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Identification of Novel Mutation delC336 and insC376 in Exon 4-of *LDLR* Gene in Vietnamese Patients with High-Blood-Cholesterol

Phuong Kim Truong[1], Thuan Duc Lao[2] and Thuy Ai Huyen Le[3*]

Department of Pharmaceutical and Medical Biotechnology, Faculty of Biotechnology, Ho Chi Minh City Open University, Ho Chi Minh City, Vietnam
Phone: [1]<+84.90.309.3390>, [2]<+84.76.544.9770>, [3]<+84.905.784.471>
Email: [1]<phuong.tk@ou.edu.vn>, [2]<ducthuanlao87@gmail.com>, [2]<thuan.ld@ou.edu.vn>, [3]<thuy.lha@ou.edu.vn>

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ABSTRACT The *LDLR* (*Low density lipoprotein receptor*) gene encodes the low density lipoprotein receptor. It has been reported to function as a carrier of cholesterol in the blood. Further, exon 4 of *LDLR* gene has been found to be a hotspot of mutation. However, *LDLR* gene mutation has not been studied in Vietnam. Therefore, the aim of the current study is to identify mutations located in exon 4 of *LDLR* gene in Vietnamese high blood cholesterol patients by PCR-sequencing method. Based on the results, two novel mutations including (1) deletion "C" at position 336 and (2) insertion "C" at position 376 (insC376) were detected in 2 of 17 clinical samples (accounting for 11.76%) and 14 of 17 clinical samples (accounting for 82.35%), respectively. Both delC336 and insC376 are located at the binding domain repeat III. The observed mutations were separately predicted to cause a translational frameshift leading to the abnormal LDLR proteins that may have lost their function, resulting in the high blood cholesterol levels in patients.