Stanford

Stuart A. Scott

Professor of Pathology

CONTACT INFORMATION

- **Administrative Contact**

  Mirela Puljic - Administrative Associate

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Bio

ACADEMIC APPOINTMENTS

- Professor - University Medical Line, Pathology
- Member, Maternal & Child Health Research Institute (MCHRI)

ADMINISTRATIVE APPOINTMENTS

- Director, Clinical Genomics Laboratory, (2020- present)

HONORS AND AWARDS

- William K. Bowes Jr Award in Medical Genetics, Partners HealthCare Center for Personalized Genetic Medicine, Harvard Medical School (2011)
- Dr. Harold and Golden Lamport Research Award, Icahn School of Medicine at Mount Sinai (2012)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Steering Committee, Pharmacogene Variation Consortium (PharmVar) (2016 - present)
- Member, Association for Molecular Pathology (AMP) Pharmacogenomics (AMP PGx) Working Group (2016 - present)
- Steering Committee, Clinical Pharmacogenetics Implementation Consortium (CPIC) (2019 - present)
- Scientific Editor, Medical Genetics Summaries (MGS); NCBI/NIH, National Library of Medicine (NLM) (2019 - present)

PROFESSIONAL EDUCATION

- Ph.D., University of Saskatchewan, Saskatoon, SK, Canada , Department of Pathology (2005)
- Fellowship, Icahn School of Medicine at Mount Sinai, New York, NY , Clinical Molecular Genetics and Genomics (2005)
- Board Certification, American Board of Medical Genetics and Genomics (ABMGG) , Clinical Molecular Genetics and Genomics (2007)
- Fellowship, Icahn School of Medicine at Mount Sinai, New York, NY , Clinical Cytogenetics and Genomics (2007)
- Board Certification, American Board of Medical Genetics and Genomics (ABMGG) , Clinical Cytogenetics and Genomics (2009)
- M.S., Icahn School of Medicine at Mount Sinai, New York, NY , Clinical Research Training Program (CRTP) (2016)

LINKS

- Pharmacogenomics, 2nd Edition: Challenges and Opportunities in Therapeutic Implementation. Elsevier, 2018.: https://www.sciencedirect.com/book/9780128126264/pharmacogenomics

Publications

PUBLICATIONS

- **Pharmacogenomic polygenic risk score for clopidogrel responsiveness among Caribbean Hispanics: A candidate gene approach.** *Clinical and translational science*

  Duconge, J., Santiago, E., Hernandez-Suarez, D. F., Monero, M., Lopez-Reyes, A., Rosario, M., Renta, J. Y., Gonzalez, P., Ileana Fernandez-Morales, L., Antonio Velez-Figueroa, L., Arce, O., Marin-Maldonado, F., Nunez, et al

  2021

- **PharmVar GeneFocus: CYP2C9.** *Clinical pharmacology and therapeutics*

  Sangkuhl, K., Claudio-Campos, K., Cavallari, L. H., Agundez, J., Whirl-Carrillo, M., Duconge, J., Del Tredici, A. L., Wadelius, M., Botton, M. R., Woodahl, E. L., Scott, S. A., Klein, T. E., Pratt, et al

  2021

- **Deletion of ERF and CIC causes abnormal skull morphology and global developmental delay.** *Cold Spring Harbor molecular case studies*

  Singh, R., Cohen, A. S., Poulton, C., Hjortshoj, T. D., Akahira-Azuma, M., Mendiratta, G., Khan, W. A., Azmanov, D. N., Woodward, K. J., Kirchhoff, M., Shi, L., Edelmann, L., Baynam, et al

  2021; 7 (3)

- **Multi-Institutional Implementation of Clinical Decision Support for APOL1, NAT2, and YEATS4 Genotyping in Antihypertensive Management.** *Journal of personalized medicine*

  Schneider, T. M., Eadon, M. T., Cooper-DeHoff, R. M., Cavanaugh, K. L., Nguyen, K. A., Arwood, M. J., Tillman, E. M., Pratt, V. M., Dexter, P. R., McCoy, A. B., Orlando, L. A., Scott, S. A., Nadkarni, et al

  2021; 11 (6)

- **Pharmacogenomic education among genetic counseling training programs in North America.** *Journal of genetic counseling*

  Loudon, E., Scott, S. A., Rigobello, R., Scott, E. R., Zinberg, R., Naik, H.

  2021

- **Pharmacogenomic Determinants of Interindividual Drug Response Variability: From Discovery to Implementation.** *Genes*

  Scott, S. A., Swen, J. J.

  2021; 12 (3)

- **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 : JMD*

  Pratt, V. M., Cavallari, L. H., Del Tredici, A. L., Gaedigk, A., Hachad, H., Ji, Y., Kalman, L. V., Ly, R. C., Moyer, A. M., Scott, S. A., van Schaik, R. H., Whirl-Carrillo, M., Weck, et al

  2021

- **Novel Pharmacogenomic Locus Implicated in Angiotensin-Converting Enzyme Inhibitor-Induced Angioedema.** *Journal of the American College of Cardiology*

  Scott, S. A., Nicoletti, P.

  2021; 78 (7): 710-712

- **Phased Haplotype Resolution of the SLC6A4 Promoter Using Long-Read Single Molecule Real-Time (SMRT) Sequencing.** *Genes*

  Botton, M. R., Yang, Y., Scott, E. R., Desnick, R. J., Scott, S. A.

  2020; 11 (11)

- **Digital Health Applications for Pharmacogenetic Clinical Trials.** *Genes*

  Naik, H., Palaniappan, L., Ashley, E. A., Scott, S. A.

  2020; 11 (11)

- **Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing.** *Clinical pharmacology and therapeutics*

  Lima, J. J., Thomas, C. D., Barbarino, J., Desta, Z., Van Driest, S. L., El Rouby, N., Johnson, J. A., Cavallari, L. H., Shakhnovich, V., Thacker, D. L., Scott, S. A., Schwab, M., Uppugunduri, et al

  2020

- **Recommendations for Clinical Warfarin Genotyping Allele Selection A Report of the Association for Molecular Pathology and the College of American Pathologists** *JOURNAL OF MOLECULAR DIAGNOSTICS*

  Pratt, V. M., Cavallari, L. H., Del Tredici, A. L., Hachad, H., Ji, Y., Kalman, L. V., Ly, R. C., Moyer, A. M., Scott, S. A., Whirl-Carrillo, M., Weck, K. E.

  2020; 22 (7): 847–59

- **PharmVar GeneFocus: CYP2C19.** *Clinical pharmacology and therapeutics*

  Botton, M. R., Whirl-Carrillo, M., Del Tredici, A. L., Sangkuhl, K., Cavallari, L. H., Agundez, J. A., Duconge, J., Lee, M. T., Woodahl, E. L., Claudio-Campos, K., Daly, A. K., Klein, T. E., Pratt, et al

  2020

- **Development and Analytical Validation of a 29 Gene Clinical Pharmacogenetic Genotyping Panel: Multi-Ethnic Allele and Copy Number Variant Detection.** *Clinical and translational science*

  Scott, S. A., Scott, E. R., Seki, Y. n., Chen, A. J., Wallsten, R. n., Owusu Obeng, A. n., Botton, M. R., Cody, N. n., Shi, H. n., Zhao, G. n., Brake, P. n., Nicoletti, P. n., Yang, et al

  2020

- **VarCover: Allele Min-Set Cover Software.** *The Journal of molecular diagnostics : JMD*

  Scott, E. R., Bansal, V. n., Meacham, C. n., Scott, S. A.

  2020; 22 (2): 123–31

- **Genetic Factors Influencing Warfarin Dose in Black-African Patients: A Systematic Review and Meta-Analysis.** *Clinical pharmacology and therapeutics*

  Asiimwe, I. G., Zhang, E. J., Osanlou, R. n., Krause, A. n., Dillon, C. n., Suarez-Kurtz, G. n., Zhang, H. n., Perini, J. A., Renta, J. Y., Duconge, J. n., Cavallari, L. H., Marcatto, L. R., Beasly, et al

  2020; 107 (6): 1420–33

- **Knowledge and attitudes on pharmacogenetics among pediatricians.** *Journal of human genetics*

  Rahawi, S. n., Naik, H. n., Blake, K. V., Owusu Obeng, A. n., Wasserman, R. M., Seki, Y. n., Funanage, V. L., Oishi, K. n., Scott, S. A.

  2020; 65 (5): 437–44

- **Haploinsufficiency of the basic helix-loop-helix transcription factor HAND2 causes congenital heart defects.** *American journal of medical genetics. Part A*

  Cohen, A. S., Simotas, C. n., Webb, B. D., Shi, H. n., Khan, W. A., Edelmann, L. n., Scott, S. A., Singh, R. n.

  2020; 182 (5): 1263–67

- **Multi-site Investigation of Genetic Determinants of Warfarin Dose Variability in Latinos.** *Clinical and translational science*

  El Rouby, N. n., Rodrigues Marcatto, L. n., Claudio, K. n., Camargo Tavares, L. n., Steiner, H. n., Botton, M. R., Lubitz, S. A., Fallon, E. N., Yee, K. n., Kaye, J. n., Scott, S. A., Karnes, J. n., Caleb Junior de Lima Santos, et al

  2020

- **Implementing a pharmacogenetic-driven algorithm to guide dual antiplatelet therapy (DAPT) in Caribbean Hispanics: protocol for a non-randomised clinical trial.** *BMJ open*

  Hernandez-Suarez, D. F., Melin, K. n., Marin-Maldonado, F. n., Nunez, H. J., Gonzalez, A. F., Gonzalez-Sepulveda, L. n., Rivas-Tumanyan, S. n., Naik, H. n., Ruaño, G. n., Scott, S. A., Duconge, J. n.

  2020; 10 (8): e038936

- **Clinical Pharmacogenetic Testing and the Posttest Counseling Conundrum.** *Clinical pharmacology and therapeutics*

  Rigobello, R. n., Rahawi, S. n., Wallsten, R. n., Cody, N. n., Nicoletti, P. n., Owusu Obeng, A. n., Naik, H. n., Dillon, M. W., Scott, S. A.

  2020; 108 (5): 924–28

- **A Call for Clear and Consistent Communications Regarding the Role of Pharmacogenetics in Antidepressant Pharmacotherapy.** *Clinical pharmacology and therapeutics*

  Hicks, J. K., Bishop, J. R., Gammal, R. S., Sangkuhl, K., Bousman, C. A., Leeder, J. S., Llerena, A., Mueller, D. J., Ramsey, L. B., Scott, S. A., Skaar, T. C., Caudle, K. E., Klein, et al

  2019

- **Effect of CYP4F2, VKORC1, and CYP2C9 in Influencing Coumarin Dose: A Single-Patient Data Meta-Analysis in More Than 15,000 Individuals** *CLINICAL PHARMACOLOGY & THERAPEUTICS*

  Danese, E., Raimondi, S., Montagnana, M., Tagetti, A., Langaee, T., Borgiani, P., Ciccacci, C., Carcas, A. J., Borobia, A. M., Tong, H. Y., Davila-Fajardo, C., Botton, M., Bourgeois, et al

  2019; 105 (6): 1477–91

- **Pharmacogene Variation Consortium Gene Introduction: NUDT15** *CLINICAL PHARMACOLOGY & THERAPEUTICS*

Yang, J. J., Whirl-Carrillo, M., Scott, S. A., Turner, A. J., Schwab, M., Tanaka, Y., Suarez-Kurtz, G., Schaeffeler, E., Klein, T. E., Miller, N. A., Gaedigki, A.
2019; 105 (5): 1091–94

- **Development of a Genomic Data Flow Framework: Results of a Survey Administered to NIH-NHGRI IGNITE and eMERGE Consortia Participants.** *AMIA ... Annual Symposium proceedings. AMIA Symposium*
  Dexter, P. n., Ong, H. n., Elsey, A. n., Bell, G. n., Walton, N. n., Chung, W. n., Rasmussen, L. n., Hicks, K. n., Owusu-Obeng, A. n., Scott, S. n., Ellis, S. n., Peterson, J. n.
  2019; 2019: 363–70

- **Standardizing CYP2D6 Genotype to Phenotype Translation: Consensus Recommendations from the Clinical Pharmacogenetics Implementation Consortium and Dutch Pharmacogenetics Working Group.** *Clinical and translational science*
  Caudle, K. E., Sangkuhl, K. n., Whirl-Carrillo, M. n., Swen, J. J., Haidar, C. E., Klein, T. E., Gammal, R. S., Relling, M. V., Scott, S. A., Hertz, D. L., Guchelaar, H. J., Gaedigk, A. n.
  2019

- **Essential Characteristics of Pharmacogenomics Study Publications** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Thorn, C. F., Whirl-Carrillo, M., Hachad, H., Johnson, J. A., McDonagh, E. M., Ratain, M. J., Relling, M. V., Scott, S. A., Altman, R. B., Klein, T. E.
  2019; 105 (1): 86–91

- **Recommendations for Clinical CYP2C9 Genotyping Allele Selection: A Joint Recommendation of the Association for Molecular Pathology and College of American Pathologists.** *The Journal of molecular diagnostics : JMD*
  Pratt, V. M., Cavallari, L. H., Del Tredici, A. L., Hachad, H. n., Ji, Y. n., Moyer, A. M., Scott, S. A., Whirl-Carrillo, M. n., Weck, K. E.
  2019

- **Characterization of Reference Materials for Genetic Testing of CYP2D6 Alleles: A GeT-RM Collaborative Project.** *The Journal of molecular diagnostics : JMD*
  Gaedigk, A. n., Turner, A. n., Everts, R. E., Scott, S. A., Aggarwal, P. n., Broeckel, U. n., McMillin, G. A., Melis, R. n., Boone, E. C., Pratt, V. M., Kalman, L. V.
  2019; 21 (6): 1034–52

- **Structural variation at the CYP2C locus: Characterization of deletion and duplication alleles.** *Human mutation*
  Botton, M. R., Lu, X. n., Zhao, G. n., Repnikova, E. n., Seki, Y. n., Gaedigk, A. n., Schadt, E. E., Edelmann, L. n., Scott, S. A.
  2019; 40 (11): e37–e51

- **Prenatal cytogenomic identification and molecular refinement of compound heterozygous STRC deletion breakpoints.** *Molecular genetics & genomic medicine*
  Shi, L. n., Bai, Y. n., Kharbutli, Y. n., Oza, A. M., Amr, S. S., Edelmann, L. n., Mehta, L. n., Scott, S. A.
  2019; 7 (8): e806

- **Interpreting and Implementing Clinical Pharmacogenetic Tests: Perspectives From Service Providers.** *Clinical pharmacology and therapeutics*
  Hachad, H. n., Ramsey, L. B., Scott, S. A.
  2019; 106 (2): 298–301

- **Multi-site investigation of strategies for the clinical implementation of CYP2D6 genotyping to guide drug prescribing.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Cavallari, L. H., Van Driest, S. L., Prows, C. A., Bishop, J. R., Limdi, N. A., Pratt, V. M., Ramsey, L. B., Smith, D. M., Tuteja, S. n., Duong, B. Q., Hicks, J. K., Lee, J. C., Obeng, et al
  2019; 21 (10): 2255–63

- **Familial inheritance of the 3q29 microdeletion syndrome: case report and review.** *BMC medical genomics*
  Khan, W. A., Cohen, N. n., Scott, S. A., Pereira, E. M.
  2019; 12 (1): 51

- **Integrated CYP2D6 interrogation for multiethnic copy number and tandem allele detection.** *Pharmacogenomics*
  Qiao, W. n., Martis, S. n., Mendiratta, G. n., Shi, L. n., Botton, M. R., Yang, Y. n., Gaedigk, A. n., Vijzelaar, R. n., Edelmann, L. n., Kornreich, R. n., Desnick, R. J., Scott, S. A.
  2019; 20 (1): 9–20

- **Pharmacogene Variation Consortium Gene Introduction: NUDT15.** *Clinical pharmacology and therapeutics*
  Yang, J. J., Whirl-Carrillo, M., Scott, S. A., Turner, A. J., Schwab, M., Tanaka, Y., Suarez-Kurtz, G., Schaeffeler, E., Klein, T. E., Miller, N. A., Gaedigk, A.
  2018

- **The effect of CYP4F2, VKORC1 and CYP2C9 in influencing coumarin dose. A single patient data meta-analysis in more than 15,000 individuals.** *Clinical pharmacology and therapeutics*
  Danese, E., Raimondi, S., Montagnana, M., Tagetti, A., Langaee, T., Borgiani, P., Ciccacci, C., Carcas, A. J., Borobia, A. M., Tong, H. Y., Davila-Fajardo, C., Botton, M. R., Bourgeois, et al
  2018

- **Essential characteristics of pharmacogenomics study publications.** *Clinical pharmacology and therapeutics*
  Thorn, C. F., Whirl-Carrillo, M., Hachad, H., Johnson, J. A., McDonagh, E. M., Ratain, M. J., Relling, M. V., Scott, S. A., Altman, R. B., Klein, T. E.
  2018

- **Multi-ethnic genome-wide association study for atrial fibrillation** *NATURE GENETICS*
  Roselli, C., Chaffin, M. D., Weng, L., Aeschbacher, S., Ahlberg, G., Albert, C. M., Almgren, P., Alonso, A., Anderson, C. D., Aragam, K. G., Arking, D. E., Barnard, J., Bartz, et al
  2018; 50 (9): 1225-+

- **Pharmacogenetic association study on clopidogrel response in Puerto Rican Hispanics with cardiovascular disease: a novel characterization of a Caribbean population.** *Pharmacogenomics and personalized medicine*
  Hernandez-Suarez, D. F., Botton, M. R., Scott, S. A., Tomey, M. I., Garcia, M. J., Wiley, J. n., Villablanca, P. A., Melin, K. n., Lopez-Candales, A. n., Renta, J. Y., Duconge, J. n.
  2018; 11: 95–106

- **Multi-ethnic SULT1A1 copy number profiling with multiplex ligation-dependent probe amplification.** *Pharmacogenomics*
  Vijzelaar, R. n., Botton, M. R., Stolk, L. n., Martis, S. n., Desnick, R. J., Scott, S. A.
  2018; 19 (9): 761–70

- **Recommendations for Clinical CYP2C19 Genotyping Allele Selection: A Report of the Association for Molecular Pathology.** *The Journal of molecular diagnostics : JMD*
  Pratt, V. M., Del Tredici, A. L., Hachad, H. n., Ji, Y. n., Kalman, L. V., Scott, S. A., Weck, K. E.
  2018; 20 (3): 269–76

- **Effect of cilostazol on platelet reactivity among patients with peripheral artery disease on clopidogrel therapy.** *Drug metabolism and personalized therapy*
  Hernandez-Suarez, D. F., Núñez-Medina, H. n., Scott, S. A., Lopez-Candales, A. n., Wiley, J. M., Garcia, M. J., Melin, K. n., Nieves-Borrero, K. n., Rodriguez-Ruiz, C. n., Marshall, L. n., Duconge, J. n.
  2018; 33 (1): 49–55

- **Cytogenomic identification and long-read single molecule real-time (SMRT) sequencing of a Bardet-Biedl Syndrome 9 (BBS9) deletion.** *NPJ genomic medicine*
  Reiner, J. n., Pisani, L. n., Qiao, W. n., Singh, R. n., Yang, Y. n., Shi, L. n., Khan, W. A., Sebra, R. n., Cohen, N. n., Babu, A. n., Edelmann, L. n., Jabs, E. W., Scott, et al
  2018; 3: 3

- **Multisite Investigation of Strategies for the Implementation of CYP2C19 Genotype-Guided Antiplatelet Therapy.** *Clinical pharmacology and therapeutics*
  Empey, P. E., Stevenson, J. M., Tuteja, S. n., Weitzel, K. W., Angiolillo, D. J., Beitelshees, A. L., Coons, J. C., Duarte, J. D., Franchi, F. n., Jeng, L. J., Johnson, J. A., Kreutz, R. P., Limdi, et al
  2018; 104 (4): 664–74

- **Clinical Pharmacogenetics Implementation Consortium (CPIC) Guidelines for CYP2C19 and Voriconazole Therapy** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Moriyama, B., Obeng, A., Barbarino, J., Penzak, S. R., Henning, S. A., Scott, S. A., Agundez, J. G., Wingard, J. R., McLeod, H. L., Klein, T. E., Cross, S. J., Caudle, K. E., Walsh, et al
  2017; 102 (1): 45–51

- **The CYP2D6 VCF Translator** *PHARMACOGENOMICS JOURNAL*
  Qiao, W., Wang, J., Pullman, B. S., Chen, R., Yang, Y., Scott, S. A.
  2017; 17 (4): 301–3

- **Large-scale analyses of common and rare variants identify 12 new loci associated with atrial fibrillation.** *Nature genetics*
  Christophersen, I. E., Rienstra, M., Roselli, C., Yin, X., Geelhoed, B., Barnard, J., Lin, H., Arking, D. E., Smith, A. V., Albert, C. M., Chaffin, M., Tucker, N. R., Li, et al
  2017; 49 (6): 946-952

- **Clinical pharmacogenetics implementation consortium (cpic) guideline for pharmacogenetics-guided warfarin dosing: 2017 update.** *Clinical pharmacology & therapeutics*

  Johnson, J. A., Caudle, K. E., Gong, L., Whirl-Carrillo, M., Stein, C. M., Scott, S. A., Lee, M. T., Gage, B. F., Kimmel, S. E., Perera, M. A., Anderson, J. L., Pirmohamed, M., Klein, et al

  2017

- **Standardizing terms for clinical pharmacogenetic test results: consensus terms from the Clinical Pharmacogenetics Implementation Consortium (CPIC)** *GENETICS IN MEDICINE*

  Caudle, K. E., Dunnenberger, H. M., Freimuth, R. R., Peterson, J. F., Burlison, J. D., Whirl-Carrillo, M., Scott, S. A., Rehm, H. L., Williams, M. S., Klein, T. E., Relling, M. V., Hoffman, J. M.

  2017; 19 (2): 215-223

- **Modeling susceptibility to drug-induced long QT with a panel of subject-specific induced pluripotent stem cells** *ELIFE*

  Stillitano, F., Hansen, J., Kong, C., Karakikes, I., Faunck-Brentano, C., Geng, L., Scott, S., Reynier, S., Wu, M., Valogne, Y., Desseaux, C., Salem, J., Jeziorowska, et al

  2017; 6

- **DNA Methylation Profiling Using Long-Read Single Molecule Real-Time Bisulfite Sequencing (SMRT-BS).** *Methods in molecular biology (Clifton, N.J.)*

  Yang, Y. n., Scott, S. A.

  2017; 1654: 125–34

- **Institutional profile: translational pharmacogenomics at the Icahn School of Medicine at Mount Sinai.** *Pharmacogenomics*

  Scott, S. A., Owusu Obeng, A. n., Botton, M. R., Yang, Y. n., Scott, E. R., Ellis, S. B., Wallsten, R. n., Kaszemacher, T. n., Zhou, X. n., Chen, R. n., Nicoletti, P. n., Naik, H. n., Kenny, et al

  2017; 18 (15): 1381–86

- **Clinical determinants of clopidogrel responsiveness in a heterogeneous cohort of Puerto Rican Hispanics.** *Therapeutic advances in cardiovascular disease*

  Hernandez-Suarez, D. F., Scott, S. A., Tomey, M. I., Melin, K. n., Lopez-Candales, A. n., Buckley, C. E., Duconge, J. n.

  2017; 11 (9): 235–41

- **Warfarin Anticoagulation Therapy in Caribbean Hispanics of Puerto Rico: A Candidate Gene Association Study.** *Frontiers in pharmacology*

  Claudio-Campos, K. n., Labastida, A. n., Ramos, A. n., Gaedigk, A. n., Renta-Torres, J. n., Padilla, D. n., Rivera-Miranda, G. n., Scott, S. A., Ruaño, G. n., Cadilla, C. L., Duconge-Soler, J. n.

  2017; 8: 347

- **Concordance between Research Sequencing and Clinical Pharmacogenetic Genotyping in the eMERGE-PGx Study.** *The Journal of molecular diagnostics : JMD*

  Rasmussen-Torvik, L. J., Almoguera, B. n., Doheny, K. F., Freimuth, R. R., Gordon, A. S., Hakonarson, H. n., Hawkins, J. B., Husami, A. n., Ivacic, L. C., Kullo, I. J., Linderman, M. D., Manolio, T. A., Obeng, et al

  2017; 19 (4): 561–66

- **Sequencing the CYP2D6 gene: from variant allele discovery to clinical pharmacogenetic testing.** *Pharmacogenomics*

  Yang, Y. n., Botton, M. R., Scott, E. R., Scott, S. A.

  2017; 18 (7): 673–85

- **Apolipoprotein L1 Variants and Blood Pressure Traits in African Americans.** *Journal of the American College of Cardiology*

  Nadkarni, G. N., Galarneau, G. n., Ellis, S. B., Nadukuru, R. n., Zhang, J. n., Scott, S. A., Schurmann, C. n., Li, R. n., Rasmussen-Torvik, L. J., Kho, A. N., Hayes, M. G., Pacheco, J. A., Manolio, et al

  2017; 69 (12): 1564–74

- **Maternal uniparental disomy of chromosome 15 and concomitant STRC and CATSPER2 deletion-mediated deafness-infertility syndrome.** *American journal of medical genetics. Part A*

  Karger, L. n., Khan, W. A., Calabio, R. n., Singh, R. n., Xiang, B. n., Babu, A. n., Cohen, N. n., Yang, A. C., Scott, S. A.

  2017; 173 (5): 1436–39

- **Chromosomal Microarray Detection of Constitutional Copy Number Variation Using Saliva DNA.** *The Journal of molecular diagnostics : JMD*

  Reiner, J. n., Karger, L. n., Cohen, N. n., Mehta, L. n., Edelmann, L. n., Scott, S. A.

  2017; 19 (3): 397–403

- **Implementing Algorithm-Guided Warfarin Dosing in an Ethnically Diverse Patient Population Using Electronic Health Records and Preemptive CYP2C9 and VKORC1 Genetic Testing** *CLINICAL PHARMACOLOGY & THERAPEUTICS*

Obeng, A., Kaszemacher, T., Abul-Husn, N. S., Gottesman, O., Vega, A., Waite, E., Myers, K., Cho, J., Bottinger, E. P., Ellis, S. B., Scott, S. A.
2016; 100 (5): 427–30

- **Exome Sequencing of Extreme Clopidogrel Response Phenotypes Identifies B4GALT2 as a Determinant of On-Treatment Platelet Reactivity** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Scott, S. A., Collet, J., Baber, U., Yang, Y., Peter, I., Linderman, M., Sload, J., Qiao, W., Kini, A. S., Sharma, S. K., Desnick, R. J., Fuster, V., Hajjar, et al
  2016; 100 (3): 287–94

- **Genetic Variation Among 82 Pharmacogenes: The PGRNseq Data From the eMERGE Network** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Bush, W. S., Crosslin, D. R., Owusu-Obeng, A., Wallace, J., Almoguera, B., Basford, M. A., Bielinski, S. J., Carrell, D. S., Connolly, J. J., Crawford, D., Doheny, K. F., Gallego, C. J., Gordon, et al
  2016; 100 (2): 160–69

- **Standardizing terms for clinical pharmacogenetic test results: consensus terms from the Clinical Pharmacogenetics Implementation Consortium (CPIC).** *Genetics in medicine*
  Caudle, K. E., Dunnenberger, H. M., Freimuth, R. R., Peterson, J. F., Burlison, J. D., Whirl-Carrillo, M., Scott, S. A., Rehm, H. L., Williams, M. S., Klein, T. E., Relling, M. V., Hoffman, J. M.
  2016

- **Pharmacogenetic allele nomenclature: International workgroup recommendations for test result reporting.** *Clinical pharmacology & therapeutics*
  Kalman, L. V., Agúndez, J., Appell, M. L., Black, J. L., Bell, G. C., Boukouvala, S., Bruckner, C., Bruford, E., Caudle, K., Coulthard, S. A., Daly, A. K., Tredici, A. D., den Dunnen, et al
  2016; 99 (2): 172-185

- **A de novo 2.78-Mb duplication on chromosome 21q22.11 implicates candidate genes in the partial trisomy 21 phenotype.** *NPJ genomic medicine*
  Weisfeld-Adams, J. D., Tkachuk, A. K., Maclean, K. N., Meeks, N. L., Scott, S. A.
  2016; 1

- **Polygenic overlap between schizophrenia risk and antipsychotic response: a genomic medicine approach.** *The lancet. Psychiatry*
  Ruderfer, D. M., Charney, A. W., Readhead, B. n., Kidd, B. A., Kähler, A. K., Kenny, P. J., Keiser, M. J., Moran, J. L., Hultman, C. M., Scott, S. A., Sullivan, P. F., Purcell, S. M., Dudley, et al
  2016; 3 (4): 350–57

- **Analytical Validation of a Personalized Medicine APOL1 Genotyping Assay for Nondiabetic Chronic Kidney Disease Risk Assessment.** *The Journal of molecular diagnostics : JMD*
  Zhang, J. n., Fedick, A. n., Wasserman, S. n., Zhao, G. n., Edelmann, L. n., Bottinger, E. P., Kornreich, R. n., Scott, S. A.
  2016; 18 (2): 260–66

- **Association of Arrhythmia-Related Genetic Variants With Phenotypes Documented in Electronic Medical Records.** *JAMA*
  Van Driest, S. L., Wells, Q. S., Stallings, S. n., Bush, W. S., Gordon, A. n., Nickerson, D. A., Kim, J. H., Crosslin, D. R., Jarvik, G. P., Carrell, D. S., Ralston, J. D., Larson, E. B., Bielinski, et al
  2016; 315 (1): 47–57

- **Characterization of 137 Genomic DNA Reference Materials for 28 Pharmacogenetic Genes: A GeT-RM Collaborative Project.** *The Journal of molecular diagnostics : JMD*
  Pratt, V. M., Everts, R. E., Aggarwal, P. n., Beyer, B. N., Broeckel, U. n., Epstein-Baak, R. n., Hujsak, P. n., Kornreich, R. n., Liao, J. n., Lorier, R. n., Scott, S. A., Smith, C. H., Toji, et al
  2016; 18 (1): 109–23

- **Long-Read Single Molecule Real-Time Full Gene Sequencing of Cytochrome P450-2D6.** *Human mutation*
  Qiao, W. n., Yang, Y. n., Sebra, R. n., Mendiratta, G. n., Gaedigk, A. n., Desnick, R. J., Scott, S. A.
  2016; 37 (3): 315–23

- **Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Selective Serotonin Reuptake Inhibitors** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Hicks, J. K., Bishop, J. R., Sangkuhl, K., Mueller, D. J., Ji, Y., Leckband, S. G., Leeder, J. S., GRAHAM, R. L., Chiulli, D. L., Llerena, A., Skaar, T. C., Scott, S. A., Stingl, et al
  2015; 98 (1): 127-134

- **ClinGen - The Clinical Genome Resource** *NEW ENGLAND JOURNAL OF MEDICINE*
  Rehm, H. L., Berg, J. S., Brooks, L. D., Bustamante, C. D., Evans, J. P., Landrum, M. J., Ledbetter, D. H., Maglott, D. R., Martin, C. L., Nussbaum, R. L., Plon, S. E., Ramos, E. M., Sherry, et al

2015; 372 (23): 2235-2242

- **The pharmacogenetic control of antiplatelet response: candidate genes and CYP2C19.** *Expert opinion on drug metabolism & toxicology*
  Yang, Y. n., Lewis, J. P., Hulot, J. S., Scott, S. A.
  2015; 11 (10): 1599–1617
- **Quantitative and multiplexed DNA methylation analysis using long-read single-molecule real-time bisulfite sequencing (SMRT-BS).** *BMC genomics*
  Yang, Y. n., Sebra, R. n., Pullman, B. S., Qiao, W. n., Peter, I. n., Desnick, R. J., Geyer, C. R., DeCoteau, J. F., Scott, S. A.
  2015; 16: 350
- **Design and Anticipated Outcomes of the eMERGE-PGx Project: A Multicenter Pilot for Preemptive Pharmacogenomics in Electronic Health Record Systems** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Rasmussen-Torvik, L. J., Stallings, S. C., Gordon, A. S., Almoguera, B., Basford, M. A., Bielinski, S. J., Brautbar, A., Brilliant, M. H., Carrell, D. S., Connolly, J. J., Crosslin, D. R., Doheny, K. F., Gallego, et al
  2014; 96 (4): 482–89
- **An Ashkenazi Jewish SMN1 haplotype specific to duplication alleles improves pan-ethnic carrier screening for spinal muscular atrophy** *GENETICS IN MEDICINE*
  Luo, M., Liu, L., Peter, I., Zhu, J., Scott, S. A., Zhao, G., Eversley, C., Kornreich, R., Desnick, R. J., Edelmann, L.
  2014; 16 (2): 149–56
- **Incorporation of Pharmacogenomics into Routine Clinical Practice: the Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline Development Process.** *Current drug metabolism*
  Caudle, K. E., Klein, T. E., Hoffman, J. M., Muller, D. J., Whirl-Carrillo, M., Gong, L., McDonagh, E. M., Sangkuhl, K., Thorn, C. F., Schwab, M., Agundez, J. A., Freimuth, R. R., Huser, et al
  2014; 15 (2): 209-217
- **Paroxysmal kinesigenic dyskinesia caused by 16p11.2 microdeletion.** *Tremor and other hyperkinetic movements (New York, N.Y.)*
  Termsarasab, P. n., Yang, A. C., Reiner, J. n., Mei, H. n., Scott, S. A., Frucht, S. J.
  2014; 4: 274
- **Physician Attitudes toward Adopting Genome-Guided Prescribing through Clinical Decision Support.** *Journal of personalized medicine*
  Overby, C. L., Erwin, A. L., Abul-Husn, N. S., Ellis, S. B., Scott, S. A., Obeng, A. O., Kannry, J. L., Hripcsak, G. n., Bottinger, E. P., Gottesman, O. n.
  2014; 4 (1): 35–49
- **Implementation and utilization of genetic testing in personalized medicine.** *Pharmacogenomics and personalized medicine*
  Abul-Husn, N. S., Owusu Obeng, A. n., Sanderson, S. C., Gottesman, O. n., Scott, S. A.
  2014; 7: 227–40
- **Warfarin pharmacogenetic trials: is there a future for pharmacogenetic-guided dosing?** *Pharmacogenomics*
  Scott, S. A., Lubitz, S. A.
  2014; 15 (6): 719–22
- **Antiplatelet drug interactions with proton pump inhibitors.** *Expert opinion on drug metabolism & toxicology*
  Scott, S. A., Owusu Obeng, A. n., Hulot, J. S.
  2014; 10 (2): 175–89
- **Multi-ethnic cytochrome-P450 copy number profiling: novel pharmacogenetic alleles and mechanism of copy number variation formation** *PHARMACOGENOMICS JOURNAL*
  Martis, S., Mei, H., Vijzelaar, R., Edelmann, L., Desnick, R. J., Scott, S. A.
  2013; 13 (6): 558–66
- **The Electronic Medical Records and Genomics (eMERGE) Network: past, present, and future** *GENETICS IN MEDICINE*
  Gottesman, O., Kuivaniemi, H., Tromp, G., Faucett, W., Li, R., Manolio, T. A., Sanderson, S. C., Kannry, J., Zinberg, R., Basford, M. A., Brilliant, M., Carey, D. J., Chisholm, et al
  2013; 15 (10): 761–71
- **Clinical Pharmacogenetics Implementation Consortium Guidelines for CYP2C19 Genotype and Clopidogrel Therapy: 2013 Update** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Scott, S. A., Sangkuhl, K., Stein, C. M., Hulot, J., Mega, J. L., Roden, D. M., Klein, T. E., Sabatine, M. S., Johnson, J. A., Shuldiner, A. R.
  2013; 94 (3): 317-323

- **Genetic variants associated with warfarin dose in African-American individuals: a genome-wide association study.** *Lancet*
  Perera, M. A., Cavallari, L. H., Limdi, N. A., Gamazon, E. R., Konkashbaev, A., Daneshjou, R., Pluzhnikov, A., Crawford, D. C., Wang, J., Liu, N., Tatonetti, N., Bourgeois, S., Takahashi, et al
  2013; 382 (9894): 790-796

- **Multi-ethnic distribution of clinically relevant CYP2C genotypes and haplotypes** *PHARMACOGENOMICS JOURNAL*
  Martis, S., Peter, I., Hulot, J., Kornreich, R., Desnick, R. J., Scott, S. A.
  2013; 13 (4): 369–77

- **The CLIPMERGE PGx Program: Clinical Implementation of Personalized Medicine Through Electronic Health Records and Genomics-Pharmacogenomics** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Gottesman, O., Scott, S. A., Ellis, S. B., Overby, C. L., Ludtke, A., Hulot, J., Hall, J., Chatani, K., Myers, K., Kannry, J. L., Bottinger, E. P.
  2013; 94 (2): 214–17

- **Combined and independent impact of diabetes mellitus and chronic kidney disease on residual platelet reactivity** *THROMBOSIS AND HAEMOSTASIS*
  Baber, U., Bander, J., Karajgikar, R., Yadav, K., Hadi, A., Theodoropolous, K., Gukathasan, N., Roy, S., Sayeneni, S., Scott, S. A., Kovacic, J. C., Yu, J., Sartori, et al
  2013; 110 (1): 118–23

- **Warfarin pharmacogenetics: a controlled dose-response study in healthy subjects.** *Vascular medicine (London, England)*
  Kadian-Dodov, D. L., van der Zee, S. A., Scott, S. A., Peter, I. n., Martis, S. n., Doheny, D. O., Rothlauf, E. B., Lubitz, S. A., Desnick, R. J., Halperin, J. L.
  2013; 18 (5): 290–97

- **Clinical Pharmacogenomics: Opportunities and Challenges at Point of Care** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Scott, S. A.
  2013; 93 (1): 33–35

- **An allele-specific PCR system for rapid detection and discrimination of the CYP2C19#4A, #4B, and #17 alleles: implications for clopidogrel response testing.** *The Journal of molecular diagnostics : JMD*
  Scott, S. A., Tan, Q. n., Baber, U. n., Yang, Y. n., Martis, S. n., Bander, J. n., Kornreich, R. n., Hulot, J. S., Desnick, R. J.
  2013; 15 (6): 783–89

- **Frequency of the cholesteryl ester storage disease common LIPA E8SJM mutation (c.894G>A) in various racial and ethnic groups.** *Hepatology (Baltimore, Md.)*
  Scott, S. A., Liu, B. n., Nazarenko, I. n., Martis, S. n., Kozlitina, J. n., Yang, Y. n., Ramirez, C. n., Kasai, Y. n., Hyatt, T. n., Peter, I. n., Desnick, R. J.
  2013; 58 (3): 958–65

- **Impact of the CYP4F2 p.V433M Polymorphism on Coumarin Dose Requirement: Systematic Review and Meta-Analysis** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Danese, E., Montagnana, M., Johnson, J. A., Rettie, A. E., Zambon, C. F., Lubitz, S. A., Suarez-Kurtz, G., Cavallari, L. H., Zhao, L., Huang, M., Nakamura, Y., Mushiroda, T., Kringen, et al
  2012; 92 (6): 746-756

- **Identification of CYP2C19*4B: pharmacogenetic implications for drug metabolism including clopidogrel responsiveness** *PHARMACOGENOMICS JOURNAL*
  Scott, S. A., Martis, S., Peter, I., Kasai, Y., Kornreich, R., Desnick, R. J.
  2012; 12 (4): 297–305

- **Copy number variation and warfarin dosing: evaluation of CYP2C9, VKORC1, CYP4F2, GGCX and CALU** *PHARMACOGENOMICS*
  Scott, S. A., Patel, M., Martis, S., Lubitz, S. A., van der Zee, S., Yoo, C., Edelmann, L., Halperin, J. L., Desnick, R. J.
  2012; 13 (3): 297–307

- **PharmGKB summary: very important pharmacogene information for cytochrome P450, family 2, subfamily C, polypeptide 19** *PHARMACOGENETICS AND GENOMICS*
  Scott, S. A., Sangkuhl, K., Shuldiner, A. R., Hulot, J., Thorn, C. F., Altman, R. B., Klein, T. E.
  2012; 22 (2): 159-165

- **Personalizing medicine with clinical pharmacogenetics** *GENETICS IN MEDICINE*
  Scott, S. A.
  2011; 13 (12): 987–95

- **Clinical Pharmacogenetics Implementation Consortium Guidelines for CYP2C9 and VKORC1 Genotypes and Warfarin Dosing** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Johnson, J. A., Gong, L., Whirl-Carrillo, M., Gage, B. F., Scott, S. A., Stein, C. M., Anderson, J. L., Kimmel, S. E., Lee, M. T., Pirmohamed, M., Wadelius, M., Klein, T. E., Altman, et al
  2011; 90 (4): 625-629

- **Clinical Pharmacogenetics Implementation Consortium Guidelines for Cytochrome P450-2C19 (CYP2C19) Genotype and Clopidogrel Therapy** *CLINICAL PHARMACOLOGY & THERAPEUTICS*
  Scott, S. A., Sangkuhl, K., Gardner, E. E., Stein, C. M., Hulot, J., Johnson, J. A., Roden, D. M., Klein, T. E., Shuldiner, A. R.
  2011; 90 (2): 328-332

- **Experience with Carrier Screening and Prenatal Diagnosis for 16 Ashkenazi Jewish Genetic Diseases** *HUMAN MUTATION*
  Scott, S. A., Edelmann, L., Liu, L., Luo, M., Desnick, R. J., Kornreich, R.
  2010; 31 (11): 1240–50

- **Large inverted repeats within Xp11.2 are present at the breakpoints of isodicentric X chromosomes in Turner syndrome** *HUMAN MOLECULAR GENETICS*
  Scott, S. A., Cohen, N., Brandt, T., Warburton, P. E., Edelmann, L.
  2010; 19 (17): 3383–93

- **Combined CYP2C9, VKORC1 and CYP4F2 frequencies among racial and ethnic groups** *PHARMACOGENOMICS*
  Scott, S. A., Khasawneh, R., Peter, I., Kornreich, R., Desnick, R. J.
  2010; 11 (6): 781–91

- **Comparative performance of gene-based warfarin dosing algorithms in a multiethnic population** *JOURNAL OF THROMBOSIS AND HAEMOSTASIS*
  Lubitz, S. A., Scott, S. A., Rothlauf, E. B., Agarwal, A., Peter, I., Doheny, D., van der Zee, S., Jaremko, M., Yoo, C., Desnick, R. J., Halperin, J. L.
  2010; 8 (5): 1018–26

- **Detection of low-level mosaicism and placental mosaicism by oligonucleotide array comparative genomic hybridization** *GENETICS IN MEDICINE*
  Scott, S. A., Cohen, N., Brandt, T., Toruner, G., Desnick, R. J., Edelmann, L.
  2010; 12 (2): 85–92

- **Reexpression of epigenetically silenced AML tumor suppressor genes by SUV39H1 inhibition** *ONCOGENE*
  Lakshmikuttyamma, A., Scott, S. A., DeCoteau, J. F., Geyer, C. R.
  2010; 29 (4): 576–88

- **CYP2C9*8 is prevalent among African-Americans: implications for pharmacogenetic dosing** *PHARMACOGENOMICS*
  Scott, S. A., Jaremko, M., Lubitz, S. A., Kornreich, R., Halperin, J. L., Desnick, R. J.
  2009; 10 (8): 1243–55

- **Strategies to Re-Express Epigenetically-Silenced Tumor Suppressor Genes Converge on the Requirement for Inhibition of the Histone Methyltransferase SUV39H1**
  Lakshmikuttyamma, A., Scott, S., Sheridan, D. P., DeCoteau, J., Geyer, R.
  AMER SOC HEMATOLOGY.2008: 1152–53

- **Warfarin pharmacogenetics: CYP2C9 and VKORC1 genotypes predict different sensitivity and resistance frequencies in the Ashkenazi and Sephardi Jewish populations** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Scott, S. A., Edelmann, L., Kornreich, R., Desnick, R. J.
  2008; 82 (2): 495–500

- **Induction of ID1 expression and apoptosis by the histone deacetylase inhibitor (trichostatin A) in human acute myeloid leukaemic cells** *CELL PROLIFERATION*
  Yu, W., Scott, S. A., Dong, W.
  2008; 41 (1): 86–97

- **Decreased expression of the histone methyltransferase SUV39H1 in AML cells reactivates hypermethylated tumor suppressor p15INK4B in the absence of promoter demethylation**
  Scott, S., Geyer, C., DeCoteau, J. F.
  AMER SOC HEMATOLOGY.2007: 106B

- **CYP2C9, CYP2C19 and CYP2D6 allele frequencies in the Ashkenazi Jewish population** *PHARMACOGENOMICS*

Scott, S., Edelmann, L., Kornreich, R., Erazo, M., Desnick, R. J.
2007; 8 (7): 721–30

- **Zebularine inhibits human acute myeloid leukemia cell growth in vitro in association with p15INK4B demethylation and reexpression** *EXPERIMENTAL HEMATOLOGY*
  Scott, S. A., Lakshimikuttysamma, A., Sheridan, D. P., Sanche, S. E., Geyer, C., DeCoteau, J. F.
  2007; 35 (2): 263–73

- **5-Aza-2 '-deoxycytidine (decitabine) can relieve p21WAF1 repression in human acute myeloid leukemia by a mechanism involving release of histone deacetylase 1 (HDAC1) without requiring p21WAF1 promoter demethylation** *LEUKEMIA RESEARCH*
  Scott, S. A., Dong, W. F., Ichinohasama, R., Hirsch, C., Sheridan, D., Sanche, S. E., Geyer, C. R., DeCoteau, J. F.
  2006; 30 (1): 69–76

- **Methylation status of cyclin-dependent kinase inhibitor genes within the transforming growth factor beta pathway in human T-cell lymphoblastic lymphoma/leukemia** *LEUKEMIA RESEARCH*
  Scott, S. A., Kimura, T., Dong, W. F., Ichinohasama, R., Bergen, S., Kerviche, A., Sheridan, D., DeCoteau, J. F.
  2004; 28 (12): 1293–1301

- **Microsatellite mutations of transforming growth factor-beta receptor type II and caspase-5 occur in human precursor T-cell lymphoblastic lymphomas/ leukemias in vivo but are not associated with hMSH2 or hMLH1 promoter methylation** *LEUKEMIA RESEARCH*
  Scott, S., Kimura, T., Ichinohasama, R., Bergen, S., Magliocco, A., Reimer, C., Kerviche, A., Sheridan, D., DeCoteau, J. F.
  2003; 27 (1): 23–34

- **MSH2-deficient murine lymphomas harbor insertion/deletion mutations in the transforming growth factor beta receptor type 2 gene and display low not high frequency microsatellite instability** *BLOOD*
  Lowsky, R., Magliocco, A., Ichinohasama, R., Reitmair, A., Scott, S., Henry, M., Kadin, M. E., DeCoteau, J. F.
  2000; 95 (5): 1767–72