

Fast CYP2E1 genotyping using automated fluorescent detection

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KEY WORDS

CYP2E1; PCR; Single Nucleotide Primer Extension (SNUPE)

SUMMARY

Background: Among polymorphic genes coding for xenobiotic-metabolizing enzymes, the ethanol-inducible CYP2E1 gene (1667 bp) is known to play a major role in the metabolism of several chemicals. **Objectives:** In order to apply large-scale genotyping, we explored the use of a Single Nucleotide Primer Extension (SNUPE) assay coupled with automated fluorescent detection to assess the presence of low-frequency CYP2E1*5B (c2) allele. **Methods:** a classic PCR-RFLP (Polymerase Chain Reaction-Restriction Fragment Length Polymorphism) method specific for polymorphic 5'-flanking region of CYP2E1 gene was tested in conjunction with a newly developed accelerated SNUPE assay. **Results:** compared to the classic PCR-RFLP method, the accelerated SNUPE assay proved to be both sensitive and specific for fast CYP2E1 genotyping. **Conclusions:** automated fluorescent methods as SNUPE assay are useful for public health perspectives, allowing rapid genotyping of metabolic genes in large population studies in clinical or epidemiological settings.