Elizabeth S. Barrie, PhD, FACMG

Assistant Professor of Pathology
Assistant Director of Molecular Diagnostics and Cytogenetics
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Education

Post Graduate

ABMGG Fellowship in Clinical Molecular and Cytogenetics and Genomics, Nationwide Children's Hospital & The Ohio State University, Columbus, OH

Post-doctoral Research, Center for Pharmacogenomics, The Ohio State University, Columbus, OH

Graduate

PhD Biomedical Sciences, The Ohio State University, Columbus, OH.
Thesis: *Genetic Factors Regulating Expression of Dopaminergic Genes*, Advisor: Wolfgang Sadee

Undergraduate

BS Biology, Case Western Reserve University, Cleveland, OH

Licenses & Certifications

Board Certifications

Clinical Cytogenetics and Genomics, American Board of Medical Genetics and Genomics (ABMGG) 09/2019

Clinical Molecular Genetics and Genomics, American Board of Medical Genetics and Genomics (ABMGG) 09/2019

Awards & Honors

Pharmacogenomics Research Network Featured Investigator 2015

Finalist for Hayes Graduate Research Forum 2014

Chauncey D. Leake Award for Excellence in Pharmacology 2013

Distinguished University Fellowship Recipient, The Ohio State University 2013

Howard Hughes Medical Institute Grant recipient for SPUR (Summer Program in Undergraduate Research) 2007

Professional Organizations

American College of Medical Genetics and Genomics (ACMG)

Association for Molecular Pathology (AMP)

Invited Presentations

"Persistent high levels of donor cells following solid organ transplant confirm diagnosis of graft versus host disease." Association for Molecular Pathology Meeting, Virtual, 11/2020. (Senior author; Kelly Rafferty presenting author).

"Atypical Fragile X syndrome mosaicism in family of 5 boys." Association for Molecular Pathology Meeting, San Antonio, TX. 11/2018

"Hypertetraploid partial molar pregnancy identified by microarray and STR analysis, with subsequent choriocarcinoma diagnosis." Association for Molecular Pathology Meeting, Salt Lake City, UT. 11/2017

Recent Poster Presentations

Memari P, **Barrie ES**, Williamson V, Ferreira-Gonzalez A, Pillappa R, Robila V, Muthusamy S. "Concurrent STK11 and TP53 Mutations Correlate with a Unique Morphological Phenotype and Clinical Stage in Lung Adenocarcinomas." United States & Canadian Academy of Pathology's Annual Meeting, Los Angeles, CA, 03/2022

Barrie ES, Pillappa R, Olson T, Sabato F, Williamson V, Paulraj P, Turner S, Ferreira-Gonzalez A. "Harnessing Existing Genetic Results from Next Generation Sequencing of Solid Tumors Facilitates Recommendation of New Therapies as Evidenced by Approval of Sotorasib for Treating KRAS G12C Tumors." Association for Molecular Pathology Meeting, Virtual, 11/2021

Williamson VS, Bullard B, Barrick B, Silvester A, Olson T, Morris P, Chhoa M, Zhang Y, Margaritini C, Sene M, Sabato M, Paulraj P, **Barrie ES**, Turner S, Ferreira-Gonzalez A. "Use of a Gaussian Mixed Model for Setting Viable Quality Control Cutoffs in RNA-sequencing." Association for Molecular Pathology Meeting, Virtual, 11/2021

Sábato MF, Beard A, Zhang Y, Sene M, Turner S, **Barrie ES**, Paulraj P, Ferreira-Gonzalez A. "Analytical Validation of a Modified FDA-cleared System for the Diagnosis and Monitoring of Chronic Myeloid Leukemia." Association for Molecular Pathology Meeting, Virtual, 11/2021

Lee W, Zhang Y, Olson T, Memari P, Garcia P, Sabato F, Williamson V, **Barrie E**, Paulraj P, Ferreira-Gonzalez A, Sayeed S, Turner S. "Determination of cytology smears adequacy for use in comprehensive genomic profiling of solid tumors by next generation sequencing." Association for Molecular Pathology Meeting, Virtual, 11/2021

Rafferty K, **Barrie ES**, Jackson-Cook C, Paulraj P. "A Rare 2p11.2 Microdeletion in an Infant with T cell Lymphopenia and an Abnormal Newborn Screen for Severe Combined Immunodeficiency." ACMG Annual Clinical Genetics Meeting, Digital Edition, 04/2021

Catic A, **Barrie ES**, Turner S, Ferreira-Gonzalez A, Jackson-Cook C, Lai G, Dalton J, Paulraj P. "Trisomy 3 and CBFA2T3-GLIS2 fusion in a child with acute megakaryoblastic leukemia without Down syndrome." ACMG Annual Clinical Genetics Meeting, Digital Edition, 04/2021

Paulraj P, **Barrie ES**, Jackson-Cook C. "Optical Genomic Mapping Reveals Balanced and Unbalanced Cytogenetic Findings Associated with Tumor-forming Potential in a Prostate Cancer Cell line (M2205)." ACMG Annual Clinical Genetics Meeting, Digital Edition, 04/2021

Barrie ES, Yu X, Sabato-Charreun F, Olson T, Ganesh V, Williamson VS, Bullard B, Gonzalez I, Barrick B, Paulraj P, Turner SA, Ferreira-Gonzalez A. "Validation of a Comprehensive, Targeted, Next-Generation Sequencing Panel for Solid Tumors." Association for Molecular Pathology Meeting, Virtual. 11/2020

Williamson VS, Bullard B, Gonzalez I, Barrick B, Yu X, Sabato-Charreun F, **Barrie ES**, Paulraj P, Turner SA, Ferreira-Gonzalez A. "Use and feasibility of Multi-algorithmic Consensus-based Bioinformatics pipelines in the detection of fusions in FFPE treated samples." Association for Molecular Pathology Meeting, Virtual. 11/2020

Barrie ES, Overwater E, van Haelst MM, Motazacker MM, Truxal K, Crist E, Mostafavi R, Pivnick EK, Choudhri AF, Narumanchi T, Castelluccio V, Walsh LE, Garganta C, Gastier-Foster J, "Expanding the spectrum of *CEP55*-associated

disease to viable phenotypes." ACMG Annual Clinical Genetics Meeting, Digital Edition. 03/2020

Morgan A, Koboldt DC, **Barrie ES**, Crist E, Mezzavilla M, Faletra F, Mihalic-Mosher T, Wilson RK, Manickam K, Gasparini P, Dell'Orco D, Girotto G. "Mutations in *PLS1*, encoding fimbrin, cause autosomal dominant non-syndromic hearing loss (ADNSHL)." European Human Genetics Conference, Gothenburg, Sweden. 06/2019

Barrie ES, Cottrell CE, Gastier-Foster JM, Hickey S, Patel A, Santoro S, Alfaro MP. "Genotype-Phenotype Correlation; Inheritance and Mutation-Type Infer Pathogenicity of Variants in *IQSEC2* Gene." ACMG Annual Clinical Genetics Meeting, Seattle, WA. 04/2019

Publications

Papers Published in Peer Reviewed Journals

Rose NC*, **Barrie ES***, Malinowski J, Jenkins G, McClain MR, LaGrave D, Leung ML, ACMG Professional Practice and Guidelines Committee. "Systematic evidence-based review: the application of noninvasive prenatal screening using cell free DNA in general risk pregnancies." *In revision* at *Genetics in Medicine*.

Rafferty KA, **Barrie ES**, Turner SA, Ferreira-Gonzalez A, Jackson-Cook C, Paulraj P. "Severe Graft-Versus-Host Disease Following Solid Organ Transplant Confirmed by Chimerism Studies and Cytogenetic Analyses." Experimental and Clinical Transplantation. *Accepted*.

Barrie ES, Overwater E, van Haelst MM, Motazacker MM, Truxal KV, Crist E, Mostafavi R, Pivnick EK, Choudhri AF, Narumanchi T, Castelluccio V, Walsh LE, Garganta C, Gastier-Foster JM. Expanding the spectrum of CEP55-associated disease to viable phenotypes. Am J Med Genet A. 2020 Feb 25. PMID: 32100459

Morgan A, Koboldt DC, **Barrie ES**, Crist ER, García García G, Mezzavilla M, Faletra F, Mihalic Mosher T, Wilson RK, Blanchet C, Manickam K, Roux AF, Gasparini P, Dell'Orco D, Girotto G. Mutations in PLS1, encoding fimbrin, cause autosomal

dominant nonsyndromic hearing loss. Hum Mutat. 2019 Dec; 40(12):2286-2295. PMID: 31397523

Barrie ES, Cottrell CE, Gastier-Foster J, Hickey SE, Patel AD, Santoro SL, Alfaro MP. Genotype-phenotype correlation: Inheritance and variant-type infer pathogenicity in IQSEC2 gene. Eur J Med Genet. 2019 Aug 12; :103735. PMID: 31415821

Barrie ES, Alfaro MP, Pfau RB, Goff MJ, McBride KL, Manickam K, Zmuda EJ. De novo loss-of-function variants in *NSD2* (*WHSC1*) associate with a subset of Wolf-Hirschhorn syndrome. Cold Spring Harb Mol Case Stud. 2019 Aug; 5(4). PMID: 31171569

Barrie ES, Li Y, Lamb-Thrush D, Hashimoto S, Matthews T, Mouhlas D, Pyatt R, Reshmi SC, Gastier-Foster JM, Pfau R, Astbury C. Pericentromeric regions of homozygosity on the X chromosome: Another likely benign population variant. Eur J Med Genet. 2018 Jul; 61(7):416-420. PMID: 29572065

Barrie ES, Pinsonneault JK, Sadee W, Hollway JA, Handen BL, Smith T, Arnold LE, Butter E, Hansen-Kiss E, Herman GE, Aman MG. Testing genetic modifiers of behavior and response to atomoxetine in autism spectrum disorder with ADHD. J Dev Phys Disabil. 2018 Jun; 30(3):355-371. PMID: 30197492

Barrie ES, Lee SH, Frater JT, Kataki M, Scharre DW, Sadee W. Alpha-synuclein mRNA isoform formation and translation affected by polymorphism in the human SNCA 3'UTR. Mol Genet Genomic Med. 2018 May 6. PMID: 29730891

Barrie ES, Hartmann K, Lee SH, Frater JT, Seweryn M, Wang D, Sadee W. The CHRNA5/CHRNA3/CHRNB4 Nicotinic Receptor Regulome: Genomic Architecture, Regulatory Variants, and Clinical Associations. Hum Mutat. 2017 Jan; 38(1):112-119. PMID: 27758088

Sanford JC, Wang X, Shi J, **Barrie ES**, Wang D, Zhu HJ, Sadee W. Regulatory effects of genomic translocations at the human carboxylesterase-1 (CES1) gene locus. Pharmacogenet Genomics. 2016 May; 26(5):197-207. PMID: 26871237

Cubells JF, Schroeder JP, **Barrie ES**, Manvich DF, Sadee W, Berg T, Mercer K, Stowe TA, Liles LC, Squires KE, Mezher A, Curtin P, Perdomo DL, Szot P, Weinshenker D. Human Bacterial Artificial Chromosome (BAC) Transgenesis Fully

Rescues Noradrenergic Function in Dopamine ß-Hydroxylase Knockout Mice. PLoS One. 2016; 11(5):e0154864. PMID: 27148966

Barrie ES, Lodder M, Weinreb PH, Buss J, Rajab A, Adin C, Mi QS, Hadley GA. Role of ITGAE in the development of autoimmune diabetes in non-obese diabetic mice. J Endocrinol. 2015 Mar; 224(3):235-43. PMID: 25525188

Barrie ES, Weinshenker D, Verma A, Pendergrass SA, Lange LA, Ritchie MD, Wilson JG, Kuivaniemi H, Tromp G, Carey DJ, Gerhard GS, Brilliant MH, Hebbring SJ, Cubells JF, Pinsonneault JK, Norman GJ, Sadee W. Regulatory polymorphisms in human DBH affect peripheral gene expression and sympathetic activity. Circ Res. 2014 Dec 5; 115(12):1017-25. PMID: 25326128

Kempkes R, Stofko E, Lam K, Snell EH. Glycerol concentrations required for the successful vitrification of cocktail conditions in a high-throughput crystallization screen. Acta Crystallogr D Biol Crystallogr. 2008 Mar; 64(Pt 3):287-301. PMID: 18323624

Coling DE, Ding D, Young R, Lis M, Stofko E, Blumenthal KM, Salvi RJ. Proteomic analysis of cisplatin-induced cochlear damage: methods and early changes in protein expression. Hear Res. 2007 Apr; 226(1-2):140-56. PMID: 17321087

Editorials, Reviews, Commentaries, Proceedings, Invited Published Papers in Peer Reviewed Journals

1. **Barrie ES**, Smith RM, Sanford JC, Sadee W. mRNA transcript diversity creates new opportunities for pharmacological intervention. Mol Pharmacol. 2012 May; 81(5):620-30. PMID: 22319206

Books, Book Chapters and Monographs

1. Lee SH, **Barrie ES**, Sadee W, Smith RM. "Nicotine dependence and the *CHRNA5/CHRNA3/CHRNB4* nicotinic receptor regulome." In Victor R. Predy (Ed.), Neuroscience of Nicotine: Mechanisms and Treatment. 2019 Mar: 347-353.