



## Medical Genetics Summaries Expert Reviewers

Laura Dean, MD<sup>1</sup>

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Alzghari, Saeed, MS, MBA, PharmD; Gulfstream Genomics, Dallas, TX, USA

- Carbamazepine Therapy and *HLA* Genotype
- Carvedilol Therapy and *CYP2D6* Genotype
- Warfarin Therapy and *VKORC1* and *CYP* Genotype

Amstutz, Ursula, PhD; University of Bern, Bern, Switzerland

- Carbamazepine Therapy and *HLA* Genotype

Appell, Malin Lindqvist, PhD; Linköping University, Linköping, Sweden

- Thioguanine Therapy and *TPMT* Genotype

Beam, Teresa, PhD; Manchester University, North Manchester, IN, USA

- Sofosbuvir Therapy and *IFNL4* Genotype

Beckers, Albert, MD, PhD; University of Liège, Liège, Belgium

- McCune-Albright Syndrome

Bitner-Glindzicz, Maria, PhD, FRCP; University College London, London, UK

- Gentamicin Therapy and *MT-RNR1* Genotype

Brauch, Hiltrud, PhD; Dr. Margarete Fischer-Bosch-Institute of Clinical Pharmacology (IKP), Stuttgart, Germany

- Tamoxifen Therapy and *CYP2D6* Genotype

Burstein, Harold, MD, PhD; Harvard Medical School, Boston, MA, USA

- Tamoxifen Therapy and *CYP2D6* Genotype

Callaghan, John, MD, PhD; Indiana University School of Medicine, Indianapolis, IN, USA

- Carisoprodol Therapy and *CYP2C19* Genotype
- Propafenone Therapy and *CYP2D6* Genotype
- Risperidone Therapy and *CYP2D6* Genotype

Carpenter, William, MD; University of Maryland School of Medicine, Baltimore, MD, USA

- Clozapine Therapy and *CYP2D6*, *CYP1A2*, and *CYP3A4* Genotype

Cavallari, Lisa, PharmD; University of Florida, Gainesville, FL, USA

- Clopidogrel Therapy and *CYP2C19* Genotype
- Metoprolol Therapy and *CYP2D6* Genotype

Chapman, Paul, MD; Memorial Sloan Kettering Cancer Center, New York, NY, USA

- Vemurafenib Therapy and *BRAF* and *NRAS* Genotype

Crews, Kristine, PharmD; St. Jude Children's Research Hospital, Memphis, TN, USA

- Codeine Therapy and *CYP2D6* Genotype

Cronin-Fenton, Deirdre, PhD; Aarhus University, Aarhus, Denmark

- Tamoxifen Therapy and *CYP2D6* Genotype

Daly, Adrian, MBBch, PhD; University of Liège, Liège, Belgium

- McCune-Albright Syndrome

Ehret, Megan, MS, PharmD; Fort Belvoir Community Hospital, Fort Belvoir, VA, USA

- Aripiprazole Therapy and *CYP2D6* Genotype
- Diazepam Therapy and *CYP2C19* Genotype

Esquivel, Bernard, MD, MHA, PhD; Latin American Association for Personalized Medicine, Mexico

- Carvedilol Therapy and *CYP2D6* Genotype
- Esomeprazole Therapy and *CYP2C19* Genotype
- Warfarin Therapy and *VKORC1* and *CYP* Genotype

Eugene, Andy; MD, PhD; Bernard J. Dunn School of Pharmacy, Shenandoah University - Fairfax Inova Center for Personalized Health, Fairfax, VA, USA

- Lesinurad Therapy and *CYP2C9* Genotype

Formea, Christine, PharmD, BCPS, FCCP, FASHP; Mayo Clinic College of Medicine and Science, Rochester, MN, USA

- Warfarin Therapy and *VKORC1* and *CYP* Genotype

Gaedigk, Andrea, MS, PhD; University of Missouri, Kansas City, MO, USA

- Aripiprazole Therapy and *CYP2D6* Genotype
- Atomoxetine Therapy and *CYP2D6* Genotype

Gage, Brian, MD, MSc; Washington University, St. Louis, MO, USA

- Warfarin Therapy and *VKORC1* and *CYP* Genotype

Grond, Stefan, MD; Klinikum Lippe, Lemgo, Germany

- Tramadol Therapy and *CYP2D6* Genotype

Gupta, Anita, DO, PharmD; Drexel University College of Medicine, Philadelphia, PA, USA

- Celecoxib Therapy and *CYP2C9* Genotype

Hachad, Houda, PharmD; Oregon Health & Science University, Portland, OR, USA

- Flurbiprofen Therapy and *CYP2C9* Genotype
- Lesinurad Therapy and *CYP2C9* Genotype
- Piroxicam Therapy and *CYP2C9* Genotype
- Risperidone Therapy and *CYP2D6* Genotype

Hardison, Matthew, PhD, FACMG; Aegis Sciences Corporation, Nashville, TN, USA

- Dabrafenib Therapy and *BRAF* and *G6PD* Genotype
- Vemurafenib Therapy and *BRAF* and *NRAS* Genotype

Henrich, Timothy, MD; Brigham and Women's Hospital, Cambridge, MA, USA

- Maraviroc Therapy and *CCR5* Genotype

Henricks, Linda, PharmD; The Netherlands Cancer Institute, Amsterdam, Netherlands

- Capecitabine Therapy and *DPYD* Genotype

Hickey, Scott, MD; Ohio State University, Columbus, OH, USA

- Methylenetetrahydrofolate Reductase Deficiency

Holsappel, Inge, MSc; Royal Dutch Pharmacists Association (KNMP), The Hague, Netherlands

- Abacavir Therapy and *HLA-B\*57:01* Genotype
- Carbamazepine Therapy and *HLA* Genotype
- Carvedilol Therapy and *CYP2D6* Genotype
- Clopidogrel Therapy and *CYP2C19* Genotype
- Esomeprazole Therapy and *CYP2C19* Genotype
- Tamoxifen Therapy and *CYP2D6* Genotype
- Warfarin Therapy and *VKORC1* and *CYP* Genotype

Hicks, David, MD; University of Rochester Medical Center, Rochester, NY, USA

- Trastuzumab (Herceptin) Therapy and *ERBB2 (HER2)* Genotype

Hudis, Clifford, MD; Weill Cornell Medical College, New York, NY, USA

- Trastuzumab (Herceptin) Therapy and *ERBB2 (HER2)* Genotype

Iwuchukwu, Otito F., RPh, PhD; Fairleigh Dickinson University School of Pharmacy, Florham Park, NJ, USA

- Irinotecan Therapy and *UGT1A1* Genotype

Johnson, Douglas, MD; Vanderbilt University Medical Center, Nashville, TN, USA

- Dabrafenib Therapy and *BRAF* and *G6PD* Genotype

Jönsson, Erik, MD, PhD; Karolinska Institutet, Stockholm, Sweden

- Schizophrenia

Joshi, Avadhut, PhD; Translational Software, Bellevue, WA, USA

- Dabrafenib Therapy and *BRAF* and *G6PD* Genotype

Kawedia, Jitesh, PhD; University of Texas MD Anderson Cancer Center, Houston, TX, USA

- Diazepam Therapy and *CYP2C19* Genotype
- Simeprevir Therapy and *IFNL3* Genotype

Kaye, Alan, MD, PhD; Louisiana State University Health Sciences Center, New Orleans, LA, USA

- Tramadol Therapy and *CYP2D6* Genotype

Kim, Hyun, PharmD; Boston Children's Hospital, Boston, MA, USA

- Carbamazepine Therapy and *HLA* Genotype

Kisor, David, PharmD; Manchester University, North Manchester, IN, USA

- Amitriptyline Therapy and *CYP2D6* and *CYP2C19* Genotype
- Imipramine Therapy and *CYP2D6* and *CYP2C19* Genotype
- Sofosbuvir Therapy and *IFNL4* Genotype
- Thioridazine Therapy and *CYP2D6* Genotype

Kong, Ben, PharmD; Oregon Health & Science University, Portland, OR, USA

- Brivaracetam Therapy and *CYP2C19* Genotype
- Carisoprodol Therapy and *CYP2C19* Genotype
- Irinotecan Therapy and *UGT1A1* Genotype
- Lacosamide Therapy and *CYP2C19* Genotype
- Propafenone Therapy and *CYP2D6* Genotype
- Risperidone Therapy and *CYP2D6* Genotype

Laan, Ellen T.M., PhD; Amsterdam University Medical Centre, Amsterdam, Netherlands

- Flibanserin Therapy and *CYP2C19* Genotype

Lagging, Martin, MD, PhD; University of Gothenburg, Gothenburg, Sweden

- Sofosbuvir Therapy and *IFNL4* Genotype

Langer, Nicolas, PhD; Child Mind Institute, New York, NY, USA

- ACHOO Syndrome

Lee, Seok-Yang, PharmD; Sungkyunkwan University, Seoul, Republic of Korea

- Flurbiprofen Therapy and *CYP2C9* Genotype

Leeder, Steven, PharmD, PhD; Children's Mercy Hospital, Kansas City, MO, USA

- Aripiprazole Therapy and *CYP2D6* Genotype

Lennard, Lynne, PhD; University of Sheffield, Sheffield, UK

- Thioguanine Therapy and *TPMT* Genotype

Linder, Mark, PhD; University of Louisville, Louisville, KY, USA

- Risperidone Therapy and *CYP2D6* Genotype

Lipkowitz, Stanley, MD, PhD; National Cancer Institute, Bethesda, MD, USA

- Trastuzumab (Herceptin) Therapy and *ERBB2 (HER2)* Genotype

Lively, Tracy, PhD; National Cancer Institute, Bethesda, MD, USA

- Trastuzumab (Herceptin) Therapy and *ERBB2 (HER2)* Genotype

Malhotra, Anil, MD; Hofstra Northwell School of Medicine, Hempstead, NY, USA

- Clozapine Therapy and *CYP2D6, CYP1A2, and CYP3A4* Genotype

Manzi, Shannon, PharmD, BCPPS; Boston Children's Hospital, Boston, MA, USA

- Gentamicin Therapy and *MT-RNR1* Genotype

McGill, Neil William, MD; Royal Prince Alfred Hospital, Sydney, NSW, Australia

- Lesinurad Therapy and *CYP2C9* Genotype

Merl, Man Yee, PharmD, BCOP; Yale-New Haven Hospital, New Haven, CT, USA

- Irinotecan Therapy and *UGT1A1* Genotype

Molden, Espen, PhD; University of Oslo, Oslo, Norway

- Risperidone Therapy and *CYP2D6* Genotype

Morton, Jenny, PhD, ScD; University of Cambridge, Cambridge, UK

- Deutetrabenazine Therapy and *CYP2D6* Genotype

Mukerjee, Gouri, PhD; Geneyouin Inc., Toronto, Canada

- Brivaracetam Therapy and *CYP2C19* Genotype
- Carisoprodol Therapy and *CYP2C19* Genotype
- Lacosamide Therapy and *CYP2C19* Genotype
- Propafenone Therapy and *CYP2D6* Genotype

Müller, Daniel, MD, PhD; University of Toronto, Canada

- Clozapine Therapy and *CYP2D6, CYP1A2, and CYP3A4* Genotype

Murphy, Michael, MD; University of Oxford, Oxford, UK

- ABO Blood Group

Nagy, Mohamed, BPharm; Children's Cancer Hospital, Cairo, Egypt

- Amitriptyline Therapy and *CYP2D6* and *CYP2C19* Genotype
- Capecitabine Therapy and *DPYD* Genotype
- Carbamazepine Therapy and *HLA* Genotype
- Carisoprodol Therapy and *CYP2C19* Genotype
- Fluorouracil Therapy and *DPYD* Genotype
- Flurbiprofen Therapy and *CYP2C9* Genotype
- Imipramine Therapy and *CYP2D6* and *CYP2C19* Genotype
- Lesinurad Therapy and *CYP2C9* Genotype
- Phenytoin Therapy and *HLA-B\*15:02* and *CYP2C9* Genotype
- Piroxicam Therapy and *CYP2C9* Genotype
- Prasugrel Therapy and *CYP* Genotype
- Propafenone Therapy and *CYP2D6* Genotype
- Risperidone Therapy and *CYP2D6* Genotype

- [Thioridazine Therapy and \*CYP2D6\* Genotype](#)

O'Brien, Thomas, MD, MPH; National Cancer Institute, Bethesda, MD, USA

- [Sofosbuvir Therapy and \*IFNL4\* Genotype](#)

Obeng, Aniwaa Owusu, PharmD; Icahn School of Medicine at Mount Sinai, New York, NY, USA

- [Maraviroc Therapy and \*CCR5\* Genotype](#)
- [Prasugrel Therapy and \*CYP\* Genotype](#)

Ostrov, David A., PhD; University of Florida College of Medicine, Gainesville, FL, USA

- [Abacavir Therapy and \*HLA-B\\*57:01\* Genotype](#)

Patel, Jai N., PharmD, BCOP; Levine Cancer Institute, Charlotte, NC, USA

- [Irinotecan Therapy and \*UGT1A1\* Genotype](#)

Patrinos, George, PhD; University of Patras, Patra, Greece

- [Fluorouracil Therapy and \*DPYD\* Genotype](#)
- [Risperidone Therapy and \*CYP2D6\* Genotype](#)
- [Warfarin Therapy and \*VKORC1\* and \*CYP\* Genotype](#)

Pauli, Emily, PharmD; Clearview Cancer Institute, Huntsville, AL, USA

- [Capecitabine Therapy and \*DPYD\* Genotype](#)
- [Phenytoin Therapy and \*HLA-B\\*15:02\* and \*CYP2C9\* Genotype](#)

Pawloski, Pamala, PharmD; HealthPartners Institute, Bloomington, MN, USA

- [Dabrafenib Therapy and \*BRAF\* and \*G6PD\* Genotype](#)
- [Vemurafenib Therapy and \*BRAF\* and \*NRAS\* Genotype](#)

Phillips, Elizabeth, MD, FIDSA; Vanderbilt University Medical Center, Nashville, TN, USA

- [Abacavir Therapy and \*HLA-B\\*57:01\* Genotype](#)

Pirmohamed, Munir, MD, PhD; University of Liverpool, Liverpool, UK

- [Abacavir Therapy and \*HLA-B\\*57:01\* Genotype](#)

Pollock, Bruce, MD, PhD; University of Toronto, Toronto, Canada

- [Venlafaxine Therapy and \*CYP2D6\* Genotype](#)

Pratt, Victoria, PhD, FACMG; Indiana University School of Medicine, Indianapolis, IN, USA

- [Fluorouracil Therapy and \*DPYD\* Genotype](#)
- [Irinotecan Therapy and \*UGT1A1\* Genotype](#)
- [Maraviroc Therapy and \*CCR5\* Genotype](#)
- [Prasugrel Therapy and \*CYP\* Genotype](#)

Purcell, Anthony, PhD; Monash University, Melbourne, Australia

- [Abacavir Therapy and \*HLA-B\\*57:01\* Genotype](#)

Rahman, Shamima, PhD, FRCP; University College London, London, UK

- Gentamicin Therapy and *MT-RNR1* Genotype

Rashed, Rashed, BCNSP, MSc; Children's Cancer Hospital, Cairo, Egypt

- Irinotecan Therapy and *UGT1A1* Genotype

Rogawski, Michael, MD, PhD; University of California, Davis, CA, USA

- Brivaracetam Therapy and *CYP2C19* Genotype
- Carbamazepine Therapy and *HLA* Genotype
- Clobazam Therapy and *CYP2C19* Genotype
- Phenytoin Therapy and *HLA-B\*15:02* and *CYP2C9* Genotype

Rosenzweig, Mark, PhD; Foundation Medicine Inc., Cambridge, MA, USA

- Dabrafenib Therapy and *BRAF* and *G6PD* Genotype
- Vemurafenib Therapy and *BRAF* and *NRAS* Genotype

Ross, Lisa, MS; ViiV Healthcare, Durham, NC, USA

- Abacavir Therapy and *HLA-B\*57:01* Genotype

Saab, Yolande, PharmD, PhD; Lebanese American University, Beirut, Lebanon

- Amitriptyline Therapy and *CYP2D6* and *CYP2C19* Genotype
- Imipramine Therapy and *CYP2D6* and *CYP2C19* Genotype
- Thioridazine Therapy and *CYP2D6* Genotype

Saruwatari, Junji, PhD; Kumamoto University, Kumamoto, Japan

- Clobazam Therapy and *CYP2C19* Genotype

Schellens, Jan, MD, PhD; The Netherlands Cancer Institute, Amsterdam, Netherlands

- Capecitabine Therapy and *DPYD* Genotype

Schneeweiss, Andreas, MD; Heidelberg University Hospital, Heidelberg, Germany

- Pertuzumab Therapy and *ERBB2 (HER2)* Genotype

Schroth, Werner, PhD; Institute of Clinical Pharmacology, Stuttgart, Germany

- Tamoxifen Therapy and *CYP2D6* Genotype

Schulman, Sol, MD; Harvard Medical School, Boston, MA, USA

- Warfarin Therapy and *VKORC1* and *CYP* Genotype

Scott, Stuart, PhD, ABMGG; Icahn School of Medicine at Mount Sinai, New York, NY, USA

- Allopurinol Therapy and *HLA-B\*58:01* Genotype
- Amitriptyline Therapy and *CYP2D6* and *CYP2C19* Genotype
- Azathioprine Therapy and *TPMT* Genotype
- Celecoxib Therapy and *CYP2C9* Genotype
- Clopidogrel Therapy and *CYP2C19* Genotype
- Deutetrabenazine Therapy and *CYP2D6* Genotype
- Diazepam Therapy and *CYP2C19* Genotype
- Esomeprazole Therapy and *CYP2C19* Genotype



- Flibanserin Therapy and *CYP2C19* Genotype
- Gentamicin Therapy and *MT-RNR1* Genotype
- Imipramine Therapy and *CYP2D6* and *CYP2C19* Genotype
- Lacosamide Therapy and *CYP2C19* Genotype
- Mercaptopurine Therapy and *TPMT* Genotype
- Omeprazole Therapy and *CYP2C19* Genotype
- Phenytoin Therapy and *HLA-B\*15:02* and *CYP2C9* Genotype
- Simeprevir Therapy and *IFNL3* Genotype
- Tamoxifen Therapy and *CYP2D6* Genotype
- Thioguanine Therapy and *TPMT* Genotype
- Venlafaxine Therapy and *CYP2D6* Genotype

Siasos, Gerasimos, MD, PhD, FCCP, FACC; Brigham and Women's Hospital, Boston, MA, USA

- Clopidogrel Therapy and *CYP2C19* Genotype

Skaar, Todd, PhD; Indiana University, Bloomington, IN, USA

- Codeine Therapy and *CYP2D6* Genotype

Stockis, Armel, UCB Pharma, Brussels, Belgium

- Brivaracetam Therapy and *CYP2C19* Genotype

Sukasem, Chonlaphat, PhD; Mahidol University, Bangkok, Thailand

- Carbamazepine Therapy and *HLA* Genotype
- Risperidone Therapy and *CYP2D6* Genotype

Sweatt, J David, PhD; Vanderbilt University, Nashville, TN, USA

- Pitt-Hopkins Syndrome

Thirumaran, Ranjit, MPharm, PhD; Genelex Labs, Seattle, WA, USA

- Imipramine Therapy and *CYP2D6* and *CYP2C19* Genotype

Thompson, Alex, MD, PhD; Duke University Medical Centre, Durham, NC, USA

- Simeprevir Therapy and *IFNL3* Genotype

Trenk, Dietmar, PhD; Albert Ludwig University of Freiburg, Freiburg, Germany

- Clopidogrel Therapy and *CYP2C19* Genotype

Tsunedomi, Ryouichi, PhD; Yamaguchi University Graduate School of Medicine, Yamaguchi, Japan

- Irinotecan Therapy and *UGT1A1* Genotype

Uppugunduri, Chakradhara, PhD; University of Geneva, Geneva, Switzerland

- Amitriptyline Therapy and *CYP2D6* and *CYP2C19* Genotype
- Celecoxib Therapy and *CYP2C9* Genotype
- Flurbiprofen Therapy and *CYP2C9* Genotype
- Diazepam Therapy and *CYP2C19* Genotype
- Imipramine Therapy and *CYP2D6* and *CYP2C19* Genotype
- Lesinurad Therapy and *CYP2C9* Genotype
- Piroxicam Therapy and *CYP2C9* Genotype



- [Warfarin Therapy and \*VKORC1\* and \*CYP\* Genotype](#)

van Rhenen, Mandy, MSc; Royal Dutch Pharmacists Association (KNMP), The Hague, Netherlands

- [Amitriptyline Therapy and \*CYP2D6\* and \*CYP2C19\* Genotype](#)
- [Imipramine Therapy and \*CYP2D6\* and \*CYP2C19\* Genotype](#)
- [Metoprolol Therapy and \*CYP2D6\* Genotype](#)
- [Propafenone Therapy and \*CYP2D6\* Genotype](#)
- [Risperidone Therapy and \*CYP2D6\* Genotype](#)

Visk, DeeAnn, PhD; San Diego, CA, USA

- [Propafenone Therapy and \*CYP2D6\* Genotype](#)

Wadelius, Mia, PhD; Uppsala University, Uppsala, Sweden

- [Allopurinol Therapy and \*HLA-B\\*58:01\* Genotype](#)
- [Atomoxetine Therapy and \*CYP2D6\* Genotype](#)
- [Esomeprazole Therapy and \*CYP2C19\* Genotype](#)
- [Irinotecan Therapy and \*UGT1A1\* Genotype](#)
- [Omeprazole Therapy and \*CYP2C19\* Genotype](#)

Wainberg, Mark, PhD, OC, OQ, FRSC; McGill University, Montreal, Quebec, Canada

- [Maraviroc Therapy and \*CCR5\* Genotype](#)

Wikstrand, John, MD, PhD; University of Gothenburg, Gothenburg, Sweden

- [Metoprolol Therapy and \*CYP2D6\* Genotype](#)

Yin, Ji-Ye; Xiangya Hospital, Central South University, Changsha, Hunan Province, China

- [Irinotecan Therapy and \*UGT1A1\* Genotype](#)

Zujewski, Jo Anne, MD; National Cancer Institute, Bethesda, MD, USA

- [Pertuzumab Therapy and \*ERBB2 \(HER2\)\* Genotype](#)

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