



## Genetic analysis of STR markers on chromosome 21 in a Han population from southeast China

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Genet. Mol. Res. 14 (1): 1718-1725 (2015)  
Received February 10, 2014  
Accepted August 4, 2014  
Published March 6, 2015  
DOI <http://dx.doi.org/10.4238/2015.March.6.18>

**ABSTRACT.** Short tandem repeats (STRs) are highly polymorphic sequences and have been extensively used as genetic markers in mapping studies, disease diagnosis, and human identity testing. In this study, 11 STR markers on chromosome 21, including D21S1432, D21S11, D21S1246, D21S1412, D21S1437, D21S1442, D21S2039, D21S1270, D21S1435, D21S1409, and D21S1446, were analyzed in 740 unrelated Han individuals from southeast China. A total of 132 alleles, ranging from 7-21 for each locus, were named according to the guidelines of the International Society for Forensic Haemogenetics. The distributions of allelic frequencies for the 11 STRs and population genetic parameters were determined. All 11 STR markers showed high polymorphism and heterogeneity in the southeast Han population, with polymorphism information content of 0.61-0.87, heterogeneity of 64.5-86.1%, and power of discrimination of 0.835-0.973. Among the 11 STR markers, D21S1412, D21S1270, D21S11, and D21S1442 showed relatively higher heterogeneity. Their combination was relatively informative and was used in a quantitative fluorescence-polymerase chain reaction

assay to diagnose Down syndrome (trisomy 21) in a southeast Chinese Han population. The genetic information and population data for these 11 STRs may be used not only in quantitative fluorescence-polymerase chain reaction assays but also in forensic studies and other genetic tests.

**Key words:** Southeast China; Down syndrome; Han population; Quantitative fluorescence polymerase chain reaction; Chromosome 21; Short tandem repeats