An HLA imputation was conducted to explore the relationship between HLA and patients with moyamoya disease (MMD) in the Chinese Han population.

In this study of Wan et al. performed an association analysis of the major histocompatibility complex region in 2,786 individuals of Chinese Han ancestry (2,031 controls and 755 patients with MMD), through a widely used HLA imputation method.

They identified that the variant rs3129731 (odds ratio [OR] = 1.79, p = 3.69 × 10-16) located between the MTCO3P1 and HLA-DQA2 is a major genetic risk factor for MMD. In addition to this variant, found in the conditional association analysis, we also detected another independent signal, rs1071817 (OR = 0.62, p = 1.20 × 10-11), in HLA-B.

This research suggests that the genetic polymorphism of HLA-DQA2 and HLA-B could be a genetic predisposing factor for MMD in Chinese Han. This may provide some evidence for further HLA-related studies of patients with MMD of Chinese Han ethnicity and indicates that MMD is an immune-related disease [1)].

1)
Wan J, Ling W, Zhengshan Z, Xianbo Z, Lian D, Kai W. Association of HLA-DQA2 and HLA-B With Moyamoya Disease in the Chinese Han Population. Neurol Genet. 2021 Jun 2;7(3):e592. doi: 10.1212/NXG.0000000000000592. PMID: 34095496; PMCID: PMC8176556.