Intellectual Property (Non-confidential)





Rapid Mutation Scanning in DNA

DESCRIPTION

Single-strand conformation polymorphism (SSCP) is one of the most widely used methods for mutation scanning in DNA. Single-base sequence changes can be detected using electrophoresis in a nondenaturing gel. The secondary structure of the DNA strands varies between the wild type sequence and each of the possible single-base mutations, including sequences with multiple single-base mutations. The difference in secondary structures causes the DNA strands to migrate along the gel at different rates, thus allowing mutations to be easily detected. However, sensitivity for SSCP is heavily affected by the variance in mobility as well as whether the wild type represents the mode.

City of Hope has developed a novel method variation of SSCP by identifying a carefully selected set of parameters, under which 100% sensitivity and 90% specificity has been experimentally demonstrated using factor IX (hemophilia B) as a test case. Electrophoresis parameters, such as temperature, buffer, gel matrix, and additives were

meticulously tested and five conditions were selected with sufficient redundancy to detect all mutations within a tested set. This method, $SSCP_5$ is estimated to be five times faster than fluorescent DNA sequencing. $SSCP_5$ is also capable of detecting very low counts of mutated sequences within a predominantly wild-type sample. As genetic testing and personalized medicine become more prevalent, the ability to quickly and accurately detect mutations will be a valuable addition to the field of medical diagnostics.

KEY ASPECTS

- Single-strand conformation polymorphism method with parameters maximized for speed and accuracy in gene mutation identification
- 5x faster than fluorescent DNA sequencing
- 100% sensitivity, 90% specificity experimentally demonstrated for mutations in genes related to hemophilia

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Title	US Patent Number	lssued
Multi-Conditional SSCP (SSCP ₅): A Rapid Method for Mutation Scanning with Virtually 100% Sensitivity	6,287,441	9/11/2001

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