Association Between IRF6 Variants and Nonsyndromic Cleft Lip With or Without Cleft Palate in Chile

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Abstract

The aim of this study was to assess the association betweenIRF6single nucleotide polymorphisms and nonsyndromic cleft lip, with or without cleft palate (NSCL/P), in a Chilean population, based on a case-control sample and confirmed in a case-parent trio population. In a sample of 150 Chilean case-parent trios and 164 controls (cohort 1), we evaluated the association between three commonIRF6variants (rs764093, rs2236909, rs2235375) and NSCL/P using odds ratio (OR) for case-control and case-parent trios and in a combined OR of both designs. To confirm associations from the cohort 1, we increased the sample size to 215 triads and 320 controls (cohort 1 + cohort 2). The combined OR for the cohort 1 reveals that the rs2235375 C allele is associated with NSCL/P in Chile (OR 1.34;p = 0.013), which was supported by the results for the two cohorts (OR 1.29;p = 0.006). Bioinformatic prediction showed that this variant, located 27 bp downstream fromIRF6exon 6, potentially alters the splicing process and based on functional annotations is associated with a decrease of gene expression. We propose that the C allele of rs2235375 fromIRF6gene seems to be a risk factor for NSCL/P in a Chilean population. However, we cannot discard a population stratification bias in our findings. On the other hand, further studies are necessary to confirm the biological role of rs2235375 inIRF6function at craniofacial development level.

Keywords

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