CURRICULUM VITAE

Kimberly (Splinter) LeBlanc, MS, CGC

Director, Undiagnosed Diseases Network (UDN) Coordinating Center
Department of Biomedical Informatics
Harvard Medical School
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EDUCATION

Undergraduate: Boston College, *Chestnut Hill, MA* *Sept 2008-May 2012*
B.A. in Human Development, Minor in Biology

Graduate: Stanford University, *Palo Alto, CA* *Sept 2012- June 2014*
M.S. in Human Genetics and Genetic Counseling
Master's Thesis: "Parents report impaired health-related quality of life in children with methylmalonic acidemia and perceive improvement following liver transplantation"
Advisor: Jonathan Bernstein, MD, PhD

CERTIFICATION AND LICENSURE

American Board of Genetic Counseling Certification *Feb 2015-Dec 2025*

Massachusetts Genetic Counselor Licensure *Jan 2015-Jan 2023*

PROFESSIONAL EXPERIENCE

Genetic Counselor/Project Coordinator *June 2014-Apr 2015*
Undiagnosed Diseases Network Coordinating Center
Boston Children's Hospital, Boston, MA

Genetic Counselor/Project Manager *Apr 2015-May 2017*
Undiagnosed Diseases Network Coordinating Center
Harvard Medical School, Boston, MA

Genetic Counselor *Aug 2016-Aug 2017*
PWNHealth
New York, NY

Guest Researcher *July 2015-Pres*
National Human Genome Research Institute
National Institutes of Health, Bethesda, MD

Associate Director of Research Operations *May 2017-July 2021*
Undiagnosed Diseases Network Coordinating Center
Harvard Medical School, Boston, MA

Assistant Professor *Oct 2020-Pres*
Department of Genetic Counseling, School of Health and Rehabilitation Sciences
MGH Institute of Health Professions, Boston, MA

Director *July 2021-Pres*
Undiagnosed Diseases Network Coordinating Center
Harvard Medical School, Boston, MA

MEMBERSHIPS IN PROFESSIONAL SOCIETIES

National Society of Genetic Counselors *Dec 2012-Pres*

PUBLICATIONS

Splinter, K., Niemi, A. K., Cox, R., Platt, J., Shah, M., Enns, G. M., Kasahara, M., & Bernstein, J. A. (2016). Impaired Health-Related Quality of Life in Children and Families Affected by Methylmalonic Acidemia. Journal of genetic counseling, 25(5), 936–944. https://doi.org/10.1007/s10897-015-9921-x

Splinter, K., Hull, S. C., Holm, I. A., McDonough, T. L., Wise, A. L., Ramoni, R. B., & Members of the Undiagnosed Diseases Network (2018). Implementing the Single Institutional Review Board Model: Lessons from the Undiagnosed Diseases Network. Clinical and translational science, 11(1), 28–31. https://doi.org/10.1111/cts.12512

Palmer, C., McConkie-Rosell, A., Holm, I. A., **LeBlanc, K.**, Sinsheimer, J. S., Briere, L. C., Dorrani, N., Herzog, M. R., Lincoln, S., Schoch, K., Spillmann, R. C., Brokamp, E., & Undiagnosed Diseases Network (2018). Understanding Adult Participant and Parent Empowerment Prior to Evaluation in the Undiagnosed Diseases Network. Journal of genetic counseling, 27(5), 1087–1101. https://doi.org/10.1007/s10897-018-0228-6

Walley, N. M., Pena, L., Hooper, S. R., Cope, H., Jiang, Y. H., McConkie-Rosell, A., Sanders, C., Schoch, K., Spillmann, R. C., Strong, K., McCray, A. T., Mazur, P., Esteves, C., **LeBlanc, K.**, Undiagnosed Diseases Network, Wise, A. L., & Shashi, V. (2018). Characteristics of undiagnosed diseases network applicants: implications for referring providers. BMC health services research, 18(1), 652. https://doi.org/10.1186/s12913-018-3458-2

Splinter, K., Adams, D. R., Bacino, C. A., Bellen, H. J., Bernstein, J. A., Cheatle-Jarvela, A. M., Eng, C. M., Esteves, C., Gahl, W. A., Hamid, R., Jacob, H. J., Kikani, B., Koeller, D. M., Kohane, I. S., Lee, B. H., Loscalzo, J., Luo, X., McCray, A. T., Metz, T. O., Mulvihill, J. J., … Undiagnosed Diseases Network (2018). Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. The New England Journal of Medicine, 379(22), 2131–2139. https://doi.org/10.1056/NEJMoa1714458

Macnamara, E. F., Schoch, K., Kelley, E. G., Fieg, E., Brokamp, E., Undiagnosed Diseases Network, Signer, R., **LeBlanc, K.**, McConkie-Rosell, A., & Palmer, C. (2019). Cases from the Undiagnosed

Diseases Network: The continued value of counseling skills in a new genomic era. Journal of Genetic Counseling, 28(2), 194–201. https://doi.org/10.1002/jgc4.1091

Studwell, C. M., Kelley, E. G., Undiagnosed Diseases Network, Sinsheimer, J. S., Palmer, C., & **LeBlanc, K.** (2021). Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. Journal of Genetic Counseling, 30(2), 439–447. https://doi.org/10.1002/jgc4.1329

Kobren, S. N., Baldridge, D., Velinder, M., Krier, J. B., **LeBlanc, K**., Esteves, C., Pusey, B. N., Züchner, S., Blue, E., Lee, H., Huang, A., Bastarache, L., Bican, A., Cogan, J., Marwaha, S., Alkelai, A., Murdock, D. R., Liu, P., Wegner, D. J., Paul, A. J., … Kohane, I. S. (2021). Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine. Advance online publication. https://doi.org/10.1038/s41436-020-01084-8

Yates, J., Gutiérrez-Sacristán, A., Jouhet, V., **LeBlanc, K.**, Esteves, C., DeSain, T. N., Benik, N., Stedman, J., Palmer, N., Mellon, G., Kohane, I., & Avillach, P. (2021). Finding commonalities in rare diseases through the undiagnosed diseases network. Journal of the American Medical Informatics Association: JAMIA. Advance online publication. https://doi.org/10.1093/jamia/ocab050

LeBlanc, K., Kelley, E. G., Nagy, A., Bater, J., Berro, T., McGuinness, M. A., Studwell, C., Undiagnosed Diseases Network, & Might, M. (2021). Rare disease patient matchmaking: development and outcomes of an internet case-finding strategy in the Undiagnosed Diseases Network. Orphanet Journal of Rare Diseases, 16(1), 210. https://doi.org/10.1186/s13023-021-01825-1

McCray, A. T., LeBlanc, K., & Undiagnosed Diseases Network (2021). Patients as Partners in Rare Disease Diagnosis and Research. The Yale journal of biology and medicine, 94(4), 687–692.

PRESENTATIONS

"Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia", Medical Genetics Grand Rounds, Stanford, CA, September 27, 2013

"Problems in unraveling- a cause of autism?", Human Genetics Journal Club, Stanford, CA, November 6, 2013

"Genetic counseling in the primary care setting", Primary Care Lunch Conference (Pierre Ezzi, MD, Agnes Jimenez, MD and Associates), Beverly, MA, March 13, 2014

"Health-related quality of life in children with methylmalonic acidemia", 2014 NSGC Annual Conference, New Orleans, LA, September 20, 2014

"Overview of the Undiagnosed Diseases Network & How to Apply", National Organization for Rare Disorders Webinar, June 3, 2016

"UDN Data Sharing", Brigham Genomic Medicine Meeting, Boston, MA, September 26, 2016.

"The Multidisciplinary Clinical and Research Evaluation of Patients with Rare and Undiagnosed Conditions", New England Regional Genetics Group Genetic Counselors' Conference, Danvers, MA, May 4, 2018

"Strategies for Rare Disease Diagnosis and Discovery – The Undiagnosed Diseases Network Experience", CAGC Annual Education Conference, St. John's Newfoundland, CA, October 10, 2018

"Searching for Answers in Rare Disease: The Undiagnosed Diseases Network", New York State Genetics Task Force Quarterly Meeting, New York, NY, February 27, 2019

"The role of genetic counselors in educating rare disease patients and families about genetic testing, data and diagnosis", World Orphan Drug Congress, Oxon Hill, MD, April 11, 2019

"Hyperindividualized Treatments" (Panel Discussion), Precision Medicine 2019, Boston, MA, June 18, 2019

"Rare Disease Diagnosis and Discovery – The Undiagnosed Diseases Network Experience", Brigham and Women's Hospital Center for Advanced Molecular Diagnostics Conference, Boston, MA, October 24, 2019

"Rare and Undiagnosed Conditions", The Human Genetics Association of New Jersey, New Brunswick, NJ, January 14, 2020

"Whole Genome Sequencing as a First-Tier Diagnostic Test. Are We Ready?", NSGC Virtual Annual Conference, November 19, 2020

"From Diagnosis to Driving Research" (Moderator), NORD RareLaunch Research Ready Virtual Workshop, April 28, 2021

ABSTRACTS

1. Green R.C., Lautenbach D.M., Kalia S.S., **Splinter K**., Beggs A.H. (2014) The MedSeq and BabySeq Projects: Clinical trials that explore the impact of genomic sequencing in adults and newborns. Presented at the Annual Meeting of the European Society of Human Genetics, Milan, Italy, May 31-June 3.
2. **Splinter K.**, Eastwood R., Might M. (2016) Accelerating Rare Disease Discovery: Does the Internet Hold the Key? Presented at the 2016 ACMG Annual Clinical Genetics Meeting, Tampa, Florida, March 8-12.

3. **Splinter K.,** Might M., Esteves C., Kohane I., Ramoni R. (2016) Helping Patients to Navigate the Clinical-Research Divide. Presented at the RDCRN Conference on Clinical Research for Rare Diseases, Washington, D.C., November 3.
4. Esteves C., **Splinter K.**, Ramoni R., Members of the UDN (2017) Diversity in Genomic Research: The Undiagnosed Diseases Network Experience. Presented at the 2017 ACMG Annual Clinical Genetics Meeting, Phoenix, Arizona, March 21-25.
5. **Splinter K.**, Eastwood R., Esteves C., Might M., Ramoni R., Members of the UDN (2017) Does This Patient Sound like Someone You Know? Piloting an Internet Case Finding Strategy in the Undiagnosed Diseases Network. Presented at the 2017 ACMG Annual Clinical Genetics Meeting, Phoenix, Arizona, March 21-25.
6. **LeBlanc K.**, Esteves C., Undiagnosed Diseases Network, Berro T., Kohane I., Wagner N., Might M. (2018) Implementing a Patient Research Navigator (PRN) Process in the Undiagnosed Diseases Network (UDN). Presented at the 2018 ACMG Annual Clinical Genetics Meeting, Charlotte, North Carolina, April 10-14.
7. Esteves C., **LeBlanc K.,** Undiagnosed Diseases Network. (2018) Leveraging Online Social Networks to Increase Engagement in Rare Disease Research. Presented at the 2018 ACMG Annual Clinical Genetics Meeting, Charlotte, North Carolina, April 10-14.
8. Studwell C., **LeBlanc K.**, Kelley E., Undiagnosed Diseases Network. (2019) Understanding the Practice of Genetic Result Communication to Extended Family Members by Participants in the Undiagnosed Diseases Network (UDN). Presented at the 2019 NSGC Annual Conference, Salt Lake City, Utah, November 5-8.
9. Frazier Z., Suslovitch V., Redfield S., **LeBlanc K**. (2021) Genetic Counselor Perceptions of Antisense Oligonucleotide Therapies. Presented at the 2021 NSGC Annual Conference, Virtual, September 22-26.
10. Rosenfeld L.E.*, **LeBlanc K.***, Nagy A, Ego B.K., Undiagnosed Diseases Network, McCray A.T. (2021) Participating in a national diagnostic research study: The patient perspective. Presented at the NORD Rare Diseases and Orphan Products Breakthrough Summit, Virtual, October 18-19. *Co first authors.

TEACHING

Courses

Advanced Clinical and Medical Genetics (GC-713), MGH Institute of Health Professions, Fall 2020

Advanced Clinical and Medical Genetics (GC-713), MGH Institute of Health Professions, Fall 2021

Course lectures

"Translational genomics", Genomics (Bio 374) class, Suffolk University, February 12, 2014

"Strategies for Rare Disease Diagnosis and Discovery – The Undiagnosed Diseases Network Experience," Genetic Counseling Seminar, Boston University School of Medicine, October 17, 2018

"Strategies for Rare Disease Diagnosis and Discovery – The Undiagnosed Diseases Network Experience," Genetic Counseling Seminar, MGH Institute of Health Professions, November 14, 2019

"Rare Disease Diagnosis," Project Success, Harvard Medical School, July 7, 2020

"The Undiagnosed Diseases Network," Genetic Counseling Seminar, Boston University School of Medicine, December 2, 2020

"Evaluation and Genetic Testing Approaches in the Undiagnosed Diseases Network," Laboratory Applications, Brandeis University, February 19, 2021

"Strategies for Rare Disease Diagnosis and Discovery – The Undiagnosed Diseases Network Experience," Project Success, Harvard Medical School, July 28, 2021

"Genetic Counselors in Emerging Specialties," Genetic Counseling in the 21st Century, Rutgers University, February 2, 2022

"Evaluation and Genetic Testing Approaches in the Undiagnosed Diseases Network," Laboratory Applications, Brandeis University, April 8, 2022

Supervision

Genetic counseling students, Boston Children's Hospital, Boston, MA *Oct 2014-Apr 2015*
Project Success student, Harvard Medical School, Boston, MA *August 2020*
Genetic counseling students, Harvard Medical School, Boston, MA *Mar 2021-Pres*

Capstone committees

Genetic counseling student, Boston University School of Medicine, Boston, MA *May 2018-May 2019*
Genetic counseling student, Boston University School of Medicine, Boston, MA *Mar 2020-May 2021*
Genetic counseling student, MGH Institute of Health Professions, Boston, MA *Jan 2021-May 2022*
Genetic counseling students (2), MGH Institute of Health Professions, Boston, MA. *Jan 2022-May 2024*

COMMITTEE AND VOLUNTEER SERVICE

Boston College *Oct 2014*
Panelist for "Careers in Healthcare" event

MCPHS University *Oct 2015*
Genetic Counseling Program Advisory Board Member

National Society of Genetic Counselors
New Student/Member SIG volunteer *Aug 2013-June 2016*
Abstract Workgroup member *April 2020-Pres*

Journal Reviewer

Journal of Genetic Counseling	*Mar 2017*
European Journal of Human Genetics	*Dec 2018*
Journal of Medical Genetics	*Sept 2021*

Veterans Affairs *Sept 2019-Pres*
Genomic Medicine Program Advisory Committee Member

National Organization for Rare Disorders *Jan 2020-Pres*
Undiagnosed Rare Disease Registry Advisory Committee Member