

SIMPLE DETECTION AND SCREENING METHOD OF HETEROPLASMY ON THE MITOCHONDRIAL DNA HV1 REGION

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By the present, techniques of sequencing mitochondrial DNA for individual identification have been reported all over the world. But the heteroplasmy of mitochondrial DNA sequence may bring confusion in decision of the sequence or sometimes may cause misjudgment of comparing the sequences of samples. We tried SSCP analysis for PCR products of HV1 region as a method to detect such heteroplasmy simply.

Mitochondrial DNA HV1 region (16,190 ~ 16,420) was amplified with TAMRA labeled primer set and electrophoresed using ABI Prism® 310 genetic analyzer. The data of electrophoresis were analyzed using GeneScan® analysis software. As a result of analysis, single peaks were detected from homoplasmic samples and multiple peaks were detected from heteroplasmic samples as expected. By this technique, an existence of heteroplasmy was predicted very simply.

Using this technique, DNA samples extracted from 102 of Japanese were analyzed on mitochondrial HV1 region. As a result multiple peaks were recognized in electropherogram of PCR product from 3 of 102 origins, and existence of heteroplasmy was predicted. The sequences of these three samples were analyzed and the existence of each heteroplasmy was confirmed expectedly.