

# Colleen Jackson-Cook, PhD



Professor, Pathology and OB/GYN  
Director, Molecular Cytogenetics Lab Division of Molecular Diagnostics  
Department of Pathology 1991 - present  
Program Director, Laboratory Genetics & Genomics Fellowship  
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## Education

1988 Clinical Cytogenetics Fellowship, Virginia Commonwealth University, Richmond, VA

1985 PhD, Virginia Commonwealth University, Richmond, VA

1980 MS, West Virginia University

1978 BS, University of Charleston, West Virginia

## Licenses & Certifications

Clinical Cytogeneticist, American Board of Medical Genetics

PhD Medical Geneticist, American Board of Medical Genetics

## Selected Awards & Honors

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

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

## Recent Grants & 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 are 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 is to identify biological changes related to cytoplasmic self-DNA that are acquired with age (somatic chromosomal changes, telomere shortening, senescence, and/or epigenetic alterations) due to 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 were 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

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

## Professional Service (outside)

2025 - 2026 Treasurer-Elect for Board of Directors of American Board of Medical Genetics & Genomics

2021 - 2027 Member of Board of Directors of American Board of Medical Genetics & Genomics

2023 - present NIH Program Consultant for Somatic Mosaicism Across Human Tissues Network

2025 - 2026 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

2000 -present Cancer & Leukemia Group B (CALGB); Cytogenetics Working Group

## 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

2026 "Unmasking trisomy 21-specific biological alterations by comparing isogenic trisomic and disomic cells from people with mosaic Down syndrome: A longitudinal study", Trisomy 21 Research Society Annual Meeting, Denver, CO

2025 "Understanding Mosaic Down Syndrome: Live Q&A with Dr. Jackson-Cook", IMDSA Webinar

2025 "Unmasking Genetic and Epigenetic Changes Associated with Trisomy 21 Across the Lifespan: Studies of People with Mosaic Down Syndrome", Clinical Genomics Grand Rounds, Mayo Clinic, Rochester, MN

2025 "Mosaic Down Syndrome 101", International Mosaic Down Syndrome Association Research & Retreat Conference, Keynote Address, Niagara Falls, Ontario, Canada

2024 "Studies of Cytosolic DNAs in the Interactions of Aging Hallmarks", NIH Aging Hallmarks Consortium of Grantees' Meeting, Seattle, WA

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

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

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

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

## Selected Publications

### *Peer Reviewed Papers (subset of 10 selected)*

Brown RC, D'Aguilar A, Hurshman Q, Nailor-Zee R, York TP, Capone G, Amstadter AB, **Jackson-Cook C**. Internalizing Psychiatric Symptoms in People With Mosaicism for Trisomy 21. *Am J Med Genet B Neuropsychiatr Genet*. Jul;198(5):e33022. PMID: 39821956, 2025.

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

### *Book Chapters and Monographs*

**Jackson-Cook C**, Barrie E, Lyalin D, Gwilliam K, Martin K, Alhareeri A, Lawce H: Chromosome Stains. in eds. P Hu and J Smith, *The AGT Cytogenetics Laboratory Manual*, 5th edition, John Wiley & Sons, Inc., Hoboken, NJ, 2025

**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

## *Patents*

Provisional Patent Application entitled, "Amplification-free detection of genetic, epigenetic, and structural variations with single molecule imaging from genomic DNA", Reed J, Gheghiani L, Mikheykin A, Jackson-Cook C, Riethman H, Fall 2025

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 (subset of 10 selected)*

Gheghiani L, Mikheykin A, Riethman HC, **Jackson-Cook C**, Reed J. Seeing is Believing: Physical Ground-Truthing of Telomeres and Subtelomeres via Nano-Imaging. Workshop on Long-Read Sequencing, Farmington, CT, May 2026

Gheghiani L, Olaosebikan T, Rafferty K, Archer KJ, **Jackson-Cook C**. Micronuclei Profiling of Isogenic Trisomic/Disomic Cells from People with Mosaic Down Syndrome Reveals a Trisomy 21- and Age-Associated Increase in Genome-wide Chromosomal Instability, Trisomy 21 Research Society Meeting, Denver, CO, June 2026

Alaali L, York TP, **Jackson-Cook C**. Analysis of Telomere Dysfunction in Isogenic Trisomic and Disomic Cells from People with Mosaic Down Syndrome. Trisomy 21 Research Society Meeting, Denver, CO, June 2026

Ashton VS, Gheghiani L, Dozmorov M, **Jackson-Cook C**. Unmasking Trisomy 21-Related Transcriptomic Dysregulation & Enhanced Senescence Using a Mosaic Down Syndrome Model, Trisomy 21 Research Society Meeting, Platform presentation, Denver, CO, June 2026

Gheghiani L, Mikheykin A, Riethman HC, Reed J, **Jackson-Cook C**. Advancing Telomere Length Analysis: A Single-molecule Precision Approach Using High-speed Atomic Force Microscopy (HS-AFM). Poster presentation at the American Cytogenomics Consortium, San Diego, CA, May 2026

Ashton VS, Gheghiani L, Dozmorov M, **Jackson-Cook C**. Unmasking Trisomy 21-Related Transcriptomic Dysregulation & Enhanced Senescence Using a Mosaic Down Syndrome Model, platform presentation at the American Cytogenomics Consortium, San Diego, CA, May 2026

Gwilliam K, Chang J, Colon-Cartagena L, Zot P, Smith S, **Jackson-Cook C**. Development and clinical utility of a whole genome SNP/Copy number microarray for assessment of mesenchymal tumors, selected for poster presentation, Association of Molecular Pathologists, Boston, MA, November 2025

Gheghiani L, Mikheykin A, Johnson J, Riethman HC, Reed J, **Jackson-Cook C**. Single-molecule profiling of telomeres and subtelomeres using high-speed atomic force microscopy and CRISPR-directed labeling, American Society of Human Genetics, Boston, MA, October 2025

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