



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, Cardiovascular Institute
- 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 Lampion 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

- Innovations in Genomic Diagnostics: <https://onlinelibrary.wiley.com/toc/10981004/2022/43/11>
- Pharmacogenomic Determinants of Interindividual Drug Response Variability: From Discovery to Implementation: https://www.mdpi.com/journal/genes/special_issues/Pharmacogenomic_Determinants
- Pharmacogenomics: Challenges and Opportunities in Therapeutic Implementation: <https://www.sciencedirect.com/book/9780128126264/pharmacogenomics>
- Research Laboratory: <https://stuartscottlab.org/>

Publications

PUBLICATIONS

- **Knowledge and perspectives on pharmacogenomic-guided antidepressant treatment among psychiatrists and primary care practitioners.** *Pharmacogenetics and genomics*
Schmidt, K. T., Naik, H., Nguyen, T. P., Bishop, J. R., Bousman, C. A., Singh, A. B., Klein, T. E., Scott, S. A.
2026
- **Analytical validation of short-read genome sequencing for diagnostic panel and exome testing.** *The Journal of molecular diagnostics : JMD*
Yang, Y., Hammond, N. A., Tong, P. W., Ho, C., Watson, N., Liao, L., Tan, T., Chen, L., Dumas, K., Fisk, D., Grove, M. E., Hale, C., Ng, et al
2026
- **Delivering pharmacogenomic results via electronic health record patient portals: Impacts of health literacy and genetics knowledge.** *Patient education and counseling*
Kile, S. L., Owusu-Obeng, A., Scott, S. A., Naik, H.
2026; 149: 109654
- **De Novo Variants Associated With Autosomal Recessive Conditions: Case Series and Implications for Genetic Testing and Counseling.** *American journal of medical genetics. Part A*
Niehaus, A. D., Bonner, D. E., Carter, J., Avello, K., Jacob, N., Neu, M. B., Mendez, R., Qiao, W., Scott, S. A., Levy, R. J., Mattas, L., Schymick, J., Van Andel, et al
2026
- **Multiplexed antigen panel analysis identifies B cell phenotype and receptor genetic contributions to antibody breadth.** *Immunity*
Wirz, O. F., Kotagiri, P., Haraguchi, E., Röltgen, K., Hunter, M., Craig, E., Afaghani, J., Lee, J. Y., Nielsen, S. C., Hoh, R. A., Pursell, T., Arunachalam, P. S., Manohar, et al
2026
- **Transient Neonatal Zinc Deficiency due to Maternal Variants in SLC30A2: An Emerging and Atypical Candidate Gene for Maternal Carrier Screening.** *American journal of medical genetics. Part A*
Carter, C., Luz, D., Smith, E. E., Scott, S. A., Srinivas, N. S., Shi, R. Z., Tise, C. G.
2026: e70131
- **Biallelic Variants in RNU6ATAC Result in a Minor Spliceopathy Characterized by Transcriptome-Wide Minor Intron Retention Events and Short Stature with Variable Multisystem Manifestations.** *HGG advances*
Mendez, R., Arriaga, T. M., Ma, J., Bonner, D. E., Emami, S., Levy, R. J., Alsagheir, A., Alhaddad, B., Bakur, K., Ungar, R. A., Matalon, D. R., Miller, A. M., Nguyen, et al
2026: 100588
- **Pathogenic variants in myoferlin (*MYOF*) cause arrhythmogenic cardiomyopathy**
Kirillova, A., AYTEKIN, M., Chan, I., Mina, A., Shao, Y., Kirdar, A., Romito, O., Kalepalli, N., Zhang, D., Al Aaraj, Y., Reuter, C., Annis, J., Yue, et al
LIPPINCOTT WILLIAMS & WILKINS.2025
- **GREGoR: accelerating genomics for rare diseases.** *Nature*
Dawood, M., Heavner, B., Wheeler, M. M., Ungar, R. A., LoTempio, J., Wiel, L., Berger, S., Bernstein, J. A., Chong, J. X., Délot, E. C., Eichler, E. E., Lupski, J. R., Shojai, et al
2025; 647 (8089): 331-342
- **Drivers of differential antibody breadth in SARS-CoV-2 mRNA vaccination and infection**

- Fabian Wirz, O., Kotagiri, P., Haraguchi, E., Roltgen, K., Hunter, M., Craig, E., Afaghani, J., Lee, J., Nielsen, S. C. A., Hoh, R. A., Pursell, T., Arunachalam, P. S., Chang, et al
OXFORD UNIV PRESS.2025
- **Long-Read Sequencing Unveils an Intronic ASPA Retrotransposon Variant Implicated in Canavan Disease.** *Neurology. Genetics*
Qiao, W., Scott, S.
2025; 11 (5): e200300
 - **Transcriptome-wide outlier approach identifies individuals with minor spliceopathies.** *American journal of human genetics*
Arriaga, T. M., Mendez, R., Ungar, R. A., Bonner, D. E., Matalon, D. R., Lemire, G., Goddard, P. C., Padhi, E. M., Miller, A. M., Nguyen, J. V., Ma, J., Smith, K. S., Scott, et al
2025
 - **Towards an integrated resource for pharmacogenomics (PGx): Survey findings from the genomic medicine communities.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Klein, C. J., Gong, L., Caudle, K. E., Naik, H., Empey, P. E., Hoffman, J. M., Scherer, S., Iwuchukwu, O. F., Gregornik, D., Monte, A. A., Swen, J. J., Scott, S. A., Whirl-Carrillo, et al
2025: 101529
 - **Progress in Pharmacogenomics Implementation in the United States: Barrier Erosion and Remaining Challenges.** *Clinical pharmacology and therapeutics*
Smith, D. M., Douglas, M. P., Aquilante, C. L., Deverka, P. A., Devine, B., Dunnenberger, H. M., Empey, P. E., Hertz, D. L., Monte, A. A., Moyer, A. M., Patel, J. N., Pratt, V. M., Saulsberry, et al
2025
 - **Analytical validation of germline small variant detection using long-read HiFi genome sequencing.** *Genome research*
Hammond, N., Liao, L., Tong, P. W., Ng, Z., Nguyen, T. P., Ho, C., Yang, Y., Scott, S. A.
2025
 - **Discovery of ancestry-specific variants associated with clopidogrel response among Caribbean Hispanics.** *NPJ genomic medicine*
Yang, G., Gonzalez, P., Monero, M., Carrasquillo, K., Renta, J. Y., Hernandez-Suarez, D. F., Botton, M. R., Melin, K., Scott, S. A., Ruano, G., Roche-Lima, A., Alarcon, C., Ritchie, et al
2025; 10 (1): 20
 - **A Guide for Implementing DPYD Genotyping for Systemic Fluoropyrimidines into Clinical Practice.** *Clinical pharmacology and therapeutics*
Ho, T. T., Smith, D. M., Aquilante, C. L., Cicali, E. J., El Rouby, N., Hertz, D. L., Imanirad, I., Patel, J. N., Scott, S. A., Swain, S. M., Tuteja, S., Hicks, J. K.
2025
 - **Integrating Pharmacogenomics into the Broader Construct of Genomic Medicine: Efforts by the ClinGen Pharmacogenomics Working Group (PGxWG).** *Clinical chemistry*
Gong, L., Klein, C. J., Caudle, K. E., Moyer, A. M., Scott, S. A., Whirl-Carrillo, M., Klein, T. E.
2025; 71 (1): 36-44
 - **Transcriptome-wide outlier approach identifies individuals with minor spliceopathies.** *medRxiv : the preprint server for health sciences*
Arriaga, M. T., Mendez, R., Ungar, R. A., Bonner, D. E., Matalon, D. R., Lemire, G., Goddard, P. C., Padhi, E. M., Miller, A. M., Nguyen, J. V., Ma, J., Smith, K. S., Scott, et al
2025
 - **Twenty-Five Years of Germline Genetic Testing and What May Lie Ahead.** *The Journal of molecular diagnostics : JMD*
Pratt, V. M., Akhavanfard, S., Houldsworth, J., Laffin, J. J., Moyer, A. M., Reddi, H. V., Scott, S. A., Lebo, M. S.
2024; 26 (12): 1038-1041
 - **Implementing a pharmacogenomic-driven algorithm to guide antiplatelet therapy among Caribbean Hispanics: a non-randomised clinical trial.** *BMJ open*
Nuñez-Medina, H. J., Monero, M., Torres, L. M., Leal, E., Gonzalez-Sepulveda, L., Mayor, Á. M., Renta, J. Y., González-García, E. R., González, A., Melin, K., Scott, S. A., Ruaño, G., Hernandez-Suarez, et al
2024; 14 (9): e084119
 - **DPYD Genotyping Recommendations: A Joint Consensus Recommendation of the Association for Molecular Pathology, American College of Medical Genetics and Genomics, Clinical Pharmacogenetics Implementation Consortium, College of American Pathologists, Dutch Pharmacogenetics Working Group of the Royal Dutch Pharmacists Association, European Society for Pharmacogenomics and**

- Personalized Therapy, Pharmacogenomics Knowledgebase, and Pharmacogene Variation Consortium.** *The Journal of molecular diagnostics : JMD*
Pratt, V. M., Cavallari, L. H., Fulmer, M. L., Gaedigk, A., Hachad, H., Ji, Y., Kalman, L. V., Ly, R. C., Moyer, A. M., Scott, S. A., Turner, A. J., van Schaik, R. H., Whirl-Carrillo, et al
2024
- **Strategies for DPYD testing prior to fluoropyrimidine chemotherapy in the US.** *Supportive care in cancer : official journal of the Multinational Association of Supportive Care in Cancer*
Tracksdorf, T., Smith, D. M., Pearse, S., Cicali, E. J., Aquilante, C. L., Scott, S. A., Ho, T. T., Patel, J. N., Hicks, J. K., Hertz, D. L.
2024; 32 (8): 497
 - **Applications of genome sequencing as a single platform for clinical constitutional genetic testing.** *Genetics in medicine open*
Yang, Y., Del Gaudio, D., Santani, A., Scott, S. A.
2024; 2: 101840
 - **Nonlinear Mixed-Effects Model of Z-Endoxifen Concentrations in Tamoxifen-Treated Patients from the CEPAM Cohort.** *Clinical pharmacology and therapeutics*
Mc Laughlin, A. M., Helland, T., Klima, F., Koolen, S. L., van Schaik, R. H., Mathijssen, R. H., Neven, P., Swen, J. J., Guchelaar, H. J., Dalenc, F., White-Koning, M., Michelet, R., Mikus, et al
2024
 - **Knowledge and attitudes on implementing cardiovascular pharmacogenomic testing.** *Clinical and translational science*
Russell, C., Champion, M., Grove, M. E., Matsuda, K., Klein, T. E., Ashley, E., Naik, H., Wheeler, M. T., Scott, S. A.
2024; 17 (3): e13737
 - **From sample to star alleles: a long-read pharmacogenomics pipeline powered by Twist target enrichment and PacBio HiFi sequencing**
Holt, S., Harting, J., Han, T., Arbiza, L., Kingan, S., Soupe, A., Zhang, S., Baybayan, P., Lambert, C., Ferrao, H., Li, B., Sangkuhl, K., Woon, et al
SPRINGER NATURE.2024: 701-702
 - **FRAGILE X SYNDROME DUE TO A RARE <i>FMR1</i> SEQUENCE VARIANT**
Keehan, L., Hale, C., Fisk, D. G., Dykzeul, N., Scott, S. A., Tise, C. G.
SAGE PUBLICATIONS LTD.2024: 487
 - **Applications of genome sequencing as a single platform for clinical constitutional genetic testing** *Genetics in Medicine Open*
Yang, Y., del Gaudio, D., Santani, A., Scott, S. A.
2024
 - **Pharmacogenomic knowledge and awareness among diverse patients treated with angiotensin converting enzyme inhibitors.** *Pharmacogenomics*
Naik, H., O'Connor, M. Y., Sanderson, S. C., Pinnell, N., Dong, M., Wiegand, A., Obeng, A. O., Abul-Husn, N. S., Scott, S. A.
2023
 - **Implementing a Pharmacogenomic-driven Algorithm to Guide Antiplatelet Therapy among Caribbean Hispanics: A non-randomized prospective cohort study.** *medRxiv : the preprint server for health sciences*
Nuñez-Medina, H., Monero, M., Torres, L. M., Leal, E., González-Sepúlveda, L., Mayor, Á. M., Renta, J. Y., González-García, E. R., González, A., Melin, K., Scott, S. A., Rúaño, G., Hernandez-Suarez, et al
2023
 - **Using Genotyping-Based Mendelian Error Analysis to Identify and Minimize the Risk of Sample Misidentification in Trio Exome Sequencing Service**
Ho, C., Liao, L., Yang, Y., Scott, S., Qiao, W.
ELSEVIER SCIENCE INC.2023: S25
 - **Two epilepsy-associated variants in KCNA2 (KV1.2) at position H310 oppositely affect channel functional expression.** *The Journal of physiology*
Minguez-Vinas, T., Prakash, V., Wang, K., Lindstrom, S. H., Pozzi, S., Scott, S. A., Spiteri, E., Stevenson, D. A., Ashley, E. A., Gunnarsson, C., Pantazis, A.
2023
 - **Discovery of Ancestry-specific Variants Associated with Clopidogrel Response among Caribbean Hispanics.** *medRxiv : the preprint server for health sciences*

- Yang, G., González, P., Moneró, M., Carrasquillo, K., Renta, J. Y., Hernandez-Suarez, D. F., Botton, M. R., Melin, K., Scott, S. A., Rúaño, G., Roche-Lima, A., Alarcon, C., Ritchie, et al
2023
- **Highly Scalable Pharmacogenomic Panel Testing with Hybrid Capture and Long-Read Sequencing**
Gonzaludo, N., Kingan, S., Harting, J., Baybayan, P., Han, T., Arbiza, L., Li, B., Sangkuhl, K., Woon, M., Whaley, R., Whirl-Carrillo, M., Yang, Y., Klein, et al
LIPPINCOTT WILLIAMS & WILKINS.2023: 183
 - **Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for *CYP2D6*, *CYP2C19*, *CYP2B6*, *SLC6A4*, and *HTR2A* Genotypes and Serotonin Reuptake Inhibitor Antidepressants**
Bousman, C. A., Stevenson, J. M., Ramsey, L. B., Sangkuhl, K., Hicks, J., Strawn, J. R., Singh, A. B., Ruano, G., Mueller, D. J., Tsermpini, E., Brown, J. T., Bell, G. C., Leeder, et al
LIPPINCOTT WILLIAMS & WILKINS.2023: 181-183
 - **Beyond the exome: What's next in diagnostic testing for Mendelian conditions.** *American journal of human genetics*
Wojcik, M. H., Reuter, C. M., Marwaha, S., Mahmoud, M., Duyzend, M. H., Barseghyan, H., Yuan, B., Boone, P. M., Groopman, E. E., Délot, E. C., Jain, D., Sanchis-Juan, A., Starita, et al
2023; 110 (8): 1229-1248
 - **The Genetic Testing Reference Materials (GeT-RM) Coordination Program: Over 10 years of support for pharmacogenomic testing.** *The Journal of molecular diagnostics : JMD*
Scott, S. A.
2023
 - **CYP3A4 and CYP3A5 Genotyping Recommendations: A Joint Consensus Recommendation of the Association for Molecular Pathology, Clinical Pharmacogenetics Implementation Consortium, College of American Pathologists, Dutch Pharmacogenetics Working Group of the Royal Dutch Pharmacists Association, European Society for Pharmacogenomics and Personalized Therapy, and Pharmacogenomics Knowledgebase.** *The Journal of molecular diagnostics : JMD*
Pratt, V. M., Cavallari, L. H., Fulmer, M. 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
2023
 - **Response to the FDA Decision Regarding DPYD Testing Prior to Fluoropyrimidine Chemotherapy.** *Clinical pharmacology and therapeutics*
Hertz, D. L., Smith, D. M., Scott, S. A., Patel, J. N., Hicks, J. K.
2023
 - **Clinical Pharmacogenomic MT-RNR1 Screening for Aminoglycoside-Induced Ototoxicity and the Post-Test Counseling Conundrum.** *Clinical pharmacology and therapeutics*
Rigobello, R., Shaw, J., Ilg, D., Zimmerman, R., Edelmann, L., Kornreich, R., Scott, S. A., Cody, N.
2023
 - **Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants.** *Clinical pharmacology and therapeutics*
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, et al
2023
 - **Re-envisioning community genetics: community empowerment in preventive genomics.** *Journal of community genetics*
Wand, H., Martschenko, D. O., Smitherman, A., Michelson, S., Pun, T., Witte, J. S., Scott, S. A., Cho, M. K., Ashley, E. A., Preventive Genomics Program Co-Design Working Group, Goldberg, E., Knepper, L., Michelson, S., et al
2023
 - **An efficient genotyper and star-allele caller for pharmacogenomics.** *Genome research*
Hari, A., Zhou, Q., Gonzaludo, N., Harting, J., Scott, S. A., Qin, X., Scherer, S., Sahinalp, S. C., Numanagic, I.
2023
 - **Prescriber Adoption of SLCO1B1 Genotype-Guided Simvastatin Clinical Decision Support in a Clinical Pharmacogenetics Program.** *Clinical pharmacology and therapeutics*
Owusu Obeng, A., Scott, S. A., Kaszemacher, T., Ellis, S. B., Mejia, A., Gomez, A., Nadukuru, R., Abul-Husn, N. S., Vega, A., Waite, E., Gottesman, O., Cho, J., Bottinger, et al
2022

- **Characterization of Reference Materials for TPMT and NUDT15: A GeT-RM Collaborative Project**
Pratt, V., Wang, W., Boone, E., Broeckel, U., Cody, N., Edelmann, L., Gaedigk, A., Lynnes, T., Medeiros, E., Mitchell, M., Moyer, A., Scott, S., Starostik, et al
ELSEVIER SCIENCE INC.2022: S6
- **Human Mutation special issue on Innovations in Genomic Diagnostics.** *Human mutation*
Scott, S. A., Wang, K., Spinner, N. B.
2022
- **Long-read HiFi sequencing of NUDT15: Phased full-gene haplotyping and pharmacogenomic allele discovery.** *Human mutation*
Scott, E. R., Yang, Y., Botton, M. R., Seki, Y., Hoshitsuki, K., Harting, J., Baybayan, P., Cody, N., Nicoletti, P., Moriyama, T., Chakraborty, S., Yang, J. J., Edelmann, et al
2022
- **Characterization of Reference Materials for TPMT and NUDT15 - A GeT-RM Collaborative Project.** *The Journal of molecular diagnostics : JMD*
Pratt, V. M., Wang, W. Y., Boone, E. C., Broeckel, U., Cody, N., Edleman, L., Gaedigk, A., Lynnes, T. C., Medeiros, E., Moyer, A. M., Mitchell, M. M., Scott, S. A., Starostik, et al
2022
- **TPMT and NUDT15 Genotyping Recommendations: A Joint Consensus Recommendation of the Association for Molecular Pathology, Clinical Pharmacogenetics Implementation Consortium, College of American Pathologists, Dutch Pharmacogenetics Working Group of the Royal Dutch Pharmacists Association, European Society for Pharmacogenomics and Personalized Therapy, and Pharmacogenomics Knowledgebase.** *The Journal of molecular diagnostics : JMD*
Pratt, V. M., Cavallari, L. H., Fulmer, M. 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
2022
- **Attitudes on pharmacogenomic results as secondary findings among medical geneticists.** *Pharmacogenetics and genomics*
Bartos, M. N., Scott, S. A., Jabs, E. W., Naik, H.
2022
- **Effects of Testing and Disclosing Ancestry-Specific Genetic Risk for Kidney Failure on Patients and Health Care Professionals: A Randomized Clinical Trial.** *JAMA network open*
Nadkarni, G. N., Fei, K., Ramos, M. A., Hauser, D., Bagiella, E., Ellis, S. B., Sanderson, S., Scott, S. A., Sabin, T., Madden, E., Cooper, R., Pollak, M., Calman, et al
2022; 5 (3): e221048
- **Clinical pharmacogenomic testing and reporting: A technical standard of the American College of Medical Genetics and Genomics (ACMG).** *Genetics in medicine : official journal of the American College of Medical Genetics*
Tayeh, M. K., Gaedigk, A., Goetz, M. P., Klein, T. E., Lyon, E., McMillin, G. A., Rentas, S., Shinawi, M., Pratt, V. M., Scott, S. A., ACMG Laboratory Quality Assurance Committee. Electronic address: documents@acmg.net,,
2022
- **Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2C19 Genotype and Clopidogrel Therapy: 2022 Update.** *Clinical pharmacology and therapeutics*
Lee, C. R., Luzum, J. A., Sangkuhl, K., Gammal, R. S., Sabatine, M. S., Stein, C. M., Kisor, D. F., Limdi, N. A., Lee, Y. M., Scott, S. A., Hulot, J. S., Roden, D. M., Gaedigk, et al
2022
- **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
 - **Machine Learning for Prediction of Stable Warfarin Dose in US Latinos and Latin Americans.** *Frontiers in pharmacology*
Steiner, H. E., Giles, J. B., Patterson, H. K., Feng, J., El Rouby, N., Claudio, K., Marcatto, L. R., Tavares, L. C., Galvez, J. M., Calderon-Ospina, C. A., Sun, X., Hutz, M. H., Scott, et al
2021; 12: 749786
 - **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.
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