

## Genetic Pharmacology Services Report References

### Psychiatry Pharmacogenetics Expanded Panel<sup>1-19</sup>

1. Bråten LS, Haslemo T, Jukic MM, Ingelman-Sundberg M, Molden E, Kringen MK. Impact of CYP2C19 genotype on sertraline exposure in 1200 Scandinavian patients. *Neuropsychopharmacology*. 2020;45(3):570-576. doi:10.1038/s41386-019-0554-x
2. Brown JT, Abdel-Rahman SM, Van Haandel L, Gaedigk A, Lin YS, Leeder JS. Single dose, CYP2D6 genotype-stratified pharmacokinetic study of atomoxetine in children with ADHD. *Clin Pharmacol Ther*. 2016;99(6):642-650. doi:10.1002/cpt.319
3. Hicks JK, Swen JJ, Thorn CF, et al. Clinical pharmacogenetics implementation consortium guideline for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants. *Clin Pharmacol Ther*. 2013;93(5):402-408. doi:10.1038/clpt.2013.2
4. Hicks JK, Bishop JR, Sangkuhl K, et al. Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for CYP2D6 and CYP2C19 genotypes and dosing of selective serotonin reuptake inhibitors. *Clin Pharmacol Ther*. 2015;98(2):127-134. doi:10.1002/cpt.147
5. Hicks JK, Sangkuhl K, Swen JJ, et al. Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2D6 and CYP2C19 genotypes and dosing of tricyclic antidepressants: 2016 update. *Clin Pharmacol Ther*. 2017;102(1):37-44. doi:10.1002/cpt.597
6. Jukić MM, Haslemo T, Molden E, Ingelman-Sundberg M. Impact of CYP2C19 genotype on escitalopram exposure and therapeutic failure: A retrospective study based on 2,087 patients. *Am J Psychiatry*. 2018;175(5):463-470. doi:10.1176/appi.ajp.2017.17050550
7. Mrazek DA, Lerman C. Facilitating Clinical Implementation of Pharmacogenomics. 2011;306(3):304-305.
8. Pratt VM, Del Tredici AL, Hachad H, et al. Recommendations for Clinical CYP2C19 Genotyping Allele Selection: A Report of the Association for Molecular Pathology. *J Mol Diagnostics*. 2018;20(3):269-276. doi:10.1016/j.jmoldx.2018.01.011
9. Stingl JC, Brockmüller J, Viviani R. Genetic variability of drug-metabolizing enzymes: The dual impact on psychiatric therapy and regulation of brain function. *Mol Psychiatry*. 2013;18(3):273-287. doi:10.1038/mp.2012.42
10. Botton MR, Whirl-Carrillo M, Del Tredici AL, et al. PharmVar GeneFocus: CYP2C19. *Clin Pharmacol Ther*. 2021;109(2):352-366. doi:10.1002/cpt.1973
11. Brown JT, Bishop JR, Sangkuhl K, et al. Clinical Pharmacogenetics Implementation Consortium Guideline for Cytochrome P450 (CYP)2D6 Genotype and Atomoxetine Therapy. *Clin Pharmacol Ther*. 2019;106(1):94-102. doi:10.1002/cpt.1409
12. Caudle KE, Sangkuhl K, Whirl-Carrillo M, et al. Standardizing CYP2D6 Genotype to Phenotype Translation: Consensus Recommendations from the Clinical Pharmacogenetics Implementation Consortium and Dutch Pharmacogenetics Working Group. *Clin Transl Sci*. 2020;13(1):116-124. doi:10.1111/cts.12692
13. CDER FDA. HIGHLIGHTS OF PRESCRIBING INFORMATION for ABILIFY.  
[https://www.accessdata.fda.gov/drugsatfda\\_docs/label/2014/021436s038,021713s030,021729s022,021866s023lbl.pdf](https://www.accessdata.fda.gov/drugsatfda_docs/label/2014/021436s038,021713s030,021729s022,021866s023lbl.pdf). Published 2014. Accessed March 16, 2020.

## Genetic Pharmacology Services Report References

14. Chang M, Tybring G, Dahl ML, Lindh JD. Impact of Cytochrome P450 2C19 Polymorphisms on Citalopram/Escitalopram Exposure: A Systematic Review and Meta-Analysis. *Clin Pharmacokinet*. 2014;53(9):801-811. doi:10.1007/s40262-014-0162-1
15. DailyMed - ATOMOXETINE- atomoxetine hydrochloride capsule.  
<https://dailymed.nlm.nih.gov/dailymed/drugInfo.cfm?setid=48980c85-2fa0-4c9e-b182-227b0d057f6b>.  
Accessed May 28, 2020.
16. DailyMed - THIORIDAZINE HYDROCHLORIDE tablet, film coated.  
<https://dailymed.nlm.nih.gov/dailymed/drugInfo.cfm?setid=56b3f4c2-52af-4947-b225-6808ae9f26f5>.  
Accessed May 28, 2020.
17. Falzoi M, Mossa A, Congeddu E, Saba L, Pani L. Multiplex genotyping of CYP3A4, CYP3A5, CYP2C9 and CYP2C19 SNPs using MALDI-TOF mass spectrometry. *Pharmacogenomics*. 2010;11(4):559-571.  
doi:10.2217/pgs.09.172
18. Falzoi M, Pira L, Lazzari P, Pani L. Genotyping of CYP2D6 Polymorphisms by MALDI-TOF Mass Spectrometry in Sardinian People. *ISRN Genet*. 2013:609797. doi:10.5402/2013/609797
19. Bousman, C. A., Stevenson, J. M., Ramsey, L. B., Sangkuhl, K., Hicks, J. K., Strawn, J. R., Singh, A. B., Rúaño, G., Mueller, D. J., Tsermpini, E. E., Brown, J. T., Bell, G. C., Leeder, J. S., Gaedigk, A., Scott, S. A., Klein, T. E., Caudle, K. E., & Bishop, J. R. (2023). Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. *Clinical pharmacology and therapeutics*, 114(1), 51–68.

### CYP2D6 Opioid Panel<sup>1-9</sup>

1. Falzoi M, Mossa A, Congeddu E, Saba L, Pani L. Multiplex genotyping of CYP3A4, CYP3A5, CYP2C9 and CYP2C19 SNPs using MALDI-TOF mass spectrometry. *Pharmacogenomics*. 2010;11(4):559-571.  
doi:10.2217/pgs.09.172
2. Falzoi M, Pira L, Lazzari P, Pani L. Genotyping of CYP2D6 Polymorphisms by MALDI-TOF Mass Spectrometry in Sardinian People. *ISRN Genet*. 2013:609797. doi:10.5402/2013/609797
3. Nofziger C, Turner AJ, Sangkuhl K, et al. PharmVar GeneFocus: CYP2D6. *Clin Pharmacol Ther*. 2020;107(1):154-170. doi:10.1002/cpt.1643
4. Crews KR, Caudle KE, Dunnenberger HM, Sadhasivam S, Skaar TC. Considerations for the utility of the CPIC guideline for CYP2D6 genotype and codeine therapy. *Clin Chem*. 2015.  
doi:10.1373/clinchem.2014.237412
5. Caudle KE, Sangkuhl K, Whirl-Carrillo M, et al. Standardizing CYP2D6 Genotype to Phenotype Translation: Consensus Recommendations from the Clinical Pharmacogenetics Implementation Consortium and Dutch Pharmacogenetics Working Group. *Clin Transl Sci*. 2020;13(1):116-124. doi:10.1111/cts.12692
6. Crews KR, Gaedigk A, Dunnenberger HM, et al. Clinical pharmacogenetics implementation consortium guidelines for cytochrome P450 2D6 genotype and codeine therapy: 2014 Update. *Clin Pharmacol Ther*. 2014. doi:10.1038/clpt.2013.254

## Genetic Pharmacology Services Report References

7. Crews KR, Gaedigk A, Dunnenberger HM, et al. Clinical pharmacogenetics implementation consortium (CPIC) guidelines for codeine therapy in the context of cytochrome P450 2D6 (CYP2D6) genotype. *Clin Pharmacol Ther.* 2012;91(2):321-326. doi:10.1038/clpt.2011.287
8. FDA (Food and Drug Administration). *Safety Review Update of Codeine Use in Children; New Boxed Warning and Contraindication on Use after Tonsillectomy and/or Adenoidectomy.* <https://www.fda.gov/media/85072/download>. Accessed March 31, 2020.
9. Crews KR, Monte AA, Huddart R, et al. Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for CYP2D6 , OPRM1 , and COMT genotype and select opioid therapy . *Clin Pharmacol Ther.* January 2021. doi:10.1002/cpt.2149

### CYP2D6/2C19 Genotype Analysis<sup>1-5</sup>

1. Falzoi M, Mossa A, Congeddu E, Saba L, Pani L. Multiplex genotyping of CYP3A4, CYP3A5, CYP2C9 and CYP2C19 SNPs using MALDI-TOF mass spectrometry. *Pharmacogenomics.* 2010;11(4):559-571. doi:10.2217/pgs.09.172
2. Falzoi M, Pira L, Lazzari P, Pani L. Genotyping of CYP2D6 Polymorphisms by MALDI-TOF Mass Spectrometry in Sardinian People. *ISRN Genet.* 2013:609797. doi:10.5402/2013/609797
3. Nofziger C, Turner AJ, Sangkuhl K, et al. PharmVar GeneFocus: CYP2D6. *Clin Pharmacol Ther.* 2020;107(1):154-170. doi:10.1002/cpt.1643
4. Botton MR, Whirl-Carrillo M, Del Tredici AL, et al. PharmVar GeneFocus: CYP2C19. *Clin Pharmacol Ther.* 2021;109(2):352-366. doi:10.1002/cpt.1973
5. Caudle KE, Sangkuhl K, Whirl-Carrillo M, et al. Standardizing CYP2D6 Genotype to Phenotype Translation: Consensus Recommendations from the Clinical Pharmacogenetics Implementation Consortium and Dutch Pharmacogenetics Working Group. *Clin Transl Sci.* 2020;13(1):116-124. doi:10.1111/cts.12692

### CYP3A5 Genotype Analysis (tacrolimus)<sup>1-8</sup>

1. Birdwell KA, Decker B, Barbarino JM, et al. Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for CYP3A5 genotype and tacrolimus dosing. *Clin Pharmacol Ther.* 2015;98(1):19-24. doi:10.1002/cpt.113
2. Brunet M, van Gelder T, Åsberg A, et al. Therapeutic Drug Monitoring of Tacrolimus-Personalized Therapy. *Ther Drug Monit.* 2019;41(3):261-307. doi:10.1097/FTD.0000000000000640
3. Elens L, Capron A, Van Schaik R, et al. Impact of CYP3A4\*22 allele on tacrolimus pharmacokinetics in early period after renal transplantation: Toward updated genotype-based dosage guidelines. *Ther Drug Monit.* 2013;35(5):608-616. doi:http://dx.doi.org/10.1097/FTD.0b013e318296045b
4. Hooper DK, Fukuda T, Gardiner R, et al. Risk of tacrolimus toxicity in CYP3A5 nonexpressors treated with intravenous nicardipine after kidney transplantation. *Transplantation.* 2012;93(8):806-812. doi:10.1097/TP.0b013e318247a6c7

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5. Hooper DK, Carle AC, Schuchter J, Goebel J. Interaction between tacrolimus and intravenous nicardipine in the treatment of post-kidney transplant hypertension at pediatric hospitals. *Pediatr Transplant*. 2011;15(1):88-95. doi:10.1111/j.1399-3046.2010.01417.x
6. Tang JT, Andrews LM, van Gelder T, et al. Pharmacogenetic aspects of the use of tacrolimus in renal transplantation: recent developments and ethnic considerations. *Expert Opin Drug Metab Toxicol*. 2016;12(5):555-565. doi:10.1517/17425255.2016.1170808
7. Min S, Papaz T, Lafreniere-Roula M, et al. A randomized clinical trial of age and genotype-guided tacrolimus dosing after pediatric solid organ transplantation. *Pediatr Transplant*. 2018;22(7):1-9. doi:10.1111/ptr.13285
8. Saint-Marcoux F, Woillard J-B, Jurado C, Marquet P. Lessons From Routine Dose Adjustment of Tacrolimus in Renal Transplant Patients Based on Global Exposure. *Ther Drug Monit*. 2013;35(3):322-327. doi:10.1097/FTD.0b013e318285e779

### Thiopurine Pharmacogenetics Analysis<sup>1-5</sup>

1. Falzoi, M., Pira, L., Lazzari, P. & Pani, L. Genotyping of CYP2D6 Polymorphisms by MALDI-TOF Mass Spectrometry in Sardinian People. *ISRN Genet*. 609797 (2013) doi:10.5402/2013/609797.
2. Falzoi, M., Mossa, A., Congeddu, E., Saba, L. & Pani, L. Multiplex genotyping of CYP3A4, CYP3A5, CYP2C9 and CYP2C19 SNPs using MALDI-TOF mass spectrometry. *Pharmacogenomics* 11, 559–571 (2010).
3. Lennard, L. Implementation of TPMT testing. *Br. J. Clin. Pharmacol*. 77, 704–714 (2014).
4. Relling, M. V. et al. Clinical Pharmacogenetics Implementation Consortium Guideline for Thiopurine Dosing Based on TPMT and NUDT15 Genotypes: 2018 Update. *Clin. Pharmacol. Ther*. 105, 1095–1105 (2019).
5. Yang, J. J. et al. Inherited NUDT15 variant is a genetic determinant of mercaptopurine intolerance in children with acute lymphoblastic leukemia. *J. Clin. Oncol*. 33, 1235–1242 (2015).

### Warfarin Pharmacogenetics<sup>1-5</sup>

1. Falzoi M., Mossa, A., et al. (2010). Multiplex genotyping of CYP3A4, CYP3A5, CYP2C9 and CYP2C19 SNPs using MALDI-TOF mass spectrometry. *Pharmacogenomics* 11(4):559-71
2. Pratt V.M., Cavallari, L.H., et al. (2019) Recommendations for clinical CYP2C9 genotyping allele selection. *J Mol Diagn* 21(5):746-755
3. Johnson J.A., Caudle K.E., et al. (2017) Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Pharmacogenetics-Guided Warfarin Dosing: 2017 Update. *Clin Pharmacol Ther* 102(3):397–404
4. Hamberg, A.K., Wadelius, M., et al. (2014) Characterizing variability in warfarin dose requirements in children using modelling and simulation. *Br J Clin Pharmacol* 78:158–169
5. Shaw, K., Amstutz, U., et al. (2014). VKORC1 and CYP2C9 genotypes are predictors of warfarin-related outcomes in children. *Pediatr Blood Cancer* 61(6):1055-62