



Differences in frequencies of UGT1A9, 1A7, and 1A1 genetic polymorphisms in Chinese Tibetan versus Han Chinese populations

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ABSTRACT. As part of a series of pharmacogenomics studies of the Chinese population, we investigated genetic polymorphisms of some UGT1A regions. The three genes that were analyzed were UGT1A9, 1A7, and 1A1; we sequenced their exons, together with promoters, surrounding introns and 3'-untranslated regions (3'UTR) in 100 unrelated-healthy Chinese Tibetan individuals. We compared the data with information on Han Chinese of the same region, which we downloaded from the HapMap database. We identified 40 polymorphisms; 16 of them were shared by the two populations. We then analyzed their linkage disequilibrium map. The UGT1As cluster can be divided into two linkage blocks in the Tibetan population: Block 1 (UGT1A9, UGT1A7), Block 2 (3'-UTR). Furthermore, we identified haplotypes and selected their tagSNPs. In exon 1 of UGT1A7 gene, 393G>A (Arg131Gln, rs17868324) was found at a frequency of 44.4% in the Tibetan population, compared to only 0.7% in the Han population. The linkage blocks in the Han Chinese sample differed from that of the Chinese Tibetan group; the former had Block 1 (UGT1A9,

UGT1A7), Block 2 (UGT1A7), and Block 3 (3'-UTR). These findings provide fundamental information for future molecular genetic studies of the UGT1A gene cluster as well as for personalized medicine in Chinese.

Key words: *UGT1A*; Tibetan; Chinese Han in Beijing, China (CHB); Linkage disequilibrium; Haplotype