# Ann R. Seman, MS, CGC

## Work Experience

July 2020-present

**Director of Clinical Education, MS Genetic Counseling Program**
**Assistant Professor, College of Health & Rehabilitation Sciences**
**MGH Institute of Health Professions, Boston, MA**

- Coordinate, monitor, and evaluate students' clinical and non-clinical fieldwork experiences
- Develop annual plans for assuring an appropriate variety and number of cases for each student
- Cultivate and maintain relationships with fieldwork placement sites
- Oversee and evaluate effectiveness of each supervisor
- Orient new clinical supervisors to their responsibilities
- Monitor accreditation standards to ensure program compliance in terms of fieldwork requirements
- Plan and implement clinical supervisor CEU training experiences including annual conference
- Design, provide instruction, and evaluate student performance for Fieldwork and Embryology/Teratology courses
- Coordinate, monitor, and modify standardized patient sessions
- Participate in student recruitment and admissions process
- Faculty advisor to five students per cohort
- Advise Capstone Committees
- Equity advocate for faculty appointment search committees
- Child Development Day committee member

Sept 2007-June 2020

**Senior Genetic Counselor**
**Division of Genetics and Genomics, Boston Children's Hospital, Boston, MA**

- Provide genetic counseling services in general pediatrics clinic in-person and via telemedicine
- Conduct whole exome sequencing consents
- Triage genetics clinic inquiries on an on-call basis
- Develop and implement triage workflow procedure for patients referred for connective tissue disorder evaluation
- Dictate clinic notes via Dragon voice recognition software
- Write letters of medical necessity
- Supervise genetic counseling students in clinic
- Meet with potential genetic counseling students to provide career advisement and information about the field

Sept 2007-June 2017

**Laboratory Genetic Counselor**
**DNA Diagnostic Laboratory, Boston Children's Hospital, Boston, MA/Claritas Genomics, Cambridge, MA**

- Classify variants per ACMG guidelines as identified by various testing platforms by utilizing published scientific literature and genetics databases
- Write interpretations and assemble reports for exome sequencing, NGS, SNP chromosomal microarray analysis, Sanger sequencing, and MLPA

- Act as liaison between laboratory personnel and clinicians by providing education regarding appropriate test ordering and information about the tests offered
- Collaborate with product development and marketing team
- Develop and implement workflow job aids, clinical utility documents, report templates, and educational materials
- Assist marketing team at educational conferences

Aug 2005-Aug 2007 **Genetic Counselor**
**Genetics Program, Specialty Pediatrics, Eastern Maine Medical Center, Bangor, ME**

- Provide genetic counseling services in adult, pediatrics, and preconception clinics
- Participate in multidisciplinary clinics including Metabolic, Muscular Dystrophy, Cleft Lip and Palate, and Hemophilia
- Co-lead telemedicine services to patients in rural areas of Maine
- Provide consult service for NICU, PICU, and pediatrics floors
- Active member of State Newborn Screening Joint Advisory Committee
- Recipient of Splash Award for excellent teamwork (co-worker nominated)
- Genetics Program representative at Maine's Biggest Baby Shower

## Certification

2009-present **Commonwealth of Massachusetts Board of Registration of Genetic Counselors (GC036)**
2007-present **American Board of Genetic Counseling**

## Professional Memberships

July 2019-present Program Review Committee member, Accreditation Council for Genetic Counseling
Dec 2011-Dec 2020 American College of Medical Genetics and Genomics
Jan 2004-Dec 2010 National Society of Genetic Counselors
May 1999-present Philanthropic Education Organization (P.E.O.)

- Served on Massachusetts State Board from April 2013 to April 2017
- Convention co-chair for Massachusetts State Convention April 2020
- Served as president of local chapter and member of state and local committees

## Education

Sept 2003-Sept 2005 **M.S. Medical Genetics**
University of Cincinnati and Cincinnati Children's Hospital Medical Center, Cincinnati, OH

Sept 1999-May 2003 **B.S. Molecular, Cellular, and Developmental Biology**
University of New Hampshire, Durham, NH

- Summa Cum Laude Graduate
- Honors Biology Graduate
- Phi Beta Kappa

## Teaching Experience

June 2019-June 2020 **Term Lecturer/Adjunct Faculty**
MGH Institute of Health Professions, Boston, MA

- Design team-based learning Embryology/Teratology course

- Develop readiness assessment tests and cumulative assessment test
- Participate in the University of Cincinnati online embryology and teratology courses
- Review and assess students' pediatric/adult standardized patient sessions
- Provide guest lecture regarding chromosomal microarray analysis and MLPA for Lab Methods course

July 2018 **Guest Lecturer**
*Genetic Testing and Adult Clinical Genetics,* Northeastern University Physician Assistant Program, Boston, MA
- Prepare PowerPoint presentation
- Develop examination questions

Summer 2012 **Volunteer Teacher**
Camp Harbor View, Boston, MA
- Designed and implemented activity to teach importance of family history
- Promoted field of genetic counseling

Oct 2008-March 2020 **Guest Facilitator**
Clinical Genetics Course, Genetic Counseling Program, Brandeis University, Waltham, MA
- Facilitate and lead clinical case study regarding a craniosynostosis diagnosis
- Share genetic counseling field experiences
- Provide lecture about copy number variant and sequence variant analysis

Sept 2004-June 2005 **Teaching Assistant**
Introduction to Genetic Counseling, University of Cincinnati, Cincinnati, OH

Jan 2004-March 2004 **Teaching Assistant**
Honors Biology, University of Cincinnati, Cincinnati, OH

## Research Experience

Sept 2021-present **Committee Member, MGH Institute of Health Professions' Genetic Counseling Student's Capstone Project, Boston, MA**
*A qualitative investigation of Ehlers-Danlos syndrome genetics triage*

Sept 2020-April 2022 **Advisor, MGH Institute of Health Professions' Genetic Counseling Student's Capstone Project, Boston, MA**
*The Utilization and Impact of Clinical Dysmorphology on Racially Diverse Patients: A Scoping Review*
**Committee Member, MGH Institute of Health Professions' Genetic Counseling Student's Capstone Project, Boston, MA**
*Information needs of parents of children with surfactant dysfunction disorders: What parents know about genetics and how they communicate it to their children*

Sept 2011-April 2012 **Project Reader, Boston University Genetic Counseling Student's Thesis**
*Genetic Counselors' Laboratory Experiences: Perspectives from Recent Graduates*

Oct 2008-May 2009 **Advisor, Brandeis University Genetic Counseling Student's Thesis**

*Challenges in Counseling for Rare Chromosome Conditions: Genetic Counselors' Perspective*

Jan 2004-June 2005 **Primary Investigator, Master's Thesis**
University of Cincinnati, Cincinnati, OH

- *Perinatal Hospice: An Important Option for Families Continuing Pregnancies with Lethal Fetal Abnormalities*
- The purpose of this research study was to perform a preliminary assessment of the perinatal hospice in the Cincinnati area with a focus on grieving and functioning of the families after their loss
- Submitted for publication to *American Journal of Obstetrics and Gynecology*

June 2004 Myriad Genetic Laboratories General Genetics Research Grant

## Publications

Dec 2020 Duncan AR, Vitobello A, Collins SC, Vancollie VE, Lelliott CJ, Rodan L, Shi J, **Seman AR**, Agolini E, Novelli A, Prontera P, Guillen Sacoto MJ, Santiago-Sim T, Trimouille A, Goizet C, Nizon M, Bruel AL, Philippe C, Grant PE, Wojcik MH, Stoler J, Genetti CA, van Dooren MF, Maas SM, Alders M, Faivre L, Sorlin A, Yoon G, Yalcin B, Agrawal PB. Heterozygous variants in KDM4B lead to global developmental delay and neuroanatomical defects. *Am J Hum Genet*. 107(6):1170-1177.

Oct 2020 Drivas TG, Li D, Nair D, Alaimo JT, Alders M, Altmüller J, Barakat TS, Bebin EM, Bertsch NL, Blackburn PR, Blesson A, Bouman AM, Brockmann K, Brunelle P, Burmeister M, Cooper GM, Denecke J, Dieux-Coëslier A, Dubbs H, Ferrer A, Gal D, Bartik LE, Gunderson LB, Hasadsri L, Jain M, Karimov C, Keena B, Klee EW, Kloth K, Lace B, Macchiaiolo M, Marcadier JL, Milunsky JM, Napier MP, Ortiz-Gonzalez XR, Pichurin PN, Pinner J, Powis Z, Prasad C, Radio FC, Rasmussen KJ, Renaud DL, Rush ET, Saunders C, Selcen D, **Seman AR**, Shinde DN, Smith ED, Smol T, Snijders Blok L, Stoler JM, Tang S, Tartaglia M, Thompson ML, van de Kamp JM, Wang J, Weise D, Weiss K, Woitschach R, Wollnik B, Yan H, Zackai EH, Zampino G, Campeau P, Bhoj E. A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. *Eur J Hum Genet*. 28(10):1422-1431.

Jan 2020 Mak CCY, Doherty D, Lin AE, Vegas N, Cho MT, Viot G, Dimartino C, Weisfeld-Adams JD, Lessel D, Joss S, Li C, Gonzaga-Jauregui C, Zarate YA, Ehmke N, Horn D, Troyer C, Kant SG, Lee Y, Ishak GE, Leung G, Barone Pritchard A, Yang S, Bend EG, Filippini F, Roadhouse C, Lebrun N, Mehaffey MG, Martin PM, Apple B, Millan F, Puk O, Hoffer MJV, Henderson LB, McGowan R, Wentzensen IM, Pei S, Zahir FR, Yu M, Gibson WT, **Seman A**, Steeves M, Murrell JR, Luettgen S, Francisco E, Strom TM, Amlie-Wolf L, Kaindl AM, Wilson WG, Halbach S, Basel-Salmon L, Lev-El N, Denecke J, Vissers LELM, Radtke K, Chelly J, Zackai E, Friedman JM, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics, Reid RR, Devriendt K, Chae JH, Stolerman E, McDougall C, Powis Z, Bienvenu T, Tan TY, Orenstein N, Dobyns WB, Shieh JT, Choi M, Waggoner D, Gripp KW, Parker MJ, Stoler J, Lyonnet S, Cormier-Daire V, Viskochil D, Hoffman TL, Amiel J, Chung BHY, Gordon CT. MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. *Brain*. 143(1):55-68.

Feb 2016 Al-Maawali A, Barry BJ, Rajab A, El-Quessny M, **Seman A**, Coury SN, Barkovich AJ, Yang E, Walsh CA, Mochida GH, and Stoler JM. Novel loss-of-function variants in *DIAPH1* associated with syndromic microcephaly, blindness, and early onset seizures. *Am J Med Genet A*. 170A(2):435-440.

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| Dec 2014 | Geng J, Picker J, Zheng Z, Zhang X, Wang J, Hisama F, Brown DW, Mullen MP, Harris D, Stoler J, **Seman A**, Miller DT, Fu Q, Roberts AE, and Shen Y. Chromosomal microarray testing for patients with congenital heart defects reveals novel disease-causing loci and high diagnostic yield. *BMC Genomics.* 15:1127. |
| March 2012 | Parental Testing for Chromosomal Microarray Variants of Uncertain Significance Abstract presented as a platform presentation at the ACMG Annual Clinical Genetics Meeting by Dr. David Miller. |
| April 2009 | Miller DT, Shen Y, Weiss LA, Korn J, Anselm I, Bridgemohan C, Cox GF, Dickinson H, Gentile J, Harris DJ, Hegde V, Hundley R, Khwaja O, Kothare S, Luedke C, Nasir R, Poduri A, Prasad K, Raffalli P, **Reinhard A**, Smith SE, Sobeih MM, Soul JS, Stoler J, Takeoka M, Tan WH, Thakuria J, Wolff R, Yusupov R, Gusella JF, Daly MJ, and Wu BL. Microdeletion/duplication at 15q13.2q13.3 among individuals with features of autism and other neuropsychiatric disorders. *J Med Genet*. 46(4):242-8. |

## Presentations

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| March 2016 | *Mosaicism for SRY-Positive Y Chromosome in Karyotype and Chromosomal Microarray Analysis in a Female with Turner syndrome Presentation,* Poster Presentation, ACMG Annual Clinical Genetics Meeting, Tampa, FL |
| Oct 2015 | *UPD1 in a Child with Multiple Congenital Anomalies,* Poster Presentation, ASHG Annual Meeting, Baltimore, MD |
| Feb 2012 | *FAQ on CMA: Update on Chromosomal Microarray Testing for Neurology,* Neurology staff meeting, Boston Children's Hospital, Boston, MA |
| March 2011 | *Genetic Counselors' Role in the DNA Lab: Result Interpretation and Report Preparation*, Longwood Genetic Counselors' Monthly Meeting, Boston, MA |
| Feb 2008 | *Introduction to Chromosomes and Genetics Concepts*, Nursing students, Boston Children's Hospital, Boston, MA |
| Nov 2006 | *The Genetic Evaluation and Hearing Loss Syndromes*, Governor Baxter School for the Deaf, Falmouth, ME |
| Dec 2005 | Clinical Presentation at the New England Regional Genetics Group Annual Meeting, Durham, NH |
| Nov 2005 | *Perinatal Hospice: An Important Option for Families Continuing Pregnancies with Lethal Fetal Abnormalities,* Poster Presentation, NSGC Annual Education Conference, Los Angeles, CA |

## References

Available upon request