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Mutation analysis of *PVRL1* in patients with non-syndromic cleft of the lip and/or palate in Guangdong

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ABSTRACT. Non-syndromic cleft of the lip and/or palate (NSCLP) is a very common birth defect; the poliovirus receptor-like 1 gene (*PVRL1*) has been identified as a genetic risk factor for NSCLP in patients from Norway, the Philippines, and South America. Given the considerable variation in allele frequencies across these geographical regions, this study explored the relationship between NSCLP and mutations of *PVRL1* in patients from Guangdong, China. We recruited 171 NSCLP patients and 100 volunteers, and divided our samples into 2 groups: a sequencing group and a mass spectrometry group. In the sequencing group, we screened for mutations in exons 2 and 5 of *PVRL1* by polymerase chain reaction and direct sequencing in 71 NSCLP patients and 100 volunteers. In the mass spectrometry group, we screened for amino acid mutations in α-spliced transcript codons 112, 131, and 395, and in the β-spliced transcript codon 1082 using matrix-assisted laser desorption/ionization time-of-flight mass spectrometry analysis in 100 NSCLP patients and 100 volunteers. No mutations were detected in either

PVRL1 exons 2 or 5 in the 71 NSCLP patients and 100 volunteers, nor did we find mutations of α-spliced transcript codons 112, 131, 395 and the β-spliced transcript codon 1082 in any of the 100 NSCLP patients and 100 volunteers. Thus, mutations in exons 2 and 5 of *PVRL1*, and T334A, A391T, G1183A in the α-spliced transcript, and G1082T in the β-spliced transcript do not participate in the development of NSCLP in patients from Guangdong.

Key words: Mutation; Non-syndromic cleft of the lip and/or palate; *PVRL1*; Exons

