rs10757278

Genome-wide association study (GWAS) has identified that rs10757278 in chromosome 9p21 was a risk loci of ischemic stroke (IS). Interferon-beta 1 (IFNB1), located on 9p21, has a protective role in IS.

Han et al. aimed to investigate whether the rs10757278, rs9333358 and rs1051922 in IFNB1 were related to the risk of IS. The 3 polymorphisms were genotyped using a TaqMan allelic discrimination assay in 505 patients with IS and 652 controls with frequencies matched to cases regarding age, gender, living area, and ethnicity. The IFNB1 mRNA levels were determined using quantitative polymerase chain reaction and relative luciferase activity was measured using the Dual Luciferase reporter assay. An increased risk of IS was found for both the rs10757278 (GG vs. AA: adjusted OR = 1.80, 95% CI: 1.27-2.55, P < 0.001; GG vs. AA/AG: adjusted OR = 1.57, 95% CI: 1.18-2.10, P = 0.002; G vs. A: adjusted OR = 1.35, 95% CI: 1.13-1.61, P < 0.001) and the rs9333358 (GG vs. AA: adjusted OR = 1.91, 95% CI: 1.27-2.88, P = 0.002; GG vs. AA/AG: adjusted OR = 1.81, 95% CI: 1.23-2.68, P = 0.003; G vs. A: adjusted OR = 1.35, 95% CI: 1.12-1.64, P = 0.002). Similarly, a higher risk of IS was also observed in the combined genotypes of the rs10757278 AG/GG and rs9333358 AG/GG (95% CI: 1.34-2.83, P < 0.001). Moreover, individuals carrying the rs9333358 GG genotype exhibited lower levels of IFNB1 mRNA and the rs9333358 G allele displayed a reduced transcriptional activity. These findings suggest that 9p21 rs10757278-IFNB1 rs9333358 may have both single and joint effects on the development of IS [1].

1)
Han X, Wang C, Tang D, Shi Y, Gao M. Association of genetic polymorphisms in chromosome 9p21 with risk of ischemic stroke. Cytokine. 2019 Dec 3;127:154921. doi: 10.1016/j.cyto.2019.154921. [Epub ahead of print] PubMed PMID: 31810024.