

# Association recommends minimum set of pharmacogenetic alleles to guide clinical CYP2D6 genotype testing

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The Association for Molecular Pathology (AMP), the premier global, molecular diagnostic professional society, has today published consensus recommendations to aid in the design and validation of clinical CYP2D6 assays, promote standardization of testing across different laboratories and improve patient care. The manuscript, "Recommendations for Clinical CYP2D6 Genotyping Allele Selection: A Joint Consensus

Recommendation of the Association for Molecular Pathology, College of American Pathologists, Dutch Pharmacogenetics Working Group of the Royal Dutch Pharmacists Association, and European Society for Pharmacogenomics and Personalized Therapy," was released online ahead of publication in *The Journal of Molecular Diagnostics*.

The AMP Pharmacogenetics (PGx) Working Group is developing a series of guidelines designed to help standardize clinical testing for frequently used [genotyping](#) assays. The latest CYP2D6 report builds on the earlier recommendations for clinical genotyping of CYP2C19, CYP2C9, and genes important for warfarin testing. The recommendations should be implemented together with other relevant clinical guidelines such as those issued by the Clinical Pharmacogenetics Implementation Consortium (CPIC), Dutch Pharmacogenetics Working Group (DPWG), Canadian Pharmacogenomics Network for Drug Safety (CPNDS), and the American College of Medical Genetics and Genomics (ACMG), which mostly focus on the interpretation of PGx test results and therapeutic recommendations for specific drug-gene pairs.

"CYP2D6 is known to be responsible for the metabolism of many commonly prescribed medications including some antidepressants, atypical and typical antipsychotics, beta-blockers, opioids, antiemetics, and atomoxetine," said Victoria M. Pratt, Ph.D., Professor and Director of Pharmacogenetics and Molecular Genetics Laboratories, Indiana University School of Medicine, and AMP PGx Working Group Chair. "The ultimate goal of the AMP PGx Working Group is to promote standardization of PGx gene/allele testing across clinical laboratories. While the CYP2D6 gene is especially complex, with this latest report, we have been able to define a minimum set of variants that should be included in all future clinical CYP2D6 PGx genotyping assays."

Similar to the previous reports in the series, the CYP2D6 genotyping guideline offers a two-tier categorization of alleles that are

recommended for inclusion in clinical PGx genotyping assays. Using criteria such as allele frequencies in different populations and ethnicities, the availability of reference materials, and other technical considerations, the AMP PGx Working Group recommended a minimum set of alleles and their defining variants that should be included in all clinical CYP2D6 genotyping tests (Tier 1). The team also defined a Tier 2 list of optional alleles that do not currently meet one or more of the criteria for inclusion in Tier 1. These recommendations are meant to be a reference guide and not to be interpreted as a restrictive list. AMP intends to update these recommendations as new data and/or reference materials become available.

"AMP is committed to collaborating with the broader laboratory community to continuously improve professional PGx practices amidst a rapidly evolving molecular diagnostic landscape," said Antonia R. Sepulveda, MD, Ph.D., AMP President and Professor and Chair of the George Washington School of Medicine Department of Pathology. "Standardizing clinical testing for frequently used PGx genotyping assays will improve concordance across laboratories and enable healthcare professionals to provide high-quality patient care."

**More information:** Victoria M. Pratt et al, Recommendations for Clinical CYP2D6 Genotyping Allele Selection: A Joint Consensus Recommendation of the Association for Molecular Pathology, College of American Pathologists, Dutch Pharmacogenetics Working Group of the Royal Dutch Pharmacists Association, and European Society for Pharmacogenomics and Personalized Therapy, *The Journal of Molecular Diagnostics* (2021). [DOI: 10.1016/j.jmoldx.2021.05.013](https://doi.org/10.1016/j.jmoldx.2021.05.013)

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