ANALYSIS STRATEGIES OF DEFICIENT GRANDPARENT-GRANDCHILD RELATIONSHIP IDENTIFICATION

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To explore the application and strategy selection of pedigree genotype reconstruction method in deficient grandparent-grandchild relationship identification, when several reference individuals in the father's generation are available for testing.

Individuals in 8000 pedigrees were simulated by computer based on the genetic population data of Hebei Han population in China at 38 autosomal short tandem repeats (STR). Then the child's biological mother and a different number of paternal individuals were selected as reference in deficient grandparent-grandchild relationship identification. Three different strategies were applied to calculate grandparent index (GI): expected value strategy, minimum probability value strategy and the optimized minimum probability value strategy. Boundary values of combined grandparent index (CGI) were set up through diagnostic experiments, and then verified by 60 practical cases.

The number of reference samples significantly influences the judgment of such cases, and the accuracy was improved with the increase of the reference number. If 104 and 10-4 were regarded as the boundary values of inclusion and exclusion, the accuracy of the expected value strategy was more than 90% even with only 1 paternal reference, and got up to 99.8% when the number of reference individuals increased to 2. The minimum probability value strategy showed a low sensitivity and a high rate of false negative, while the results of optimized minimum probability value strategy were between the above two methods. An identical result appeared in verification test in 60 practical cases, 59 of which were correctly confirmed or excluded applying expected value strategy, only 1 case with single reference individual showed uncertain result.

Pedigree genotype reconstruction method effectively utilizes the genetic information of reference samples, improving the efficiency in specific kinship analysis. Comparing with the minimum probability value strategy, the expected value strategy showed better ability to estimate the authenticity of the alleged family. Its effectiveness and safety are sufficient to meet identification requirements in such cases.

Key words: Deficient grandparent-grandchild relationship; Pedigree genotype reconstruction; Expected value strategy; Minimum probability value strategy.