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Education

1995-1999 Bachelor of Science in Biology
Geneva College, Beaver Falls, Pennsylvania

1999-2001 Master of Science in Genetic Counseling
Arcadia University, Glenside, Pennsylvania

2005-2010 Master of Bioethics (MBE)
University of Pennsylvania School of Medicine, Philadelphia, Pennsylvania

Current education

2017-- Ph.D. Candidate, Clinical Sciences
University of Colorado Health Sciences, Aurora, Colorado
- *Anticipated graduation August 2021*

Academic appointments

2007-2010 Adjunct Professor, Genetic Counseling Program
Arcadia University, Glenside, Pennsylvania

2012-2017 Assistant Professor, Department of Pediatrics
University of Colorado School of Medicine, Aurora, Colorado

2013 -- Faculty Member, Genetic Counseling Graduate Program
University of Colorado Denver, Aurora, Colorado

2016 -- Associate Faculty (2016-2019), Faculty (2020 --)
Center for Bioethics and Humanities
University of Colorado Anschutz Medical Campus, Aurora, Colorado

2017 -- Associate Professor, Department of Pediatrics
University of Colorado School of Medicine, Aurora, Colorado

Professional positions

2001-2006 Genetic Counselor, Department of Maternal Fetal Medicine
Christiana Care Health System, Newark, Delaware

2006-2010 Genetic Counselor, Section of Biochemical Genetics
The Children's Hospital of Philadelphia, Philadelphia, Pennsylvania

2010 -- Genetic Counselor, Section of Genetics and Metabolism
Children's Hospital Colorado, Aurora, Colorado

2020 -- Healthcare Ethics Consultant, Department of Pediatrics
Children's Hospital Colorado, Aurora, Colorado

Honors, special recognitions, and awards

2015 Emmanuel Shapira Award, Society for Inherited Metabolic Disorders
- Awarded for the best research paper published in Molecular Genetics and Metabolism by a member of the Society for Inherited Metabolic Disorders

Membership in professional organizations

2000 -- National Society of Genetic Counselors
- Member (2010-2012) and Chair (2013-2015), Ethics Advisory Group
- Nominating Committee (2017)
- Director-at-large, Board of Directors (2017-2018)

2002 -- American Board of Genetic Counselors
- Item writer, Certification exam (2010-2012)
- Member, Certification Exam Committee (2013-2015)

2013 -- Society for Inherited Metabolic Disorders

2014 -- American Society for Bioethics + Humanities

2019 -- Society for the Study of Inborn Errors of Metabolism

Major committee and service responsibilities

Local

2011 -- Member (2011-2019), Core faculty (2020 --)
Ethics committee, Children's Hospital of Colorado

2018 -- Member, Ethics, Legal and Social Implications (ELSI) working group
University of Colorado Health Biobank

2020 K to R (KTR) Transition Program
Colorado Clinical and Translational Science Institute, University of Colorado

Regional

2015-2016 (2015) Ethics Advisor, (2016) Ad hoc content expert - Ethics
Newborn Screening Program,
Colorado Department Public Health and Environment

National

2012 Ad hoc committee for the position of the NSGC: Genetic testing of minors
National Society of Genetic Counselors

2013 Ad hoc committee for the position of the NSGC: Incidental Findings
National Society of Genetic Counselors

2014 -- Member, Consent And Disclosure Recommendations (CADRe) committee
The Clinical Genome Resource (ClinGen)

2017 -- Curator, *ALDH7A1* (http://www.LOVD/nl/ALDH7A1)
Global variom shared Leiden Open Variation Database (LOVD)

2018 Nominal group expert, VLCAD Nutritional Management Guidelines
Available at: https://southeastgenetics.org/npg/guidelines_vlcad.phb
Southeast Regional NBS & Genetics Collaborative
Genetic Metabolic Dietitians International

2019 -- Advisory committee member, UPenn Master's in Genetic Counseling Program
University of Pennsylvania (UPenn), Philadelphia, Pennsylvania

2020 -- Member, Practice Based Competencies (PBCs) task force
Accreditation Council for Genetic Counseling

License and board certification

2002 Certified Genetic Counselor (CGC)
American Board of Genetic Counseling
- Recertification: 2012, 2017

2020 Healthcare Ethics Consulting Certification (HEC-C)
American Society for Bioethics + Humanities

Inventions, intellectual property, and patents

"6-oxopipoclic acid quantitation by mass spectrometry." Regents of the University of Colorado
Inventors: Wempe MF, Van Hove JL, **Coughlin CR 2nd**.
- US provisional patent application No. 62/632,379, 19 Feb 2018
- PCT patent app No. PCT/US/19/18570, 19 Feb 2019

Review and referee work

Editorial boards

2018 -- Review Editor, Neurogenetics: A Frontiers in Neurology journal

Grant review committees and study sections

2004-2005 Health Resources and Service Administration (HRSA)
2018 LifeArc Philanthropic Fund, London, United Kingdom
2020-2021 Colorado Clinical and Translational Science Institute, Pilot Program
2021 ZonMw Clinical Fellows (KlinischeFellows) program,
the Netherlands Organization for Health Research and Development

Ad hoc reviewer for journals, professional societies, or scientific meetings

2011 -- Ad hoc Manuscript Reviewer, Journal of Genetic Counseling
2014 -- Ad hoc Manuscript Reviewer, Journal of Human Nutrition and Dietetics
2014 -- Ad hoc Manuscript Reviewer, Annals of Internal Medicine
2014 -- Ad hoc Manuscript Reviewer, Journal of Inherited Metabolic Disease
2014 Ad hoc Book Reviewer, Oxford University Press
2015 -- Ad hoc Manuscript Reviewer, Journal of Pediatric Genetics
2015 -- Ad hoc Manuscript Reviewer, Applied & Translational Genomics
2015 Ad hoc Guideline Reviewer, American College of Medical Genetics & Genomics

2016 -- Ad hoc Manuscript Reviewer, Journal of Pediatric Intensive Care
2016 -- Ad hoc Manuscript Reviewer, BBA – Molecular Basis of Disease
2017 -- Ad hoc Manuscript Reviewer, Journal of Community Genetics
2017 -- Ad hoc Manuscript Reviewer, Journal of Pediatrics
2017 -- Ad hoc Manuscript Reviewer, BMC Medical Genetics
2017 -- Ad hoc Manuscript Reviewer, Journal of Biological Inorganic Chemistry
2018 -- Ad hoc Manuscript Reviewer, Cell Reports
2018 -- Ad hoc Manuscript Reviewer, Frontiers in Neurology
2018 -- Ad hoc Manuscript Reviewer, Journal of Pediatric Neurology
2019 -- Ad hoc Manuscript Reviewer, Scientific Reports
2019 -- Ad hoc Manuscript Reviewer, Orphanet Journal of Rare Diseases
2019 -- Ad hoc Manuscript Reviewer, Molecular Genetics and Metabolism
2019 -- Ad hoc Manuscript Reviewer, Developmental Medicine & Child Neurology
2019 -- Ad hoc Manuscript Reviewer, Bioanalysis
2019 -- Ad hoc Manuscript Reviewer, Therapeutic Advances in Neurological Disorders
2020 -- Ad hoc Manuscript Reviewer, Metabolites
2020 -- Ad hoc Manuscript Reviewer, Pediatric Neurology
2020 -- Ad hoc Manuscript Reviewer, Biochimie
2020 -- Ad hoc Manuscript Reviewer, JIMD Reports
2021 -- Ad hoc Manuscript Reviewer, Frontiers in Genetics

Invited extramural lectures, presentations and visiting professorships

Commercial or pharmaceutical audience, invited lectures

1. *Inborn errors of intermediary metabolism: An overview and prospects for treatment*. PTC Therapeutics. Presented to the R&D team and medical director (2010).
2. *Genetics and genomics: The current landscape of genetic testing.* Recordati Rare Diseases Group. Provided to medical and commercial team (2018).

Local conference, invited lectures

1. *An approach to the dysmorphic neonate: The importance of genetic counseling during the neonatal investigation.* Neonatology grand rounds, Thomas Jefferson University, Philadelphia, Pennsylvania (2005)
2. *Genetics and ethics: From eugenics to reproductive autonomy.* Ethics Elective, Temple University Medical School, Philadelphia, Pennsylvania (2006)
3. *Pyridoxine dependent epilepsy: Improving developmental outcome in a metabolic encephalopathy.* Section of Neurology, University of Colorado (2016)
4. *Pyridoxine dependent epilepsy: A common and treatable metabolic encephalopathy. Section of Neurology*. University of Colorado (2017)

Regional conference, invited lectures

1. *Risk Assessment and Common Genetic Syndrome, Incorporating Genetics into Obstetrical Care*. Christiana Care, Newark, Delaware (2003)
2. *Our Genes; Our Choices.* Adult Education Series, Elkton, Maryland (2003)
3. *How to present topics in genetics that will relate to your students' future.* Delaware Technical and Community College, Georgetown, Delaware (2004)
4. *Clinical Genetics: Introduction to the field of clinical genetics*. Delaware science teacher's continuing education, Dover, Delaware (2004; 2005)

5. *Genetics and ethics: Important partners or strange bedfellows*. Impact of Genetics on Healthcare, Jack F. Owens Campus, Georgetown, Delaware (2005)
6. *Direct to consumer testing: An ethical perspective*. Genetic Counseling Supervisor's meeting, Arcadia University, Glenside, Pennsylvania (2008)
7. *VLCAD deficiency: Pitfalls of newborn screening*. Mountain States Genetics Regional Collaborative. Aurora, Colorado (2011)
8. *Pyridoxine dependent epilepsy: Role of a lysine restricted diet.* Mountain States Genetics Regional Collaborative. Phoenix, Arizona (2013)
9. *Genetic testing: Benefits and burdens.* Children's Colorado Annual Ethics Conference 2015, Aurora, Colorado (2015)
10. *All about that base(pair): Can genetic testing reveal ethical concerns?* Semi-Annual Colorado Genetic Counseling Symposium, Aurora, Colorado (2015)
11. *Newborn screening: From PKU to personal genomics*. The 37th Annual Perinatal Clinical Update, Children's Hospital of Colorado, Aurora, Colorado (2016)
12. *PDE: A treatable epileptic encephalopathy*. Translational Approaches to Epilepsy Research. Skaggs School of Pharmacy and Pharmaceutical Sciences Annual Retreat. Breckenridge, Colorado (2017)
13. *Personal Genetics (what happens when families diagnose themselves/their children).* 10th Annual Pediatric Advanced Practice Conference. University of Colorado College of Nursing, Colorado Rock Mountain NAPNAP, Children's Hospital Colorado, Aurora, CO (2020)
14. *Mandatory vaccinations: A case debated.* 24th Annual Ethics Conference, Children's Hospital Colorado, Aurora, CO (2020).
    ** Meeting cancelled due to the novel coronavirus disease COVID-19*

National conference, invited lectures

1. *VLCAD deficiency: Examining the practicalities and pitfalls of NBS*. Genetics Rounds, Stanford University School of Medicine, Palo Alto, California (2009)
2. *Examining ethical implications of care in lethal conditions.* National Society of Genetic Counselors 31st Annual Education Conference, Boston, Massachusetts (2012)
3. *Triple therapy for pyridoxine dependent epilepsy.* The 39th Annual Meeting of the Society for Inherited Metabolic Disorders, Ponte Vedra Beach, Florida (2016)
4. *Genetic Counselors' Perspective on Constitutional Genomic Testing.* Cancer Genomics Consortium 2016 Summer Meeting. Denver, Colorado (2016)
5. *Exploring ethical, legal, and social implications of prenatal diagnostic exome sequencing.* National Society of Genetic Counselors 35th Annual Education Conference, Seattle, Washington (2016)
6. *Clinical studies in nonketotic hyperglycinemia (NKH).* Rare Disease Day Conference. The Boler-Parseghian Center for Rare and Neglected Diseases at the University of Notre Dame. South Bend, Indiana (2017)
7. *Genetic testing of minors for adult onset conditions: An ethical perspective*. National Society of Genetic Counselors 36th Annual Education Conference. Columbus, Ohio (2017)
8. *Ethical issues in genetic counseling practice: Revising ethical principles in genomic medicine*. Center for Applied Genetics and Genomic Medicine. University of Arizona, Tucson, Arizona (2018)
9. *Life sustaining measures and lethal diagnosis: A model of shared decision making.* Integrated Genetics. Philadelphia, Pennsylvania (2018)

10. *Is low carnitine a problem? To supplement or not to supplement.* The 41st Annual Meeting of the Society for Inherited Metabolic Disorders, Bellevue, Washington (2019)
11. *Ethical issues in era of genomic medicine.* 8th Annual Sanford Imagenetics Genomic Medicine Symposium. Sanford Health. Sioux Falls, South Dakota (2019)
12. *Pyridoxine-dependent epilepsy: A treatable disorder of lysine metabolism.* Genetic Metabolic Dietitians International Conference. Austin, TX (2020)
    ** Meeting cancelled due to the novel coronavirus disease COVID-19*

International conference, invited lectures

1. *Triple therapy and newborn screening for PDE*. The Pyridoxine-Dependent Epilepsy investigator meeting. BC Children's Hospital, Vancouver, British Columbia, Canada (2016)
2. *A novel biomarker for the diagnosis of pyridoxine-dependent epilepsy.* 5th International PDE Workshop. Amsterdam University Medical Center, Amsterdam, The Netherlands (2019)
3. *Ethical issues in genomic consent for high risk patients.* 14th International Congress of Inborn Errors of Metabolism. Sydney, Australia (2021)

Teaching record

Didactic teaching, graduate, and medical school students

2007-2010	Biochemical and Developmental Genetics (Graduate students) Course director and lecturer, Arcadia University
2007-2010	Ethical Issues in Genetic Counseling; (Graduate students) Course director and lecturer, Arcadia University
2007	The Role of Families in Contemporary Bioethics (Graduate students) Lecturer, University of Pennsylvania School of Medicine
2008-2010	Reproduction Module – Ethical Issues in Reproduction (Medical students) Lecturer, University of Pennsylvania School of Medicine
2011 --	Introduction to Clinical Research (Graduate students) Lecturer (2011-2012); Co-director (2013); Director (2014-present) University of Colorado Denver
2012-2013	Embryogenetics (Graduate students) Lecturer, University of Colorado Denver
2013-14, 16, 18	Public Health Genetics (Graduate students) Lecturer, University of Colorado Denver
2013 --	Clinical Cytogenetics and Molecular Genetics (Graduate students) Lecturer, University of Colorado Denver
2014, 16, 18, 20	Human Inborn Errors of Metabolism (Graduate students) Lecturer, University of Colorado Denver
2014-2019	Introduction to Ethics (Graduate students)

Lecturer, University of Texas Genetic Counseling Program at Houston

2015 -- Integrated Clinician's Course (ICC) (Medical students)
Facilitator, University of Colorado School of Medicine
- Caring for Patients with Progressive Illness
- Transition to Residency and Beyond

2019 Ethics and Professional Conduct (Graduate students)
Lecturer, Bay Path University MS Genetic Counseling Program

2019 -- Molecules to Medicine (M2M) (Medical students)
Facilitator, University of Colorado School of Medicine
- Research Ethics

2020 InterProfessional Education & Development (AMC healthcare students)
Facilitator, University of Colorado School of Medicine

2021 Clinical Ethics (Certificate in health humanities and ethics)
Lecturer, Center for Bioethics and Humanities

<u>Didactic teaching (continuing education) and postgraduate education</u>

2012 -- Metabolic University
- *Intoxication disorders* (2012, 2013)
- *Fatty acid oxidation* (2012-13, 2016-21)
- *Metabolic laboratory tests* (2013)
- *Glutaric Aciduria type I* (2013-14)
- *Anabolism* (2015)
- *Urea Cycle Disorders* (2016-2019)
- *Solid organ transplant as therapy for IEM* (2017-2020)

2014 -- *Genetics in the NICU*, Graduate Nurse Phase Class, Section of Neonatology
Children's Hospital of Colorado, Aurora, Colorado

2019 -- *Ethics and genetics*, Leadership Education in Neurodevelopmental Disabilities
University of Colorado Anschutz Medical Center, Aurora, Colorado

2020 -- *Pediatric ethics*, Second Year Fellowship Retreat (pediatric subspecialties)
Children's Hospital of Colorado, Aurora, Colorado

2020 *Amino acid disorders II: Lysine and vitamin B_6 metabolism*
Medical biochemical genetics fellows (available to trainees at 17 US programs)
Clinical Core Seminar Series, University of Colorado Anschutz Medical Campus

2021 *Fatty acid oxidation*, Metabolic dieticians
Sponsored by Ultragenyx pharmaceutical and Metabolic Education (Met-Ed)

2021 *Pyridoxine-dependent epilepsy*, Metabolic dietitians
Sponsored by Genetic Metabolic Dietitians International (GMDI) and available to the GMDI membership

Qualifying examination committees

2014 Thuy-Mi Nguyn, Genetic Counseling M.S. Program (M.S. Candidate)
University of Colorado Denver, Aurora, Colorado

Committees concerned with medical and graduate student education

Graduate students in Genetic Counseling programs – capstone or thesis committee

2007-2008 Carolyn Heuer, Genetic Counseling Program (M.S. Candidate)
Arcadia University, Glenside, Pennsylvania

2011-2012 Katie Golden-Grant, Genetic Counseling M.S. Program (M.S. Candidate)
University of Colorado Denver, Aurora, Colorado

2012-2013 Susana San Roman Rivera, Genetic Counseling M.S. Program (M.S. Candidate)
University of Colorado Denver, Aurora, Colorado

2013-2015 Thuy-Mi Nguyen, Genetic Counseling M.S. Program (M.S. Candidate)
University of Colorado Denver, Aurora, Colorado

2015-2016 Leah Rhodes, Genetic Counseling M.S. Program (M.S. Candidate)
University of Colorado Denver, Aurora, Colorado

2019-2020 Allie Morris, Augustana-Sanford Genetic Counseling Program (M.S. Candidate)
Augustana University, Sioux Falls, South Dakota

2019-2020 Maia Borensztein, MS Human Genetics and Genetic Counseling (M.S. Candidate)
Stanford University, Stanford, California

Junior faculty mentorship

2020 -- Angela Rachubinski, Ph.D., Instructor of Pediatrics
Research area: Research ethics focused on Adults with Down Syndrome

Grants, financial support, active research studies

Grants, pending

R21HD104952 (NICHD) **Coughlin** (PI) 04/01/21-03/31/23

Newborn screening and treatment monitoring for patients with pyridoxine-dependent epilepsy

The overall goal of this study is to establish a dried blood method to measure 6-oxo-pipecoalte in patients affected with pyridoxine-dependent epilepsy. This will be the basis for future newborn screening and treatment efficacy testing.

Percentile: 16.0; Awaiting council's decision on funding

Grants, funded

U54HD061221 (NIH) Gropman (PI) 09/17/19-07/31/24
Urea Cycle Disorders Consortium
The Urea Cycle Disorders Consortium is a part of the National Institutes of Health (NIH) Rare Disease Clinical Research Network (RDCRN), supported through collaboration between the Office of Rare Diseases Research (ORDR), the National Center for Advancing Translational Science (NCATS),the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), and the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK).
Role: Co-investigator

Active research studies

Inherited disorders of lysine metabolism
The overall focus of our research is to understand the role of metabolism in the phenotype of pyridoxine-dependent epilepsy and glutaric aciduria type I. Our work aims to leverage our understanding of lysine biochemistry to improve the diagnosis and treatment of patients with these devastating neurologic disorders.
Support for this work: NCATS/CCTSI, Children's Metabolic Fund, PDE Foundation, PDE Research Fund (https://giving.cu.edu/fund/pyridoxine-dependent-epilepsy-pde-research)

Completed grants and financial support:

R01HD058567 (NICHD) Tuchman (PI) 08/05/08-02/28/17
N-carbamylglutamate in the treatment of hyperammonemia
The overall objective of this project is to determine whether treatment of acute hyperammonemia with N-carbamyl-L-glutamate improves overall trajectory of ammonia.
Role: Co-investigator

UL1R001082 (NCATS) Sokol (PI)
Child Maternal Health Pilot Grant **Coughlin** (pilot grant PI) 01/01/16-06/29/17
Dried blood spot screening for pyridoxine-dependent epilepsy
The overall objective of the study is to establish a newborn screening method for pyridoxine-dependent epilepsy.

2UC4DK063821 (NIDDK) Rewers (PI) 03/01/03-05/01/18
The Environmental Determinants of Diabetes in the Young – Colorado Clinical Center
The primary objectives are to identify infectious agents, dietary factors, environmental exposures that are associated with increased risk of autoimmunity and Type 1 diabetes
Role: Co-investigator (07/01/16-10/31/17)

U54HD061221 (NIH) Batshaw (PI) 08/25/14-07/31/19
Urea Cycle Disorders Consortium
The Urea Cycle Disorders Consortium is a part of the National Institutes of Health (NIH) Rare Disease Clinical Research Network (RDCRN), supported through collaboration between the Office of Rare Diseases Research (ORDR), the National Center for Advancing Translational Science (NCATS),the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)

R01GM120772 (NIGMS) Kwok and Shaikh (PIs) 09/22/16-05/31/20
Next Generation Mapping of Complex Genomic Regions involved in Recurrent Structural Variations
The overall goal of this application is to analyze and map genomic regions containing segmental duplications, which mediate recurrent chromosomal rearrangements.
Role: Co-investigator

Bibliography

Research publications [peer-reviewed]: Google Scholar: h-index = 21, i10-index = 36

1. DeBerardinis RJ, **Coughlin CR 2nd**, Kaplan P. Penicillamine therapy for pediatric cystinuria: experience from a cohort of American children. J Urol. 2008 Dec;180(6):2620-3. PMID: 18951580
2. Kranick SM, Ganesh J, **Coughlin CR 2nd**, Licht DJ. Child neurology: a case illustrating the role of imaging in evaluation of sudden infant death. Neurology. 2009 Sep 15;73(11):e54-6. PMID: 19752447
3. Ficicioglu C, **Coughlin CR 2nd**, Bennett MJ, Yudkoff M. Very long-chain acyl-CoA dehydrogenase deficiency in a patient with normal newborn screening by tandem mass spectrometry. J Pediatr. 2010 Mar;156(3):492-4. PMID: 20056241
4. **Coughlin CR 2nd**, Ficicioglu C. Genotype-phenotype correlations: sudden death in an infant with very-long-chain acyl-CoA dehydrogenase deficiency J Inherit Metab Dis. 2010 Dec;33 Suppl 3:129-131. PMID: 20107901
5. **Coughlin CR 2nd**, Krantz ID, Schmitt ES, Zhang S, Kerr DS, Ganesh J. Somatic mosaicism for PDHA1 mutation in a male with pyruvate dehydrogenase complex deficiency. Mol Genet Metab. 2010 Jul;100(3):296-299. PMID: 20462777
6. Sampson MG, **Coughlin CR 2nd**, Kaplan P, Conlin LK, Meyers KE, Zackai EH, Spinner NB, Copelovitch L. Evidence for a recurrent microdeletion at chromosome 16p11.2 associated with congenital anomalies of the kidney and urinary tract (CAKUT) and Hirschsprung disease. Am J Med Genet A. 2010 Oct;152A(10):2618-2622. PMID: 20799338
7. van Karnebeek CDM, Hartmann H, Jaggumantri S, Bok L, Cheng B, Connolly M, **Coughlin CR 2nd**, Das AM, Gospe S, Jackobs C, van der Lee J, Mercimek-Mahmutoglu S, Meyer U, Struys E, Sinclair G, Van Hove J, Collett JP, Plecko BR, Stockler S. Lysine restricted diet for pyridoxine-dependent epilepsy: First evidence and future trials. Mol Genet Metab. 2012 Nov;107(3):335-344. PMID: 23022070
8. Stence NV, **Coughlin CR 2nd**, Fenton LZ, Thomas JA. Distinctive pattern of restricted diffusion in a neonate with molybdenum cofactor deficiency. Pediatr Radiol. 2013 Jul;43(7):882-5. PIMD: 23250031
9. **Coughlin CR 2nd**, Hyland K, Randall R, Ficicioglu C. Dihydropteridine reductase deficiency and treatment with tetrahydrobiopterin: A case report. JIMD Rep. 2013;10:53-56. PIMD: 23430801
10. Yu HC, Sloan JL, Scharer G, Brebner A, Quintana AM, Achilly NP, Manoli I, **Coughlin CR 2nd**, Geiger EA, Schneck U, Watkins D, Suormala T, Van Hove JLK, Fowler B, Baumgartner MR, Rosenblatt DS, Venditti CP, Shaikh TH. An X-linked Cobalamin Disorder Caused by Mutations in Transcriptional Coregulator *HCFC1*. Am J Hum Genet. 2013 Sep 5;93(3):506-514. PMID: 24011988
11. Baker PR 2nd, Friederich MW, Swanson MA, Shaikh T, Bhattacharya K, Scharer GH, Aicher J, Creadon-Swindell G, Geiger E, Maclean KN, Lee WT, Deshpande C, Freckmann

ML, Shih LY, Wasserstein M, Rasmussen MB, Lund AM, Procopis P, Cameron JM, Robinson BH, Brown GK, Brown RM, Compton AG, Dieckmann CL, Collard R, **Coughlin CR 2nd**, Spector E, Wempe MF, Van Hove JL. Variant non-ketotic hyperglycinemia is caused by mutations in *LIAS*, *BOLA3* and the novel gene *GLRX5*. Brain. 2014 Feb; 137(Pt 2):366-379. PMID:24334290

12. van Karnebeek CDM, Stockler-Ipsiroglu S, Jaggumantri S, Assmann B, Baxter P, Buhas D, Bok LA, Cheng B, **Coughlin CR 2nd**, Das AM, Giezen A, Al-Hertain W, Ho G, Meyer U, Mills P, Plecko B, Struys E, Ueda K, Albersen M, Verhoeven N, Gospe SM Jr, Gallagher RC, Van Hove JKL, Hartmann H. Lysine-restricted diet as adjunct therapy for pyridoxine-dependent epilepsy: The PDE consortium consensus recommendations. JIMD Rep. 2014; 15:1-11. PMID: 24748525
13. Chatfield KC, **Coughlin CR 2nd**, Friederich MW, Gallagher RC, Hesselberth JR, Lovell MA, Ofman R, Swanson MA, Thomas JA, Wanders RJA, Wartchow EP, Van Hove JL. Mitochondrial energy failure in HSD10 disease is due to defective mtDNA transcript processing. Mitochondrion. 2015 Mar; 21:1-10. PMID: 25575635
14. **Coughlin CR 2nd**[*], Scharer GH[*], Friederich M, Geiger EA, Creadon-Swindell G, Yu HC, Collins AE, Vanlander AV, Van Coster R, Powell CA, Swanson MA, Minczuk, Van Hove JLK, Shaikh TH. Mutations in the mitochondrial cysteinyl-tRNA synthase gene, *CARS2*, lead to a severe epileptic encephalopathy and complex movement disorder. J Med Genet. 2015 Aug;52(8):532-40. PMID: 25787132
15. **Coughlin CR 2nd**, van Karnebeek CDM, Al-Hertani W, Shuen AY, Jaggumantri S, Jack RM, Gaughan S, Burns C, Mirsky DM, Gallagher RC,Van Hove JLK. Triple therapy with pyridoxine, arginine supplementation and dietary lysine restriction in pyridoxine dependent epilepsy: Neurodevelopmental outcome. Mol Genet Metab 2015 Sep-Oct; 116(1-2):35-43. PMID: 26026794
16. Ferdinandusse S, Friederich MW, Burlina A, Ruiter JPN, **Coughlin CR 2nd**, Dishop MK, Gallagher RC, Bedoyan JK, Vaz FM, Waterham HR, Gowan K, Chatfield K, Bloom K, Bennett MJ, Elpeleg O, Van Hove JL, Wanders RJ. Clinical and biochemical characterization of four patients with mutations in *ESCH1*. Orphanet J Rare Dis. 2015 Jun 18;10(1)79. PMID: 26081110
17. Swanson MA, **Coughlin CR 2nd**, Scharer GH, Szerlong HJ, Bjoraker KJ, Spector EB, Creadon-Swindell G, Mahieu V, Matthijs G, Hennermann JB, Applegarth DA, Toone JR, Tong S, Williams K, Van Hove JL. Biochemical and Molecular Predictors for Prognosis in Nonketotic Hyperglycinemia. Ann Neurol. 2015 Oct;78(4):606-18. PMID: 26179960
18. Weber DR, **Coughlin CR 2nd**, Brodsky J, Lindstrom K, Ficicioglu C, Kaplan P, Freehauf C, Levine MA. Low bone mineral density is a common finding in patients with Homocystinuria. Mol Genet Metab. 2016 Mar, 117(3):351-354. PMID: 26689745
19. Bjoraker KJ, Swanson MA, **Coughlin CR 2nd**, Christodoulou J, Tan ES, Ferguson M, Dyack S, Ahmad A, Friederich MW, Spector E, Creadon-Swindell G, Hodge MA, Gaughan S, Burns C, Van Hove JLK. Neurodevelopmental Outcome and Treatment Efficacy of Benzoate and Dextromethorphan in Siblings with Attenuated Nonketotic Hyerglycinemia. J Pediatr. 2016 Mar;170:234-239. PMID: 26749113
20. Yu HC[*], **Coughlin CR**[*], Geiger EA, Salvador BJ, Elias ER, Cavanaugh JL, Chatfield KC, Miyamoto SD, Shaikh TH. Discovery of a potentially deleterious variant in *TMEM87B* in a patient with a hemizygous 2q13 microdeletion suggests a recessive condition characterized

by severe congenital heart disease and restrictive cardiomyopathy. Cold Spring Harb Mol Case Stud. 2016 May;2(3) PMID: 27148590

21. van Karnebeek CDM, Tiebout SA, Niermeijer J, Poll-The BT, Ghani A, **Coughlin CR 2nd**, Van Hove JLK, Richter JW, Christen HJ, Gallagher R**,** Hartmann H, Stockler-Ipsiroglu S. Pyridoxine-Dependent Epilepsy: an expanding clinical spectrum. Pediatr Neurol. 2016 Jun;59:6-12. PMID: 26995068
22. **Coughlin CR 2nd**, Swanson MA, Kronquist K, Acquaviva C, Hutchin T, Rodríguez-Pombo P, Väisänen ML, Spector E, Creadon-Swindell G, Brás-Goldberg AM, Rahikkala E, Moilanen J, Mahieu V, Matthijs G, Bravo-Alonso I, Pérez-Cerdá C, Ugarte M, Vinaey-Saban C, Scharer GH, Van Hove JLK. The genetic basis of classical nonketotic hyperglycinemia due to mutations in *GLDC* and *AMT*. Genet Med. 2017 Jan;19(1):104-111. PMID: 27362913
23. Beard L, Wymore E, Fenton L, **Coughlin CR**, Weisfeld-Adams JD. Lethal neonatal hyperammonemia in severe ornithine transcarbamylase (OTC) deficiency compounded by large hepatic portosystemic shunt. J Inherit Metab Dis. 2017 Jan;40(1):159-160. PMID 27832417
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31. **Coughlin CR 2nd**, van Karnebeek CDM, Jaggumantri S, Al-Hertani W, Shuen AY, Jack RM, Gaughan S, Burns C, Gallagher RC, Mirsky DM, Van Hove JLK. Triple therapy (Arginine fortification + Lysine Restricted Diet + Pyridoxine) for pyridoxine dependent epilepsy. Presented at the Garrod Symposium**,** May 2015, Vancouver, British Columbia (Platform)
32. Jaggumantri S, **Coughlin C,** Stockler S, Barb C, Mary C, Van Hove J, van Karnebbek C. 2015 update on triple therapy for pyridoxine-dependent epilepsy. Presented at the 50th Annual Congress of the Canadian Neurologic Sciences Federation, June 2015, Toronto, Ontario (Platform). Canadian Journal of Neurological Sciences. May 2015; 42(S1):S13.
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34. Friederich MW, **Coughlin CR 2nd**, O'Rourke C, Lovell MA, Gowan K, Van Hove LK. Mutations in *NDUFB10* results in isolated complex I deficiency due to incomplete assembly of complex I holoenzyme. Presented at Mitochondrial Medicine 2015, June 2015, Washington DC (Poster). Mitochondrion 2015 Sept; 24:S26.
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36. Faucett A, Rashkin M, Brothers K, **Coughlin C**, Hercher L, Hudgins L, Jamal S, Levy H, Peay H, Roche M, Savage Stosic M, Smith M, Uhlmann W, Wain K, Ormond K. Preliminary Discussions in the Development of Recommendation for Ordering Clinicians with Minimal Genetics Background: intro to the Consent and Disclosure Recommendations (CADRe) committee. Presented at the National Society of Genetic Counselors 34th Annual Education Conference. Oct 2015, Pittsburgh, Pennsylvania (Poster)

37. Nguyen TP, Karimpour-Fard A, **Coughlin CR 2nd**, Chatfield KC. MicroRNAs as a Marker of Cardiovascular Disease in Marfan Syndrome and Marfan-related Disorders. Presented at the National Society of Genetic Counselors 34th Annual Education Conference. Oct 2015, Pittsburgh, Pennsylvania (Poster)
38. Yu HC, **Coughlin CR 2nd**, Geiger EA, Elias ER, Cavanaguh JL, Miyamoto SD, Shaikh TH. Discovery of a deleterious variant in *TMEM87B* in a patient with a hemizygous 2q13 microdeletion, uncovers a recessive condition characterized by severe congenital heart defects. Presented at 65th Annual Meeting of the American Society of Human Genetics. Oct 2015, Baltimore, Maryland (Poster)
39. **Coughlin CR 2nd**, Swanson MA, Kronquist K, Acquaviva C, Hutchin T, Rodríguez-Pombo P, Kure S, Spector E, Creadon-Swindell G, Brás-Goldberg AM, Mahieu V, Matthijs G, Bravo-Alonso I, Pérez-Cerdá C, Ugarte M, Scharer GH, Van Hove JLK. The genotypic spectrum of classic nonketotic hyperglycinemia due to mutations in *GLDC* and *AMT*. Presented at the ACMG Annual Clinical Genetic Meeting. Mar 2016, Tampa, Florida (Platform)
40. **Coughlin CR 2nd**, Swanson MA, Kronquist K, Acquaviva C, Hutchin T, Rodríguez-Pombo P, Kure S, Väisänen ML, Spector E, Creadon-Swindell G, Brás-Goldberg AM, Rahikkala E, Moilanen J, Mahieu V, Matthijs G, Bravo-Alonso I, Pérez-Cerdá C, Ugarte M, Vinaey-Saban C, Scharer GH, Van Hove JLK. The genotypic spectrum of classic nonketotic hyperglycinemia due to mutations in *GLDC* and *AMT*. Presented at the 39th Annual Meeting of the Society for Inherited Metabolic Disorders. Apr 2016, Ponte Vedra Beach, Florida (Platform) Mol Genet Metab 2016 Mar;117:236.
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42. Chen MA, Wortmann SB, Tiller GE, Wevers RA, Botto LD, **Coughlin CR**, Descartes M, Kyriss M, Maranda B, Pitt J, Potente C, Rodenburg R, Srirangan S, van Settend P, Thomas J, Vockley G, Waters PJ, Weber JL, White SM. Phenotypic Variability in UMPS Heterozygotes with Orotic Aciduria. Presented at the 39th Annual Meeting of the Society for Inherited Metabolic Disorders. Apr 2016, Ponte Vedra Beach, Florida (Poster) Mol Genet Metab 2016 Mar;117:250.
43. Ormond K, Hallquist M, Buchanan A, Cho M, Kaufman D, Brothers K, **Coughlin CR 2nd**, Hercher L, Hudgins L, Jamal S, Levy H, Peay H, Roche M, Savage Stosic M, Smith M, Uhlmann W, Wain K, Faucett A. Development of Recommendations for Ordering Clinicians with Minimal Genetics Background from the ClinGen Consortium Consent and Disclosure Recommendations (CARDe) Committee. Presented at the European Meeting on Psychosocial Aspects of Genetics. May 2016, Barcelona, Spain (Platform)
44. Larson A, Freidrich M, **Coughlin CR 2nd**, Van Hove JLK. 9032T>C is a new pathogenic variant in mtDNA resulting in impaired assembly of complex V. Presented at Mitochondrial Medicine 2016, June 2016, Seattle, Washington (Poster)
45. Faucett WA, Hallquist M, Buchanan A, Cho M, Kaufman D, Brothers K, **Coughlin CR 2nd**, Hercher L, Hudgins L, Jamal S, Levy H, Peay H, Roche M, Stosic M, Smith M, Uhlmann, Wain K, Ormond KE. The ClinGen CADRe Rubric: Developing Communication Strategies for Actionable Genes. Presented at the 2016 ClinGen/DECIPHER conference. June 2016, Cambridge, England (Poster)

46. Faucett WA, Hallquist M, Buchanan A, Cho M, Brothers K, **Coughlin CR 2nd**, Hercher L, Hudgins L, Jamal S, Levy H, Peay H, Roche M, Stosic M, Smith M, Uhlmann W, Wain K, Ormond KE. Development of tools to determine communication strategies for actionable gens from ClinGen's CARDe working group. Presented at the National Society of Genetic Counselors 35th Annual Education Conference. Sept-Oct 2016, Seattle, Washington (Poster)
47. Buchanan AH, Faucett WA, Hallquist M, Cho M, Brothers K, **Coughlin CR 2nd**, Hercher L, Hudgins L, Jamal S, Levy H, Peay H, Roche M, Stosic M, Smith M, Uhlmann W, Wain K, Ormond KE. Genetic testing for hereditary cancer predisposition: Can a targeted discussion with a non-genetics clinician provide adequate consent? Presented at the National Society of Genetic Counselors 35th Annual Education Conference. Sept-Oct 2016, Seattle, Washington (Poster)
48. Larson AA, Quintana AM, Yu HC, Geiger EA, Hernandez JA, Stence NV, Kuzniecky RI, Shaikh TH, **Coughlin CR 2nd**, Meeks NJL. Mutations in *RALGAPB* cause seizures, intellectual disability and cortical malformations. Presented at the 66th Annual Meeting of The American Society of Human Genetics, Oct 2016, Vancouver, Canada. (Poster)
49. Hallquist M, Buchanan A, Faucett WA, Cho M, Brothers K, **Coughlin CR 2nd**, Hercher L, Hudgins L, Jamal S, Levy H, Peay H, Roche M, Stosic M, Smith M, Uhlmann W, Wain K, Ormond KE. Determining critical communication issues for the genetic testing process. Presented at the 66th Annual Meeting of The American Society of Human Genetics, Oct 2016, Vancouver, Canada (Poster)
50. Jaggumantri S, **Coughlin CR**, Al-Hertani W, Shuen, A, Jack RM, Burns C, Mirsky D, Gallagher R, van Karnebeek C, Van Hove J. Triple therapy (Arginine fortification + Lysine Restricted Diet + Pyridoxine) for pyridoxine dependent epilepsy. Presented at the 66th Annual Meeting of The American Society of Human Genetics, Oct 2016, Vancouver, Canada. (Poster)
51. Mostovoy Y, Levy-Sakin M, Lam J, Lam ET, Hastie AR, Marks P, Lee J, Chu C, Lin C, Džakula Z, Cao H, Schlebusch SA, Giorda K, Schnall-Levin M, Wall JD, Meeks NJL, Chatfield KC, **Coughlin CR 2nd**, Shaikh TH, Kwok P. A hybrid approach for *de novo* human genome seq uence assembly, phasing, and detection of complex structural variation. Presented at the 66th Annual Meeting of The American Society of Human Genetics, Oct 2016, Vancouver, Canada. (Platform)
52. Reyes N, Yu HC, **Coughlin C 2nd**, Shaikh T, Herenandez J, Quintana A. Mutations in GABRA1 cause multiple congenital anomaly syndrome characterized by a severe seizure phenotype. Presented at the Summer COURI Symposia, Aug 2016, El Paso, Texas (Poster)
53. Reyes N, Yu HC, **Coughlin C 2nd**, Shaikh T, Herenandez J, Quintana A. Mutations in GABRA1 cause multiple congenital anomaly syndrome characterized by a severe seizure phenotype. Presented at the Annual Biomedical Research Conference for Minority Students, Nov 2016, Tampa, Florida (Platform)
54. Reyes-Nava N, Yu HC, **Coughlin CR 2nd**, Shaikh TH, Herenandez JA, Quintana AM. Functional analysis of GABRA1 in zebrafish. Presented at the 3rd Border Biomedical Research Center Symposium, September 2017, El Paso, Texas (Poster)
55. Stence NV, Fenton LZ, Palmer C, Tong S, **Coughlin CR 2nd**, Hennermann J, Wortmann SB, Van Hove JLK. Brain imaging in classic nonketotic hyperglycinemia. Presented at the Annual Meeting of the Society for Inherited Metabolic Disorders, Mar 2018, San Diego, California. (Poster) Mol Genet Metab 2018 Mar;123:266-7.

56. **Coughlin CR 2nd**, Swanson MA, Spector E, Meeks NJ, Kronquist K, Tsai BP, Strom SP, Gao H, Nagy P, Hyland K, van Dooren SJ, Salomons GS, Van Hove JLK. The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy. Presented at the Annual Meeting of the Society for Inherited Metabolic Disorders, Mar 2018, San Diego, California. (Poster) Mol Genet Metab 2018 Mar;123:224.
57. Hallquist MLG, Ormond KE, Tricou EP, Faucett WA, Brothers K, **Coughlin CR 2nd**, Hercher L, Hudgins L, Levy H, Peay H, Roche M, Stosic M, Smith M, Uhlmann W, Wain K, Buchanan AH. Consent for genetic testing and disclosure of results: Shifting the paradigm to non-genetics clinicians. Presented at the Annual American College of Medical Genetics Clinical Genetics Meeting, Apr 2018, Charlotte, North Carolina. (Platform)
58. Ormond KE, Hallquist MLG, Tricou EP, Faucett WA, Brothers K, **Coughlin CR 2nd**, Hercher L, Hudgins L, Levy H, Peay H, Roche M, Stosic M, Smith M, Uhlmann W, Wain K, Buchanan AH. Consent for genetic testing and disclosure of results: Shifting the paradigm to non-genetics clinicians. Presented at the European Meeting on Psychosocial Aspects of Genetics, June 2018, Milan, Italy. (Platform)
59. Reyes-Nava N, Yu HC, **Coughlin CR 2nd**, Shaikh TH, Quintana AM. Knockdown of the zebrafish orthologue of GABRA1 results in absence seizures. Presented at the Society for Developmental Biology 77th Annual Meeting, July 2018, Portland, Oregon. (Poster)
60. Castellanos BS, **Coughlin C 2nd**, Yu HC, Shaikh TH, Quintana AM. Using zebrafish as a model to understand the role of HSPG2 in early craniofacial development. Presented at the Society for Developmental Biology 77th Annual Meeting, July 2018, Portland, Oregon. (Poster)
61. Yilmaz F, Mostovoy Y, Geiger EA, Meeks NJL, Chatfield KC, **Coughlin CR 2nd**, Kwok P, Shaikh TH. A next-generation mapping approach for the assembly and detection of structural variation within genomic regions containing complex segmental duplications. Presented at the Annual Meeting of the American Society of Human Genetics, Oct 2018, San Diego, California. (Poster)
62. Reyes-Nava, Yu HC, **Coughlin CR 2nd**, Shaikh TH, Quintana AM. *Gabra1* disrupts normal brain development in a zebrafish model of hypomotility. Presented at the Southwest Regional Society for Developmental Biology Meeting, March 2019, Denver, Colorado. (Poster)
63. Tseng LA, Gospe SM Jr, Aziz VG, Bok LA, Hartmann H, Kurlemann G, **Coughlin CR 2nd**, van Karnebeek CD, Pyridoxine-dependent epilepsy in adulthood. Presented at Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Sept 2019, Rotterdam, The Netherlands. (Poster)
64. Woontner M, **Coughlin C 2nd**, Goodman S. Novel biomarkers of late-onset ornithine transcarbamylase deficiency. Presented at the 42nd Annual Meeting of the Society for Inherited Metabolic Disorders, Apr 2020, Austin, Texas. (Poster)
    ** Meeting cancelled due to the novel coronavirus disease COVID-19*
65. Ormond K, Borenzstein, M, Buchanan A, Cho M, **Coughlin CR 2nd**, Faucett A, Hallquist M, Peay H, Smith M, Stosic M, Tricou E, Uhlmann W, Wain K. Targeted consent discussions for genetic testing – a consensus study to define relevant elements. Presented at the 5th ELSI Congress, June 2020, New York, New York. (Flash presentation)
    ** Meeting cancelled due to the novel coronavirus disease COVID-19*
66. Ormond KE, Borenzstein M, Buchanan AH, Faucett WA, Hallquist MLG, Peay HL, Smith ME, Tricou EP, Uhlmann WR, Wain K, **Coughlin CR 2nd**. Critical components of informed

consent for genetic testing: Results of a Delphi Consensus process. Presented at the European Human Genetics Conference, June 2020, Berlin, Germany. (Oral presentation).

67. Borensztein M, Hallquist M, **Coughlin CR 2nd**, Buchanan A, Faucett WA, Peay H, Smith ME, Tricou E, Uhlmann W, Wain K, Ormond KE. Defining the critical components of informed consent for genetic testing. Presented at the National Society of Genetic Counselors 39th Annual Education Conference. Nov 2020, Nashville, Tennessee. (Poster)
68. Baker II PR, Ables H, Bedoyan J, Feldman G, Keegan C, Lichter-Konecki U, Longo N, McDonald M, Merideth M, Venditti C, Merritt L, Shinawi M, Sutton R, Vernon H, Wilcox W, El-Gharbawy A, Koeller D, Steiner R, Barshop B, **Coughlin C II**, Koacchar A, Van Hove J, Thomas J, Larson A, McCandless S. Opportunities for fellowship education: The first year of the medical biochemical genetics clinic core seminar series. Presented at the ACMG Annual Clinical Genetics Meeting. April 2021. Virtual conference. (Platform presentation)

Media appearances

1. "Designer babies: The genetic editing experiment." *Denver 7 News,* American Broadcast Company. Originally broadcast on 28 November 2018.
    - Available at: https://www.thedenverchannel.com/news/360/designer-babies-the-genetic-editing-experiment-that-has-caused-recent-controversy
2. "Pyridoxine-dependent epilepsy." The JIMD Podcast. January 2021
    - Peter Clayton, Emma Footitt and **Curtis Coughlin** join us to discuss the new consensus guidelines for PDE-ALDH7A1 disease.
    - Available at: https://onlinelibrary.wiley.com/page/journal/15732665/podcasts and https://soundcloud.com/user-109006120