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## Salt Balance via CFTR and Evolution of *CFTR* Gene: Comparison between Japanese and Chinese

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### Summary

In order to understand polymorphic backgrounds of the *cystic fibrosis transmembrane conductance regulator* (*CFTR*) gene in Japanese and Chinese, we examined three polymorphic loci (poly T, TG repeats, and M470V) in normal subjects in Nagoya (159), Changchun (64), and Nanjing (55). Genomic DNA was extracted from the whole blood. Following PCR poly-T and TG repeats were directly sequenced and the M470V polymorphism was identified by Hph I restriction enzyme.

The T7 allele was the most common haplotype in Changchun (0.938), Nanjing (0.927), and Nagoya (0.975). The T5 allele was present in only 7 subjects in Changchun, 3 in Nanjing, and 2 in Nagoya. The (TG)<sub>11</sub> and (TG)<sub>12</sub> alleles were dominant haplotypes in both Chinese and Japanese. The (TG)<sub>10</sub> was present 2-3% in Chinese but none in Japanese. The allele frequency of the V470 in Changchun (0.633) was significantly ( $p < 0.05$ ) higher than that in Nanjing (0.500). The M470/V470 ratio in Nagoya (0.67) was intermediate between Nanjing (1.00) and Changchun (0.58). The three major haplotypes, T7-(TG)<sub>11</sub>-V470, T7-(TG)<sub>12</sub>-M470, and T7-(TG)<sub>12</sub>-V470, comprised more than 90% of both Japanese and Chinese. The T7-(TG)<sub>11</sub>-V470 was the most common haplotype in Changchun (0.500) and the T7-(TG)<sub>12</sub>-M470 was the most common in Nanjing (0.436). The haplotype frequencies of T7-(TG)<sub>11</sub>-V470 and T7-(TG)<sub>12</sub>-M470 in Nagoya were intermediate between Changchun and Nanjing, suggesting the former comes from the northern residents and the latter from a southern part of the East Asia.