

Patrick Mullen, PhD

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Education

- Doctor of Philosophy, University of Vermont, Burlington, VT** August 2021
Neuroscience Graduate Program
Advisors: Dr. Christopher Francklyn and Dr. Alicia Ebert
- Bachelor of Science, St. Lawrence University, Canton, NY** May 2013
Major: Neuroscience

Work Experience

- Graduate Research Assistant** 2015 - 2021
Departments of Biochemistry and Biology, University of Vermont, Burlington, VT
Francklyn and Ebert laboratories
- Graduate Teaching Assistant** 2015 - 2021
Courses: Medical Neuroscience (GRMD 357), Biology (BCOR 11)
- Laboratory Director** Spring Semester 2021
Physical Therapy Neuroscience (NSCI 302)
Department of Neurological Sciences, University of Vermont, Burlington, VT
- Research Assistant** 2014 - 2015
Department of Biochemistry, Boston University School of Medicine, Boston, MA
- Research Assistant/ Fellow** 2011 – 2013
Department of Biology, St. Lawrence University, Canton, NY
- Summer Intern** Summer 2011
Cerebral Palsy Association of The North Country, Canton, NY
- Cardiology Technician** 2008 - 2010
Canton-Potsdam Hospital, Potsdam, NY

Teaching Experience (UVM)

- Lab Director, Physical Therapy Neuroscience (NSCI 302) Spring Semester 2021
Graduate Student Teaching Assistant, Biology (BCOR11) Fall 2020
Graduate Student Teaching Assistant, Medical Neuroscience (GRMD 357) 2016 - 2019
Medical Student Tutoring, GRMD 357 2016 - present
Specialty Tutor for Medical Anatomy and Physiology (GRMD 354) Spring 2020
Guest Lecturer, Neurobiology (BIOL 261) Spring 2019
Guest Lecturer, Cognitive Neuroscience (CSD 281) Fall 2018
Guest Lecturer, Physical Therapy Neuroscience Course (NSCI 302) Spring 2018
UVM Anatomical Connections Outreach Day Lecturer Spring 2017

Mentoring Experience

- *Mahafuza Aktar (UVM Neuroscience Graduate Program rotation student)* – During her rotation in the Francklyn Lab, Mahafuza was trained to culture and differentiate PC12 cells, and subsequently measure and analyze neurite outgrowth. Her data on the effect of histidinol treatment on NGF-induced neurite outgrowth in PC12 cells was published in a recent manuscript (Mullen et al. 2020).
- *Christian Fjeld (UVM class of 2017)* – Christian was trained on cell culture, transfection, cell differentiation, immunostaining, and immunofluorescence imaging. His work in the Francklyn Lab culminated in a poster presentation at the 2017 *IUBMB Focused Meeting on the Aminoacyl-tRNA Synthetases* and his data on eIF2 α phosphorylation and neurite outgrowth in differentiated PC12 cells transfected with mutant histidyl-tRNA synthetase was published in a recent manuscript (Mullen et al. 2020).
- *Pryce Patterson (UVM class of 2018)* – Pryce was trained on basic procedures in the Francklyn lab, including cell culture, western blotting, immunofluorescence imaging, and reverse transcriptase polymerase chain reaction (RT-PCR). Pryce's work led to receipt of a UVM APLE award, and completion of an Honors College Thesis.
- *DéJenaé See (UC Berkeley class of 2020)* – DéJenaé received a NIH funded summer neuroscience undergraduate research fellowship (SNURF) award and spent a summer in the Francklyn lab. I trained DéJenaé on basic procedures including cell culture, western blotting, immunofluorescence imaging, RT-PCR gel electrophoresis, qRT-PCR, and pharmacological treatment of patient fibroblasts. Her work culminated in presentation of her data at the UVM SNURF poster session and will be included in a manuscript that is in preparation.
- *Joshua Appelbaum (UVM class of 2020)* – I trained Joshua on basic procedures in the Francklyn Lab including cell culture, transfection, and cell imaging. Josh co-transfected HEK cells with plasmids encoding disease-associated premature stop codon mutations in AIMP2 and anticodon engineered “suppressor” tRNAs designed to promote premature stop codon readthrough. He performed western blotting and immunofluorescence imaging to analyze AIMP2 levels and assess the efficiency of stop codon readthrough. Josh received a UVM FUSE award and his work culminated in a presentation at the 2020 *UVM Neuroscience Behavior and Health Forum*.
- *Margot Kaminski (UVM class of 2020)* – I trained Margot to perform radioactivity-based enzymatic assay that we are using to investigate the consequences of AIMP2 mutations on tRNA charging in patient cell extracts.

Honors and Awards

<i>Rodney Parsons Anatomy and Neurobiology Award</i>	January 8, 2021
<i>UVM Graduate Alumni Dissertation Fellowship Award</i>	October 22, 2020
<i>Outstanding Teaching Assistant of the Year Award</i>	January 24, 2019
<i>UVM Neuroscience Behavior and Health Forum Outstanding Platform Talk</i>	January 10, 2021

Peer-Reviewed Publications

- * Indicates equal contribution
- ** Indicates undergraduate trainee co-author
- **Mullen P**, Abbott JA, Wellman T, Aktar M, Fjeld C**, Demeler B, Ebert AM, Francklyn CS. Neuropathy-associated histidyl-tRNA synthetase variants attenuate protein synthesis in vitro and disrupt axon outgrowth in developing zebrafish. *FEBS Journal*. 2021 Jan;288(1):142-159. doi: 10.1111/febs.15449. Epub 2020 Jul 6. PMID: 32543048; PMCID: PMC7736457.

- Galatolo D, Kuo ME, **Mullen P**, Meyer-Schuman R, Doccini S, Battini R, Lieto M, Tessa A, Filla A, Francklyn C, Antonellis A, Santorelli FM. Bi-allelic mutations in HARS1 severely impair histidyl-tRNA synthetase expression and enzymatic activity causing a novel multisystem ataxic syndrome. *Human Mutation*. 2020 Jul;41(7):1232-1237. doi: 10.1002/humu.24024. Epub 2020 Apr 29. PMID: 32333447; PMCID: PMC7323910.
- Royer-Bertrand B*, Tsouni P*, **Mullen P***, Campos Xavier B, Mittaz Crettol L, Lobrinus AJ, Ghika J, Baumgartner MR, Rivolta C, Superti-Furga A, Kuntzer T, Francklyn C, Tran C. Peripheral neuropathy and cognitive impairment associated with a novel monoallelic *HARS* variant. *Annals of Clinical and Translational Neurology*. 2019 May 24;6(6):1072-1080. doi: 10.1002/acn3.791. PMID: 31211171; PMCID: PMC6562026.
- Francklyn CS, **Mullen P**. Progress and challenges in aminoacyl-tRNA synthetase-based therapeutics. *Journal of Biological Chemistry*. 2019 Apr 5;294(14):5365-5385. doi: 10.1074/jbc.REV118.002956. Epub 2019 Jan 22. PMID: 30670594; PMCID: PMC6462538.
- Siekierska A, Stamberger H, Deconinck T, Oprescu SN, Partoens M, Zhang Y, Sourbron J, Adriaenssens E, **Mullen P**, Wiencek P**, Hardies K, Lee JS, Giong HK, Distelmaier F, Elpeleg O, Helbig KL, Hersh J, Isikay S, Jordan E, Karaca E, Kecskes A, Lupski JR, Kovacs-Nagy R, May P, Narayanan V, Pendziwiat M, Ramsey K, Rangasamy S, Shinde DN, Spiegel R, Timmerman V, von Spiczak S, Helbig I; C4RCD Research Group; AR working group of the EuroEPINOMICS RES Consortium, Weckhuysen S, Francklyn C, Antonellis A, de Witte P, De Jonghe P. Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. *Nature Communications*. 2019 Feb 12;10(1):708. doi: 10.1038/s41467-018-07953-w. PMID: 30755616; PMCID: PMC6372652.
- Chen CD, Zeldich E, Khodr C, Camara K, Tung TY, Lauder EC, **Mullen P**, Polanco TJ, Liu YY, Zeldich D, Xia W, Van Nostrand WE, Brown LE, Porco JA, Abraham CR. Small Molecule Amyloid- β Protein Precursor Processing Modulators Lower Amyloid- β Peptide Levels via cKit Signaling. *Journal of Alzheimer's Disease*. 2019;67(3):1089-1106. doi: 10.3233/JAD-180923. PMID: 30776010; PMCID: PMC6474660.
- Abraham CR, **Mullen P**, Tucker-Zhou T, Chen CD, Zeldich E. Klotho Is a Neuroprotective and Cognition-Enhancing Protein. *Vitamins and Hormones*. 2016;101:215-38. doi: 10.1016/bs.vh.2016.02.004. Epub 2016 Mar 22. PMID: 27125744.
- Heckman Heckman KL, DeCoteau W, Estevez A, Reed KJ, Costanzo W, Sanford D, Leiter JC, Clauss J, Knapp K, Gomez C, **Mullen P**, Rathbun E, Prime K, Marini J, Patchefsky J, Patchefsky AS, Hailstone RK, Erlichman JS. Custom cerium oxide nanoparticles protect against a free radical mediated autoimmune degenerative disease in the brain. *ACS Nano*. 2013 Dec 23;7(12):10582-96. doi: 10.1021/nn403743b. Epub 2013 Nov 27. PMID: 24266731.

Selected Poster Presentations

** Indicates undergraduate trainee co-author

- Mullen P**, Appelbaum J, ** See D, ** Kaminski M, ** Patterson P, ** Ebert A, Maroofian R, Lakhani S, Ross M, Francklyn C. Bi-allelic mutations in aminoacyl-tRNA synthetase interacting multifunctional protein 2 are linked to severe developmental encephalopathy and cause attenuation of protein synthesis and impaired cell cycle progression. *UVM Neuroscience Behavior and Health Forum*, 2020.
- See D, ** **Mullen P**, Francklyn C. Characterization of AIMP2 expression in rare presentation of developmental encephalopathy. Characterization of AIMP2 expression in rare presentation of developmental encephalopathy. *UVM Summer Neuroscience Undergraduate Research Fellowship Poster Session*, 2019.

- **Mullen P**, Fjeld C,** Abbott J, Antonellis A, Ebert A, Francklyn C. Novel CMT associated HARS variants disrupt aminoacylation and neurite outgrowth. *IUBMB Focused Meeting on the Aminoacyl-tRNA Synthetases*, 2017.
- Chen C, **Mullen P**, Zeldich E, Brown L, Porco J, Abraham C. Deciphering the Mechanisms by which Inhibiting APP Dimerization Reduced ABeta Production. *Massachusetts Alzheimer's Disease Research Center Annual Scientific Poster Symposium*, 2015.
- DeCoteau W, Heckman K, **Mullen P**, Lipps J, Erlichman J. The Biological Effects of Nanoceria in a Model of Multiple Sclerosis. *Society for Neuroscience*, 2013.