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# A Rare Case of Left Ventricular Noncompaction in LEOPARD Syndrome

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**KEY WORDS:** LEOPARD syndrome · Left ventricular non-compaction · Atrial fibrillation · PTPN11.

A 40-year-old man presented with palpitation for 2 days. His past medical history was unremarkable. Physical examination revealed that multiple lentigines of 5 mm to 15 mm that were black-brown in color, macule, flat, and scattered on the face, neck, trunk, and on both hands (Fig. 1A). Hypertelorism was found but there were no evidences of deafness, genital anomaly, and other dysmorphic features including face. His electrocardiography (ECG) indicated atrial fibrillation, left ventricular hypertrophy with a strain pattern (Fig. 1B). Transthoracic echocardiography and cardiac magnetic resonance imaging revealed left ventricular noncompaction (Fig. 1D, E, and F, Supplementary Movies 1, 2, and 3). Skin biopsy had no evidences of malignancies (Fig. 1C). In addition, other disease with hyper-pigmented skin lesions including Addison's disease, hemochromatosis, and hyperthyroidism, were excluded. A p.Typ279Cys mutation in the Exon 7 of the PTPN 11 gene on Chromosome 12q24.1 was verified by polymerase chain reaction sequencing with a total of 15 exons. Finally, he was diagnosed as LEOPARD syndrome (LS) according to criteria proposed by Voron et al.[1)]

LS is an autosomal dominant congenital disorder.[2)] The prevalence and incidence of LS are unclear. "LEOPARD" is an umbrella term for seven characteristic features: lentigines, electrocardiographic abnormalities, ocular hypertelorism, pulmonary valve stenosis, abnormalities of the genitals, retarded growth, and deafness. Voron et al.[1)] proposed diagnostic criteria for LS that include 1 major criterion of multiple lentigines, at least 2 minor criteria (cardiac, ECG, genitourinary, endocrine, neurologic, cephalofacial or skeletal abnormalities), or 3 minor criteria. Hypertrophic cardiomyopathy, abnormal ECG, and pulmonary stenosis were common findings in LS. Interestingly, in 2007, Limongelli et al.[3)] reported just a case of left ventricular noncompaction among 26 patients with LS. To our knowledge, our case is the second report of a patient with LS and left ventricular noncompaction since Limongelli et al.[3)] LS is rare disease. Furthermore, LS with noncompaction is very rarity. Notably, LS present with diverse manifestation from asymptomatic to severe. Cardiac involvement in LS would deem to determine prognosis.[2)3)] Therefore, it is necessary to find out cardiac involvements with multi-image modalities.

## Supplementary Movie Legends

Movie 1. Transthoracic echocardiogram, apical four chamber view, showing noncompaction on the apex and inferoseptal segments.

Movie 2. Transthoracic echocardiogram, parasternal short axis, showing noncompaction, two-layered structure of myocardium on the lower mid-level segments.

Movie 3. Transthoracic echocardiogram, parasternal short axis, color doppler showing communication between myocardial recess and left ventricle on the lower mid-level segments.

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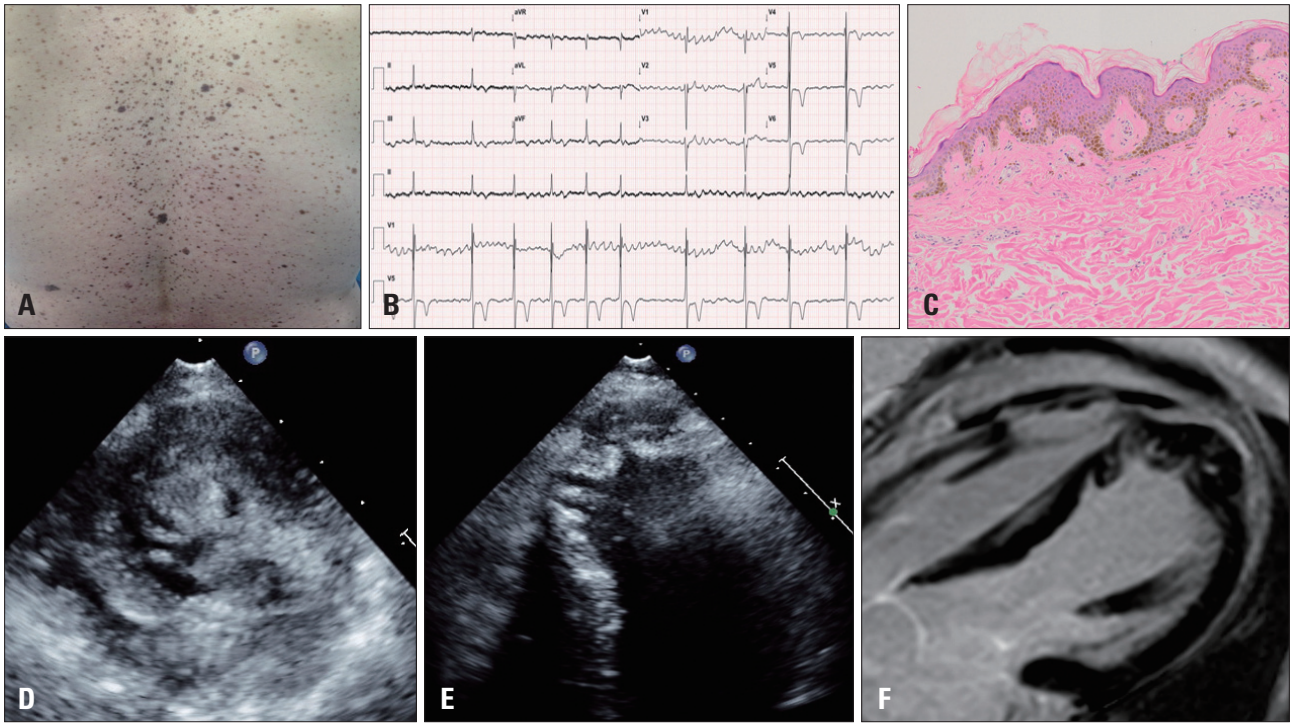


**Fig. 1.** LEOPARD syndrome clinical presentation. A: Numerous black-brown macules (lentigo) with clearly defined edge over back. B: ECG showed atrial fibrillation and left ventricular strain pattern. C: Microscopic examination showed lots of melanocytes without atypia were concentrated in the deep epidermis layer and the number of melanocytes was markedly elevated (H&E staining, × 200). D: In the parasternal short axis view, the ratio of noncompact/compact layers > 2. E: Apical four chambers view. F: Cardiac MRI revealed that multiple trabeculations in the apical segments. There were no evidences of delayed gadolinium enhancement.