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学位の種類	博士 (医学)
報告番号	甲第1549号
学位記番号	第1106号
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授与年月日	平成 28年 11月 30日
学位論文の題名	<p>Genotyping analysis of the factor V Nara mutation, Hong Kong mutation, and 16 SNPs including the R2 haplotype, and the involvement of factor V activity in patients with recurrent miscarriage</p> <p>(習慣流産と Factor V Nara/Hong Kong 変異、FV R2 ハプロタイプと FV 活性値の関連性)</p> <p>Blood Coagulation & Fibrinolysis. Accepted August 10, 2016</p>
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Abstracts

OBJECTIVES: Recurrent miscarriage (RM) can arise from a large diversity of causes and the factors responsible have not been fully clarified. The coagulation factor V (FV) R506Q (Leiden) mutation is a well-known risk factor for RM, although it has not been found in Japanese populations. We examined whether the FV Nara and Hong Kong mutations, the FV gene (*F5*) 16 single-nucleotide polymorphisms (SNPs) including the FV R2 haplotype, and plasma FV activity (FV:C) were risk factors for RM.

METHODS: A cross-sectional study was conducted among 88 patients with a history of unexplained RM and 95 fertile controls.

RESULTS: None of the patients or controls was homozygous or heterozygous for the FV Nara or Hong Kong mutation. In the 16 SNPs of *F5*, frequencies of the G/T and T/T genotypes at Ser156Ser were significantly lower in patients than in controls (OR 0.45, 95%CI 0.22-0.91, OR 0.32, 95%CI 0.14-0.72) and the allele frequency of C at Leu1288Leu was significantly higher in patients than that in controls (OR 1.66, 95%CI 1.02-2.71). The mean FV:C values were not significantly different between patients and controls. However, the prevalence of patients with a high or low FV:C (>95th or <5th percentile) was significantly greater than the controls (OR 3.59, 95%CI 1.11-11.60; OR 3.94, 95% CI 1.23-12.60).

CONCLUSIONS: These results suggest that some SNPs of *F5* and a high or low FV:C level might be associated with RM.