

Nonsyndromic orofacial clefts in Chile: LINE-1 methylation and MTHFR variants

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Abstract

Aim: To evaluate the risk of nonsyndromic orofacial clefts (NSOFCs) associated with LINE-1 methylation, as a marker of global DNA methylation, and the effect of MTHFR functional variants on this variable. **Patients & methods:** LINE-1 methylation was evaluated by bisulfite modification coupled to DNA pyrosequencing in 95 NSOFC cases and 95 controls. In these subjects, MTHFR genotypes for variants c.C677T (rs1801133) and c.A1298C (rs1801131) were obtained. **Results:** Middle levels (second tertile) of LINE-1 methylation increase the risk of NSOFCs. In addition, LINE-1 methylation depends on c.A1298C genotypes in controls but not in cases. **Conclusion:** A nonlinear association between global DNA methylation and NSOFCs was detected in this Chilean population, which appears to be influenced by MTHFR functional variants.

Palabras clave

Palabras clave de autor: [DNA methylation](#); [LINE-1](#); [MTHFR](#); [nonsyndromic orofacial clefts](#)

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