



Short communication

Novel variant of *CYP2D6**6 is undetected by a commonly used genotyping procedure

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Abstract:

We report the identification of a novel and defective variant of the gene encoding cytochrome P450 2D6 (*CYP2D6*). This novel variant is a subtype of *CYP2D6**6 that was undetected by a commercially available 5' exonuclease-based assay. Because the novel variant was found in only one of 609 individuals, it represents a rare subtype of *CYP2D6**6 that may be restricted to a single family or a subpopulation. A procedure for the identification of the novel *CYP2D6**6 variant using restriction enzyme treatment of amplified fragments was developed.

Key words:

CYP2D6 genotyping, novel variant, 5' exonuclease assay
