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Genetic Variants Associated with Recurrent Pregnancy Loss in Uzbek Women: A Genome-Wide Association Study

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Background: Recurrent pregnancy loss (RPL) is a condition characterised by the loss of two or more consecutive pregnancies before the 20th week of gestation, affecting approximately 15% of all pregnancies. Despite significant advancements in infertility diagnostics and reproductive medicine, the underlying cause remains unknown in 35-60% of RPL cases. This suggests a complex interplay of genetic, epigenetic, and environmental factors contributing to its pathogenesis. In this study, we conducted a genetic analysis to identify variants associated with RPL among women in Uzbekistan, aiming to enhance our understanding of its genetic basis.
Material and Methods: This study included 161 patients diagnosed with RPL and 511 healthy individuals. Genetic screening was performed using the Global Screening Array v.3.0. Hardy-Weinberg equilibrium test and logistic regression analyses were conducted using the SNPassoc software package to identify statistically significant associations between genetic variants and disease risk under an additive genetic model.
Results: Multiple genetic variants demonstrated significant associations with RPL. Notably, rs4871372 (LOC105375725 , p = 3.7 × 10-5, OR = 0.58) and rs2704035 (SULF1 , p = 3.9 × 10-4, OR = 0.63) were identified as protective factors, potentially reducing the risk of RPL. In contrast, rs4916848 (ADGRV1 , p = 1.8 × 10-6, OR = 1.82) and rs478993 (TENM4 , p = 4.5 × 10-4, OR = 1.58) were associated with an increased susceptibility to RPL. These findings highlight the potential role of these genetic variants in influencing pregnancy outcomes and warrant further investigation to elucidate their biological significance.
Conclusion: Our study identified novel genetic associations with RPL in Uzbek women. The IncRNA LOC105375725 (rs4871372) may function as an epigenetic regulator, while rs4916848 likely acts as an enhancer. Additionally, TENM4 has been implicated in mesoderm induction during early embryogenesis. Therefore, these variants can be used as diagnostic and prognostic markers. Further research is needed to understand their functional role of these variants in RPL pathogenesis.