

Frequencies of the *JK^{null}* (IVS5-1g>a) allele in Taiwanese, Fujian, Filipino, and Indonesian populations

The *Jk_{null}* [Jk(a-b-)] phenotype, identified in Taiwanese populations during our previous investigation,¹ appears to have a relatively higher prevalence among indigenous Taiwanese persons. In this study, we analyzed the coding exon regions (Exons 4-11) of the *JK* gene of a *Jk_{null}* Paiwan indigene to determine the molecular basis of the *Jk_{null}* phenotype in different Taiwanese populations. The Exon 4 to 11 region of the *JK* gene was divided into four segments, Exons 4 and 5, Exons 6 and 7, Exons 8 and 9, and Exons 10 and 11. Each segment was amplified by polymerase chain reaction (PCR) using specifically designed primer pairs (not shown) flanking for each specific exon region. Genomic DNA from the *Jk_{null}* individual served as the template and the amplified DNA segment was then directly sequenced. The sequencing results demonstrated that each coding exon of the *JK* gene of this *Jk_{null}* Paiwan indigene possessed the wild-type sequence, but was homozygous for a g>a mutation at the 3' acceptor splice site of intron 5 (IVS5-1g>a) of the *JK* gene.

Using PCR amplification and subsequent direct sequencing, we next analyzed the frequency of the *JK* IVS5-1g>a mutation among various Taiwanese population groups, including Taiwan's two major population groups, Minnan and Hakka, and the country's nine indigenous Austronesian-speaking groups. Because each individual has two *JK* alleles, we calculated allele frequencies by the formula "number of alleles identified/number of persons tested \times 2." The *JK*IVS5-1g>a* allele frequencies among Minnan and Hakka Taiwanese were 1.0 percent (1/50, number of alleles identified/number of persons tested) and 2.0 percent (2/50), respectively. The frequencies among the nine Taiwanese indigenous groups were as follow: Atayal 2.9 percent (3/52), Saisiat 1.0 percent (1/51), Tsou 0 percent (0/51), Bunun 3.0 percent (3/50), Paiwan 7.8 percent (8/51), Rukai 1.0 percent (1/50), Ami 4.8 percent (5/52), Puyuma 5.0 percent (5/50), and Yami 2.0 percent (2/49). Thus, the *JK*IVS5-1g>a* allele has been identified in every population group in Taiwan with the exception of the Tsou tribe. The frequency of the allele varies among the different population groups, most likely due to the historical isolation of the tribes and genetic drift, with the highest prevalence (7.8%) in the Paiwan tribe. We also analyzed the *JK*IVS5-1g>a* allele frequencies in three Southeast Asian populations, Fujian (south-

east coast of China), Filipino, and Indonesian, which were 2.5 (3/61), 9.0 (9/50), and 1.0 (1/50), respectively.

The *Jk_{null}* phenotype is very rare in ethnic populations worldwide; however, it has been found to be relatively prevalent in Polynesian population groups with phenotype frequencies of 0.1 to 1.4 percent.² The IVS5-1g>a mutation of the *JK* gene, first identified in a *Jk_{null}* Chinese person,³ is the main molecular basis for the *Jk_{null}* phenotype in Polynesian persons. A subsequent study demonstrated a frequency of 8.7 percent (8/46) for the *JK*IVS5-1g>a* allele in this population.⁴ Our previous investigations on human leukocyte antigens and the mitochondria DNA of Taiwanese indigenous groups demonstrated a relationship between indigenous Taiwanese and Polynesian populations.⁵ The results of this study, which reveal a relatively high frequency of the *JK*IVS5-1g>a* allele in a number of Taiwanese indigenous groups, support the above hypothesis of a historical relationship between these ethnic groups.

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