Discordances Among CYP2D6 Genetic Test Results and Actual Patient Metabolic Phenotype

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Abstract

Cytochrome P450 2D6 (CYP2D6) is responsible in the metabolism of 25% of our clinically used drug. The gene is also highly polymorphic which can make accurately determining the correct metabolic phenotype difficult. Having the correct phenotype leads to greater clinical outcomes and can improve medication safety by preventing toxicities for patients. Several different factors can impact the predicated phenotype from a genetic assay. Drug induced phenoconversion is the primary route that discordances may appear. But other reasons such as disease states, hybrid alleles, copy number variations, or missing heritability can also be a reason. All these explanations can alter gene expression that our current genetic assays may not consider. This can lead to patients being mistreated and not having optimal pharmacotherapy.