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The polymorphisms LDB3 (rs 45618633), TAZ (rs 104894941), LMNA (rs28928903), DTNA (rs 7243528) and TNNT2 G/T (rs 2365652) and its association with non compacted cardiomyopathy in Mexican mestizo family

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Background: Noncompacted cardiomyopathy (NCC) has been attributed to the mutation in multiple genes leading to interruption in the endomyocardial compaction process. In the affected families, associations with mutations without T gene of cardiac troponin (TNNT2) and of the genes LDB3, MYH7 and ACTC, dystrobrevin (DTNA) and tafazzin (TAZ) in some with recessive inheritance and others of dominant type were found.

Objective: The objective of this work was to identify gene variants that participated in the development of NCC in a Mexican mestizo family.

Material and methods: A total of 36 (85.7%) individuals from a family of 42 members were studied. All patients underwent clinical history, transthoracic echocardiography (according to the criteria of Jenni et al) and the polymorphisms: LDB3, TAZ, LMNA, DTNA and TNNT2 G/T were obtained from whole blood through a commercial kit (WizardGenomic DNA Isolation Kit: Promega, Wisconsin).

Results: The NCC was present in 36.1% of the patients. Seven are first-generation relatives, of whom 6 presented NCC. Thirty-three second-generation relatives of whom 6 patients were positive for the disease and 2 third-generation individuals without NCC. When evaluating those with NCC vs those without NCC, differences in diastolic diameter, systolic diameter, left atrium size, left ventricular ejection fraction, and pulmonary artery systolic pressure were found. The 100% of patients with NCC presented the A and C alleles in the polymorphisms (rs 45618633) of the LDB3 gene and (rs 104894941) of the TAZ gene. While, for the GG and TT genotype in LMNA polymorphisms (rs 28928903) and DTNA (rs 7243528) no patients with NCC were found. At follow-up, 4/13 patients with ventricular noncompaction died.

Conclusions: 36.1% of patients in the Mexican mestizo family studied developed NCC and 100% presented the A and C alleles in the polymorphisms (rs 45618633) of the LDB3 gene and (rs 104894941) of the TAZ gene. At follow-up, 30.7% of patients with NCC died.

Biography

Nilda Espinola-Zavaleta is a Cardiologists and echocardiographer of the National Institute of Cardiology "Ignacio Chavez" in Mexico City. She is a national researcher and her work is focused in pulmonary hypertension, right ventricular function, congenital heart disease, ischemic heart disease, valvular heart disease, cardiomyopathies and new techniques of echocardiography.

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