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Control/Tracking Number: 2025-A-375-ESHG
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Current Date/Time: 3/19/2025 11:32:32 PM

Identifying genetic variants associated with recurrent pregnancy loss in the Uzbek population

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Abstract:
Background: Recurrent pregnancy loss (RPL) is defined as the loss of two or more pregnancies before the 20th week of gestation and affects approximately 15% of pregnancies. Despite advances in infertility diagnostics, 35–60% of RPL cases remain unexplained, suggesting genetic, epigenetic, and environmental contributions. In this study, we analysed the genetic factors associated with RPL among women in Uzbekistan.
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: Several genetic variants showed significant associations with RPL. Protective effects were observed for rs4871372 (*LOC105375725*, $p = 3.7 \times 10^{-5}$, OR = 0.58) and rs2704035 (*SULF1*, $p = 3.9 \times 10^{-4}$, OR = 0.63). Increased RPL risk was linked to rs4916848 (*ADGRV1*, $p = 1.8 \times 10^{-6}$, OR = 1.82) and rs478993 (*TENM4*, $p = 4.5 \times 10^{-4}$, OR = 1.58).
Conclusion: Our study identified novel genetic associations with RPL in Uzbek women. The lncRNA *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.

Author Disclosure Information:

A. Punko: None. **A. Abdurakhimov:** None. **Y. Kapralova:** None. **D. Zakirova:** None. **A. Rakhmatullayev:** None. **G. Esonova:** None. **X. Meylikov:** None. **A. Abdullaev:** None. **D. Dalimova:** None. **S. Turdikulova:** None.
Topic (Complete): 18. Large scale genomics (GWAS) and other -omics association studies
Keyword (Complete): recurrent pregnancy loss ; Uzbek population
Presentation Preference (Complete): Hybrid Poster
Additional information (Complete):
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