Abstract: Around 10% of males with idiopathic azoospermia or oligozoospermia, which are important causes of male infertility, have partial deletions on the long arm of the Y chromosome. To develop a rapid and accurate detection system for screening major Y deletions found in infertile men, we developed a multiplex PCR system that can simultaneously amplify five loci on the Y chromosome, SRY, AMELY, DBY, RBMY, DAZ and one locus on the X chromosome, AMELX. The size of the PCR products was designed to increase gradually from the distal Yp to the distal Yq. Our system could detect deletions of three major candidate regions for the azoospermic factor, AZFa, AZFb and AZFc on the Y chromosome together with sex. The gradual increase in the size of the PCR products was convenient for imaging the location of deletions on the Y chromosome. Moreover, the multiplex PCR system was combined with microchip-based electrophoresis, and the PCR products derived from each locus were separated within 4 min. Our system is useful for screening Y chromosomes bearing the structural anomalies including three major AZF deletions found among azoospermic or oligozoospermic males. J. Med. Invest. 53:147-152, February, 2006

Keywords: Y chromosome, AZF, multiplex PCR, microchip electrophoresis, azoospermia
Subjects

Subjects

Multiplex PCR

Electrophoresis with a microchip device
Generation of a multiplex PCR system for detecting Y deletions

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Combination of multiplex PCR with microchip-based electrophoresis for detecting Y deletions

- Figure a shows the electrophoresis patterns for the multiplex PCR system. Peaks 1, 2, 3, 4, 5, and 6 represent different DNA fragments.
- Figure f illustrates the electrophoresis patterns for a sample with Y deletions. Peaks 1, 2, 3, 4, 5, and 6 are observed, with peak 4 being absent in the sample with Y deletions.
- Figure g shows the electrophoresis patterns for a control sample. Peaks 1, 2, 3, 4, 5, and 6 are all present.