



Association study between Y-chromosome haplogroups and susceptibility to spermatogenic impairment in Han People from southwest China

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ABSTRACT. The non-recombining portion of the Y-chromosome contains numerous polymorphisms; therefore, it is now the most informative haplotyping system with wide-ranging applications. Idiopathic azoospermia and oligospermia are among the most important causes of male infertility. Different haplogroups may have different genetic backgrounds, which may be either susceptible or unsusceptible to idiopathic azoospermia or oligospermia. This study investigated the possible association between Y-chromosome haplogroup distribution and susceptibility to spermatogenic impairment. Peripheral blood was collected from 193 men with normozoospermia, 193 men with idiopathic azoospermia and 105 men with idiopathic oligospermia. All of the subjects underwent karyotyping, azoospermia factor (*AZF*) deletion analysis by 15 *AZF*-specific sequence-tagged sites and Y-chromosome haplotype analysis by 17 binary markers. Excluding men with *AZF*

deletions and abnormal karyotypes, the remainder of these 3 groups was named Group i, Group ii, and Group iii, respectively. The comparisons of 17 Y-haplogroup distributions between Group i and Group ii, Group iii or Group ii + iii were performed with the SPSS V.18.0 software. Significantly different Y-haplogroup distributions were observed between Group i and Group ii in N1* (P = 0.002), between Group i and Group iii in F*, K*, P*, and O3* (P = 0.002, 0.001, 0.004, and 0.007, respectively), and between Group i and Group ii + iii in K*, N1* and O3* (P = 0.008, 0.012, and 0.009, respectively). These results suggest that Y-chromosome haplogroups play a role in spermatogenic impairment.

Key words: Y-chromosome haplogroups; Spermatogenic impairment; *AZF* deletion; Idiopathic azoospermia/oligospermia