STEPHANIE (Newton) COURY, M.S., C.G.C.

Licensed Genetic Counselor,
Boston Children's Hospital

EDUCATION

University of Pittsburgh, Pittsburgh, PA **2006**
Master of Science (M.S.), Genetic Counseling, Department of Human Genetics
Thesis: "Use of a matching algorithm in the admission process of genetic counseling programs"

Providence College, Providence, RI **2003**
Bachelor of Science (B.S.), Biology, graduated Magna Cum Laude (3.47 GPA)
Inducted into Alpha Epsilon Delta, the national health pre-professional honor society

BOARD CERTIFICATION and LICENSURE

Licensed Massachusetts Genetic Counselor (GC071) **2009- Present**
Board-Certified Genetic Counselor, American Board of Genetic Counseling (12535) **2007- Present**

PROFESSIONAL EXPERIENCE

Adjunct Associate Professor in the MGH Institute of Health Professions **February 2019- June 2022**

Senior Genetic Counselor **November 2013- Present**
Boston Children's Hospital, Division of Genetics and Genomics
Division Chief: Christopher Walsh, M.D., Ph.D.

- Course co-director of the Harvard Medical School Genetics Training Program (HMSGTP) Advanced Human Genetics Course
- Pediatric genetic counselor: counsel patients and families in clinic, coordinate testing, write clinic notes
- Study coordinator for industry-sponsored autism research study: enroll patients, coordinate study visits, maintain study documents and records
- Advanced Fetal Care Center (AFCC): Provide genetic counseling, and coordinate the genetics consultative service for the AFCC
- BEST (Bringing Expertise to Selecting Tests) committee member
- Pediatric Cancer Risk Program team member
- Supervise genetic counseling student clinical rotations and observations, and teach several classes each semester to genetic counseling students
- Provide coverage for in-patient genetics service for 2-4 weeks per year

Genetic Counselor II **January 2011- October 2013**
Boston Children's Hospital, Division of Genetics, Metabolism Program
Director: Gerard Berry, M.D., F.A.C.M.G., Metabolism Program

- Metabolic genetic counselor: counsel patients and families in clinic, coordinate testing, write clinic notes
- Supervise genetic counseling students, and teach several classes each semester to genetic counseling students
- Provide writing and editing assistance for galactosemia research manuscripts, grant applications, and other scientific publications
- Assist with FDA IND applications, and IRB submissions for two compassionate use protocols
- Manage and coordinate research and clinical teams, including the nurse practitioner, resident and fellow trainees; administrative coordinator for the metabolism program

Genetic Counselor **June 2006- December 2010**
Center for Human Genetics, Boston University School of Medicine, Boston, MA
Co-Directors: Aubrey Milunsky, M.D., D.Sc., F.A.C.M.G.; Jeff Milunsky, M.D., F.A.C.M.G.

- General genetic counselor: including pediatric, adult, preconception, and prenatal genetic counseling
- Primary rotation supervisor of second year genetic counseling students in the general genetics clinic
- Teach several classes each semester to genetic counseling students
- Provide counseling for patients regarding education about genetic conditions, inheritance, testing options, syndromes, risk assessment; assess and address individual and/or family psychosocial needs, coordinate sending out testing, write physician letters
- DNA diagnostics laboratory coordinator: review incoming samples for molecular testing, communicate with health care providers regarding testing information, status, and results, and writing of molecular testing reports. Primary coordinator of international samples
- Attend and present at weekly clinical genetics case conference and journal club

Graduate Student Researcher **September 2004-April 2006**
Department of Human Genetics, University of Pittsburgh
Director: Robin Grubs, PhD, CGC, and Elizabeth Gettig, MS, CGC

- Provided teaching assistance for the Co-Directors of the genetic counseling program
- Compiled an endnotes reference library of over 300 primary literature sources on genetic counseling and related issues
- Conducted thesis research on the use of a matching algorithm in the admission process of genetic counseling programs

Research Assistant **May 2003 – August 2004**
Women and Infants Hospital, **Providence, RI**
Director: Dr. Barbara Stonestreet, MD, Department of Pediatrics

- Examine the effects of dexamethasone and ischemia on ovine fetal blood brain barrier development
- Responsible for laboratory bench work, teaching students in the lab, performing statistical analysis, and helping prepare grants and papers for publication
- Assist in performing operations on pregnant ewes and their fetuses

TEACHING AND EDUCATION

Teaching for Boston University
Metabolism curriculum advisor, 2011-2016

- "Third time's a charm (i.e. How many geneticists does it take to make a diagnosis?)" GMS GC 606, 09/12/2018
- "When you go phishing…Expect the unexpected", GMS GC 606, 12/07/16
- "Biochemical and Metabolic Disorders", GMS GC605, 11/14/12, 11/20/13, 11/18/14, 12/01/15
- "Glycogen Storage Disorders", GMS GC712, 04/22/15
- "Carbohydrate disorders", GMS GC712, 04/22/15
- "Introduction to Inborn Errors of Metabolism (IEM) & Newborn Screening (NBS)", GMS GC712, 03/04/11, 03/04/13
- "Zebras, horses, and genetics", GMS GC 607, 01/29/14
- "Applications of Counseling Techniques for Genetic Counselors", GMS MH703, 11/15/09
- Special situations class, GMS GC711, 12/17/09,12/09/10
- Religion and Genetic Counseling, GMS GC711, 05/05/11, 04/15/2010
- "Genetic Counseling: Clinical Perspective", GMS MS 406S/506S, 06/26/2007
- "What IS Genetic Counseling, anyway? (and how it relates to your Dental practice!!)" SDM MB511, 11/30/09, 11/15/10

Additional Teaching

- "Introduction to clinical genetics", Northeastern University Physician Assistant Program, Principles of Medicine 3, 07/09/2018
- "Inheritance patterns and principles", Harvard Medical School Genetics Training Program (HMSGTP) Advanced Human Genetics Course, 08/23/2017
- "Inheritance patterns and principles", Harvard Medical School Genetics Training Program (HMSGTP) Advanced Human Genetics Course, 02/01/2017
- "Metabolic case discussion", Brandeis University, BIOL 204b, 03/20/13, 3/18/15, 3/11/16
- "Think like a geneticist", Harvard Medical School Genetics Training Program (HMSGTP) Advanced Human Genetics Course, 08/26/2015
- "Pedigree Analysis", Harvard Medical School Genetics Training Program (HMSGTP) Advanced Human Genetics Course, 08/27/2014
- "Genetic counseling in a brave new world; The role of a GC in personalized and Direct-to-Consumer genetic testing", Providence College, HON480, 10/17/2011

Genetic Counseling Masters Capstone Advisor- Boston University

- Amanda Harding- Practical aspects and implications of variant interpretation in the clinical setting: What is being done and what is being communicated to patients? (2017)
- Alex Ing- Examining Current Practices of Genetic Counselors Regarding Multi-Ethnic Couples and the Future of Carrier Screening (2015)
- Elizabeth Smith- An Exploration of the Association between Anxiety and/or Depression and Relationship Attachment in Individuals Affected with Galactosemia (2014)
- Erin Hoffman- Case Identification of Under-Diagnosed Monogenic Disease within a Large Clinical Database (2011)
- Mari Morse- A Comparison of Genetic Counseling Processes for Fetal Corpus Callosal Anomalies Among Prenatal Service Locations (2010)
- Molly McGinniss- Duchenne and Becker Muscular Dystrophy: Parental Knowledge and Perspectives on Genetic Counseling (2009)

Genetic Counseling Masters Thesis Advisor- Brandeis University

- Danielle Hayes- Now What Happens? An Exploration of the Genetic Counselor's Role in Transition Planning (2016)
- Chelsea Thompson- Hidden on the X: Psychosocial Implications of Ornithine Transcarbamylase Deficiency in Female Carriers (2016)

Genetic counseling fieldwork supervisor, first and second year students **2006 – Present**

- Boston University and Brandeis University students

GENETICS EDUCATION OUTREACH

- Genetic Counseling Career Seminar at UMass Lowell, presenter, Lowell, MA, 09/24/2018
- Panelist, "Genetic counseling tools and best practices", Precision Medicine Summit, Boston Children's Hospital, 09/20/2017
- "Why I think Genetic Counseling is awesome, and you should too!"- Lecture on Genetic Counseling, given at the Boston Leadership Institute. Part of an introductory course on DNA/genetics for middle and high school students, 07/05/2017 and 07/11/2018, at Gann Academy, Waltham, MA.
- "All in the family tree"- Lecture on genetics evaluation and pedigree analysis, given to the Reproductive Endocrinology faculty and staff at Brigham and Women's Hospital, 10/24/14
- Invited blog post for New England Regional Genetics Group: "Genetics on a global scale: it's a small world after all!" 11/2013, http://nergg.org/blog/
- Authored a Trait Profile for "Classic Galactosemia" 04/2013, https://www.my46.org/learning-center/traits

- "Metabolic laboratory studies." Presentation given to phlebotomy staff at Boston Children's Hospital, 12/07/2012
- "Genotype-Phenotype Correlations In Galactosemia" Presentation at the Galactosemia Foundation Conference, Dallas, TX, 07/2012
- Host monthly informative lunches for prospective genetic counseling students to learn about the career, at Boston Children's Hospital, 2011- present

PUBLICATIONS

- Okur V, Cho MT, van Wijk R, van Oirschot B, Picker J, Coury SA, Grange D, Manwaring L, Krantz I, Muraresku CC, Hulick PJ, May H, Pierce E, Place E, Bujakowska K, Telegrafi A, Douglas G, Monaghan KG, Begtrup A, Wilson A, Retterer K, Anyane-Yeboa K, Chung WK. De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. Eur J Hum Genet. 2019 Feb 18.
- Zhong C, Song H, Weiss A, Tan WH, Coury S, Huang J. Myofibromatosis presenting as reticulated vascular changes and subcutaneous atrophy in patient with somatic mosaicism of *PDGFRB* mutation. Br J Dermatol. Br J Dermatol. 2018 Jul 12.
- Coury SA, Schneider KA, Schienda J, Tan WH. Recognizing and Managing Children with a Pediatric Cancer Predisposition Syndrome: A Guide for the Pediatrician. Pediatr Ann. 2018 May 1;47(5):e204-e216.
- Lee BH, Reijnders MRF, Abubakare O, Tuttle E, Lape B, Minks KQ, Stodgell C, Bennetto L, Kwon J, Fong C-T, Gripp KW, Marsh ED, Smith WE, Huq AM, Coury SA, Tan W-H, Solis O, Mehta RI, Leventer RJ, Baralle D, Hunt D, Paciorkowski AR. Expanding the neurodevelopmental phenotype of *PURA* syndrome. Am J Med Genet A. 2018 Jan;176(1):56-67.
- Al-Maawali A, Barry BJ, Rajab A, El-Quessny M, Seman A, Coury SN, Barkovich AJ, Yang E, Walsh CA, Mochida GH, Stoler JM. Novel loss-of-function variants in DIAPH1 associated with syndromic microcephaly, blindness, and early onset seizures. Am J Med Genet A. 2016 Feb;170A(2):435-40.
- Torres A, Newton SA, Crompton B, Borzutzky A, Neufeld EJ, Notarangelo L, Berry GT. CSF 5-methyltetrahydrofolate serial monitoring to guide treatment of congenital folate malabsorption due to proton-coupled folate transporter (PCFT) deficiency. JIMD Rep. 2015 May 26. [Epub ahead of print]
- Joshi M, Eagan J, Desai NK, Newton SA, Towne MC, Marinakis NS, Esteves KM, De Ferranti S, Bennett MJ, McIntyre A, Beggs AH, Berry GT, Agrawal PB. A compound heterozygous mutation in GPD1 causes hepatomegaly, steatohepatitis and hypertriglyceridemia. Eur J Hum Genet. 2014 Oct;22(10):1229-32.
- Jamuar SS, Newton SA, Prabhu SP, Hecht L, Costas KC, Wessel AE, Harris DJ, Anselm I, Berry GT. Rhabdomyolysis, acute renal failure, and cardiac arrest secondary to status dystonicus in a child with glutaric aciduria type I. Mol Genet Metab. 2012 Aug;106(4):488-90.
- Kim CR, Sadowska GB, Newton SA, Merino M, Petersson KH, Padbury JF, Stonestreet BS. Na^+,K^+-ATPase Activity and Subunit Protein Expression: Ontogeny and Effects of Exogenous and Endogenous Steroids on the Cerebral Cortex and Renal Cortex of Sheep. Reprod Sci. 2011 Apr;18(4):359-73.
- Jamal SM, Basran RK, Newton S, Wang Z, Milunsky JM. Novel de novo PCDH19 Mutations in Three Unrelated Females with Epilepsy Female Restricted Mental Retardation Syndrome. Am J Med Genet A. 2010 Oct;152A(10):2475-81.
- Huang XL, Zou YS, Maher TA, Newton S, Milunsky JM. A de novo balanced translocation breakpoint truncating the autism susceptibility candidate 2 (AUTS2) gene in a patient with autism. Am J Med Genet A. 2010 Aug;152A(8):2112-4.
- Zou YS, Newton S, Milunsky JM. A Complex Maternal Rearrangement Results in a Pure 10.8 Mb Duplication of the 5q13.1-q14.1 Region in an Affected Son. Am J Med Genet Part A. 2010 Feb;152A(2): 498-503.
- Zhang C, Milunsky J, Newton S, Ko J, Zhao G, Maher T, Tager-Flusberg H, Bolliger M, Carter A, Boucard A, Powell C, Sudhof T. A Neuroligin-4 Missense Mutation Associated With Autism Impairs Neuroligin-4 Folding and ER Export. J Neurosci. 2009 Sep 2;29(35):10843-54.

- Zou YS, Huang XL, Ito M, Newton S, Milunsky JM. 2008 Further delineation of the critical region for the 9p-duplication syndrome. Am J Med Genet A. 2009 Feb;149A(2):272-6.
- Sciamanna CN, Clark MA, Diaz JA, Newton S. Filling the gaps in physician communication. The role of the Internet among primary care patients. *Int J Med Inform.* 2003 72(1-3):1-8

PRESENTED ABSTRACTS

International meetings

- Milanese C, Vandervore L, Fry A, Bahi-Buisson N, Keren B, Nava C, Afenjar A, Renaldo F, Coury S, Tan W, Fornerod M, Mastroberardino P, Mancini GMS. Mutations in the thioredoxin related gene TMX2 cause primary microcephaly, polymicrogyria and severe neurodegeneration with impaired mitochondrial energy metabolism. Oral presentation at the European Society of Human Genetics (ESHG) meeting, Milan, Italy, June, 2018.
- Newton SA, Agrawal P, Desai N, deFerranti SD, Joshi M, Eagan J, Rohr F, Connolly M, Bennett MJ, Berry GT. An ultra-rare defect due to glycerol-3-phosphate dehydrogenase deficiency redirects hepatic lipid metabolism causing hypertriglyceridemia and massive hepatic steatosis. Platform presentation at the International Congress of Inborn Errors of Metabolism (ICIEM), Barcelona, Spain, 2013.
- Newton SA, Poduri A, Bergin AM, Prabhu SP, Chopra S, Sahin M, Picker J, Kothare S, Berry GT. Myo-inositol treatment reduces seizures and improves clinical outcome in a new patient with the ultrarare phospholipase C beta 1 deficiency. Platform presentation at the Society for the Study of Inborn Errors of Metabolism (SSIEM) Annual Symposium, Geneva, Switzerland, 2011.

National meetings

- Heterozygous UBA2 variants are associated with variable aplasia cutis congenita, limb, genital, growth and developmental abnormalities, features of 19q13.11 microdeletions. Schnur R, Scarano M, Douglas G, Henderson L, Monaghan K, Wentzensen I, Tan WH, Coury S, Chung W, Juusola J. Poster presented at American College of Medical Genetics (ACMG) Annual Meeting, 2017.
- White A, Zetzsche L, Pino GB, Studinski A, Melver CV, Grunseich C, Neilan E, Coury SA, Shinawi M, Fassi E, MacNamara E, Wolf L, Lam C, Hertecant JL, MD, Oglesbee D, Gavrilov D, Raymond K, Tortorelli S, Rinaldo P, Matern D. An unexpected reversal of roles: The increasing need for biochemical genetics testing to validate large scale genomic testing. Platform presentation at the American College of Medical Genetics (ACMG) Annual Meeting, 2016.
- Barry BJ, Al-Maawali A, Rajab A, El-Quessny M, Seman A, Newton Coury S, Barkovich AJ, Yang E, Walsh CA, Mochida G, Stoler JM. Opening Pandora's Box: Finding Answers Amongst Incidentals and Refining the Phenotype of *DIAPH1* Mutations. Poster presented at the American College of Medical Genetics (ACMG) Annual Meeting, 2015.
- Smith E, Whalen-Campion MA, Lichten L, Newton S. An Exploration of the Association between Anxiety and/or Depression and Relationship Attachment in Individuals Affected with Galactosemia. Poster presented at the Southeast Newborn Screening & Genetics Regional Collaborative/ Southeastern Regional Genetics Group (SERC/SERGG) Annual Meeting, 2014.
- Berry GT, Newton SA, Bennett MJ. SCHAD deficiency with lethal hepatic phenotype may only be diagnosed through enzyme analysis. Poster presented at the International Congress of Inborn Errors of Metabolism, 2013.
- Abbott MA, Berry GT, Garganta C, Newton SA, Spence S, and Sullivan J. Phenotypic variability in brothers with HSD10 disease. Presented at the American College of Medical Genetics (ACMG) Annual Meeting, 2013.
- Joshi M, Eagan J, Newton SA, Connolly M, Desai NK, DeFerranti S, Berry GT, Agrawal PB. Severe Hypertriglyceridemia, Fatty Liver and Massive Hepatomegaly in a Patient is Caused by Glycerol-3-Phosphate Dehydrogenase Deficiency. Poster presented at Pediatric Academic Societies (PAS) Annual Meeting, 2013.

- Newton SA, Agrawal PB, Desai NK, deFerranti SD, Connolly M, Berry GT. The power of whole exome sequencing: Diagnosis of an ultra-rare glycolytic defect due to glycerol-3-phosphate dehydrogenase deficiency causing hepatomegaly with NASH. Presentation at the New England Regional Genetics Group (NERGG) Annual Meeting, 2012.
- Newton SA, Jamuar SS, Hecht L, Harris D, Anselm I, and Berry GT. Acute rhabdomyolysis following status dystonicus, renal failure, and cardiopulmonary arrest in a patient with glutaric aciduria type 1. Poster presented at the Society for Inherited Metabolic Disorders (SIMD) Annual Meeting, 2012.
- Newton SA, Poduri A, Neilan E,Bergin AM, Prabhu SP, Chopra S, Sahin M, Picker J, Kothare S, Berry GT. Ultrarare deficiency of PLCB1 gene: Second case report demonstrating a new molecular etiology for malignant migrating partial epilepsy of infancy (MMPEI). Presentation at the New England Regional Genetics Group (NERGG) Annual Meeting, 2011.
- Toledo DM, Newton SA, Milunsky JM. Familial cerebral and thoracic aortic aneurysms. Presentation at the New England Regional Genetics Group (NERGG) Annual Meeting, 2011.
- Newton S, Maher TA, Wang Z, Basran RK, Milunsky JM. Custom sequencing yields a molecular diagnosis for salla disease allowing informative genetic counseling. Presentation at the New England Regional Genetics Group (NERGG) Annual Meeting, 2010.
- Jamal SM, Basran RK, Newton S, Wang Z, Milunsky JM. Novel PCDH19 mutations causing EFMR. Poster presented at American Society of Human Genetics (ASHG) Annual Meeting, 2010.
- Morse M, Newton S, Dobson L. A Comparison of Genetic Counseling Processes for Fetal Corpus Callosal Anomalies Among Prenatal Service Locations. Poster presented at National Society of Genetic Counselors (NSGC) Annual Meeting, 2010. Recipient of Best Poster award.
- Newton S, Milunsky JM, Basran RK. When two is better than one: The diagnostic utility of testing multiple tissue types to make a molecular diagnosis. Poster presented at American College of Medical Genetics (ACMG) Annual Meeting, 2010.
- Newton S, Milunsky JM. Isolated skeletal findings in Marfan syndrome and the R2726W FBN1 mutation. Poster presented at National Society of Genetic Counselors (NSGC) Annual Meeting, 2009.
- McGinniss M, DeChene ET, Newton S, Milunsky J. Duchenne and Becker Muscular Dystrophy: Parental Knowledge and Perspectives on Genetic Counseling. Platform presentation at National Society of Genetic Counselors (NSGC) Annual Meeting, 2009.
- Newton S, Zou Y, Wang Z, Milunsky JM. Molecular analysis provides diagnostic resolution in a family with Neurofibromatosis-Noonan syndrome. Presentation at the New England Regional Genetics Group (NERGG) Annual Meeting, 2008.
- Newton S, Zhao G, Tager-Flusberg H, Milunsky JM. Autism and germline mosaicism for a NLGN4 alteration: Genetic Counseling implications. Poster presented at the American Society of Human Genetics (ASHG) Annual Meeting, 2008.
- Huang X, Zou YS, Maher TA, Newton S, Milunsky JM. A New de novo Balanced Translocation Breakpoint Truncating the Autism Susceptibility Candidate 2 (AUTS2) Gene in an Autistic Patient. Poster presented at the American Society of Human Genetics (ASHG) Annual Meeting, 2008.
- Newton SA, Ito M, Huang XL, Milunsky JM. Whole genome 500K SNP microarray delineates duplication/deletion of 8p in a child with MR/MCA. Poster presented at American Society of Human Genetics (ASHG) Annual Meeting, 2007.
- Newton SA, Gettig E, Grubs RE, Feingold E, and Wilson JW. Use of a matching algorithm in the admission process of genetic counseling programs. Poster presented at National Society of Genetic Counselors (NSGC) Annual Meeting, 2006.

PROFESSIONAL ACTIVITIES

- National Society of Genetic Counselors, Full Member, 2005-present
- New England Regional Genetics Group, member.
- New England Genetics Collaborative: Advisory Committee, Transition Work Group, Quality Improvement learning collaborative work group member, 2011-2017

- Advisory Board Member for the Massachusetts General Hospital (MGH) Institute of Health Professions (IHP) Genetic Counseling Program, 2017-present
- Executive Committee Member, Autism Spectrum Center at Boston Children's Hospital, 2017-present
- Telegenetics training program, hosted by Western States Genetic Services Collaborative and Southwest Telehealth Resource Center. Tucson, AZ. April, 2017.

COMMUNITY SERVICE

- Dear Jack Foundation, February 2019
- Cuddler Program, Boston Children's Hospital, November 2014- March 2017
- Mix104.1 Radiothon to benefit Boston Children's Hospital, 2013, 2014, 2015, 2016, 2017
- NSTAR Walk for Boston Children's Hospital, Miracle Maker (fundraise >$1,000), 2011-2015
- Old South Ringers handbell choir member, Old South Church, Boston, MA. 2008- 2017