

Elizabeth S. Barrie, PhD, FACMG



Assistant Professor of Pathology
Assistant Director of Molecular Diagnostics and Cytogenetics
Virginia Commonwealth University
Office: (804) 628-3662
elizabeth.barrie@vcuhealth.org

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

"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

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

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, Faletta 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/CHRNA4 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 β -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, Hebring 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/CHRNA4* nicotinic receptor regulome." In Victor R. Preedy (Ed.), *Neuroscience of Nicotine: Mechanisms and Treatment.* 2019 Mar: 347-353.