DEADLINE WEDNESDAY 2 JANUARY 2019

Genetic polymorphisms in KCNJ11 (E23K, rs5219) and SDF-1β (G801A, rs1801157) genes are associated with the risk of type 2 diabetes mellitus. Rizvi S, Raza ST, Mahdi F, Singh SP, Raijput M, Rahman Q. Br J Biomed Sci 2018; 75 (3): 139–44. Assessment No G101018


01 More than 150 distinct genetic loci, with more than 120 variants, have been identified that may be involved in the pathogenesis of diabetes.

02 Type 2 diabetes has a multifactorial aetiology, where genetic factors in combination with environmental factors confer risk of disease development and progression.

03 Genetic alterations in KATP are associated with diabetes due to the effect of KCNJ11 channels on insulin secretion.

04 Genomic DNA was extracted from peripheral blood mononuclear cells using the standard phenol-chloroform extraction method.

05 Fasting blood sugar was measured by the glycerol phosphate oxidase-peroxidase amidopipine method.

06 Cases and controls were matched for age (45 and 46 years, respectively) and gender (99 males/101 females and 98 males/102 females, respectively).

07 Mean creatinine level was 70 μmol/L in healthy controls and 50 μmol/L in cases.

08 Polymorphisms in KCNJ11 result in neonatal diabetes and congenital hypoinosinemia.

09 Studies have shown that the rs5219 variant may alter the charge of the ATP-binding region and decrease channel sensitivity to ATP.

10 Stromal cell derived factor-1 (SDF-1), also known as CXCL12, is a peptide chemokine that is coded for by a gene on chromosome 10q11.1.

11 Many of the published links between polymorphisms in KCNJ11 and SDF-1β and diabetes consider multiple genotype models.

12 Figure 1 shows a 3% agarose gel picture of MspI-digested products.

13 A number of risk alleles for diabetes and mutations in several genes may add up and predispose an individual to increased risk of disease.

14 Distribution of SDF-1β (G801A, rs1801157) genotypes according to dominant, recessive and additive models showed no significant differences in dominant and additive models between cases and controls.

15 Genetic factors are now regarded as the leading cause of diabetes.

16 ATP-sensitive potassium channels (KATP) are transmembrane proteins present on beta cells.

17 Genotyping data were compared between cases and controls using the y2 test.

18 In this study, genetic polymorphism analysis was performed by loop-mediated isothermal amplification.

19 G801A polymorphism has been studied in various diseases, including diabetes, HIV infection and cancer.

20 In this study, the frequency of AA genotype in cases was 4.5%, which was lower than reported in Iranian diabetics.

REFLECTIVE LEARNING

01 Type 2 diabetes mellitus (T2DM) is a global health problem resulting from the interaction of environmental and genetic factors. Based on the results of a literature search, what other genetic factors have been implicated in T2DM?

02 Explain the difference between the terms dominant, recessive and additive in relation to the effects of different genotype models.