. A and B). The electrocardiogram showed sinus rhythm with low voltage, flat T waves in  $V_1$ - $V_3$  and inverted T waves in  $V_4$ - $V_6$  (Fig. C). No epsilon waves were found. The 24-hour Holter ECG did not reveal any arrhythmia. There was severe cardiomegaly on the chest X-ray. Echocardiography revealed severe right atrial

and right ventricular dilatation with trabecular configuration, global hypokinesia of both ventricles with a left ventricular ejection fraction of 23% (Fig. D, Supplementary video files 1 and 2\*). Cardiac magnetic resonance imaging showed dilatation and hypokinesia of both ventricles and trabecular disarray of the right ventricular myocardium in T2 weighted images (Fig. E). On late-enhancement imaging, a transmural contrast enhancement of the left ventricular apex and free wall observed in T1-weighted spectral presaturation with inversion recovery images, which actually corresponds to a region of fibrosis and inflammation (Fig. F). Due to the presentation of woolly hair, palmoplantar keratosis and right ventricular dysplasia, the diagnosis of Naxos disease was suggested. The remaining living members of the family were healthy and did not present this phenotype. The patient was discharged with maximal medical therapy for heart failure.



**Figures–** A 23-year-old female with (A) woolly hair and (B) palmoplantar hyperkeratosis. (C) Electrocardiogram revealed low voltage, flat T waves in  $V_1$ - $V_3$  and inverted T waves in  $V_4$ - $V_6$ . (D) Apical 4-chamber view showing severe right ventricular dilatation with trabecular configuration. (E) Magnetic resonance imaging showed trabecular disarray of the right ventricular myocardium and (F) transmural contrast enhancement of the left ventricular apex and free wall. \*Supplementary video files associated with this presentation can be found in the online version of the journal.

