Elizabeth S. Barrie, PhD, FACMG

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Associate Director of Cytogenetics
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Education

2019	Fellowship: ABMGG Fellowship in Clinical Molecular and Cytogenetics and Genomics, Nationwide Children's Hospital & The Ohio State University, Columbus, OH.
2014	Doctoral: PhD Biomedical Sciences, The Ohio State University, Columbus, OH.
2008	Undergraduate: BS Biology, Case Western Reserve University, Cleveland, OH.

Licenses & Certifications

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

Selected Awards & Honors

2015	Pharmacogenomics Research Network Featured Investigator
2014	Finalist for Hayes Graduate Research Forum
2013	Chauncey D. Leake Award for Excellence in Pharmacology
2013	Distinguished University Fellowship Recipient, The Ohio State University
2007	Howard Hughes Medical Institute Grant recipient for SPUR (Summer Program in Undergraduate Research)

Professional Service (outside)

Clinical & Laboratory Standards Institute Document Development Committee (CLSI DDC) on Interpretive Framework Heritable Disorders (MM29)

Systematic evidence-based review on behalf of the ACMG Professional Practice and Guidelines Committee

Recent Invited Presentations

"Rare patient with an interstitial 13q deletion derived from a paternal chromosome 13 paracentric inversion." Association for Molecular Pathology (AMP) Meeting, Salt Lake City, UT, November 2023. (Senior author; Divya Vinjamur was presenting author).

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

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

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

Selected Publications

Peer Reviewed Papers

Paulraj P, **Barrie ES**, Jackson-Cook C. "Optical genome mapping reveals balanced and unbalanced genetic changes associated with tumor-forming potential in an early-stage prostate cancer epithelial subline (M2205)." Molecular Genetics & Genomic Medicine. Epub 2023 Oct 30. DOI: 10.1002/mgg3.2307. PMID: 37902189.

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." Genetics in Medicine. 2022 Jul;24(7):1379-1391. Epub 2022 May 24. PMID: 35608568. *equal contribution.

Gillam J, Catic A, Paulraj P, Dalton J, Lai G, Jackson-Cook C, Turner S, Ferreira-Gonzalez A, **Barrie E**. "Acute megakaryoblastic leukemia with trisomy 3 and CBFA2T3::GLIS2: A case report." Genes Chromosomes Cancer. 2022 Aug;61(8):491-496. Epub 2022 Mar 16. DOI: 10.1002/gcc.23039. PMID: 35294081.

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." Exp Clin Transplant. Epub 2022 Mar 15. DOI: 10.6002/ect.2021.0460. PMID: 35297330.

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." American Journal of Medical Genetics Part A. 2020 May;182(5):1201-1208. Epub 2020 Feb 25. DOI: 10.1002/ajmg.a.61512. PMID: 32100459.

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." European Journal of Medical Genetics. 2020 Mar;63(3):103735. Epub 2019 Aug 12:103735. DOI: 10.1016/j.ejmg.2019.103735. PMID: 31415821.

Morgan A, Koboldt DC, **Barrie ES**, Crist E, García GG, 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 non-syndromic hearing loss." Human Mutation. 2019 Dec;40(12):2286-2295. Epub 2019 Aug 9. DOI: 10.1002/humu.23891. PMID: 31397523.

Barrie ES, Alfaro MP, Pfau RB, McBride K, Manickam K, Zmuda EJ. "De novo loss-of-function variants in NSD2 (WHSC1) associate with Wolf- Hirschhorn-like phenotype." CSH Molecular Case Studies. 2019 Aug;5(4). Epub 2019 Jun 06. DOI: 10.1101/mcs.a004044. PMID: 31171569.

Barrie ES, Li Y, Lamb-Thrush D, Hashimoto S, Matthews T, Mouhlas D, Pyatt R, Reshmi S, Gastier-Foster JM, Pfau R, Astbury C. "Pericentromeric Regions of Homozygosity on the X Chromosome are Likely Benign Population Variation." European Journal of Medical Genetics, 2018 Jul;61(7):416-420. Epub 2018 Mar 20. DOI: 10.1016/j.ejmg.2018.02.008. PMID: 29572065.

Book Chapters and Monographs

Barrie ES and Ferreira-Gonzalez A. "Advances in Cell-Free DNA." In Gregory J. Tsongalis (Ed.) Advances in Molecular Pathology. 2022 Nov: Volume 5, Issue 1, 141 – 148. DOI: 10.1016/j.yamp.2022.08.002.