Identification of deletions and duplications in the CYP2E1 gene in a population in Goiânia – GO by MLPA

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Abstract/Resumo

Human CYPs are important in physiology, drug metabolism, and pathogenesis of certain diseases, and there is a strong need to explore the genetic variations and regulatory mechanisms of these enzymes. Mutations in CYP genes leading to deficiency in the enzymes result in a wide spectrum of human diseases. The aim of the present study was to investigate, the importance of CYP2E1 genetic polymorphisms in the health population in Goiânia. The study was performed acording approved by the ethics committee of Federal University of Goias (approval CEP: 895.552). In the current survey, 100 patients (35M/65F) of both genders and aged 18-90 (M: 30.2) years old were enrolled after receiving their informed consent. The Multiplex ligation-dependent probe amplification (MLPA) was performed using the P128 CYP450 MLPA kit (Version C1; MRC-Holland, Amsterdam, The Netherlands) according to the manufacturer's instructions. This gene is located on chromossome 10 and has 9 exons, but only 3 (5, 6 and 8) were analyzed. In the exon 5 was found (2 duplications and 3 deletions), exon 6 (2 duplications and 2 deletions) and the exon 8 (3 duplications and 2 deletions). This gene is the second responsible for the metabolism of alcohol in the body, after the enzyme alcohol dehydrogenase and therefore the importance of checking what these deletions and duplications can cause in the body. Is the first time this methodology is used, and we believe it is a great tool for future studies with other genes.

Keyword/Palavras-chave: Cytochrome P450; CYP2E1; Alcohol

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