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Exploring the association between genetic variants and the risk of developing type 2 diabetes in Uzbekistan

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Abstract:

Background: Type 2 Diabetes (T2D) affects nearly half a billion people worldwide, yet genetic studies largely exclude Central Asian populations. This study investigates T2D-associated genetic variants in Uzbeks, aiming to enhance the understanding of T2D genetics and underscore the importance of including diverse populations in research to improve global disease models and precision medicine.

Material and Methods: This study encompassed 96 patients diagnosed with T2D and 96 healthy individuals of Uzbek ethnicity. Genetic screening was performed using the Global Screening Array v3.0, and statistical analyses were conducted using SNPAssoc to examine genotype-disease associations under common genetic models.

Results: Under the additive model, several variants showed significant associations with T2D. Protective effects were found for rs4660329 (*MAST2*, $p = 1.91 \times 10^{-5}$, OR = 0.40), rs5909292 (*MAP3K15*, $p = 1 \times 10^{-4}$, OR = 0.24), and rs5909301 (*MAP3K15*, $p = 1.6 \times 10^{-4}$, OR = 0.36). Conversely, increased T2D risk was associated with rs16872304 (*ZFPM2*, $p = 3.30 \times 10^{-5}$, OR = 3.56), rs3859563 (*FUT6*, $p = 2.25 \times 10^{-4}$, OR = 2.08), and rs11002834 (*ZMIZ1*, $p = 5.43 \times 10^{-4}$, OR = 3.43).

Conclusion: Variants in *MAST2*, *ZFPM2*, *MAP3K15*, and *ZMIZ1* have been associated with T2D in previous studies, as documented in the GWAS catalog, highlighting their potential role in disease susceptibility and warrant further investigation to understand their biological significance.

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