Melissa Ann Kelly

Previous Names: Melissa Ann Diederichs, Melissa Ann Wilk (*she/her*)
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EDUCATION

Medical College of Wisconsin, Milwaukee, Wisconsin
PhD, Cell & Developmental Biology
Concentrations: Neuroscience, Vision Science
January 2017; GPA 3.84/4.0
Dissertation: Using Imaging and Genetics to Characterize Visual System Structure Across the Pigmentation Spectrum

Carroll University, Waukesha, Wisconsin
BS, Human Biology
Concentrations: Physiology, Neuroscience, Psychology
May 2011; GPA 3.877/4.0

CERTIFICATION/LICENSURE

American Board of Bioanalysis (ABB), St. Louis, Missouri – Certification ID#: 20213064
High-Complexity Clinical Laboratory Director (HCLD)
Specialty: Molecular Diagnostics
Certification Date: December 2022
Clinical Consultant (CC)
Certification Date: August 2023

New York State Department of Health, Albany, New York – CQ Code: KELLM12
Certificate of Qualification
Category: Genetic Testing (limited to molecular); Oncology (Molecular & Cellular Tumor Markers)
Effective Date: December 22, 2023 (*initial*)
Expiration Date: December 22, 2025

MEMBERSHIPS

Association for Molecular Pathology (AMP)
Member (2024 – present)

American Society of Human Genetics (ASHG)
Member (2023 – present)

American of Medical Genetics and Genomics (ACMG)
Affiliate Member (2022 – present)

American Association of Bioanalysts (AAB)
Member (2021 – present)

National Honor Society of Phi Kappa Phi
Member (2010 – present)

Medical Genome Initiative
Steering Committee Member (2022 – 2023)

EMPLOYMENT

The Jackson Laboratory for Genomic Medicine, Farmington, Connecticut
Clinical Laboratory Director (June 2023 – present)

- Ensure the laboratory is staffed with qualified and trained personnel.
- Provide laboratory conditions that are appropriate for the testing performed by effecting a safe environment which protects employees from physical, chemical, and biological hazards.
- Monitor test methods are correct, accurate, appropriately verified and consistent with standard operating procedures (SOPs) before patient reports are issued to authorized ordering healthcare providers.
- Review laboratory personnel performing tests such that methods are executed in accordance with SOPs and quality systems throughout pre-analytical, analytical, and post-analytical phases of testing.
- Establish and lead processes of periodic review to ensure the JAX Advanced Precision Medicine

- Laboratory test catalogue, for both clinical and RUO offerings, are up-to-date and using the most appropriate, cost-effective technologies to provide accurate results to customers.
- Collaborate with the Manager of Clinical Quality and Compliance to enforce a quality-centric service that monitors staff competency to process specimens, perform test procedures, and report test results promptly and proficiently, and identify needs for remedial training or continuing education to improve skills when necessary.
- Work with the Director of CLIA Research & Development, JAX and external scientific collaborators, laboratory customers, and Clinical Genomics business leaders to identify new assay and business opportunities.
- Work with the R&D organization to define assays and formulate development plans that deliver optimized assays appropriate for clinical validation, including any informatics requirements.
- Lead all aspects of assay validation in the clinical laboratory, including quality systems compliance, and sign-off on the completed test to be added to the JAX Advanced Precision Medicine Laboratory test catalogue.

Associate Director of Dry Lab & Quality (April 2023 – May 2023)

- Supervision of a team of clinical analysts and bioinformaticists.
- Review of clinical genomics data and sign-out of clinical reports.
- Strategic planning for CLIA lab offerings.
- Validation of new or modified laboratory developed tests.
- Coordination and implementation of compliance and accreditation activities.

HudsonAlpha Clinical Services Lab, LLC, Huntsville, Alabama

Assistant Director (January 2023 – September 2023)

- Review of clinical genomics data, including array-based methods, whole genome sequencing, and Sanger sequencing.
- Sign-out of clinical reports.
- Supervision of a team of clinical analysts.
- Analysis of whole genome sequencing data for the diagnosis of rare disease and array data for disease risk screening.
- Validation of new or modified laboratory developed tests.
- Tracking of projects, samples, data, and reporting in the laboratory.
- Client development and customer relations for whole genome sequencing.
- Coordination and implementation of compliance and accreditation activities.
- Liaison for the development of clinical software applications.

Clinical Genomic Variant Scientist/Clinical Genomics Supervisor (March 2019 – December 2022)

- Supervision of a team of clinical analysts.
- Analysis of whole genome sequencing data for the diagnosis of rare disease and array data for disease risk screening.
- Data processing via validated clinical pipelines.
- Validation of new or modified laboratory developed tests.
- Tracking of projects, samples, data, and reporting in the laboratory.
- Client development and customer relations for whole genome sequencing.
- Coordination and implementation of compliance and accreditation activities.
- Liaison for the development of clinical software applications.

HudsonAlpha Institute for Biotechnology, Huntsville, Alabama

Lab Supervisor, Software Development & Informatics (May 2018 – March 2019)

- Analysis of whole genome sequencing data for the identification of disease-causing, modifier, and pharmacogenomic variants.
- Participation in clinical genome analysis through the HudsonAlpha Clinical Services Lab, LLC, for patients from the Undiagnosed Diseases Network.
- Management of projects, tracking samples and data, and reporting of outcomes.
- Stakeholder relations for software development team.
- Preparation of grant and Institutional Review Board proposals.
- Supervision of day-to-day research activities.

Post-Doctoral Fellow, Worthey & Jacob Labs (November 2016 – May 2018)

- Analysis of whole genome sequencing data for the identification of disease-causing, modifier, and pharmacogenomic variants.

TEACHING EXPERIENCE

Medical College of Wisconsin, Milwaukee, Wisconsin

Lecturer
- Fundamentals of Neuroscience (Spring 2015, Spring 2016)

Teaching Assistant
- Fundamentals of Neuroscience (Spring 2013)

Carroll University, Waukesha, Wisconsin

Adjunct Instructor
- Neuroscience Foundations for Occupational Therapy Practice (Spring 2016)
- Neuroscience Foundations for Occupational Therapy Practice Laboratory (Spring 2016)
- Introduction to Human Anatomy & Physiology I Laboratory (Fall 2014)
- Human Physiology Laboratory (Spring 2013, Summer 2013, Spring 2014)

RESEARCH EXPERIENCE

HudsonAlpha Institute for Biotechnology, Huntsville, Alabama

Post-Doctoral Fellow (November 2016 – May 2018)

Advisors: Elizabeth Worthey, PhD & Howard Jacob, PhD
- Identification of genetic modifiers and relevant pharmacogenomic variants in cystic fibrosis, pulmonary arterial hypertension, and Duchenne muscular dystrophy; genetics of rare disease.

Medical College of Wisconsin, Milwaukee, Wisconsin

PhD Candidate (July 2011 – October 2016)

Advisor: Joseph Carroll, PhD, Department of Ophthalmology
- Dissertation research on normal retinal morphology; visual deficits and their retinal correlates in albinism; retino-cortical relationships in albinism using high-resolution retinal imaging and function magnetic resonance imaging (fMRI); genotype-phenotype relationships in retinal pigment; and assessment of melanin and its effect on high-resolution retinal imaging.

PROFESSIONAL ACTIVITIES & SERVICE

» Graduate Student Association (GSA), Medical College of Wisconsin, Milwaukee, Wisconsin
- President (2015 - 2016)
- Treasurer (2013 - 2015)
- Neuroscience Representative (2012 - 2013)

» National Organization for Albinism & Hypopigmentation (NOAH) – Event Organizer; Milwaukee Mini-Conference (Summer 2015)

» Cold Spring Harbor Laboratory, Lloyd Harbor, New York – Course Attendee; Vision: A Platform for Linking Circuits, Perception, and Behavior (June 2013)

GRANTS & AWARDS

» Outstanding Dissertation Award, Medical College of Wisconsin, Milwaukee, Wisconsin (2017)

» Knights Templar Eye Foundation Travel Grant, Association for Research in Vision & Ophthalmology (2016)

» Graduate Student Association Travel Award, Medical College of Wisconsin, Milwaukee, Wisconsin (2013 – 2016)

» Vision Science Training Grant, National Institutes of Health (2012 – 2015)

PUBLICATIONS *(NCBI Bibliography)*

Nakouzi G, Bick D, Cochran M, Greve V, Lyon E, **Kelly MA**, East K, Kelley W. (In preparation). Clinical Genome Sequencing Cases: The HudsonAlpha Institute for Biotechnology Collection. In Rifai N, Chiu RWK, Young I, Burnham CAD, Wittwer CT (Eds.), *Tietz Textbook of Laboratory Medicine* (7th Edition). Elsevier.

Rehm HL, Alaimo JT, Aradhya S, Bayrak-Toydemir P, Best H, Brandon R, Buchan JG, Chao EC, Chen E, Clifford J, Cohen ASA, Conlin LK, Das S, Davis KW, Gaudio DD, Del Viso F, DiVincenzo C, Eisenberg M, Guidugli L, Hammer MB, Harrison SM, Hatchell KE, Dyer LH, Hoang LU, Holt JM, Jobanputra V, Karbassi ID, Kearney HM, **Kelly MA**, Kelly JM, Kluge ML, Komala T, Kruszka P, Lau L, Lebo MS, Marshall CR, McKnight D, McWalter K, Meng Y, Nagan N, Neckelmann CS, Neerman N, Niu Z, Paolillo VK, Paolucci SA, Perry D, Pesaran T, Radtke K, Rasmussen KJ, Retterer K, Saunders CJ, Spiteri E, Stanley C, Szuto A, Taft RJ, Thiffault I, Thomas BC, Thomas-Wilson A, Thorpe E, Tidwell TJ, Towne MC, Zouk H; Medical Genome Initiative. The landscape of reported VUS in multi-gene panel and genomic testing: Time for a change. Genet Med. 2023 Jul 30:100947. doi: 10.1016/j.gim.2023.100947. Epub ahead of print. PMID: 37534744.

Felker SA, Lawlor JMJ, Hiatt SM, Thompson ML, Latner DR, Finnila CR, Bowling KM, Bonnstetter ZT, Bonini KE, Kelly NR, Kelley WV, Hurst ACE, Rashid S, **Kelly MA**, Nakouzi G, Hendon LG, Bebin EM, Kenny EE, Cooper GM. Poison exon annotations improve the yield of clinically relevant variants in genomic diagnostic testing.

Genet Med. 2023 May 6;25(8):100884. doi: 10.1016/j.gim.2023.100884. [Epub ahead of print] PubMed PMID: 37161864.

Rehm HL, Alaimo JT, Aradhya S, Bayrak-Toydemir P, Best H, Brandon R, Buchan JG, Chao EC, Chen E, Clifford J, Cohen AS, Conlin LK, Das S, Davis KW, del Gaudio D, Del Viso F, DiVincenzo C, Eisenberg M, Guidugli L, Hammer MB, Harrison SM, Hatchell KE, Dyer LH, Hoang LU, Holt JM, Jobanputra V, Karbassi ID, Kearney HM, **Kelly MA**, Kelly JM, Kluge ML, Komala T, Kruszka P, Lau L, Lebo MS, Marshall CR, McKnight D, McWalter K, Meng Y, Nagan N, Neckelmann CS, Neerman N, Niu Z, Paolillo VK, Paolucci SA, Perry D, Pesaran T, Radtke K, Rasmussen KJ, Retterer K, Saunders CJ, Spiteri E, Stanley CM, Szuto A, Taft RJ, Thiffault I, Thomas BC, Thomas-Wilson A, Thorpe E, Tidwell TJ, Towne MC, Zouk H. The landscape of reported VUS in multi-gene panel and genomic testing: Time for a change. medRxiv 2022.09.21.22279949; doi: 10.1101/2022.09.21.22279949

Hiatt SM, Trajkova S, Sebastiano MR, Partridge EC, Abidi FE, Anderson A, Ansar M, Antonarakis SE, Azadi A, Bachmann-Gagescu R, Bartuli A, Benech C, Berkowitz JL, Betti MJ, Brusco A, Cannon A, Caron G, Chen Y, Cochran ME, Coleman TF, Crenshaw MM, Cuisset L, Curry CJ, Darvish H, Demirdas S, Descartes M, Douglas J, Dyment DA, Elloumi HZ, Ermondi G, Faoucher M, Farrow EG, Felker SA, Fisher H, Hurst ACE, Joset P, **Kelly MA**, Kmoch S, Leadem BR, Lyons MJ, Macchiaiolo M, Magner M, Mandrile G, Mattioli F, McEown M, Meadows SK, Medne L, Meeks NJL, Montgomery S, Napier MP, Natowicz M, Newberry KM, Niceta M, Noskova L, Nowak CB, Noyes AG, Osmond M, Prijoles EJ, Pugh J, Pullano V, Quélin C, Rahimi-Aliabadi S, Rauch A, Redon S, Reymond A, Schwager CR, Sellars EA, Scheuerle AE, Shukarova-Angelovska E, Skraban C, Stolerman E, Sullivan BR, Tartaglia M, Thiffault I, Uguen K, Umaña LA, van Bever Y, van der Crabben SN, van Slegtenhorst MA, Waisfisz Q, Washington C, Rodan LH, Myers RM, Cooper GM. Deleterious, protein-altering variants in the transcriptional coregulator ZMYM3 in 27 individuals with a neurodevelopmental delay phenotype. Am J Hum Genet. 2022 Dec 21:S0002-9297(22)00541-9. doi: 10.1016/j.ajhg.2022.12.007. Epub ahead of print. PMID: 36586412.

Felker SA, Lawlor JM, Hiatt SM, Thompson ML, Latner DR, Finnila CR, Bowling KM, Bonnstetter ZT, Bonini KE, Kelly NR, Kelley WV, Hurst AC, **Kelly MA**, Nakouzi G, Hendon LG, Bebin EM, Kenny EE, Cooper GM. Poison exon annotations improve the yield of clinically relevant variants in genomic diagnostic testing. bioRxiv. 2023 Jan 13;. doi: 10.1101/2023.01.12.523654. PubMed PMID: 36711854; PubMed Central PMCID: PMC9882217.

Banka S, Bennington A, Baker MJ, Rijckmans E, Clemente GD, Ansor NM, Sito H, Prasad P, Anyane-Yeboa K, Badalato L, Dimitrov B, Fitzpatrick D, Hurst ACE, Jansen AC, **Kelly MA**, Krantz I, Rieubland C, Ross M, Rudy NL, Sanz J, Stouffs K, Xu ZL, Malliri A, Kazanietz MG, Millard TH. Activating RAC1 variants in the switch II region cause a developmental syndrome and alter neuronal morphology. Brain. 2022 Dec 19;145(12):4232-4245. doi: 10.1093/brain/awac049. PubMed PMID: 35139179; PubMed Central PMCID: PMC9762944.

Bowling KM, Thompson ML, Kelly MA, Scollon S, Slavotinek AM, Powell BC, Kirmse BM, Hendon LG, Brothers KB, Korf BR, Cooper GM, Greally JM, Hurst ACE. Return of non-ACMG recommended incidental genetic findings to pediatric patients: considerations and opportunities from experiences in genomic sequencing. Genome Med. 2022 Nov 21;14(1):131. doi: 10.1186/s13073-022-01139-2. PMID: 36414972; PMCID: PMC9682742.

Cochran M, East K, Greve V, **Kelly M**, Kelley W, Moore T, Myers RM, Odom K, Schroeder M, Bick D. A study of elective genome sequencing and pharmacogenetic testing in an unselected population. Mol Genet Genomic Med. 2021;00:e1766. doi: 10.1002/mgg3.1766. PubMed PMID: 34313030.

Holt JM, **Kelly M**, Sundlof B, Nakouzi G, Bick D, Lyon E. Reducing Sanger confirmation testing through false positive prediction algorithms. Genet Med. 2021 Jul;23(7):1255-1262. doi: 10.1038/s41436-021-01148-3. PubMed PMID: 33767343.

Woertz EN, **Wilk MA**, Duwell EJ, Mathis JR, Carroll J, DeYoe EA. The relationship between retinal cone density and cortical magnification in human albinism. J Vis. 2020 Jun 3;20(6):10. doi: 10.1167/jov.20.6.10. PubMed PMID: 32543650.

Wilk MA, Braun AT, Farrell PM, Laxova A, Brown DM, Holt JM, Birch CL, Sosonkina N, Wilk BM, Worthey EA. Applying whole-genome sequencing in relation to phenotype and outcomes in siblings with cystic fibrosis. Cold Spring Harb Mol Case Stud. 2020 Feb;6(1). doi: 10.1101/mcs.a004531. PubMed PMID: 32014855; PubMed Central PMCID: PMC6996517.

Hurst AC, **Wilk MA**, Holt JM, Lyon E, Bick D. Clinical Diagnosis through Whole-Genome Sequencing. Clin Chem. 2020 January; 66(1):261–262. doi: 10.1093/clinchem.2019.310128.

Holt JM, Wilk B, Birch CL, Brown DM, Gajapathy M, Moss AC, Sosonkina N, **Wilk MA**, Anderson JA, Harris JM, Kelly JM, Shaterferdosian F, Uno-Antonison AE, Weborg A, Worthey EA. VarSight: prioritizing clinically reported variants with binary classification algorithms. BMC Bioinformatics. 2019 Oct 15;20(1):496. doi: 10.1186/s12859-019-3026-8. PubMed PMID: 31615419; PubMed Central PMCID: PMC6792253.

Holt JM, Birch CL, Brown DM, Gajapathy M, Sosonkina N, Wilk B, **Wilk MA**, Spillman RC, Strong N, Lee H, Huang AY, Bonner D, Kohler JN, Macnamara EF, Undiagnosed Diseases Network, Nelson SF, Shashi V, Worthey EA. Identification of Pathogenic Structural Variants in Rare Disease Patients through Genome Sequencing. bioRxiv. 2019 May. doi: 10.1101/627661.

Lee DJ, Woertz EN, Visotcky A, **Wilk MA**, Heitkotter H, Linderman RE, Tarima S, Summers CG, Brooks BP, Brilliant MH, Antony BJ, Lujan BJ, Carroll J. The Henle Fiber Layer in Albinism: Comparison to Normal and Relationship to Outer Nuclear Layer Thickness and Foveal Cone Density. Invest Ophthalmol Vis Sci. 2018 Nov 1;59(13):5336-5348. doi: 10.1167/iovs.18-24145. PubMed PMID: 30398625; PubMed Central PMCID: PMC6219160.

Huckenpahler A, **Wilk M**, Link B, Carroll J, Collery R. Repeatability and Reproducibility of In Vivo Cone Density Measurements in the Adult Zebrafish Retina. Adv Exp Med Biol. 2018;1074:151-156. doi: 10.1007/978-3-319-75402-4_19. PubMed PMID: 29721939; PubMed Central PMCID: PMC6363109.

Wilk MA, Huckenpahler AL, Collery RF, Link BA, Carroll J. The Effect of Retinal Melanin on Optical Coherence Tomography Images. Transl Vis Sci Technol. 2017 Apr;6(2):8. doi: 10.1167/tvst.6.2.8. eCollection 2017 Apr. PubMed PMID: 28392975; PubMed Central PMCID: PMC5381330.

Wilk MA, Dubis AM, Cooper RF, Summerfelt P, Dubra A, Carroll J. Assessing the spatial relationship between fixation and foveal specializations. Vision Res. 2017 Mar;132:53-61. doi: 10.1016/j.visres.2016.05.001. Epub 2016 Jun 18. PubMed PMID: 27286921; PubMed Central PMCID: PMC5164985.

Huckenpahler AL, **Wilk MA**, Cooper RF, Moehring F, Link BA, Carroll J, Collery RF. Imaging the adult zebrafish cone mosaic using optical coherence tomography-CORRIGENDUM. Vis Neurosci. 2017 Jan;34:E005. doi: 10.1017/S0952523817000025. PubMed PMID: 28965523.

Wilk MA, Wilk BM, Langlo CS, Cooper RF, Carroll J. Evaluating outer segment length as a surrogate measure of peak foveal cone density. Vision Res. 2017 Jan;130:57-66. doi: 10.1016/j.visres.2016.10.012. Epub 2016 Dec 2. PubMed PMID: 27887888; PubMed Central PMCID: PMC5186335.

Patterson EJ, **Wilk M**, Langlo CS, Kasilian M, Ring M, Hufnagel RB, Dubis AM, Tee JJ, Kalitzeos A, Gardner JC, Ahmed ZM, Sisk RA, Larsen M, Sjoberg S, Connor TB, Dubra A, Neitz J, Hardcastle AJ, Neitz M, Michaelides M, Carroll J. Cone Photoreceptor Structure in Patients With X-Linked Cone Dysfunction and Red-Green Color Vision Deficiency. Invest Ophthalmol Vis Sci. 2016 Jul 1;57(8):3853-63. doi: 10.1167/iovs.16-19608. PubMed PMID: 27447086; PubMed Central PMCID: PMC4968428.

Cooper RF, **Wilk MA**, Tarima S, Carroll J. Evaluating Descriptive Metrics of the Human Cone Mosaic. Invest Ophthalmol Vis Sci. 2016 Jun 1;57(7):2992-3001. doi: 10.1167/iovs.16-19072. PubMed PMID: 27273598; PubMed Central PMCID: PMC4898203.

Razeen MM, Cooper RF, Langlo CS, Goldberg MR, **Wilk MA**, Han DP, Connor TB Jr, Fishman GA, Collison FT, Sulai YN, Dubra A, Carroll J, Stepien KE. Correlating Photoreceptor Mosaic Structure to Clinical Findings in Stargardt Disease. Transl Vis Sci Technol. 2016 Mar;5(2):6. doi: 10.1167/tvst.5.2.6. eCollection 2016 Mar. PubMed PMID: 26981328; PubMed Central PMCID: PMC4790429.

Huckenpahler AL, **Wilk MA**, Cooper RF, Moehring F, Link BA, Carroll J, Collery RF. Imaging the adult zebrafish cone mosaic using optical coherence tomography. Vis Neurosci. 2016 Jan;33:E011. doi: 10.1017/S0952523816000092. PubMed PMID: 28177275; PubMed Central PMCID: PMC5659228.

McCafferty BK, **Wilk MA**, McAllister JT, Stepien KE, Dubis AM, Brilliant MH, Anderson JL, Carroll J, Summers CG. Clinical Insights Into Foveal Morphology in Albinism. J Pediatr Ophthalmol Strabismus. 2015 May-Jun;52(3):167-72. doi: 10.3928/01913913-20150427-06. PubMed PMID: 26053207; PubMed Central PMCID: PMC4948980.

Wilk MA, Dubra A, Curcio CA, Brilliant MH, Summers CG, Carroll J. Author response: relationship between foveal cone specialization and pit morphology in albinism. Invest Ophthalmol Vis Sci. 2014 Sep 18;55(9):5923. doi: 10.1167/iovs.14-15470. PubMed PMID: 25237180; PubMed Central PMCID: PMC4687793.

Liu BS, Tarima S, Visotcky A, Pechauer A, Cooper RF, Landsem L, **Wilk MA**, Godara P, Makhijani V, Sulai YN, Syed N, Yasumura G, Garg AK, Pennesi ME, Lujan BJ, Dubra A, Duncan JL, Carroll J. The reliability of parafoveal cone density measurements. Br J Ophthalmol. 2014 Aug;98(8):1126-31. doi: 10.1136/bjophthalmol-2013-304823. Epub 2014 May 22. PubMed PMID: 24855115; PubMed Central PMCID: PMC4112420.

Wilk MA, McAllister JT, Cooper RF, Dubis AM, Patitucci TN, Summerfelt P, Anderson JL, Stepien KE, Costakos DM, Connor TB Jr, Wirostko WJ, Chiang PW, Dubra A, Curcio CA, Brilliant MH, Summers CG, Carroll J. Relationship between foveal cone specialization and pit morphology in albinism. Invest Ophthalmol Vis Sci. 2014 May 20;55(7):4186-98. doi: 10.1167/iovs.13-13217. PubMed PMID: 24845642; PubMed Central PMCID: PMC4098060.

Godara P, Cooper RF, Sergouniotis PI, **Diederichs MA**, Streb MR, Genead MA, McAnany JJ, Webster AR, Moore AT, Dubis AM, Neitz M, Dubra A, Stone EM, Fishman GA, Han DP, Michaelides M, Carroll J. Assessing retinal structure in complete congenital stationary night blindness and Oguchi disease. Am J Ophthalmol. 2012 Dec;154(6):987-1001.e1. doi: 10.1016/j.ajo.2012.06.003. Epub 2012 Sep 7. PubMed PMID: 22959359; PubMed Central PMCID: PMC3498541.

VOLUNTEER ACTIVITIES

Cattyshack, Inc., Huntsville, Alabama

Foster Liaison (July 2022 – present)

Forgotten Felines, Huntsville, Alabama

Adoption Center Volunteer/Volunteer Coordinator (July 2021 – present)