Colleen Jackson-Cook, PhD

Professor, Pathology, Human & Molecular Genetics, and OB/GYN
Director, Molecular Cytogenetics Lab Division of Molecular Diagnostics
Department of Pathology 1991 - present
Program Director, LGG Fellowship, VCU 2015 - present
Office: 804-628-2992 | Colleen.Jackson-Cook@vcuhealth.org
Clinical Specialities: Molecular cytogentics

Education

University of Charleston, WV, BS, 1978
West Virginia University, MS, 1980
Virginia Commonwealth University, PhD, 1985
Clinical Cytogenetics Fellowship, Virginia Commonwealth University, 1988

Licenses & Certifications

American Board of Medical Genetics Clinical Cytogeneticist
American Board of Medical Genetics PhD Medical Geneticist

Selected Awards & Honors

Professional Achievement Award, WISDM, School of Medicine, VCU, 2024

Outstanding Teacher, M1 Human Genetics Course or Department of Human Genetics, 2000, 2004, 2007, 2013, 2017, 2018, 2019, 2020, 2022, 2023

Virginia Commonwealth University Alumni Star Award, VCU, 2005

Gallery of Outstanding Alumni Achievement (Founding Fellow), University of Charleston, 1992

American Society of Human Genetics Predoctoral Research Award, 1985

Recent Grants and Funding

Currently active:

R01AG082811 (Jackson-Cook) 09/15/2023 – 06/30/2028
NIH/NIA
Cytosolic DNA, Telomeres/Subtelomeres, and Epigenetics: A Longitudinal Twin Study to Assess the Role of Genetics and Environment on Their Frequency and Inter-relationships

The primary aims for this project was to determine the frequency, genetic contents, and stability of cytosolic self-DNA over time and determine if these attributes are associated with other age-related biological changes. By longitudinally studying identical and fraternal twins, we will also discover if cytosolic DNA arises in somatic cells due to a genetic predisposition or from environmental factors.

R01HD111943 (Jackson-Cook) 05/01/2023 - 03/31/2028
NIH/NICHD/NIA
A mosaic Down syndrome model system comparing isogenic trisomic/disomic cells to unmask trisomy 21-related genomic, epigenomic, and senescence changes acquired across the lifespan.

The overall goal of this study was to identify biological changes related to cytoplasmic self-DNA that are acquired with age (somatic chromosomal changes, telomere shortening, senescence, and/or epigenetic alterations) as a result of a trisomic imbalance for chromosome 21 in people with mosaic Down syndrome.

Previous (subset of 3 selected):

R13HD112166-01 (Jackson-Cook) 04/01/2023 – 03/31/2024
NIH/NICHD
2023 International Mosaic Down Syndrome Association Community-Empowered Research and Retreat Weekend: Increasing Partnerships, Cohorts, and Diversity for Research Related to Down Syndrome

The primary goals for this conference were to share knowledge and provide research participation opportunities related to mosaic and non-mosaic Down syndrome in a collaborative and supportive setting, with a focus on recruiting participants and scientific trainees from minority groups.

R03HD092640 (Bhattacharyya and Jackson-Cook) 04/01/2018 – 02/31/2021
NICHD
Generation of isogenic trisomy 21 iPSC resource
The primary aim of this proposal was to generate isogenic trisomy 21 and euploid iPSCs from somatic cells of individuals with mosaic DS and to make these cells readily available to other researchers who wish to better understand biological changes related to Down syndrome

RNR0112667A (Lyon and Jackson-Cook) 09/29/2010 - 07/31/2016
Epigenetics and Psychoneurologic Symptoms in Women with Breast Cancer
The primary aims of this proposal are to quantify the frequency of acquired chromosomal and epigenetic changes that arise during and following chemotherapy treatment and to determine if there was a relationship between the acquisition and/or persistence of fully characterized psychoneurologic symptoms with the acquisition/retention of genetic/epigenetic changes.

Editorial Advisory Boards

Co-Editor, Frontiers in Genetic Research Topic, "Down Syndrome: Genetic and Epigenetic Influences on this Multi-faceted Condition", 2021-23

Professional Service (outside)

2021 – 2026	Member of Board of Directors of American Board of Medical Genetics and Genomics
2023 – 2024	NIH Program Consultant for Somatic Mosaicism Across Human Tissues Network
2024	Assistant Book Chief, Certification Exam, American Board Medical Genetics & Genomics
2000 – present	Scientific Advisor, International Mosaic Down Syndrome Association

2020 – 2022	AGCMG LGG Milestones 2.0 Quality Assurance Project
2020	American Board Medical Genetics & Genomics, Board of Directors Nominating Committee, Chair
2019	ACGME LGG Milestones Committee
2019 – 2020	NIH INCLUDE Down Syndrome Minimal Common Dataset Working Group
2019 – 2020	NIH INCLUDE Down Syndrome Community Outreach Working Group
2019 – 2020	NIH INCLUDE Down Syndrome Existing Cohorts Working Group
2019 – 2020	NIH INCLUDE Down Syndrome Biobanking and Specimen Collection Working Group
2000 – present	Cancer & Leukemia Group B (CALGB); Cytogenetics Working Group
1998 – 2002	Advisor to NCCLS Subcommittee on FISH Methods for Medical Genetics

VCU Professional Service

2023	Mid-Atlantic Twin Registry Director Search Committee
2021 – present	Mid-Atlantic Twin Registry Advisory Committee
2019 – 2021	Virginia Commonwealth University Promotion and Tenure Appeal Committee
2021 – 2022	Chair, Virginia Commonwealth University Promotion and Tenure Appeal Committee
1985 – present	Genetics Departmental Curriculum Committee

Recent Invited Presentations (subset of 10 selected)

"NMASKK Repository", International Mosaic Down Syndrome Association Research & Retreat Conference, San Diego, CA, July 28, 2023.

"Unmasking Cellular Changes in Trisomy 21: Studies of Isogenic Trisomic/Disomic Cells from People with Mosaicism, Virtual NIH INCLUDE Investigators Annual Workshop, October 12, 2022.

"Unmasking Cellular changes Associated with Trisomy 21: Studies of People with Mosaic Down Syndrome or Down Syndrome Regression Disorder", Southeastern Regional Genetics Group (SERGG), Asheville, NC, July 15, 2022.

"Mosaic Down Syndrome and Precocious Aging", International Mosaic Down Syndrome Research Retreat, Quarryville, Pennsylvania, July 16, 2021.

"Reproductive senescence in Turner syndrome as a model of aging", Turner Resource Network Symposium "Turner Science in the 21st Century", Arlington, Virginia, July 15-17, 2018.

"Increased frequencies of age-related, trisomy 21-associated somatic chromosomal instability and epigenetic alterations unmasked using an isogenic trisomic/disomic mosaic model system", Keystone Symposium, Biology of Down Syndrome: Impacts Across the Biomedical Spectrum (A4), Santa Fe, New Mexico, January 24-27, 2016.

"Acquired chromosomal and epigenetic changes in healthy cells following chemotherapy for breast cancer", Affymetrix American Cytogenetics Conference Symposium, Sunriver, Oregon, June 27, 2016.

"The Chromosomal and Epigenetic Make-up of Our Somatic Cells: Is it Black and White, or Fifty Shades of Grey?", Annual Virginia State Genetics Education Meeting, Richmond, VA, May 4, 2015.

"Pragmatic Aspects of Using DNA Copy Number and SNP Microarrays to Detect Cytogenetic Abnormalities in Solid Tumors", NIH sponsored by Affymetrix, Bethesda, MD, July 30, 2015.

"Studies of Discordant Identical Twins Reveal the Presence of Acquired Chromosomal Instability and Epigenetic Alterations", Association of Genetic Technologists Annual meeting, Savannah, Georgia, June 5, 2015.

Selected Publications

Peer Reviewed Papers (subset of 10 selected)

Deignan JL, Aggarwal V, Bale AE, Bellissimo DB, Booker JK, Cao Y, Crooks KR, Deak KL, delGaudio D, Funke B, Hoppman NL, Horner V, Hufnagel RB, **Jackson-Cook C**, Koduru P, Leung ML, Li S, Liu P, Luo M, Mao R, Mason-Suares H, Mikhail FM, Moore SR, Naeem RC, Pollard LM, Repnikova EA, Shao L, Shaw BM, Shetty S, Smolarek TA, Spiteri E, Ziffle JV, Vance GH, Vnencak-Jones CL, Williams ES. The challenges and opportunities of offering and integrating training in clinical molecular genetics and clinical cytogenetics: a survey of LGG Fellowship Program Directors, Gen Med., 2024

Paulraj P, Barrie E, **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). Mol Genet Genomic Med. Jan;12(1):e2307. PMID: 37902189, 2024

Fidler DJ, Riggs N, Esbensen AJ, **Jackson-Cook C**, Rosser T, Cohen A. Outreach and engagement efforts in research on Down syndrome: An NIH INCLUDE working group consensus statement. Int Rev Res Dev Disabi. 63: 247-267, PMID 36545326, 2022

Yang GS, Yang K, Weaver MT, Kelly DL, Dorsey SG, **Jackson-Cook C**, Lyon D. Exploring the relationship between DNA methylation age measures and psychoneurological symptoms in women with early-stage breast cancer. Support Care Cancer, 31 (1): 65, PMID 36538110, 2022

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

Martin D, Smith SC, Chesney A, **Jackson-Cook C**, Raghavendra P. Too Sensitive, or Just Right: A Comparison of Two ALK Antibodies (D5F3 and ALK1) for Assessment of Anaplastic Large Cell Lymphoma (ALCL), Am J Clin Pathology, 158(2):283-290, 2022

Rafferty K, Archer KJ, Turner K, Brown R, **Jackson-Cook C**. Trisomy 21-associated increases in chromosomal instability are unmasked by comparing isogenic

trisomic/disomic leukocytes from people with mosaic Down syndrome. PLoS One Jul 20;16(7):e0254806. PMID: 34283872, 2021

Lancaster EE, Lapato DM, **Jackson-Cook C**, Strauss JF, Roberson-Nay R, York TP: Maternal biological age assessed in early pregnancy is associated with gestational period, Scientific Reports 11(1): 15440. PMID34326348, 2021

Gheghiani L, Wang L, Zhang Y, Moore X, Zhang J, Smith SC, Tian Y, Wang L, Turner K, **Jackson-Cook C**, Mukhopadhyay N, Fu Z: PLK1 induces chromosomal instability and overrides cell cycle checkpoints to drive tumorigenesis. Cancer Res. 81(5):1293-1307, 2021

Alhareeri AA, Archer, KJ, Fu H, PhD; Lyon DE, Elswick RK, Kelly DL, Starkweather AR, Elmore LW, **Jackson-Cook CK**. Telomere lengths in women treated for breast cancer show associations with chemotherapy, pain symptoms, and cognitive domain measures: A longitudinal study. Breast Cancer Res. 2020 Dec 4;22(1):137, 2020

Book Chapters and Monographs

Jackson-Cook C, Ponnala S: Application of Chromosomal Microarrays in Molecular Oncology Testing for Solid Tumors: A Pragmatic Approach, eds. Idowu, MO, Dumur CI, Garrett C, Springer, New York, pp. 181-217, 2015.

Jackson-Cook C: Constitutional and acquired autosomal aneuploidy, Clin Lab Med Dec 31(4):481-511 (PMID 22118733), 2011.

Quillin John, **Jackson-Cook Colleen,** Bodurtha Joann: The link between providers and patients: How laboratories can ensure quality results with genetic testing, Clinical Laboratory Management, Nov/Dec 2003.

Riley RS, Massey D, **Jackson-Cook C**, Idowu M, Romagnoli G: Immunophenotypic Analysis of Acute Lymphocytic Leukemia in Hematology/Oncology Clinics of North America, 16(2): 245-299, 2002.

Patents

Licensing of P69SV40T prostate cancer cell line with Applied Biological Materials Inc., 2021

VCU Invention License Application (VCU No. 06-15) entitled, “A High Throughput for the Identification of compounds which block prostate tumor progression”, Zehner Z, Holmes M, Ware, J, and Jackson-Cook C

Media

International Mosaic Down Syndrome Association, Information for Families and Professionals, 2021

S Waters-Freyer, C Jackson-Cook, Documentary “A Diagnosis of Down Syndrome: The First Steps”, 2015

Posters/Abstracts

Barrie ES, Turner SA, **Jackson-Cook C**. Enhancing Transparency in Fellowship Opportunities: A Resource for Applicants, Association of Professors of Human & Medical Genetics, Stevenson, WA. May 2024.

Vinjamur DS, **Jackson-Cook C**, Barrie E. Rare case of parental chromosome 13 paracentric inversion leading to unbalanced meiotic recombination and a child with interstitial 13q deletion. Association for Molecular Pathology, Early Bird Case Study Presentation, Salt Lake City, UT, Nov 2023.

Turner SA, Guo G, **Jackson-Cook C**, Barrie E, Ferreira-Gonzalez A. Applications of molecular techniques in the clinical laboratory: An immersive educational experience for clinical learners. Presented in poster session at APHMG meeting, Kiawah Island, May 2023.

Bhattacharyya A, Yin Y, Soref C, Strand B, Xu K, Held D, **Jackson-Cook C**. Generation of Isogenic Trisomy 21 iPSC Resources. International Society for Stem Cell Research, 2022.

Rafferty K, Lapato D, Turner K, Skotko BG, Torres A, Santoro SL, Oreskovic N, Haugen K, Krell K, Brown R, **Jackson-Cook C**. Increased micronuclei frequency and DNA methylation patterns associated with Down Syndrome Regression Disorder, American Cytogenetics Consortium, 2022.

Saadeh-Jackson S, King K, Al Saif H, **Jackson-Cook C**, Schwartz S, Schleede J, Couser N. “Eye, Ocular Adnexa, and Facial Manifestations of Tetrasomy 18p: Case Report and Literature Review”, ACMGG 2021 Virtual Meeting, submitted in 2020.
Rafferty K, **Jackson-Cook C**, Paulraj P, Turner S, Gonzalez P, Ferreira-Gonzalez A,

Barrie E. “Persistent high levels of donor cells following solid organ transplant confirm diagnosis of graft versus host disease, Association for Molecular Pathology, Virtual Meeting, 2020.

Koebley SR, Mikheyken A, **Jackson-Cook C**, Reed J. “Nanomapping with Cas9: A new high- resolution and efficient method for characterizing chromosomal rearrangements”, submitted in Dec 2019 for American Cytogenetics Conference in Hilton Head, SC, 2020.

Jackson-Cook C, Rafferty K, Bhattacharyya A. “A new isogenic trisomy 21 iPSC resource”, submitted in Dec 2019 for American Cytogenetics Conference in Hilton Head, SC, 2020.

Yang C, Smith S, Akbarzadeh C, Keiser J, **Jackson-Cook C**. “Genome-wide copy number abnormalities in lipomatous neoplasms detected with chromosomal microarray: Is gain of MDM2 necessary for 12q amplification characterization and prognostication?”, submitted in 2019 for American Cytogenetics Conference in Hilton Head, SC, 2020.