Juliann M. Savatt Curriculum Vitae

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Education:

May 2013	Bachelor of Science – Biology (*suma cum laude*) Allegheny College, Meadville, Pennsylvania
May 2015	Master of Science – Genetic Counseling The University of North Carolina at Greensboro, North Carolina
June 2020	Statistical Reasoning in Public Health I and II, Certificate of Completion Johns Hopkins Summer Institute, Baltimore, Maryland

Certification: American Board of Genetic Counseling, 2015; recertified 2020

License: Pennsylvania Medical Board (Genetic Counseling), 2015-present

Honors and Awards:

2009 – 2010	Doane Distinguished Scholar
2009 – 2012	Distinguished Alden Scholar-Dean's List
2012	Outstanding Biology Major
2013	Phi Beta Kappa, Allegheny College
2013 – 2014	Graduate Assistant (Competitive Stipend)
2013 – 2014	Graduate Student Dean's Award

Professional Experience:

June 2015 – July 2017	Genetic Counselor/ Research Coordinator, Genomic Medicine Institute, Geisinger, Danville, Pennsylvania
July 2017 – July 2022	Genetic Counselor II, Autism & Developmental Medicine Institute, Geisinger Lewisburg, Pennsylvania
July 2022 – Present	Assistant Professor, Department of Genomic Health, Geisinger, Danville, Pennsylvania
October 2022 – Present	Co-Director, MyCode Genomic Screening and Counseling Program, Geisinger, Danville, Pennsylvania

Professional Society Memberships

2014 – Present	*National Society of Genetic Counselors* Member*, Cancer SIG,* 2015-2017 Member, *Neurogenetics SIG,* 2017-2019 Member, *Precision Medicine SIG*, 2020-Present
2020 – Present	*Pennsylvania Association of Genetic Counselors*
2020, 2022 – Present	*American Society of Human Genetics*

RESEARCH SUPPORT:

Active Support:

June 2015 – Present	NIH/NHGRI,"A Unified Clinical Genomics Database" 1U41HG006834-01-A1 (09/23/2013- 7/31/2026) MPIs: Christa Lese Martin, Heidi L. Rehm, Erin Rooney Riggs, 60%. Roles: Lead ClinGen's patient data sharing efforts including GenomeConnect, the ClinGen patient registry, participate in gene curation efforts, chair the Data Access, Protection, and Confidentiality Working Group. The goal of this project is to build an authoritative central resource that defines the clinical relevance of genes and variants for use in precision health and research. A main aim is to develop a public domain for the sharing of genotypic and phenotypic information from laboratories, clinicians, and patients to support evidence-based curation of genes and genomic variants.
October 2019 – 2020 2022 – Present	NIH/NHRGI, "Reporting Adult-Onset Genomic Results to Pediatric Biobank Participants and Parents" 5R01HG009671-02 (07/1/2019 – 06/30/2023), PI: Adam H. Buchanan, 20%. Roles: 2019-2020: Summarized the study's methodology in a methods publication, 2022-Present: Lead data collection for aim 2 focused on recommended management performance post results disclosure.
November 2022 – Present	The Leona M. and Harry B. Helmsley Charitable Trust, "Preventing Type 1 Diabetes and Familial Hypercholesterolemia through Well Child Screening" Helmsley Foundation Grant #2305-06052 (11/2022 - 04/2024), PI: Laney Jones, 15% Roles: Contribute to qualitative data collection from patients and payers.

Previous Support:

December 2020 – 2022	NIH/NIGMS: "Discovering Splicing Defects in Human Genes" 5R01GM127472-02 (08/23/2018-04/30/2020); 20% effort, MPI: William Fairbrother, Geisinger PI: Natasha Strande, 20%. Roles: Facilitate the creation and validation of automated data pulls of patient phenotype data from the electronic health record, complete necessary manual chart reviews, and collaborate on data analysis. The goal of this project is to discover and validate disease causing splicing mutations in patients.
October 2019 – 2020	NIH/NHRGI, "Reporting Adult-Onset Genomic Results to Pediatric Biobank Participants and Parents" 5R01HG009671-02 (07/1/2019 – 06/30/2023), MPI: Adam H. Buchanan, 10%. Role: Summarized the study's methodology in a methods publication.
October 2019 – 2020	NIH/NHGRI, "EMR-Linked Biobank for Translational Genomics" 3U01HG008679-04S1 (04/29/2019-03/31/2020) MPIs: Marc S. Williams, 5%. Role: Completed necessary manual chart reviews to collect phenotype data.
October 2019 –2021	NIH/NHGRI, A Unified Clinical Genomics Database" U41HG009650 (08/01/2017- 07/31/2021) MPI: Jonathan S Berg, 5%. Role: Participated in the Consent & Disclosure Recommendations working group to apply the consent and disclosure model to testing indications.

PEER-REVIEWED PUBLICATIONS:

1. Rocha, H. M., **Savatt, J. M**., Riggs, E. R., Wagner, J. K., Faucett, W. A., & Martin, C. L. (2017). Incorporating Social Media into your Support Toolbox: Points to Consider from Genetics-Based Communities. *Journal of Genetic Counseling 27(2):*470-480. doi:10.1007/s10897-017-0170-z. PMID: 29130143.

2. Wain, K.E., Palen, E., **Savatt, J.M.,** Finucane, B., Seeley, A., Challman, T.D., Myers, S.M., Martin, C.L. (2018). The value of genomic variant ClinVar submissions from clinical providers: Beyond the addition of novel variants. *Human Mutation 39(11):*1660-1667. doi: 10.1002/humu.23607. PMID: 30311381

3. Kandamurugu, M., Buchanan, A.H., Schwartz, M.L.D., Hallquist, M.H., Williams, J.L., Kulchak Rahm, A., Rocha, H., **Savatt, J.M.,** Evans, A.E., Butry, L.M., Lazzeri, A.L., Lindbuchler, D.M., Flansburg, C.N., Leeming, R., Vogel, V.G., Lebo, M.S., Mason-Suares, H.M., Hoskinson, D.C., Abul-Husn, N.S., Dewey, F.E., Overton, J.D., Reid, J.G., Baras, A., Willard, H.F., McCormick, C.Z., Krishnamurthy, S.B., Hartzel, D.N., Kost, K.A., Lavage, D.R., Sturm, A.C., Frisbie, L.R., Person, T.N., Metpally, R.P., Giovanni, M.A., Lowry, L.E., Leader, J.B., Ritchie, M.D., Carey, D.J., Justice, A.E., Kirchner, H.L., Faucett, W.A., Williams, M.S., Ledbetter, D.H., Murray, M.F. (2018). Exome Sequencing–Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants. *JAMA Network Open.* doi: 10.1001/jamanetworkopen.2018.2140. PMID : 30646163.

4. **Savatt, J.M.,** Azzariti D.R., Faucett, W.A., Harrison, S., Hart, J., Kattman, B., Landrum, M.J., Ledbetter, D.H., Miller, V.R, Palen, E., Rehm, H., Rhode, J., Turner, S., Vidal, J.A., Wain, K.E., Riggs, E.R., Martin, C.L. (2018). ClinGen's GenomeConnect Registry Enables Patient-Centered Data Sharing. *Human Mutation 39(11)*:1668-1676*.* doi: 10.1002/humu.23633. PMID: 30311371.

5. Vandervore, L.V., Schot, R., Milanese, C., Smits, D.J., Kasteleijn, E., Fry, A.E., Pilz, D.T., Brock, S., Börklü-Yücel, E., Post, M., Bahi-Buisson, N., Sánchez-Soler, M.J., van Slegtenhorst, M., Keren, B., Afenjar, A., Coury, S.A., Tan, W.H., Oegema, R., de Vries, L.S., Fawcett, K.A., Nikkels, P.G.J., Bertoli-Avella, A., Al Hashem, A., Alwabel, A.A., Tlili-Graiess, K., Efthymiou, S., Zafar, F., Rana, N., Bibi, F., Houlden, H., Maroofian, R., Person, R.E., Crunk, A., **Savatt, J.M.**, Turner, L., Doosti, M., Karimiani, E.G., Saadi, N.W., Akhondian, J., Lequin, M.H., Kayserili, H., van der Spek, P.J., Jansen, A.C., Kros, J.M., Verdijk, R.M., Milošević, N.J., Fornerod, M., Mastroberardino, P.G., Mancini, G.M.S. (2019). TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. *Am J Hum Genet*., *105*(6):1126–1147. doi:10.1016/j.ajhg.2019.10.009. PMID: 31735293.

6. Yan, K., Rousseau, J., Machol, K., Cross, L.A., Agre, K.E., Gibson, C.F., Goverde, A., Engleman, K.L., Verdin, H., De Baere, E., Potocki, L., Zhou, D., Cadieux-Dion, M., Bellus, G.A., Wagner, M.D., Hale, R.J., Esber, N., Riley, A.F., Solomon, B.D., Cho, M.T., McWalter, K., Eyal, R., Hainlen, M.K., Mendelsohn, B.A., Porter, H.M., Lanpher, B.C., Lewis, A.M., **Savatt, J.**, Thiffault, I., Callewaert, B., Campeau, P.M., Yang, X.J. (2020). Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. *Sci Adv*. 2020;6(4):eaax0021. doi:10.1126/sciadv.aax002. PMID: 32010779.

7. Connaughton, D.M., Dai, R., Owen, D.J., Marquez, J., Mann, N., Graham-Paquin, A.L., Nakayama, M., Coyaud, E., Laurent, E.M.N., St-Germain, J.R., Blok, L.S., Vino, A., Klämbt, V., Deutsch, K., Wu, C.W., Kolvenbach, C.M., Kause, F., Ottlewski, I., Schneider, R., Kitzler, T.M., Majmundar, A.J., Buerger, F., Onuchic-Whitford, A.C., Youying, M., Kolb, A., Salmanullah, D., Chen, E., van der Ven, A.T., Rao, J., Ityel, H., Seltzsam, S., Rieke, J.M., Chen, J., Vivante, A., Hwang, D.Y., Kohl, S., Dworschak, G.C., Hermle, T., Alders, M., Bartolomaeus, T., Bauer, S.B., Baum, M.A., Brilstra, E.H., Challman, T.D., Zyskind, J., Costin, C.E., Dipple, K.M., Duijkers, F.A., Ferguson, M., Fitzpatrick, D.R., Fick, R., Glass, I.A., Hulick, P.J., Kline, A.D., Krey, I., Kumar, S., Lu, W., Marco, E.J., Wentzensen, I.M., Mefford, H.C., Platzer, K., Povolotskaya, I.S., **Savatt, J.M.,** Shcherbakova, N.V., Senguttuvan, P., Squire, A.E., Stein, D.R., Thiffault, I., Voinova, V.Y., Somers, M.J.G., Ferguson, M.A., Traum, A.Z., Daouk, G.H., Daga, A.,

Rodig, N.M., Terhal, P.A., van Binsbergen, E., Eid, L.A., Tasic, V., Rasouly, H.M., Lim, T.Y., Ahram, D.F., Gharavi, A.G., Reutter, H.M., Rehm, H.L., MacArthur, D.G., Lek, M., Laricchia, K.M., Lifton, R.P., Xu, H., Mane, S.M., Sanna-Cherchi, S., Sharrocks, A.D., Raught, B., Fisher, S.E., Bouchard, M., Khokha, M.K., Shril, S., Hildebrandt, F. (2020). Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. *Am J Hum Genet*. 2020 Oct 1;107(4):727-742. doi: 10.1016/j.ajhg.2020.08.013. PMID: 32891193.

8. **Savatt, J.M.,** Wagner, J.K., Joffe, S., Rahm, A.K., Williams, M.S., Bradbury, A.R., Davis, F.D. Hergenrather, J., Hi, Y., Kelly, M.A., Kirchner, H.L., Meyer, M.N., Mozersky, J., O'Dell, S.M., Pervola, J., Seeley, A., Sturm, A.C., Buchanan, A.H. (2020) Pediatric Reporting of Genomic Results Study (PROGRESS): A mixed-methods, longitudinal, observational cohort study protocol to explore disclosure of actionable adult- and pediatric- onset genomic variants to minors and their parents. *BMC Pediatrics*. 20, 222. doi:10.1186/s12887-020-02070-4. PMID: 32414353.

9. Wain, K.E., Azzariti, D.R., Goldstein, J.L., Johnson, A.K., Krautscheid, P., Lepore, B., O'Daniel, J., Ritter, D., **Savatt, J.M**., Riggs, E.R., Martin, C.L. (2020). Variant interpretation is a component of clinical practice among genetic counselors in multiple specialties. *Genet Med* 22, 785–792. doi:10.1038/s41436-019-0705-9. PMID: 31754268.

10. Hallquist, M.L.G., Tricou, E.P., Hallquist, M.N., **Savatt, J.M.**, Rocha, H., Evans, A.E., Deckard, N., Hu, Y., Kirchner, H.L., Pervola, J., Rahm, A.K., Rashkin, M., Schmidlen, T.J., Schwartz, M.L.B., Williams, J.L., Williams, M.S., Buchanan, A.H. (2020). Positive impact of genetic counseling assistants on genetic counseling efficiency, patient volume, and cost in a cancer genetics clinic. *Genet Med.* Aug;22(8):1348-1354. doi: 10.1038/s41436-020-0797-2. Epub 2020 Apr 30. PMID: 32350418.

11. Hallquist, M.L.G., Tricou, E.P., Ormond, K.E**. Savatt, J.M.,** Coughlin, C.R., Faucett, W.A., Hercher, L., Levy, H.P., O'Daniel, J.M., Peay, H.L., Stosic, M., Smith, M, Uhlmann, W.R., Wand, H., Wain, K.E., Buchahan, A.H (2021). Application of a framework to guide genetic testing communication across clinical indications. *Genome Med* 13, 71. doi.org/10.1186/s13073-021-00887-x. PMID: 33926532.

12. Rodríguez-Palmero, A., Boerrigter, M.M., Gómez-Andrés, Aldinger, K.A, Marcos-Alcalde, M., Popp, B., Everman, D.B., Kern Lovgren, A., Arpin, S, Bahrambeigi, V., Beunders, G., Bisgaard, A.M., Bjerrgaard, V.A., Bruel, A.L., Challman, T.D., Congé, B., Coubes, C., de Man, S.A., Denommé-Pichon, A.S., Dye, T.J, Slmslie, F., Feuk, L., García-Miñaúr,, S., Gertler, T., Giorgio, E., Gruchy, N., Haack, T.B., Haldeman-Engert, C.R., Haukanes, B.I., Hoyer, J., Hurst, A.C.E., Isidor, B., Johansson Soller, M., Kushary, S., Kvarnun, M., Landau, Y.E, Leppig, K.A., Lindstrand, A., Kleinendorst, L., MacKenzie, A., Mandrile, G., Mendelsohn, B.A., Moghadasi, S., Morton, J.E., Moutton, S., Müller,A.J., O'Leary, M., Pacio-Míguez, M., Palomares-Bralo, M., Parikh, S., Pfundt, R., Pode-Shakked, B., Rauch, A., Repnikova, E., Revah-Politi, A., Ross, M.J., Claudia A. L. Ruivenkamp, C.A.L., Sarrazin, E., **Savatt, J.M.,** Schlüter, A., Schönewolf-Greulich, B., Shad, Z., Shaw-Smith, C., Shieh, J.T., Shohat, M., Spranger, S., Thiese, H., Mau-Them, F.T., van Bon, B., van de Burgt, I., van de Laar, I.M.B.H., van Drie, E., van Haelst, M. M., van Ravenswaaij-Arts, C.M., Verdura, E., Vitobello, A., Waldmüller, S., Whiting, S., Zweier, C., Prada, C.E., de Vries, B.B.A., Dobyns, W.B., Reiter, S.F., Gómez-Puertas, P., Pujol, A., & Tümer, Z. (2021). *DLG4*-related synaptopathy: a new rare brain disorder. *Genet Med* 23, 888–899. doi:10.1038/s41436-020-01075-9. PMID: 33597769.

13. Johannesen, K.M., Liu, Y., Koko, M., Gjerulfsen, C.E., Sonnenberg, L., Schubert, J., Fenger, C.D., Eltokhi, A., Rannap, M., Koch, N.A., Lauxmann, S., Krüger, J., Kegele, J., Canafoglia, L., Franceschetti, S., Mayer, T., Rebstock, J., Zacher, P., Ruf, S., Alber, M., Sterbova, K., Lassuthová, P., Vlckova, M., Lemke, J.R., Platzer, K., Krey, I., Heine, C., Wieczorek, D., Kroell-Seger, J., Lund, C., Klein, K.M., Billie Au, P.Y., Rho, J.M., Ho, A.W., Masnada, S., Veggiotti, P., Giordano, L., Accorsi, P., Hoei-Hansen, C.E., Striano, P., Zara, F., Verhelst, H., Verhoeven, J.S., van der Zwaag, B., Harder, A.V.E., Brilstra, E., Pendziwiat, M., Lebon, S., Vaccarezza, M., Minh Le, N., Christensen, J., Grønborg, S., Scherer, S.W., Howe, J., Fazeli, W., Howell, K.B., Leventer, R., Stutterd, C., Walsh, S., Gerard, M., Gerard, B., Matricardi, S., Bonardi, C.M., Sartori, S., Berger, A., Hoffman-Zacharska, D., Mastrangelo, M., Darra,

F., Vøllo, A., Motazacker, M.M., Lakeman, P., Nizon, M., Betzler, C., Altuzarra, C., Caume, R., Roubertie, A., Gélisse, P., Marini, C., Guerrini, R., Bilan, F., Tibussek, D., Koch-Hogrebe, M., Perry, M.S., Ichikawa, S., Dadali, E., Sharkov, A., Mishina, I., Abramov, M., Kanivets, I., Korostelev, S., Kutsev, S., Wain, K.E., Eisenhauer, N., Wagner, M., **Savatt, J.M.**, Müller-Schlüter, K., Bassan, H., Borovikov, A., Nassogne, M.C., Destrée, A., Schoonjans, A.S., Meuwissen, M., Buzatu, M., Jansen, A., Scalais, E., Srivastava, S., Tan, W.H., Olson, H.E., Loddenkemper, T., Poduri, A., Helbig, K.L., Helbig, I., Fitzgerald, M.P., Goldberg, E.M., Roser, T., Borggraefe, I., Brünger, T., May, P., Lal, D., Lederer, D., Rubboli, G., Heyne, H.O., Lesca, G., Hedrich, U.B.S., Benda, J., Gardella, E., Lerche, H., Møller, R.S. (2021). Genotype- phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. *Brain*, 25:awab321. doi: 10.1093/brain/awab321. Epub ahead of print. PMID: 34431999.

14. Finucane, B., **Savatt, J.M.,** Shimelis, H., Girirajan, S., Myers, S.M. (2021). Birt-Hogg-Dubé symptoms in Smith- Magenis syndrome include pediatric-onset pneumothorax. *Am J Med Genet A;*185(6):1922-1924. doi: 10.1002/ajmg.a.62159. PMID: 33666332.

15. **Savatt, J.M**., Azzariti, D.R., Ledbetter, D.H., Palen. E., Rehm, H.L, Riggs, E.R., Martin, C.L. (2021). Recontacting registry participants with genetic updates through GenomeConnect, the ClinGen patient registry. *Genet Med*. doi:10.1038/s41436-021-01197-8. PMID: 34007001

16. Barrett, K.S., Masnick, M., Hatchell, K.E., **Savatt, J.M.,** Banet, N., Buchanan, A.H., Willard, H.F. (2022). Clinical Validation of Genomics Functional Screen Data: Analysis of Observed BRCA1 Variants in an Unselected Population Cohort. *Human Genetics and Genomics Advances.* doi: 10.1016/j.xhgg.2022.100086.

17. Angelozzi, M., Karvande, A., Molin, A.N., Ritter, A.L., Leonard, J., **Savatt, J.M**., Douglass, K., Myers, S.M., Fuoppa, M., Tolchin, D., Zackai, E., Donoghue, S., Hurst, A.C.E., Descartes, M., Smith, K., Valasco, D., SChmanski, A., Crunk, A., Tokita, M.J., de Lange, I., van Gassen, K., Robinson, H., Guegan, K., Suri, M., Patel, C., Bournez, M., Faivre, L., Baker, J., Fabie, N., Weaver, K.N., Shillington, A., Hopkin, R., Barge-Schaapvled, D.Q.C.M., Ruivenkamp, C.A.L., Bökenkamp,R., Schrier Vergano, S.A., Noelia Seco, M., Misra, V., Rogers, C., Graziano, C., Ahrens-Nicklas, R.C., Lefebvre, V. (2022). Identification of novel SOX4 variants broadening the clinical and genetic spectrum of Coffin-Siris syndrome 10. *Journal of Medical Genetics.* Published Online First: 01 March 2022. doi: 10.1136/jmedgenet-2021-108375

18. Asif, M., Kaygusuz, E., Shinawi, M., Nickelsen, A., Hsieh, T.C., Wagle, P., Budde, B.S., Hochscherf, J., Abdullah, U., Höning, S., Nienberg, C., Lindenblatt, D., Noegel, A.A., Altmüller, J., Thiele, H., Motameny, S., Fleischer, N., Segal, I., Pais, L., Tinschert, S., Samra, N.N., **Savatt, J.M**., Rudy, N.L., De Luca, C., Fortugno, P., White, S.M., Krawitz, P., Hurst, A.C.E., Niefind, K., Jose, J., Brancati, F., Nürnberg, P., Hussain, M.S. (2022). De novo variants of *CSNK2B* cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway. *Human Genetics and Genomics Advances,* 3(3). doi.org/10.1016/j.xhgg.2022.100111.

19. Rooney Riggs, E., Bingaman, T.I., Barry, C.A., Behlmann, A., Bluske, K., Bostwick, B., Bright, A., Chen, C.A., Clause, A.R., Dharmadhikari, A.V., Ganapathi, M., Gonzaga-Jauregui, C., Grant, A., Hughes, M.Y., Kim, S.R., Krause, A., Liao, J., Lumaka, A., Mah, M., Maloney, C.M., Mohan, S., Osei-Owusu, I.A., Reble, E., Rennie, O**., Savatt, J.M.,** Shimelis, H., Siegert, R., Sneddon, T.P., Thaxton, C., Toner, K.A., Trung Tran, K., Webb, R., Wilcox, E.H., Yin, J., Zhuo, X., Znidarsic, M., Martin, C.L., Betancur, C., Vorstman, J.A.S., Miller, D.T., Schaaf, C.P. (2022). Clinical validity assessment of genes frequently tested on intellectual disability/autism sequencing panels. *Genetics in Medicine.* I

20. Jones, L.K., Strande, N.T., Calvo, E.M., Chen, J., Rodriguez, G., McCormick, C.Z., Hallquist, M.L.G., **Savatt, J.M.,** Rocha, H., Williams, M.S., Sturm, A.C., Buchanan, A.H., Glasgow, R.E., Martin, C.L., Rahm, A.K. (2022). A RE-AIM framework analysis of DNA-based population screening: using

implementation science to translate research into practice in a healthcare system. *Frontiers in Genetics.*

21. **Savatt, J.M.,** Ortiz, N.M., Thone, G.M., McDonald, W.S., Kelly, M.A., Berry, A.S.F., Alvi, M.M., Hallquist, M.L.G., Malinowski, J., Purdy, N.C., Williams, M.S., Sturm, A.C., Buchanan, A.H. (2022). Observational Study of Population Genomic Screening for Variants Associated with Endocrine Tumor Syndromes in a Large, Healthcare-Based Cohort. *BMC Medicine.*

22. **Savatt, J.M.,** Shimelis, H., Moreno-De-Luca, A., Strande, N.T., Ledbetter, D.H., Martin, C.L., Myers, S.M., Finucane, B.M. (2022). Frequency of *FLCN* Loss of Function Variants and Birt-Hogg-Dubé-Associated Phenotypes in a Healthcare System Population. *Genetics in Medicine*.

23. Pichardo, P., Hellums, R.N., Hao, H., Hassen, D., **Savatt, J.M.,** Pellitteri, P.K., Alvi, M., Buchanan, A.H., Purdy, N.C. Thyroidectomy outcomes in patients identified with RET pathogenic variants through a Population Genomic Screening Program. Submitted to *JAMA Otolaryngology-Head & Neck Surgery.*

REVIEW ARTICLES:

1. **Savatt, J.,** Pisieczko, C.J., Zhang, Y., Lee, M.T., Faucett, W.A., & Williams, J.L. (2019). Biobanks in the Era of Genomic Data. *Current Genetic Medicine Reports*, 7, 153 - 161. doi: 10.1007/s40142-019-00171-w.

2. **Savatt, J.M.** & Myers, S.M. (2021). Genetic Testing in Neurodevelopmental Disorders. *Frontiers* 9. doi: 10.3389/fped.2021.52677.

MANUSCRIPTS IN PROGRESS:

1. **Savatt, J.M.,** Johns, A., Schwartz, M.L.B., McDonald, W.S., Salvati, Z.M., Ortiz, N.M., Masnick, M., Hatchell, K., Hao, J., Buchanan, A.H., Williams, M.S. Homozygous *HFE* C282Y related hemochromatosis disclosure in an unselected health care system population: a cross-sectional study of clinical burden and health behaviors. Submitted, Under Review – *JAMA Internal Medicine*.

2. Schwiter, R., Rocha, H., Johns, A., **Savatt, J.M.,** Diehl, D., Kelly, M.A., Williams, M.S., Buchanan, A.H. Low adenoma burden in unselected patients with a pathogenic *APC* variant. Submitted and Under Review – *Genetics in Medicine.*

3. Chopra, M. Savatt, J.M., Bingaman, T., Good, M.E., Mahida, S., Lanzotti, V., Baldridge, D., Pive, j., Hazlett, H., Sahin, M., Payne, P., Riggs, E.R., Constantino, J. on behalf of the Brain Gene Registry Consortium. Harnessing Clinical Variants for Brain Gene Curation: A Test of Principle.

PEER-REVIEWED PRESENTATIONS:

1. Leeming, R., Maney, E., Fan, A., Rocha, H., **Koenig, J.M.**, Kulchak Rahm, A., Snyder, S., Hao, J., Pittcavage, J. Evaluation and Risk Assessment for Breast Cancer: An Integrated Health System Approach. Presented at the 2016 America Society of Breast Surgeons (ASBS) Annual Meeting. April 13-17th, 2016. Dallas, Texas.

2. **Koenig, J.M.** GenomeConnect - Making Connections and Engaging Patients in Genomic Discovery (Platform Presentation). Presented at Stanford Medicine X, September 7, 2016, Stanford, California.

3. **Savatt, J.M**. Azzariti, D.R, Faucett, W.A., Ledbetter, D.H., Martin, C.L. Rangel Miller, V. Rehm, H., Wain, K., Riggs, E. Contacting Patients with Updated Variant Interpretations: An Innovative Approach Utilizing GenomeConnect, an Online Patient Registry; (Abstract #32, Platform Presentation). Presented at the 2017 Annual Meeting of the American College of Medical Genetics, March 24, 2017, Phoenix, Arizona.

4. Hindoff, L., Blout, C., Scollon, S., Fayer, S., O'Daniel, J., Pollin, T., Orlando, L., Smith, M., Hoell, C., Azzariti, D.R, **Savatt, J.M.** Unlocking the Acronyms: Research Genetic Counselors and the NIH Partnering Together to Improve Patient Care; (Program A05, Pre-Conference Symposium). Presented at the 36th Annual Education Conference for the National Society of Genetic Counselors, September 13, 2017, Columbus, Ohio.

5. **Savatt, J.M.**, Azzariti, D.A., Faucett, W.A., Landrum, M.J., Ledbetter, D.H., Martin, C.L., Rangel Miller, V., Palen, E., Rehm, H., Rhode, J., Turner, S., Riggs, E.R. The impact of sharing patient-derived data in ClinVar via GenomeConnect; (Program 231, Platform Presentation). Presented at the Annual Meeting of the American Society of Human Genetics, October 20, 2017, Orlando, Florida.

6. Wain K.E., Azzariti D.R., Goldstein J., Knight Johnson A., Krautscheid P., O'Daniel J.M., Ritter D., **Savatt J.M**., Martin C.L., Rooney Riggs E., on behalf of the ClinGen Education Working Group. Variant Interpretation Practice Amongst Clinical Genetic Counselors: Assessment of Training and Resource Needs to Support Clinical Practice. Platform presentation. Presented at the Annual Meeting of the American Society of Human Genetics, October 17, 2018, San Diego, California.

7. **Savatt, J.M.,** Azzariti, D.R., Palen, E., Hart, J., Kattman, B., Landrum, M.J., Miller, V.R., Rhode, J., Vidal, J.A., Ledbetter, D.H., Rehm, H., Faucett, W.A., Riggs, E.R., Martin, C.L. Patient-data sharing of whole exome sequencing results with GenomeConnect informs variant interpretation and gene-disease relationships (Program 123, Platform Presentation). Presented at the Annual Meeting of the American Society of Human Genetics, October 17, 2018, San Diego, California.
8. Wain K.E., Azzariti D.R., Goldstein J., Knight Johnson A., Krautscheid P., O'Daniel J.M., Ritter D., **Savatt J.M.,** Martin C.L., Rooney Riggs E., on behalf of the ClinGen Education Working Group. Variant Interpretation is a Wide-Spread and Valuable Practice Amongst Clinical Genetic Counselors Across Multiple Specialties. Platform presentation. Presented at the National Society of Genetic Counselors, November 2018, Atlanta, Georgia.

9. **Savatt, J.M.,** Azzartiti, D.R., Ledbetter, D.H., Miller, V.R., Palen, E., Rehm, H., Rhode, J., Vidal, J.A., Riggs, E.R., Martin, C.L. Sharing Patient-derived Genetic and Phenotypic Data Benefits Patients and the Genomics Community, Oral Presentation. Presented at the 2020 Annual Meeting of the Pennsylvania Association of Genetic Counselors, March 12, 2020, Pittsburgh, Pennsylvania.

10. Williams, M.S., Buchanan, A.H., Sturm, A.C., Schwartz, M., **Savatt, J.,** Sundaresan, A., Bellus, G., McCromick, C.Z., Hatchell, K.E., Masnick, M., Barrett, K.M., Deverka, P. Underrecognition of the Disease Burden of Hereditary Hemochromatosis: A Population Screening Study. Platform Presentation. Presented at the 2020 Annual Meeting of the American College of Medical Genetics, March 20, 2020, San Antonio, Texas. Withdrawn due to COVID-19 pandemic.

11. **Savatt, J.M.,** Azzariti, D.R., Faucett, W.A., Ledbetter, D.H., Miller, V.R., Palen, E., Rehm, H., Rhode, J., Toner, K., Vidal, J.A., Riggs, E.R., Martin, C.L. Contributing to Gene-disease Discovery Through GenomeConnect, ClinGen's Online Patient Registry," Poster Presentation and Oral Rapid Poster Presentation. Presented at the 2020 Annual Meeting of the American College of Medical Genetics, March 18, 2020, San Antonio, Texas.

12. **Savatt, J.M.,** Shimelis, H., Myers, S.M., Moreno-De-Luca, A., Finucane, B. Identifying loss of function FLCN variants and Birt-Hogg-Dube-associated phenotypes in a large, healthcare-based cohort. Platform Presentation. Presented at the 2021 Annual Meeting of the American College of Medical Genetics, April 14, 2021, Virtual.

13. **Savatt, J.M.,** Paul, L.D., Smith, R., Walsh, L., Wain, K., Azzariti, A., Ledbetter, D.L.., Rehm, H.L., Taylor, C., Riggs, E.E., Martin, C.L. Expanding patient data sharing: ClinGen's partnership with Simons Searchlight to submit patient-provided genetic and health information to ClinVar. Platform Presentation.

Presented at the 2021 Annual Meeting of the American College of Medical Genetics and Genomics, April 14, 2021, Virtual.

14. **Savatt, J.M.,** Azzartiti, D.R., Ledbetter, D.H., Rehm, H., Riggs, E.R., Martin, C.L. The Shared Responsibility of Recontact: Patients Obtaining Updated Genetic Test Results Through Patient Registries. Platform Presentation. Presented at the 2021 Curating the Clinical Genome Meeting, May 14, 2021, Virtual.

15. Paul, L., **Savatt J.M.,** Azzariti, D., Rehm, H.L., Riggs, E.R., Martin, C.L. Genetic Updates Returned by GenomeConnect, the ClinGen Patient Registry: a Pilot Study of Participant Experience. Presented at the 2022 Annual Meeting of the American College of Medical Genetics and Genomics, March 23, 2022, Nashville, TN.

16. Singer, T., Kelly, M.A., **Savatt, J.M**., Strande, T. Characterization of MyCode Participants with Pathogenic or Likely Pathogenic Variants in *ENG*, *ACVRL1*, or *SMAD4* Associated with Hereditary Hemorrhagic Telangiectasia. Presented as a platform presentation at the 2023 American College of Medical Genetics and Genomics Conference, March 14-18, 2023, Salt Lake City, Utah.

PEER-REVIEWED POSTERS:

1. **Koenig, J.M**., Corneliussen, K., Quinn, K., Decker, M., Culp Stewart, R. The Interface Between Genetic Counselors and Obstetricians: Education Concerning Noninvasive Prenatal Screening. Poster presentation**.** Presented at the 34th Annual Education Conference for the National Society of Genetic Counselors, October 23, 2015, Pittsburgh, Pennsylvania.

2. **Koenig, J.M**., Riggs, E.R., Azzariti, D.R, Rangel Miller, V., Rehm, H., Ledbetter, D.H., Martin, Faucett,

1. W.A. GenomeConnect Participant Matching System: Connecting Individuals with Rare Diseases or Genetic Variants. Poster presentation. Presented at the 2016 Curating the Clinical Genome Meeting, June 22, 2016, Hinxton, Cambridge, United Kingdom. **Koenig, J.M**., Azzariti, D.R, Landrum, M.J., Ledbetter, D.H., Martin, C.L. Maglott, D.R., Rangel Miller, V., Rehm, H., Riggs, E.R., Faucett, W.A. GenomeConnect: Case Level Data Submission to NCBI ClinVar. Poster presentation. Presented at the 2016 Curating the Clinical Genome Meeting, June 22, 2016, Hinxton, Cambridge, United Kingdom.

2. **Koenig, J.M.**, Azzariti, D.R., Ledbetter, D.H., Martin, C.L., Rangel Miller, V., Rehm, H., Riggs, E.R., Faucett, W.A. GenomeConnect participant matching system: Connecting individuals with rare diseases or genomic variants. Poster presentation. Presented at the 35th Annual Education Conference for the National Society of Genetic Counselors, September 31, 2016, Seattle, Washington.

3. **Savatt, J.M.,** Azzariti, D.R, Faucett, W.A., Landrum, M.J, Ledbetter, D.H., Martin, C.L, Rangel Miller, V., Palen, E., Rehm, H., Rhode, J., Turner, S., Wain, K., Riggs, E.R. Sharing Patient-Derived Data in ClinVar via GenomeConnect (Poster #44**).** Poster presentation. Presented at the 2017 Curating the Clinical Genome Meeting, June 29, 2017, Bethesda, Maryland.

4. Wain K.E., Kasparson L., Challman T., Adamson K., Douglass K., Eisenhauer N., Fisher H., Haas-Givler B., Kinney M., Mitchel M., Merz A., Myers S.M., Nelson T., Oetjens K., Palen E., Petro B., **Savatt, J.M**., Seeley A., Shuman D., Taylor C.M., Troutman K., Turner L., Wagner M., Ledbetter D., Finucane B., Martin C.L. A Genotype-First Approach for Developmental Brain Disorders: Diagnostic Yield Supports Whole Exome Sequencing as the First Step in Genomic Testing. Poster presentation (High Rank Distinction). Presented at the 2018 Annual Meeting of the American College of Medical Genetics, April 13, 2018, Charlotte, North Carolina.

5. **Savatt, J.M.,** Azzariti, D.R, Faucett, W.A., Landrum, M.J, Ledbetter, D.H., Martin, C.L, Rangel Miller, V., Palen, E., Rehm, H., Rhode, J., Turner, S., Wain, K., Riggs, E.R., Vidal, J.A., Martin, C.L. ClinGen's

GenomeConnect Registry Enables Patient-Centric Data Sharing. Poster presentation (High Rank Distinction). Presented at the 2018 Annual Meeting of the American College of Medical Genetics, April 13, 2018, Charlotte, North Carolina.

6. **Savatt, J.M.,** Azzartiti, D.R, Faucett, W.A., Harrison, S.M., Hart, J., Landrum, M.J, Ledbetter, D.H., Miller, V.R., Palen, E., Rehm, H., Rhode, J., Riggs, E.R., Vidal, J.A., Martin, C.L. Patient-derived Genomic Data from ClinGen's GenomeConnect: Advancing Genomic Knowledge and Keeping Patients Informed of Variant Classifications. Poster presentation. Presented at the 37th Annual Education Conference for the National Society of Genetic Counselors, November 17, 2018, Atlanta, Georgia.

7. Martin C.L., Wain K.E., Challman T.D., Kasparson K., Myers S.M., Palen E., **Savatt J.M.**, Finucane B., Ledbetter D.H. Routine Clinical Genetic Testing in Patients with Autism Spectrum Disorder and Related Developmental Disorders Has a Diagnostic Yield up to 40%, Improves Medical Management, and Significantly Alters Recurrence Risk Counseling. Poster presentation. Presented at the International Society for Autism Research Annual Meeting. May 2019. Montreal, Canada.

8. **Savatt, J.M.,** Azzartiti, D.R, Faucett, W.A., Ledbetter, D.H., Miller, V.R., Palen, E., Rehm, H., Rhode, J., Rogers, L., Talbird, S., Trutoiu, L., Vidal, J.A., Riggs, E.R., Martin, C.L. ClinGen's Patient Data Sharing Program: Leveraging Data Sharing Experience from GenomeConnect to Broaden Patient Data Sharing Efforts. Poster presentation. Curating the Clinical Genome Meeting. May 30, 2019. Washington, District of Columbia.

9. **Savatt, J.M,** Azzartiti, D.R, Faucett, W.A., Ledbetter, D.H., Miller, V.R., Palen, E., Rehm, H., Rhode, J., Vidal, J.A., Riggs, E.R., Martin, C.L. Reporting of Variants in Genes with Limited, Disputed, or No Evidence for a Mendelian Condition Among GenomeConnect Participants. Poster presentation (High Rank Distinction). Presented at the 2019 Annual Meeting of the American Society of Human Genetics, October 18, 2019, Houston, Texas.

10. Wain, K.E., Azzartiti, D., Barnett, S., Giordano, J., Goehringer, S., Goldstein, J., Helgeson, M., Knight Johnson, A., Kelly, M.A., Kurtz, L., Mester, J., O'Daniel, J., Ritter, D., **Savatt, J.M**., Riggs, E.R., Martin, C.L. Poster presentation. Presented at the 38th Annual Education Conference for the National Society of Genetic Counselors, November 6, 2019, Salt Lake City, Utah.

11. Miller, S., **Savatt, J.M.**, Ali, M., Doyle, L., Iannaccone, A. Identifying Ethnicity-Associated Differences in Genetic Testing Results for Patients with Suspected Inherited Retinal Degeneration. Poster presentation. Presented at the 38th Annual Education Conference for the National Society of Genetic Counselors, November 6, 2019, Salt Lake City, Utah.

12. **Savatt, J.M.,** Azzartiti, D.R, Faucett, W.A., Florin, J., Ledbetter, D.H., Miller, V.R., Palen, E., Rehm, H., Rhode, J., Rogers, L., Talbird, S., Trutoiu, L., Vidal, J.A., Waggoner, C., Riggs, E.R., Martin, C.L. Expanding Patient Data Sharing: GenomeConnect's Pilot to Engage External Registries in Data Sharing. Poster presentation. Presented at the 38th Annual Education Conference for the National Society of Genetic Counselors, November 6, 2019, Salt Lake City, Utah.

13. **Savatt, J.M.,** Azzariti, D.R., Ledbetter, D.H., Rehm, H., Riggs, E.R., Martin, C.L. Recontacting Registry Participants with Genetic Updates through GenomeConnect: The ClinGen Patient Registry. Poster presentation. Presented at the 2020 Annual Meeting of the American Society of Human Genetics, October 26, 2020, Virtual.

14. **Savatt, J.M.,** Azzariti, D.R., Ledbetter, D.H., Rehm, H., Riggs, E.R., Martin, C.L. The Role of GenomeConnect in Keeping Patients Updated about their Genetic Test Result. Poster presentation. Presented at the 2020 Annual Meeting of the National Society of Genetic Counselors, September 25, 2020, Virtual.

15. **Savatt, J.M.,** Deckard, N.M., Thone, G.M., McDonald, W.S., Alvi, M.M., Purdy, N.C., Lindemann, T.L., Sturm, A.C., Buchanan A.H. Experience Completing Population Screening for Variants Associated with Endocrine Tumor Syndromes in a Large, Healthcare-Based Cohort. Poster Presentation. Presented at the 2021 Endocrine Society Annual Meeting, March 17-20, 2021, Virtual.

16. Paul, L.D., **Savatt, J.M.,** Good, M.E., Heidlebaugh, A.R., Azzariti, D.R., Jae, D., Ledbetter, D.H., McLarney, B., Rehm, H.L., Riggs, E.R., Martin, C.L. Addressing barriers to patient data sharing: Exploring the effects of employing electronic consent to obtain genetic testing records through ClinGen's Patient Data Sharing Program. Poster Presentation. Presented at the 2021 Annual Meeting of the American Society of Human Genetics, October 18, 2021, Virtual.

17. Pichardo, P., Hellums, R.N., Hao, H., Hassen, D., **Savatt, J.M.,** Purdy, N.C. Thyroidectomy Outcomes for Patients with *RET* Mutations Detected in a Rural Tertiary Care Population Genomic Screening Program. Presented at the 2022 Triologic Society Combined Sections Meeting, January 20-22, 2022, San Diego, California.

18. **Savatt, J.M**, Buoy, C.J., Ney, S.M., Kelsey, C.R., Buchanan, A.H., Banet, N., Kelly, M.A., Puttagunta, R., Ramey, H., Fairbother, W., Strande, N.T. Validation of automated electronic health record (EHR) data capture of hereditary breast and ovarian cancer and Lynch syndrome phenotypes. Presented at the 2022 American Society of Human Genetics Meeting, October 25-30, 2022, Los Angeles, California.

19. Morgan, A., **Savatt, J.M**., Azzariti, D., Ledbetter, D.H., Rehm, H.L., Martin, C.L., Riggs, E.R. GenomeConnect - The ClinGen patient registry's case-level data can impact variant classification. Presented at the 2022 American Society of Human Genetics Meeting, October 25-30, 2022, Los Angeles, California.

20. **Savatt, J.M,** Johns, A., Schwartz, M.L.B., Salvati, Z.M., Ortiz, N.M., Masnick, M., Hatchell, K., Hao, J., Buchanan, A.H., Williams, M.S. Clinical burden and health behaviors associated with genomic screening for homozygous HFE C282Y variants in an unselected health care system. Presented as a poster with rapid poster presentation at the 2023 American College of Medical Genetics and Genomics Conference, March 14-18, 2023, Salt Lake City, Utah.

21. **Savatt, J.M.,** Shimelis, H., Buchanan, A.H., Kelly, M.A., Strande, N.T Identification of *PALB2* Variants and Associated Cancers in a Large, Unselected Healthcare Population. Presented as a poster at the 2023 American College of Medical Genetics and Genomics Conference, March 14-18, 2023, Salt Lake City, Utah.

22. Kelly, M.A. Carruth, E.D., **Savatt, J.M.**, Haggerty, C., Strande, N.T., Assessing disease burden in *TTN* and *FLNC* from population genomic screening in the Geisinger MyCode® Community Health Initiative. Presented as a poster at the 2023 American College of Medical Genetics and Genomics Conference, March 14-18, 2023, Salt Lake City, Utah.

23. Diloreto, K., **Savatt, J**.M, Yu, K., Kelly, M.A. Rates of cardiomyopathy in probands and their family members from a large, unselected healthcare cohort. Presented as a poster with rapid poster presentation at the 2023 American College of Medical Genetics and Genomics Conference, March 14-18, 2023, Salt Lake City, Utah.

NON-PEER REVIEWED ARTICLES:

1. Martin, C.L. & **Savatt, J.M.** Sharing genetic results can advance autism science, medical care. (April 9, 2019). *Spectrum*. https://www.spectrumnews.org/opinion/viewpoint/sharing-genetic-results-can-advance-autism-science-medical-care/**.**

2. **Savatt, J.M** on behalf of the GenomeConnect team. ClinGen: Enabling Patient-Driven Data Sharing through GenomeConnect. (Fall 2019). *American College of Medical Genetics Fall News Magazine.*

INVITED SCIENTIFIC PRESENTATIONS:

May 16, 2016	"Cancer Genetics Risk Assessment Clinic Overview" Annual Geisinger Gastrointestinal Conference, Danville, Pennsylvania
May 25, 2018	"GenomeConnect: Sharing Individual Level Data Through Patient Registries" Curating the Clinical Genome, Hinxton, England
April 26, 2019	"ClinVar Community Call for Clinician Submitters" Community ClinVar Call
June 14, 2019	"Patient Data Sharing of Genetic and Health Information Informs Genetic Discovery and Fuels Research" Presentation with Sandra Talbird, Rare Disease Innovation and Partnering Summit, CBI Conference, Boston, Massachusetts
October 5, 2019	"ClinGen Curation Activities and Patient Engagement" Presentation with Diane Zastrow, Northern California Coalition of Genetic Counselors Annual Meeting, Berkeley, California.
January 28, 2020	"Strategies for Data Sharing from Patient Registries" Real World Evidence, and HEOR, InformaConnect Conference, Miami, Florida
February 5, 2021	"GenomeConnect (The ClinGen Patient Registry) & Patient Registry Partnerships":
May 5, 2021	"Increasing our Understanding of Genetics and Health Through Patient Data Sharing" New York Mid-Atlantic and Caribbean Annual Meeting Workshop, Virtual
June 10, 2021	"Homozygous *HFE* C282Y Related Hemochromatosis Disclosure in an Unselected Health Care System Population: Clinical Burden and Health Behaviors" MyCode Case Conference, Geisinger
June 24, 2021	"GenomeConnect and Patient Data Sharing" Clinical Genome Resource Retreat, Virtual
April 19, 2022	"Increasing our Understanding of Genetics and Health Through Patient Data Sharing" InformedDNA Continuing Education Units Series, Virtual
March 17, 2023	"GenomeConnect and the ClinGen Patient Data Sharing Program: Enhancing Knowledge of Genetic Variants and Health through Patient Data Sharing" Learning Lounge, American Society of Medical Genetics and Genomics Annual Meeting, Salt Lake City, Utah.

ACADEMIC LECTURES:

March 9, 2018	"The Multidisciplinary Team at Geisinger Autism & Developmental Medicine Institute"

Geisinger Commonwealth School of Medicine, Scranton, Pennsylvania

2019 - 2022 *"*Genetics 101"
Annually, ADMI Summer Undergraduate Research Program

2022 – 2023 "The Clinical Genome Resource (ClinGen): An Overview"
Various Genetic Counseling Programs including Augustana University, University of California San Francisco, Washington University in St. Louis, Boise State University (Recorded), Long Island University, University of Oklahoma, University of Washington, Thomas Jefferson University, Baylor, Wayne State, University of Nebraska medical Center.

COMMUNITY PRESENTATIONS:

September 16, 2015 "Does it Run in the Family? Hereditary Cancer Overview," Geisinger Wyoming Valley Cancer Support Group, Wilkes Barre, Pennsylvania

November 8, 2016 "Does it Run in the Family? Hereditary Breast and Ovarian Cancer," Holy Spirit Breast Cancer Support Group, Harrisburg, Pennsylvania

August 7, 2020 "Genetics 101 and How to Read Your Laboratory Report"
Simons Searchlight Virtual Family Conference for *SETBP1*, *CSNK2A1*, *HIVEP2,* and *MED13L*

June 24, 2021 "How to Read Your Laboratory Report and The Importance of Genomic Data Sharing"
Association for Creatine Deficiencies, Virtual Webinar

June 27, 2021 "Genetics 101 and How to Read Your Laboratory Report"
GenomeConnect, Virtual Webinar

September 16, 2021 "How to Read Your Laboratory Report and The Importance of Genomic Data Sharing"
Cure GM1 Foundation 2021 Virtual Conference

SUPERVISORY TEACHING:

Genetic Counseling Student Rotations and Thesis/Capstone Committees:

2015 Elly Brokamp (Sarah Lawrence University), Rotation Supervisor (non-primary)
Currently a genetic counselor at Vanderbilt University Medical Center

2016 Nathan Hassel (Sarah Lawrence University), Rotation Supervisor (non-primary)
Currently a genetic counselor at Valley Children's Healthcare

2016 – 2017 Caroline Augsburger and Maira Pires, Sarah Lawrence University. Thesis Committee Member, "Developing an Alternative Model for Pre-Test Genetic Counseling for Women Diagnosed with Breast Cancer."

2017 Caitlin Bozick (Arcadia University), Rotation Supervisor (non-primary)
Currently a genetic counselor at Levine Children's Specialty Center

2018 Angelina Londono (University of Alabama at Birmingham), Rotation Supervisor (non-primary)
Currently a Bilingual Health Educator at See Yourself Health, a digital coaching company

2018 – 2019	Sam Miller (University of North Carolina at Greensboro), Thesis Committee Member, "Variant Analysis in Inherited Ocular Diseases"
2019	Jessica Davidson (University of Alabama at Birmingham), Supervisor (non-primary) Currently a genetic counselor at Northside Hospital
2022 - 2023	Nataly Abrams (Rutgers University), Thesis Committee Member, "Characterizing 'non-core' cancers in BRCA1/2 carriers through large scale genomic screening"

Other Students and Learners

2019	Bri Lepore (Bucknell University), Undergraduate intern at Geisinger Autism & Developmental Medicine Institute, Supervised creation of patient-facing materials Graduate of The Ohio State University Genetic Counseling program
2019 – 2021	Sara Stansbury (Juniata College), Undergraduate intern at Geisinger Autism & Developmental Medicine Institute, Co-mentor Awarded "Best Poster" in the Health Sciences category during the 2019 and 2021 Summer Undergraduate Research Symposiums. Currently a graduate student at Baylor College Genetic Counseling program
2020	Megan Keeney (Bucknell University), Undergraduate intern at Geisinger Autism & Developmental Medicine Institute, Mentor Currently a graduate student at Kean University Genetic Counseling program
2020 – 2022	Molly Good, (Geisinger), Genetic Counseling Assistant, Co-mentor Currently a medical student at Lake Erie College of Osteopathic Medicine
2020 – 2022	Lianna Paul (Geisinger), Genetic Counseling Assistant, Co-mentor Currently a graduate student at Vanderbilt University Genetic Counseling program
2020 – 2022	Taylor Bingaman (Geisinger), Genetic Counseling Assistant, Co-mentor Currently a GCA at Walter Reed Cancer Genetics Clinic
2021 – 2022	Zoe Lindsey Mills (Geisinger), Genetic Counseling Assistant, Mentor in the GCA Mentee/GC Mentor program
2022 – Present	Allison Rossel (Geisinger), Genetic Counseling Assistant, Co-mentor
2022 – Present	Ineke Cordova (Geisinger), Genetic Counseling Assistant, Co-mentor
2022 – Present	Bryanna VanHoute (Geisinger), Genetic Counseling Assistant, Co-mentor
2022 – Present	Mireya Jimenez-Lopez (Geisinger), Genetic Counseling Assistant, Mentor in the GCA Mentee/GC Mentor program
2023 – Present	Tyler Singer (Geisinger Commonwealth School of Medicine), Medical Student, Mentor on Independent Research
2023 – Present	John (Jack) Murray (Geisinger Commonwealth School of Medicine), Medical Student, Mentor on Independent Research

CURRICULUM DEVELOPMENT:

November 2015 — GenomeFirst Continuing Medical Education Module, Hereditary Paraganglioma and Pheochromocytoma

December 2016 — Variant of Uncertain Significance (VUS) Result Updating, American College of Medical Genetics Maintenance of Certification Module. Developed by the ClinGen Phenotype Working Group and hosted by ACMG. Approved by the American Board of Medical Genetics and Genomics for Part IV MOC

April 2018 — ClinGen Variant Interpretation Discrepancy Resolution Maintenance of Certification Module. Developed by Danielle Azzariti, Steven Harrison, Heidi Rehm, Christa Martin, Juliann Savatt, and Erin Riggs with review by the ClinGen Education, Coordination and Training Working Group. Approved by the American Board of Medical Genetics and Genomics for Part IV MOC.

June – July 2021 and 2022 — Clinical Genome Resource Clinical Genomics Career Panel Series. Developed and coordinated by Juliann Savatt and Erin Riggs to expose interested learners to genomics career paths.

June – August 2021 and 2022 — Geisinger Autism & Developmental Medicine Summer Undergraduate Research Program Presentation Series. Developed by Juliann Savatt and Cora M. Taylor to expose undergraduate interns completing summer research internships to genetics and neurodevelopment.

MANUSCRIPT REVIEWER:

Journal of Genetic Counseling
Journal of Pediatrics
Genetics in Medicine
PEC Innovation
PLOS One

COMMITTEE MEMBERSHIPS:

National and International:

2020 – Present — National Society of Genetic Counselors, Annual Education Conference Abstract Committee
- Reviewer

2022 – Present — American Board of Genetic Counseling, Research Committee
- Member

2022 – Present — Global Alliance for Genomic Health (GA4GH) Regulator & Ethics Work Stream Clinical Data Sharing and Consent Group
- Co-chair

Regional and State:

2021 – Present — Pennsylvania Association of Genetic Counselors (PAGC), Genetic Services Committee
- Member

2022 – Present — New York, Mid Atlantic, and Caribbean (NYMAC). Pennsylvania Access Team.
- Member

2016 – 2017 Facing Our Risk for Cancer Empowered (FORCE) Danville Chapter
- Genetic Counselor Liaison

Institutional:

ClinGen:

2019 – Present Data Access, Protection, and Confidentiality (DAPC) Working Group
- Co-chair 2021-Present
- Member 2019-2021

2021 – Present Intellectual Disability/Autism Gene Curation Expert Panel (Miller Group)
- Member/Biocurator

2015 – Present Education, Coordination & Training Working Group
- Member

2015 – 2016 Phenotype Working Group
- Coordinator

2017 – 2020 Variant Interpretation Education Subgroup, Member, 2017-2020
- Member

2017 – 2018 Neurodevelopmental Disorders Clinical Domain Working Group and Sub-groups
- Co-coordinator

2019 – 2021 Consent & Disclosure Recommendations
- Member

Geisinger:

2018 – Present Genetic Counselor Education Committee
- Co-chair 2020-Present
- Member 2018-2020

2021 – Present Genetic Counseling Assistant Educational Task Team
- Member

2023 – Present Institutional Review Board
- Member

2017 Social Media Working Group
- Member

University of North Carolina:

2019 Capstone Reaccreditation Subcommittee
- Member