CURTIS R. COUGHLIN II

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Education

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

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

2010 Master of Bioethics
University of Pennsylvania School of Medicine, Philadelphia, Pennsylvania

2021 Doctor of Philosophy in Clinical Science
University of Colorado Anschutz Medical Campus, Aurora, Colorado
Advisors: Stephen Goodman MD [deceased 2020], Michael Woontner PhD

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 (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

2010-2019 Genetic Counselor, Section of Genetics and Metabolism
Children's Hospital Colorado, Aurora, Colorado

2020 -- Healthcare Ethics Consultant, Ethics Core Faculty
Children's Hospital Colorado, Aurora, Colorado

Honors, special recognitions, and awards

2015 Emmanuel Shapira Award, Society for Inherited Metabolic Disorders
- Annual award for the best paper in the field of Biochemical Genetics and Metabolism published in Molecular Genetics and Metabolism

Membership in professional organizations

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

2002 -- American Board of Genetic Counselors
- Item Writer, Certification Exam (2010-2012)
- Certification Exam Committee (2013-2015)
- Nominating Committee (2022)

2013 -- Society for Inherited Metabolic Disorders
- Annual Meeting Planning Committee (2021-2023)
- Education Committee (2021 --)
- Membership Engagement Committee (2021 --)

2014 -- American Society for Bioethics + Humanities

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

Major committee and service responsibilities

Local (Anschutz Medical Campus)

2011 -- Ethics committee at the Children's Hospital of Colorado
- Annual conference planning committee (2015, 2022)
- Workgroup, Pediatric metabolic and bariatric surgery (2018-2020)
- Workgroup, Crisis standards of care (2020)
- Workgroup, Fetal precision medicine (2021 --)
- Workgroup, Interventions for neonates with Trisomy's 13 and 18 (2021 --)

2018 -- Colorado Center for Personalized Medicine (CCPM)
- Ethics, Legal and Social Implications (ELSI) working group (2018-2021)
- Biobank advisory committee (2021-)
- Biobank Enrollment And Results (BEAR) working group (2021 –)
- Pharmacogenetics Implementation Committee Colorado (PICColo) (2021 --)

2020 K to R (KTR) program, Colorado Clinical and Translational Science Institute

2022 -2023 Co-Chair, Center for Bioethics and Humanities faculty search committee

Regional

2015-2016 Ethics advisor and ad hoc content expert: newborn screening
Colorado Department Public Health and Environment

2021 Program committee and content expert: Ethics and genetics
Co-sponsored by the Center for Bioethics and Humanities and
Denver Museum of Nature and Sciences' Institute of Science & Policy

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)

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

2020-2023 Member, Practice Based Competencies (PBCs) task force
Accreditation Council for Genetic Counseling

International

2017 -- *ALDH7A1* variant curation (genetic cause of pyridoxine-dependent epilepsy)
Leiden Open Variation Database (http://www.LOVD/nl/ALDH7A1)
Human Disease Genes (https://humandiseasegenes.nl)

Scientific Advisory Boards

2021-2024 CHAnging Rare disorders of LysInE (CHARLIE) consortium
Funded by the European Joint Programme of Rare Diseases

License and board certification

2002 Certified Genetic Counselor
American Board of Genetic Counseling
- Recertification: 2012, 2017, 2022

2020 Healthcare Ethics Consulting Certification
American Society for Bioethics + Humanities

Inventions, intellectual property, and patents

"6-oxopipecolic 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 of *Neurogenetics – Frontiers in Neurology*

Grant review committees and study sections

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

Data safety monitoring board (DSMB)

2022 DSMB member, "A randomized, double blind, placebo-controlled parallel study of tolerability and efficacy of Cannabidiol (CBD) on motor symptoms in Parkinson's disease" University of Colorado AMC (COMIRB #17-2318)
2022 -- DSMB member, "A double-blind, placebo-controlled crossover study comparing the analgesic efficacy of cannabis versus oxycodone," University of Colorado AMC (COMIB #14-1909)
2023 -- DSMB member, "Mechanistic investigation of therapies for Down Syndrome regression disorder," University of Colorado AMC (COMIRB #)

Ad hoc reviewer for academic programs, professional societies, or scientific meetings

2014 Book reviewer, National Society of Genetic Counselors; Oxford University Press
2015 Guideline reviewer, American College of Medical Genetics & Genomics
2015 Genetic counseling program, Arcadia University (ethics curriculum)
2021 Abstract reviewer, Society for Inherited Metabolic Disorders Annual Meeting
2023 Genetic counseling program, University of Pennsylvania (ethics curriculum)

Ad hoc reviewer for journals (listed alphabetically)

Annals of Internal Medicine
Applied & Translational Genomics
BBA – Molecular Basis of Disease
Bioanalysis
Biochimie
BMC Medical Ethics
BMC Medical Genetics
Cancer Control
Cell Reports
Clinical Genetics
Developmental Med & Child Neurology
Epilepsy Research
European Journal of Paediatric Neurology
Frontiers in Genetics
Frontiers in Neurology
Frontiers in Nutrition
Genes
International Journal of Neonatal Screening
JIMD Reports
Journal of Biological Inorganic Chemistry
Journal of Community Genetics
Journal of Genetic Counseling
Journal of Human Nutrition and Dietetics
Journal of Inherited Metabolic Disease
Journal of Pediatric Genetics
Journal of Pediatric Intensive Care
Journal of Pediatric Neurology
Journal of Pediatrics
Journal of Personalized Medicine
Metabolites
Metabolic Brain Disease
Molecular Genetics and Metabolism
Molecular Genetics and Metabolism Reports
Orphanet Journal of Rare Diseases
Pediatric Neurology
Scientific Reports
Therapeutic Advances Neurologic Disorders

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. Presented to the 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 the 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)
5. *PDE: A treatable epileptic encephalopathy*. Translational Approaches to Epilepsy Research. Skaggs School of Pharmacy and Pharmaceutical Sciences Annual Retreat. Breckenridge, Colorado (2017)
6. *Involvement in mentorship in professional organizations.* Instructor – Senior instructor lecture series. University of Colorado (2022)
7. *A treatable intellectual disability: Lessons from pyridoxine-dependent epilepsy*. Human Medical Genetics and Genomics Program (HMGGP) Annual Retreat. Colorado Springs, Colorado (2022)

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. *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)
13. **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
14. *The importance of incorporating ethical perspectives in genetics.* Colorado Genetic Counseling Symposium, Aurora, Colorado (2022)

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
13. *Pyridoxine-dependent epilepsy and triple therapy*. Rare Disease Day Grand Rounds. Children's Health of Orange County (CHOC), Orange, CA (2023)
14. *Newborn screening for pyridoxine-dependent epilepsy.* Metabolic Grand Rounds. Boston Children's Hospital, Boston, MA (2023)

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)
4. *Pyridoxine-dependent epilepsy: Towards newborn screening.* Newborn screening symposium. United for Metabolic Diseases. Amsterdam, The Netherlands (2022)
5. *What tools does the clinician need for better future treatment?* The annual CHARLIE consortium meeting. Barcelona, Spain (2022)

Teaching record

Course director: Graduate and medical school students

2007-2010 Biochemical and Developmental Genetics (Graduate students)
Genetic Counseling Program, Arcadia University

2007-2010 Ethical Issues in Genetic Counseling, GC 557 (Graduate students)
Genetic Counseling Program, Arcadia University

2013 -- Introduction to Clinical Research, GENC 6170 (Graduate students)
Genetic Counseling Program, Graduate School, University of Colorado Denver

Lecturer: Graduate and medical school students

2007 The Role of Families in Contemporary Bioethics (Graduate students)
Penn Masters of Bioethics Program, University of Pennsylvania

2008-2010 Reproduction Module – Ethical Issues in Reproduction (Medical students)
School of Medicine, University of Pennsylvania

2011-2012 Introduction to Clinical Research, GENC 6170 (Graduate students)
Genetic Counseling Program, Graduate School, University of Colorado Denver

2012-2013 Embryo genetics, GENC 6125 (Graduate students)
Genetic Counseling Program Graduate School, University of Colorado Denver

2013-2018 Public Health Genetics, EPID 6642 (Graduate students)
School of Public Health, University of Colorado Denver

2013 -- Clinical Cytogenetics and Molecular Genetics, GENC 6120 (Graduate students)
Genetic Counseling Program, Graduate School, University of Colorado Denver

2014 -- Human Inborn Errors of Metabolism, GENC 6140 (Graduate students)
Genetic Counseling Program, Graduate School, University of Colorado Denver

2014-2019 Introduction to Ethics (Graduate students)
Genetic Counseling Program at Houston, University of Texas

2019 Ethics and Professional Conduct, PSY 535 (Graduate students)
Genetic Counseling Program, Bay Path University

2019 Ethical issues and genetic counseling (Graduate students)
Augustana-Sanford Genetic Counseling Program, Augustana University

2021 Clinical Ethics, HEHE 5850 (Certificate in health humanities and ethics students)
Center for Bioethics and Humanities, University of Colorado

Facilitator: Graduate and medical school students

2015 -- Integrated Clinicians Course, IDPT 7001, 7002, 7003 (Medical students)
School of Medicine, University of Colorado
- End of Life
- Shared Decision-Making
- Transition to Residency and Beyond
- Caring for Patients with Progressive Illness

2019 Molecules to Medicine, IPDT 5002 (Medical students)
School of Medicine, University of Colorado
- Research Ethics

2020 InterProfessional Education & Development (IPED) (AMC healthcare students)
Center for Interprofessional Practice and Education, University of Colorado

2022 Health & Society Pillar (Medical students)
School of Medicine, University of Colorado
- Introduction to Research Ethics
- Professionalism Across the Continuum of Physician Identify Formation

2022 Reproductive System and Life Cycle Course (Medical students)
School of Medicine, University of Colorado
- Controversies in Reproductive Health and Conscientious Objection

2022 Operative/Perioperative Care, IPDT 7050 (Medical students)
School of Medicine, University of Colorado
- Informed Consent: Patient-Professional Relationships

2022 OB-GYN Clerkship (Medical students)
School of Medicine, University of Colorado
- Ethical issues in the practice of Obstetrics and Gynecology

Course co-director: Continuing education and postgraduate education

2021 -- Pediatric ethics grand rounds (pediatric subspecialties)
Children's Hospital of Colorado, Aurora, Colorado

Course lecturer: Continuing education and postgraduate education

2012 -- Metabolic University (Dietitians, physicians, advanced practice providers)
- *Intoxication disorders* (2012, 2013)
- *Fatty acid oxidation* (2012-13, 2016-22)

- *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* (pediatric subspecialities and graduate students)
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* (post-doctoral fellows)
Medical Biochemical Genetics (MBG) Clinical Core Seminar Series
Supported by MBG programs across North America, www.simd.org/education

2021 *Pyridoxine-dependent epilepsy is a treatable disorder of lysine metabolism*
Genetic Metabolic Dietitians International (GMDI) webinar education series

2022 *Ethics in clinical practice* (genetic counselors)
Online review course in Medical Genetics and Genetic Counseling
Available at https://blcommunications.us

Graduate and medical student research training

Master's degree students (*primary mentor)

Date	*Student*	*Graduate Program*	*University*
2007-2008	Carolyn Heuer	Genetic Counseling	Arcadia University
2011-2012	*Katie Golden-Grant	Genetic Counseling	University of Colorado
2012-2013	*Susana San Roman Rivera	Genetic Counseling	University of Colorado
2013-2015	Thuy-Mi Nguyen	Genetic Counseling	University of Colorado
2015-2016	Leah Rhodes	Genetic Counseling	University of Colorado
2019-2020	Maia Borensztein	Genetic Counseling	Stanford University
2021-2022	Lauren Walker	Genetic Counseling	Thomas Jefferson

Medical students

Date	*Student*	*Medical School*
2021-2022	Madison Hanson	University Colorado School of Medicine
2023	Marissa George	Rocky Vista University

Grants, financial support, active research studies

Grants, funded

U54HD061221 Gropman, Le Mons, Nagamani (MPI) Total Period: 09/17/19-07/31/24
The Rare Diseases Clinical Research Consortium in Urea Cycle Disorders
Goal: The Urea Cycle Disorders Consortium focuses on the 8 related disorders essential for urea biosynthesis. The primary goal is to improve the outcome of patients with UCD through the development of new therapies and improved clinical management.
Role: Co-investigator (site co-PI)

R21HD104952 (NICHD) **Coughlin** (PI) Total Period: 06/08/22-05/31/24
Newborn screening and treatment monitoring for patients with pyridoxine-dependent epilepsy
Goal: The primary aim 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 evaluation of treatment efficacy.

U54HD061221- supplement Wilkening (PI) Total Period: 07/01/22-6/30/23
Health related quality of life: Status and contributing variables in adults with UCDs
Goal: This is a pilot study with the goal to describe adult patients' perceived ability to fulfill age-appropriate social roles and the clinical and social factors that support these endeavors. The primary aim is to establish outcome measures that are relevant, sensitive, and patient centered.
Role: Co-investigator

Grants, completed

R01HD058567 (NICHD) Tuchman (PI) Total Period: 08/05/08-02/28/17
N-carbamylglutamate in the treatment of hyperammonemia
Goal: 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 (Coughlin effort limited to 09/01/10-02/28/17)

2UC4DK063821 (NIDDK) Rewers (PI) Total Period: 06/01/13-05/31/18
The Environmental Determinants of Diabetes in the Young
Goal: 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 (Coughlin effort limited to 07/01/16-10/31/17)

U54HD061221 (NIH) Batshaw (PI) Total Period: 08/25/14-07/31/19
Urea Cycle Disorders Consortium
Goal: The Urea Cycle Disorders (UCD) Consortium focuses on the 8 related disorders essential for urea biosynthesis. The consortium is an international network of 16 academic centers that provide start-of-the-art care and conduct is innovative clinical research focused on UCDs
Role: Co-investigator

UL1R001082 (NCATS) – Pilot **Coughlin** (PI) Total Period: 01/01/16-06/29/17
Dried blood spot screening for pyridoxine-dependent epilepsy
Goal: The overall objective of the study is to establish a newborn screening method for pyridoxine-dependent epilepsy.

R01GM120772 (NIGMS) Kwok, Shaikh (MPI) Total Period: 09/22/16-05/31/20
Next Generation Mapping of Complex Genomic Regions involved in Recurrent Structural Variations
Goal: To analyze and map genomic regions containing segmental duplications, which mediate recurrent chromosomal rearrangements.
Role: Co-investigator

Active research studies
The International PDE Consortium (www.pdeonline.org)
The PDE consortium is an international collaboration among clinicians, scientists, and families with the primary goal of improving the health outcome of individuals affected by PDE. Among the ongoing research studies is the international registry for patients with pyridoxine-dependent epilepsy (PDE Registry), which is an observational natural history study. I serve as the PDE Consortium co-principal investigator along with Clara van Karnebeek (UMC Amsterdam)

Bibliography Google Scholar: h-index = 29, i10-index = 41
Papers published in peer-reviewed journals (*equal contribution)

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. **Coughlin CR 2nd**, Scharer GH, Shaikh TH. Clinical impact of copy number variation analysis using high-resolution microarray technologies: Advantages, limitations, and concerns. Genome Med. 2012 Oct 30;4(10):80. PMID: 23114084
8. 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

9. 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
10. Van Hove J, **Coughlin C 2nd**, Scharer G. Glycine Encephalopathy. In: Pagon RA, Adam MP, Bird TD, Dolan CR, Fong CT, Stephens K (eds.). *GeneReviews* [Internet] Seattle (WA): University of Washington, Seattle; 1993-2013, 2002 Nov 14 [updated 2013 Jul 11]. PMID: 20301531
11. **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
12. 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
13. 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
14. 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
15. 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
16. **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
17. **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
18. 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

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15. Brodsky J, D'Aco K, **Coughlin C**, Ficicioglu C, Pyeritz R, Levine M. Bone mineral density deficits in patients with homocystinuria. Presented at the 11th European Congress on Osteoporosis and Osteoarthritis and International Liver Congress 2011. Mar 2011, Valencia, Spain (Poster). Osteoporosis International 2011; 22:135-136.
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17. Golden-Grant KL, Scharer G, Walton CS, **Coughlin CR 2nd**. Whole exome sequencing: Are we ready? Presented at the National Society of Genetic Counselors 31st Annual Education Conference. Oct 2012, Boston, Massachusetts (Poster)

18. Scharer G, Creadon-Swindell G, Spector E, **Coughlin C**, Shaikh T. Single exon deletion in PCCA gene in a patient with Propionic Acidemia highlights challenges to clinical whole exome CNV analysis. Presented at the 62nd Annual Meeting of The American Society of Human Genetics. Nov 2012, San Francisco, California (Poster)
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20. Sloan JL, Yu HC, Scharer G, Brebner A, Quintana A, Achilly NP, Manoli I, **Coughlin CR 2nd**, Geiger EA, Schneck U, Watkins D, Van Hove JL, Fowler B, Baumgartner MR, Rosenblat D, Venditti CP, Shaikh TH. Mutations in a transcription regulator cause a novel X-linked cobalamin disorder (*cblX*) with a severe neurological phenotype. Presented at the International Congress of Inborn Errors of Metabolism. Sept 2013, Barcelona, Spain (Platform)
21. Friederich MW, Chatfield KC, **Coughlin CR 2nd**, Thomas JA, Gallagher RC, Lovell MA, Wanders RJA, Wartchow EP, Van Hove JLK. Mitochondrial energy failure in HSD10 disease also known as 2-methyl-3-hydroxybutryryl-CoA dehydrogenase deficiency is secondary to defective mtRNA processing. Presented at the International Congress of Inborn Errors of Metabolism. Sept 2013, Barcelona Spain (Platform)
22. Yu HC, Sloan JL, Scharer G, Brebner A, Quintana A, Achilly NP, Manoli I, **Coughlin CR 2nd**, Geiger EA, Schneck U, Watkins D, Van Hove JL, Fowler B, Baumgartner MR, Rosenblat D, Venditti CP, Shaikh TH. Mutations in *HCFC1* a transcription coregulator causes a novel X-linked cobalamin disorder (*cblX*) with a severe neurological phenotype. Presented at 63rd Annual Meeting of The American Society of Human Genetics. Oct 2013, Boston, Massachusetts (Platform)
23. Shuen AY, **Coughlin CR 2nd**, Lefrancois M, Srour M, Thomas JA, Gallagher RC, Waters PJ, Hyland K, Van Hove JL, Al-Hertani W. Combined triple therapy trial of pyridoxine, lysine restricted diet and arginine supplementation in four patients with pyridoxine-dependent epilepsy. Presented at 37th Annual Meeting of The Society of Inherited Metabolic Diseases, Mar 2014, Asilomar, California (Platform). Molecular Genetics and Metabolism 2014 Feb; 111(3): 234.
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25. Van Hove JL, Swanson MA, **Coughlin CR 2nd**, Bjoraker K, Spector E, Scharer G, Creadon-Swindell G, Mahieu V, Matthijs G, Hennermann J. Outcome in nonketotic hyperglycinemia. Presented at 37th Annual Meeting of The Society of Inherited Metabolic Diseases, Mar 2014, Asilomar, California (Platform). Molecular Genetics and Metabolism 2014; 111(3):238-239.
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27. Van Hove JL, Swanson MA, **Coughlin CR 2nd**, Szerlong H, Bjoraker K, Spector E, Scharer G, Creadon-Swindell G, Suhong T, Mahieu V, Matthijs G, Hennermann J. Outcome in

nonketotic hyperglycinemia. Presented at the Child Neurology Society 43rd Annual Meeting, Oct 2014, Columbus, Ohio (Platform). Annals of Neurology 2014; 76:S178-S179.

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29. Larson A, Stence NV, **Coughlin CR 2nd**, Thomas JA, Gallagher RC, Collins A, Van Hove JKL. Improvement of the pathological findings on magnetic resonance imaging of the brain with the use of ketogenic diet for pyruvate dehydrogenase complex deficiency. Presented at the 38th Annual Meeting of The Society of Inherited Metabolic Diseases, Mar 2015, Salt Lake City, Utah (Poster). Mol Genet Metb 2015 Mar; 114(3):356.
30. Stence N, Fenton L, **Coughlin C**, Van Hove J. Predictable Progression of Restricted Diffusion in Nonketotic Hyperglycinemia. Presented at the American Society of Neuroradiology 53rd Annual Meeting, Apr 2015, Chicago, Illinois (Platform)
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.
35. Scalais E, Weitzel C, De Meirleir L, Martens G, Shaikh T, **Coughlin CR 2nd,** Yu HC, Swanson M, Friederich M, Scharer G, Helbling D, Wendt-Andrea J, Acquaviva-Bourdain C, Van Hove J. Cobalamin X (HCFC1 deficiency) mimicking nonketotic hyperglycinemia (NKH) with increased CSF glycine and methylmalonic acid. Presented at the Society for the Study of Inborn Errors of Metabolism, Sept 2015, Lyon, France (Poster)
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

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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.
41. Woontner M, Goodman S, Schlichting L, Hawkins M, Noll L, Weisfeld-Adams J, **Coughlin C 2nd**. A patient with hyper-beta-alaninemia. Presented at the 39th Annual Meeting of the Society for Inherited Metabolic Disorders. Apr 2016, Ponte Vedra Beach, Florida (Poster) Mol Genet Metab, 2015 Mar 117; 294.
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.
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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. MITOCHONDRION 31, 109-110. (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, Sept 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. Vervorrt L, Demaerel W, Mostovoy Y, Yilmaz F, Paster S, Hestand M, Swillen A, Vergaelen E, Geiger A, **Coughlin CR**, Chow SK, McDonald-McGinn D, Morrow BE, Kwok P, Xiao M, Emmanuel BS, Shaikh TH, Vermeesch J. Optical mapping of 22q11.2 low copy repeats reveals structural hypervariability. Presented at the Annual meeting of the European Human Genetics Conference. Sept 2019, Gothenburg, Sweden. Eur J Hum Genet. 2019 Jun;27:1138.
63. Vervoort L, Demaerel W, Mostovoy Y, Yilmaz F, Paster S, Hestand M, Swillen A, Vergaelen E, Geiger A, **Coughlin CR**, Chow SK, McDonald-McGinn D, Morrow BE, Kwok P, Xiao M, Emmanuel BS, Shaikh TH, Vermeesch J. Optical mapping of 22q11.2 low copy repeats reveals structural hypervariability. Presented at the joint NVHG and BeSHG annual meeting. Oct 2019, Veldhoven, the Netherlands.
64. 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)
65. 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)
66. 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*

67. 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*
68. 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).
69. 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)
70. 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. Mol Genet Metab 2021 Apr;132:S290-S291. (Platform presentation)
71. Tseng LA, Abdenur JE, Anderws A, Aziz VG, Bok LA, Boyer M, Buhas D, Hartmann H, Fooitt EJ, Grønborg S, Janssen MCH, Longo N, Lunsing RJ, Wijburg FA, Gospe SM Jr, **Coughlin CR 2nd**, van Karnebeek CDM. Association between early treatment and neurodevelopmental outcome in 18 families with PDE-ALDH7A1. Presented at the 14th International Congress of Inborn Errors of Metabolism. Nov 2021, Sydney, Australia. (Platform presentation)
72. Micke K, Elfman H, Jones K, **Coughlin C**, Reynolds R, Larson A, McCandless S, Behrendt N, Galan HL, Zaretsky M. Integrating whole exome sequencing into prenatal care: piloting a multidisciplinary approach. Presented at the 42nd Annual Pregnancy Meeting for the Society for Maternal-Fetal Medicine. Jan-Feb 2022, Orlando, Florida.
73. Micke K, Zaretsky M, Elfman H, McCandless S, Larson A, Reynolds R, **Coughlin C**, Jones K. Piloting a multidisciplinary approach for fetal exome sequencing to optimize yield and patient management. Presented at the 26th International Conference on Prenatal Diagnosis and Therapy. June 2022, Montréal, Canada.
74. Khalil Y, Wilson M, Wempe M, **Coughlin C**, Footitt E, Clayton PT, Mills P. Measurement of urinary 6-oxo-pipecolae in a cohort of ALDH7A1 deficient patients. Presented at the Society for the Study of Inborn Errors of Metabolism Annual Symposium. Aug-Sept 2022. Freiburg, Germany. (Poster Presentation)
75. **Coughlin C**, Tseng L, van Karnebeek C. The time has come for newborn screening for pyridoxine-dependent epilepsy. Presented at the Society for the Study of Inborn Errors of Metabolism Annual Symposium. Aug-Sept 2022. Freiburg, Germany. (Oral Presentation)
76. Murali CN, Barber JR, McCarter R, Zhang A, Ali S**, Members of the UCD Consortium**, Burrage L, Nagamani SCS. Quality of life in children and adults with Urea Cycle Disorders. Presented at the 45th annual meeting of the Society Inherited Metabolic Disorders, Mar 2023, Salt Lake City, Utah.

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
    - Available at: https://onlinelibrary.wiley.com/page/journal/15732665/podcasts and https://soundcloud.com/user-109006120