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EDUCATION

- 1992 -1993 Postdoctoral Fellow; Department of Neurobiology and Anatomy/Sanders Brown Center on Aging; University of Kentucky;
- 1992 Ph.D. Cell Biology, Neurobiology, and Anatomy; Loyola University of Chicago
- 1981 B.S. in Physical Therapy (Magna Cum Laude); The University of Illinois - Chicago

ACADEMIC APPOINTMENTS

- 2017- present Associate Professor, Department of Anatomy & Cell Biology, RMC, Rush University, Chicago, IL.
- 2017- present Conjoint Associate Professor, Department of Neurological Sciences, RMC, Rush University, Chicago, IL.
- 2017 - 2023 Associate Professor, Graduate College, Rush University.
- 2015- 2018 Conjoint Assistant Professor, Department of Occupational Therapy, College of Health Sciences, Rush University, Chicago, IL.
- 2015- 2017 Conjoint Assistant Professor, Department of Neurological Sciences, Rush University, Chicago, IL.
- 2014- 2017 Assistant Professor, Graduate College, Rush University.
- 2009 - 2017 Assistant Professor, Department of Anatomy and Cell Biology, Rush University College of Medicine, Chicago, IL.
- 2008- 2009 Part-Time Faculty Laboratory Instructor, Human Gross Anatomy for First Year Medical students in all blocks. Rush University Medical College, Department of Anatomy and Cell Biology, Chicago, IL

- 1993- 1996 Associate Member, Graduate Faculty, Physical Therapy Program, University of Kentucky, Lexington, KY.
- 1992 - 1996 Assistant Professor, Division of Physical Therapy, Department of Clinical Sciences, University of Kentucky Medical Center, Lexington, KY.
- 1992 - 1996 Core Faculty Member, Interdisciplinary Human Development Institute Leadership Development and Training Program; University of Kentucky, Lexington, KY.

Employment/Clinical Experience

- 1996–2009 Private Physical Therapy Practitioner; Chicago, IL
- 1985-1986 Pediatric Physical Therapy Supervisor; Cook County Hospital, Chicago, IL
- 1983-1985 Pediatric Physical Therapist and Research Physical Therapist; Shriners Hospital, Chicago, IL
- 1981-1983 Staff Physical Therapist; Cook County Hospital, Chicago, IL

Licensure/ Certifications

Professional Licensure: IL State PT License # 070-003541

Early Intervention Professional Specialist; Illinois License # JO94340100P
Illinois Department of Human Services

Honors, Awards, and Fellowships

- 2024 Rush University, Rush Imaging Research Center Pilot Grant Award
- 2021-2023 Huntington's Disease Society of America Human Biology Fellowship Award
- 2017 Rush University Excellence Award in Education
- 2017 Member of the Mark Lepper Society of Teachers for Teaching Excellence
- 2016 Rush Schweppe Translational Science Consortium Pilot Grant Award
- 2014 Cohn Research Fellowship Award, Rush University
- 2012 Young Investigator Award, National Fragile X Foundation
- 1992 University of Kentucky Postdoctoral Fellowship Award for Women in Science
- 1990 Society for Neuroscience (Chicago Chapter) Graduate Student Symposium Award
- 1990-1991 Arthur J. Schmitt Fellowship Awards
- 1989 Society for Sigma Xi Research grant recipient
- 1986-1990 Recipient of Loyola University Basic Science Fellowships
- 1981 Graduated Magna Cum Laude, University of Illinois-Chicago
- 1977-1981 Dean's List University of Illinois
- 1978 Alpha Lambda Delta Honor Society
- 1977 Illinois State Scholar

International, National and Regional Scientific Society Memberships

2025-	International Fragile X Premutation Registry (IFXPR) Executive Governance Committee
2024-	International Ataxia Global Initiative; Digital Outcomes and Imaging Outcomes Working Group member
2023-	Parkinson's Study Group, Professional Member
2023-	Huntington's Study Group, Professional Member; Digital Outcomes Working Group Member
2019- 2022	President, Chicago Chapter of the Society for Neuroscience
2017-2022	Executive Committee of the Chicago Chapter of the Society for Neuroscience
2017- 2018	The International Society for the Measurement of Physical Behaviour
2014-	International Society for Posture and Gait Research
2010- 2014	Gait and Clinical Movement Analysis Society
2009-	Movement Disorder Society
2009-	American Association of Anatomists
2009- 2012	American Association of Clinical Anatomists
1991-	American Physical Therapy Association: Neurology, Pediatric and Research section member
1998-	Society for Neuroscience

TEACHING

Rush University

- 2025- present Basic Science Educator for Neuroscience, Rush Medical College; Brain, Behavior, and Cognition 1 and 2 (RMD 565); Health Across the Life Span (RMD 567); Oncology (RMD 577); Complex Cases (RMD 569); Movement & Mechanics (RMD 564); Responsibilities include Creation, Modification and Delivery of Basic and Clinical Science Education in Neurobiology/Neuroscience Content throughout the Preclerkship (M1/M2) curriculum. *Contact hours: 52 Basic Science teaching sessions, 8 Classic Neurological Clinical Case workshop hours; 16 Neuroanatomy laboratory hours; 20 laboratory and practical exam set up hours; 20% FTE effort.*
- 2015- present Course Director: Introduction to Neurobiology ANA 500 (3 credit hours); This course is for doctoral students in the Rush University College of Nursing's DNP Anesthesia program AND graduate students in Rush's Department of Anatomy and Cell Biology and Rush Graduate College (until 2022). Responsibilities include 1) Content and Objective development, 2) Delivery of all Lectures and Labs, 3) Organization and dissemination of all Blackboard/Canvas content, 4) Writing/grading of all examinations, quizzes, clinical case group presentations, discussion posts, and scientific research paper assignments, Preparation/administration of gross anatomy practical exams; *Contact hours: 30*

Lecture hours; 10 Clinical Case group presentation hours; 20 Laboratory hours; 18% FTE effort

2024	Guest Lecturer (GCC 516); “Foundations in Biomedical Sciences”; Control of Posture (3 hours).
2023-2024	Guest Lecturer (GCC 652); “The Changing Nervous System”; Gross Brain Topography 1 (3 hours)
2017- 2022	Course Director: Neuroscience for Basic and Clinical Applications GCC 650. Core cognate class for students in the Neuro track of the Rush University Integrated Biomedical Sciences MS and PhD Program. <i>Lecture hours: 30; Laboratory hours: 10; 10% effort</i>
2015- 2018	Co-Block Administrator (with Dr. Thomas Hoepfner); M1 CNS/Head and Neck Curriculum Block RMD 516. Responsibilities included: 1) Content and objective development and delivery, 2) Organization and dissemination of all Blackboard content, 3) Invitation and correspondence with Rush Clinical faculty lecturers, 4) Administration and meetings with Discipline directors, 5) Writing/grading of examinations, quizzes and research assignments, 6) Preparation and administration of gross anatomy Head and Neck and Neuroanatomy practical exams. <i>Content Hours: Lecture hours: 11; Gross anatomy lab hours: 27; 18 Neuro case application workshop hours; Small group i-human case session hours: 8</i>
2017- present	Research Faculty, OCC 582 Research Methods and Evidence-Based Practice, Department of Occupational Therapy; Covers quantitative research design, data analysis strategies, and the incorporation of evidence-based practice to clinical practice; presently a research mentor for doctoral students in Occupational Therapy.
2016	Proseminar in Neurobiology; GCC 698-1; Rush Graduate College; “Neurophysiology of Gait” and Discussion; 3 contact hours.
2011- 2017	Core Faculty: Graduate Histology ANA 511; 8 hours; Rush University Graduate College.
2011–2016	Core Faculty (lecture and laboratory) for Histology in all M1 content blocks; Rush Medical College. Lecture hours: 6; Laboratory Hours 34
2011- 2014	Core Faculty: Intro to Neurobiology ANA 500; Rush University College of Nursing and Rush Graduate College. Lecture hours: 9, Laboratory hours: 10
2010–2016	Core Faculty (laboratory) for Gross Anatomy content in all RMC M1 blocks; Rush Medical College. 110 Laboratory hours

- 2014- 2015 Lecturer, BMC 315-Kinematics of Human Motion; Rush Graduate College; “Neurological gait abnormalities”; 2 lecture hours
- 2014 Faculty; Clinical Bridge for 4th year medical students, Abdominal Anatomy; Rush Medical College; 4 hours
- 2008- 2009 Part-Time Faculty Instructor, Human Gross Anatomy and Neuroanatomy Content Courses to First Year Medical students. 110 Laboratory hours; Rush Medical College.

Other Institutions

- 1996 Course Director/ Lecturer, “Mechanics of Motor Control”, 3 credit hours; 30 lecture hours/ 20 lab hours; Physical Therapy Division, University of Kentucky.
- 1996 Core Faculty, “Seminar in Physical Therapy: Research in Clinical Practice”, 8 lecture hours; Physical Therapy Division, University of Kentucky.
- 1993-1996 Core Faculty, (1)“Interdisciplinary Approaches to the Needs of Children with Disabilities and Special Health Care Needs” (2) Interdisciplinary Supports/ Service Systems for Children with Disabilities and Their Families” (3) “Leadership Seminar Supporting Children with Disabilities and Their Families”, (4) “Transdisciplinary Services for Students with Severe/Multiple Disabilities including Deaf-Blindness” 20 lecture hours University of Kentucky.
- 1992-1996 Course Director and Lecturer “Neurophysiologic Basis for Motor Behavior and Treatment Strategies for Clients with CNS Dysfunction”; complete program revision; 40 lecture hours; 20 lab hours; Physical Therapy Division, University of Kentucky.
- 1992-1996 Faculty Advisor for 10 senior students, “Research Problems in Physical Therapy”; Physical Therapy Division, University of Kentucky.
- 1995 Director and developer of Graduate Seminar course in Physical Therapy: “Brain Injury and Mechanisms of Recovery”; 10 lecture hours; University of Kentucky.
- 1995 Core Faculty “Human Growth and Development”; 8 lecture hours; Physical Therapy Division, University of Kentucky.
- 1993-1995 Course Director and Primary Lecturer; “Pediatric Physical Therapy”; complete curricular revision; 40 lecture hours; 20 lab hours; Physical Therapy Division, University of Kentucky.
- 1993 Course Director and developer for course “Recent Advances in Neuroscience”; 10 lecture hours; Physical Therapy Division, University of Kentucky.

- 1989 Teaching Assistant/Laboratory section instructor, “Medical Histology”, Dept. of Anatomy, Loyola University Medical Center
- 1988-89 Teaching Assistant, “Medical Neuroscience”, Dept. of Anatomy, Loyola University Medical Center
- 1987 Teaching Assistant, “Medical Gross Anatomy”, Dept. of Anatomy, Loyola University Medical Center

Visiting Professorships

- 2009- 2010 Visiting Gross Anatomy Professor, University of Medicine and Health Sciences, St. Kitts, West Indies, Human Gross Anatomy- Head and Neck, conducted 5 one-week teaching sessions (all-day sessions).

Continuing Education Workshops and Other Teaching Experiences

- 2011 Development and Course Director/Coordinator of two all-day Continuing Education workshops for Occupational and Physical Therapy practitioners entitled “Upper Extremity Human Anatomy Cadaver Workshop”; conducted at RUMC Department of Anatomy
- 2012- 2014 Faculty, USMLE STEP 1 Prep Courses, Kaplan; Weekend and Week-Long (4-5 days) Neuroanatomy/Neuroscience Content Delivery.

Mentorship for Rush University Students and Trainees in Research

Primary Advisor/Mentor to PhD Students

1. Josh Sevier, AuD, LLM; Assistant Professor; Co-Director, Auditory Implant Program, Department of Otolaryngology-Head and Neck Surgery; RUMC. Primary PhD Mentor, College of Health Sciences PhD program, June, 2026 - present; Dissertation Title: *“Longitudinal changes in auditory cortical activation in cochlear implant patients and prediction of speech perception outcomes.”*
2. Emily C Timm, PhD; 2022- May 2, 2026. Dissertation title: *“Cortical Correlates of Gait and Executive Function Deficits in FXTAS.”* Recipient of a 2024 National Fragile X Foundation Summer Scholar’s award (\$5,000), a 2025 National Ataxia Foundation Graduate Student Fellowship Award (\$25,000), and several Rush platform and poster presentation awards. First author on 2 publications; co-author on 6 publications and two book chapters. Dr. Timm is presently a postdoctoral fellow in Dr. O’Keefe’s lab and has accepted a Postdoctoral Fellowship at Hines VA, Maywood, IL.
3. Nicollette Purcell, MS, PhD; Primary PhD faculty advisor, 2017 to 2023; Dissertation Title: *“Cortical activation patterns in Huntington’s Disease during cognitive and motor tasks.”* First author on 3 publications and co-author on 2 additional papers and 15

scientific abstracts. Winner, 2018 Rush Research Forum Graduate College poster award. Dr. Purcell is presently an Assistant Professor at the Illinois College of Optometry.

4. Erin Robertson, PhD, AuD; Primary faculty advisor 2013- 2018, Department of Anatomy and Cell Biology, Dissertation title: “*Balance, gait and cognitive dysfunction in FXTAS versus Essential Tremor and Parkinson’s Disease*”; recipient of a 2015 National Fragile X Foundation Summer Scholar’s award (\$2,500); first place poster award Rush Graduate College, Rush Research Forum 2017; author on 14 scientific abstracts and 8 publications. 2017 1st Place winner in the 34th Annual Rush Forum for Research and Clinical Investigation Poster Competition, Graduate College Category; Clinical Doctorate in Audiology received May 2021 from Northwestern University. Dr. Robertson is presently a Research Scientist and Digital Biomarker expert at Regeneron Pharmaceuticals.

Co-Mentor and Dissertation Committee Member - PhD Students

1. Nicolle Potter, PhD student, IBS Program in DTS, RMC; Primary Mentor: Jonathan Gustafson; Co-Mentor and Dissertation Committee member; 2025- present: Dissertation Title: “*Evaluating Function After Reverse Total Shoulder Arthroplasty: The Integration of Biomechanics, Implant Wear, and Neuroplasticity*”.
2. Jessica Karl, PA, PhD; 2017-2020, CHS, Rush University; Dissertation: “*Deep Brain Stimulation programming paradigm in Parkinson’s disease to reduce axial symptoms*”; PhD Co-mentor and dissertation committee member; Dr. Karl is presently a faculty member at Northwestern University.

PhD Dissertation Committee member

1. Annalise E. Rogalsky, PhD; PhD Committee (stepped in after the untimely death of her primary PhD Advisor, Dr. Dan Nicholson, in April 2023); Dissertation title: Synaptic Architectural Alterations in CA1 and CA3 of the Ventral Hippocampus in Age-Associated Cognitively Impaired Rats. Annalise received her PhD in April 2024.
2. Diana Abassi, MS; PhD committee chair (2019- 2024); PhD student in Rush University’s Integrated Biomedical Sciences PhD program; Project Title: Utilizing Proteomic-Based Strategies to Investigate Molecular Mechanisms Influencing Phenotypic Variability in Fragile X Syndrome. Dr. Abassi is presently a Research Scientist at the University of Illinois-Chicago.
3. Elizabeth Pierobon, DPT, part-time PhD candidate in the CHS; rotation in my lab began in September, 2019; worked on research training and potential PhD projects; decided not to pursue a PhD due to family issues.
4. David J. Marmion, M.S. 2017- 08/2020; Ph.D. student in Kordower Laboratory; Dissertation title: “*Establishment of Novel Viral Vector-Mediated Rat and Nonhuman Primate Models of Multiple System Atrophy*”; Role PhD thesis committee member; Dr. Marmion started his postdoctoral fellowship in September, 2020.

Primary Advisor to Doctoral Students in Occupational Therapy

1. Cassandra Cisneros; January, 2022 - 2023; Primary advisor for research on MRI imaging in Huntington's disease and association with cortical activity; Occupational Therapy Doctorate Student | Class of 2024
2. Emma Sims; January, 2022 - 2023; Primary advisor for research on MRI imaging in Huntington's disease and association with cortical activity; Occupational Therapy Doctorate Student | Class of 2024
3. Olyvia Gryzik; January, 2022- 2023; Primary advisor for research on MRI imaging in Huntington's disease and association with cortical activity; Occupational Therapy Doctorate Student | Class of 2024

Primary Advisor/Mentor to MS Students

1. Annam Zaidi, MMBS, MS; Primary Faculty thesis advisor for MS in Clinical Research 2021-2024; "Impact of Cognition on Balance, Gait and Falls in FXTAS"; second-authored manuscript in preparation. Annam is currently a senior Clinical Research Coordinator at Weiss Memorial Hospital in Chicago.
2. Kyle Alyssir, MS; Rush University MS in Biotechnology Program, Primary Mentor; 2022-2023; second-authored manuscript submitted. Kyle is presently a student in a Physician Assistant program.
3. Nicholas Armijo, BS; Primary Faculty thesis advisor for MS in IBS 2018 to 04/2020; *Effects of a Karate Intervention on Balance and Gait in Parkinson's Disease*. Nicholas is presently a Senior Clinical Research Associate and Clinical Research Management Expert at RUMC.
4. Sharon Arhin, Mentor for Capstone Research Project for MS in Biotechnology in Rush Graduate College; 2019-2020; applied for medical school.
5. Caitlin Bailey, 2016 - 2018; MS student in Rush University's Integrated Biomedical Sciences Master's program; Primary thesis advisor; Project entitled "*Turns in Cerebellar Ataxia*"; Winner 2018 Rush Research Forum Graduate College poster award. Caitlin is presently a Research Scientist at the Rush Alzheimer's Disease Center.
6. Stephanie Voss, MS in Occupational Therapy, 2016- 2018; Primary Research Advisor; "*Development of a normative database for the instrumented Timed and Go (i-TUG), 2 minute walk test (i-WALK) and postural sway (i-SWAY) in children and young adults*"; Winner 2018 Rush Research Sigma Xi College of Health Sciences poster award; First author on two papers in Gait & Posture; Stephanie received her PhD in Kinesiology from the University of Illinois Urbana-Champaign in 2025. She is presently a T90 Post-Doctoral Fellow at Mass General Brigham in Boston, MA.

7. Rachel Tracey, MS in Occupational Therapy, 2016-2018; Primary Research Advisor; *“Development of a normative database for the instrumented Timed and Go (i-TUG), 2 minute walk test (i-WALK) and postural sway (i-SWAY) in children and young adults”*; 1 scientific abstract and author on a paper published in Gait & Posture.
8. Alexandra Palmer, MS in Occupational Therapy, 2016-2018; Primary Research Advisor; *“Development of a normative database for the instrumented Timed and Go (i-TUG), 2 minute walk test (i-WALK) and postural sway (i-SWAY) in children and young adults”*; 1 scientific abstract and author on a paper published in Gait & Posture.
9. Nicollette Purcell, 2015- 2017; MS student in the Department of Anatomy & Cell Biology; Thesis title: *“Dual cognitive task impact on balance and gait in Huntington Disease”*; Nicollette has 2 first-authored publications from her MS research; Commencement May 25, 2017; entered Rush University’s Integrated Biomedical Sciences PhD program in September, 2017. Dr. Purcell is presently an Assistant Professor at the Illinois College of Optometry.

Co-Mentor to MS Students in Clinical Research

1. Shirin Sadeghpour, MD; Rush University Movement Disorders Fellow, Co-Mentor, MS in Clinical Research Program; *“Effects of amantadine on gait and balance in Huntington’s disease”*; 2025- present.
2. Anjali Gera, MD; MS in Clinical Research; 2018 to 2020; *“Gait and Balance Analysis of Glucocerebrosidase Mutation Carriers with Parkinson’s Disease*. Role: Co-Mentor. Anjali is a primary author on one publication. She is presently a movement disorder specialist at Northwestern Medicine.

MS in IBS program or Anatomy & Cell Biology Committee member

1. Micaelan Valesky; thesis committee member 2021-2022; MS student in Rush University’s Integrated Biomedical Sciences program; Project Title: Stimulation of separate, distinct pontomedullary reticular formation subregions yields similar “halting” behavior in mice.
2. Poojan Thakkar; thesis committee member; 2021-2022, MS student in Rush University’s Integrated Biomedical Sciences program; Project Title: Assessing Changes in Shoulder Kinematics following reverse total shoulder arthroplasty. Poojan is presently attending medical school.
3. Leora Cramer, MS; thesis committee member 2020-2021; MS student in Rush University’s Integrated Biomedical Sciences program. Project Title: Concurrent Validity and Test-Retest Reliability of an Instrumented Shoe Insole to Measure Ground Reaction Force. Co-author on one publication.

4. Brittany Witkiewicz, MS in Anatomy and Cell Biology, Master's Thesis committee member, 2013; presently an Assistant Professor in the Department of Anatomy & Cell Biology at RUMC.

MD Student Primary Research Mentor

1. Benjamin Yu, MD Candidate, M1/2 Dean's Fellowship Scholars program awardee, March, 2026; Primary Mentor; "Neural underpinnings of cognitive and gait deficits in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS)".
2. Zohaib A. Aziz, MD candidate; M2 Rush University research trainee; 2025- present; "MRI analyses and correlations with cortical activity and gait in Huntington's disease".
3. Emily T. Sullivan, MD candidate; M1/2 Rush University Dean's Fellowship Scholars program awardee; 2024- 2025; Oral presentations: "*Quantitative Balance and Gait Assessment to Determine the Safety and Efficacy of Unilateral Staged MRI-Guided Focused Ultrasound Thalamotomy for the Treatment of Essential Tremor*" at 1) RMC Executive Council Reception. October 2024 and 2) Frontiers in Translational and Clinical Research: Insights from Early-Career Medical and Science Researchers. March 2025. Poster presentation: Rush Trainee Research Day, March 2025
4. Sabrina Nguyen, MD candidate; M1/2 Rush University Dean's Fellowship Scholars program awardee; 2023; Primary Mentor: "Neural underpinnings of cognitive, balance and gait deficits in Huntington's disease".
5. Christopher Ly, MD candidate; M1/M2 Summer research 2022; worked on several of Dr. O'Keefe's projects.
6. Kathryn Wrobel, 2018 M1/2 Rush Dean's Summer Research Fellowship Program; "*Development of a normative gait and balance database using inertial sensor technology and investigating efficacy of VTS270 (2-hydroxypropyl- β -cyclodextrin) in subjects with Niemann-Pick type C on gait, balance and fine motor coordination*". Kathryn was accepted into a prestigious and very competitive NIH Medical Research Scholars Program in Bethesda for her junior year of medical school where she was immersed in a program for medical students with exceptional promise to be a clinician scientist. Won a first-place award at the Rush Research Forum 2018 for her poster presentation.
7. Joseph Guan, 2018-2020 M1/2 Rush Dean's Summer Research Fellowship Program; "*Impact of cognition on balance and gait markers for FXTAS and performing longitudinal data collection in FMR1 premutation gene carriers*". Joseph is second author on a publication in the *Cerebellum*: "The Effects of Dual Task Cognitive Interference and Fast Paced Walking On Gait and Turns in Men and Women with FXTAS"; Dr. Guan is presently an Internal Medicine MD in the Chicagoland area.

8. Timothy Yung, 2017 M1/2 Rush Dean's Summer Research Fellowship Program; *"Impact of executive function on balance and gait markers for FXTAS and performing longitudinal data collection in FMR1 premutation gene carriers"*.
9. Marie Fefferman, 2017 M1/M2 Research Intern; *"Development of a normative gait and balance database using inertial sensor technology and investigating efficacy of VTS270 (2-hydroxypropyl- β -cyclodextrin) in subjects with Niemann-Pick type C on gait, balance and fine motor coordination"*.
10. Medha Parulekar, 2016; M1/M2 Summer Research Intern: *"Development of a normative database for the instrumented Timed and Go (i-TUG), 2-minute walk test (i-WALK) and postural sway (i-SWAY) in children and young adults"*; M2; submitted 1 scientific abstract; Winner Sigma Xi poster award, Rush Research Forum, 2017; entered residency program in Child Neurology; author on 2 manuscripts
11. Alexandra Bery, 2016; Rush Dean's Summer Research Fellowship Program; *"Impact of cognition on balance and gait markers for FXTAS"*; in residency; submitted 1 scientific abstract and author on 2 publications.
12. Maija Swanson, 2015; Rush Dean's Summer Research Fellowship Program; *"Effects of Cognitive Function and dual-task interference on balance and gait in premutation carriers of the FMR1 gene"*; entered residency; author on 3 scientific abstracts and one manuscript; presently a Clinical Assistant Professor at the University of Colorado.
13. Giavanna P Enriquez, 2015; Rush Dean's Summer Research Fellowship Program with Dr. Gian Pal, *"Effects of Deep brain Stimulation in Parkinson's Disease patients who carry a GBA mutation"*; entered residency.
14. Benjamin Kuo, 2014; Rush Summer Research Intern; *"Reliability of a Markerless Motion Method to Quantify Hyperkinesia in FXS"*; entered residency; author on 1 scientific abstract.
15. Joji W Kohjima, Rush Dean's Summer Research Fellowship Program, 2013; *"Characterization of balance and gait deficits using novel inertial sensor technology in FMR1 premutation carriers"*; Family Medicine practitioner in Chicagoland area.
16. Maura Walsh, Rush Summer Research Intern, 2013; *"Characterization of balance and gait deficits in FMR1 premutation carriers and molecular correlates of disease onset"*; presently a EM and Critical Care physician and Assistant Professor at UCSF.
17. Amber Fuitko, Rush Dean's Summer Research Fellowship Program, 2012; *"Characterization of balance deficits in FMR1 premutation carriers"*, entered residency; author on one publication; presently an EM physician.
18. Avram Frint, 2011-2012, medical student, Project: *"Development of a Markerless Motion Analysis System to Quantify Hyperkinesia as an Outcome Measure for Clinical"*

Trials in Fragile X Syndrome”, Fellow in Movement Disorders, Rush University; presently a Movement Disorder neurologist at Northwestern Medicine.

19. Emily Dunn, Rush Dean’s Summer Research Fellowship Program, 2011; “*Characterization of balance deficits in FMR1 premutation carriers*”, recipient of an American Academy of Neurology travel award; author on 2 scientific abstracts and 1 manuscript; presently a Movement Disorder neurologist at the University of Cincinnati Medical Center.

Undergraduate and High School Student Primary Research Mentor

1. Ketav Prem; STEM High School student intern; 2025- 2026
2. Leslie Garcia, Lake Forest College Student Intern; URM internship; 2023-2024; author on one submitted manuscript entitled “Trunk Instability in Huntington’s disease”.
3. Alex Weiss, Undergraduate summer research intern 2022.
4. Harlem Murray Summer 2019; Primary Mentor, Rush Summer Research Scholars Program for URM students; *Fine motor coordination outcome measures for efficacy of cyclodextrin treatment in Niemann-Pick Type C1*.

Other Mentees (Junior faculty, MD fellows and Research Associates)

1. Dr. Leo Shimanovich, MD; Co-Mentor, Rush Movement Disorders Fellow; Research project: “ Proof of concept study: evaluating the use of fNIRS to assess the functional neural network in cervical dystonia”; 2026-2028.
2. Jessica Joyce, BS; Research associate 2018-2019; Author on 3 manuscripts; admitted to Rush Medical School 2022.
3. Danielle Carnes, PhD; Research Associate and PhD student in Neuropsychology 2017-2019; author on 2 publications; presently a practicing Neuropsychologist.
4. Colleen Huml, MS, 2016 - 2017; Research Associate; received her MD from Georgetown University; in residency
5. Andrew McAsey, 2015-16, Research Associate; author on 4 scientific abstracts and two publications; presently an internal medicine practitioner.

University of Kentucky, Division of Physical Therapy: Master’s Thesis Committee Member

1. Billie Schanen, 1995-1997; Chair
2. Steve Langermann, 1995 – 1997, Chair
3. Margo Van Eck, 1995- 1997
4. Ruth Anne Geiger, 1995-1997
5. Rachel Neal, 1994-1996

LEADERSHIP POSITIONS

- 2025- present Director for Neuroscience/Neurobiology Preclerkship Basic Science Education, Rush Medical College. Responsibilities include Creation, Modification and Delivery of Basic and Clinical Science Education in Neurobiology/Neuroscience Content throughout the Preclerkship (M1/M2) curriculum.
- 2015- present Course Director: Introduction to Neurobiology ANA 500. Responsibilities include all of the following: 1) Content and Objective development, 2) Delivery of all Lectures and Labs, 3) Organization and dissemination of all Blackboard/Canvas content, 4) Writing/grading of all examinations, quizzes, clinical case group presentations, discussion posts, and scientific research paper assignments, 5) preparation/administration of gross anatomy practical exams.
- 2019- 2022 President, Chicago Chapter of the Society for Neuroscience (SfN), a Midwest regional chapter serving five states and one of the largest in the country. Responsibilities included chairing/organizing 2 annual conferences (inviting prestigious Presidential Symposium and Keynote speakers, arranging venue, organizing structure of the meeting, conducting weekly meetings with the Executive Committee from September through March/April every year). Served an extra year secondary to the COVID-19 pandemic.
- 2017-2023 Executive Committee of the Chicago Chapter of the Society for Neuroscience; Chair of several scientific sessions; Chair of PhD Graduate student symposia competition; Organizer and chair of the Lifetime Achievement Award 2023; poster competition judge (annually).

COMMUNITY SERVICE

- 2025- Lecture/Workshop PD 101; Education of Rush and associated patients with Parkinson's disease (PD) about the Beneficial Effects of Exercise in persons with PD (June 07, 2025, and to continue annually).
- 2016 - 2017 "Overview of Neuroanatomy", Neuro Camp for Chicagoland High School students (2-hour lecture/workshops); Rush University Medical Center,
- 2009 - 2020 Workshops/Guided tours in Anatomy Laboratory for numerous high school groups
- 2009- 2020 Presentations to elementary and high school groups on anatomical topics and careers in science and medicine.
- 2003- 2011 Science Fair Judge, Chicago Public High Schools, Chicago, IL

1996-2000 Chair, Grant writing committee, Beaubien Elementary School, Chicago, IL; Resulted in receipt of several grants including Toyota technology grant and Chicago Artists in Residency training grants.

COMMITTEE AND ADMINISTRATIVE SERVICE

Rush University

2025- Member, Rush University Faculty Council
 2025- Member, Department of Neurological Sciences Education Committee
 2022-2025 Rush University Neuroscience Research Consortium, Executive Member
 2022- 2024 Advisory Board, Center for Academic Excellence
 2019-2023 Academic Standards Committee, Rush Graduate College
 2019- 2024 Committee on Academic Excellence; Graduate College Faculty representative
 2019-2022 Departmental Advisory Committee, Cell & Molecular Medicine, Rush University
 2018- Rush Research Mentoring Program/Rush Trainee Day; abstract and poster reviewer
 2017- Cohn Research Fellowship Award application reviewer
 2018- 2020 Graduate College Council member; elected as faculty in Integrated Biomedical Sciences PhD program; Rush Graduate College
 2018- 2019 Advisory Council, Center for Academic Excellence, Rush University
 2016- 2018 Member, M1/M2 curriculum design task force
 2015- 2018 Member, Committee on Curriculum and Evaluation, Rush Medical College
 2013-2016 Member, Committee on Senior Faculty Academic Promotions (COSFAP)
 2012-2018 Member, M1 Working Group Committee, Rush Medical College
 2012-2016 Faculty Advisor, Rush University Student American Medical Association
 2012- 2014 Mentor for Rush Medical Students' Geriatric Student Interest Group
 2011- Mentee to M1/M2 students for Rush University Dean's Fellowship and other summer research internships
 2011- 2023 Judge, Rush University Research Forum/Research Trainee Day posters

SCIENTIFIC AND SCHOLARLY ACTIVITIES

Membership or Offices in Professional Societies, Panels, and Advisory Positions

2025- International Fragile X Premutation Registry (IFXPR) Executive Governance Committee
 2019- 2022 President, Chicago Chapter of the Society for Neuroscience (Midwest regional)
 2019- Member, International FXTAS Consortium (IFC)
 2018- 2019 President-Elect, Chicago Chapter of the Society for Neuroscience
 2017- 2020 Member, Executive Committee, Chicago Chapter Society for Neuroscience; Chaired the graduate student symposium and co-chaired the Movement Disorders symposium for the 2018 annual conference.
 2012 Invited Participant, NIH Research Plan for Fragile X Syndrome and Associated Disorders

1997-99 Board of Directors, Director of Research, National Neurodevelopmental Treatment Organization.

Reviewer for Federal Funding Agencies

2025 Ad hoc reviewer for the FY25 NIH Motor Function, Speech, and Rehabilitation study section.

2024 Ad hoc reviewer for FY24 Technology/Therapeutic Development Award-Hereditary Ataxia Panel; Department of Defense Congressionally Directed Medical Research Program (CDMRP)

2024 Ad hoc reviewer for FY24 PRMRP Impact Award-Hereditary Ataxia Panel; Department of Defense Congressionally Directed Medical Research Program (CDMRP)

2019 Ad hoc Reviewer for the Parkinson's Research Program (PRP) peer review panel of the Department of Defense Congressionally Directed Medical Research Program (CDMRP)

2016- 2017 Ad hoc Reviewer for the DOD Duchenne Muscular Dystrophy Research Program panel

Leadership/Service

2026 Panel Session Chair, “*Neuroimaging correlates underpinning clinical phenotypes in FMR1 premutation carriers with and without FXTAS*”, 20th NFXF International Fragile X Conference; Louisville, KY, July 16-19, 2026

2025 Invited Panel Session Chair, Sixth International *FMR1* Premutation Conference, September 8-12, 2025, Polignano a Mare, Italy

2024 Co-Leader; October 4, 2024; Midwest Regional Fragile X Conference, Chicago, IL; organized content, oversaw platform presentations, speaker introductions, panel time management, evening venue arrangements, and award selection for posters and graduate student talks.

2024 Invited National Ataxia Foundation Webinar Speaker; “*Experience preparing a successful DOD CDMRP Hereditary Ataxia grant proposal*”; April 25, 2024

Academic Professional Service

2021- Editorial Board Member, Frontiers in Neurology (Review Editor)

Ad Hoc Reviewer for Professional Journals (representative sample)

- Communications Medicine
- Movement Disorders
- Parkinsonism and Related Disorders
- Gait & Posture
- Scientific Reports

- The Clinical Neuropsychologist
- The Cerebellum
- Genes
- Journal of Neurology and Neurosurgery
- Clinical Genetics
- Frontiers in Neurology
- Genes, Brain and Behavior
- Geriatric Psychiatry
- Annals of Neurology
- Anatomical Sciences Education
- American Association of Anatomists
- Behavioral Brain Research
- Journal of Huntington's Disease
- Journal of Physical Therapy Education
- Journal of Neurodevelopmental Disorders
- Journal of Neurological Sciences
- Neurobiology of Aging
- Neurology
- Scientific Reports

NIH and other Scientific Committees

2025-	Ataxia Global Initiative- Digital Balance and Gait Outcomes Task Force
2025-	Ataxia Global Initiative- MRI imaging Outcomes Task Force
2023-	Huntington's Study Group; Digital Outcomes Working Group Member.
2020	NPC Newborn Screening Clinical Roundtable Members
2018	Rett Syndrome (RS) Clinical trials Steering Committee for validation of the RS Hand Function Scale for use in Clinical trials in girls with Rett Syndrome
2020-2021	Orphazyme Treatment Team Steering Committee; advisory section for treatment team member's knowledge regarding the drug Arimoclomol for the treatment of Niemann-Pick type C Disease.
2018-2019	Invited Member, 2018 NIH Task Force for updating the NIH Research Plan on Fragile X Syndrome and <i>FMRI</i> -related conditions; served on the <i>FMRI</i> premutation/FXTAS workgroup; invited participant in a Fragile X strategic planning meeting at the NIH (March 8-9, 2018).
2014-15	NIH Outcome Measures Scientific Advisory Board committee for planning a Phase 2/3 clinical trial in Niemann Pick C type 1. Traveled to Washington DC several times and participated in monthly conference calls to determine the best

outcome measures for the efficacy of VTS270 (2-hydroxypropyl- β -cyclodextrin) in subjects with Niemann-Pick type C Disease.

2015 – 2017 Scientific Advisory Board, Vtesse Pharmaceuticals, clinical trial design and treatment of Niemann-Pick type C

Invited Oral Presentations at Regional, National, and International Meetings

1. **O’Keefe, JA.** Rehabilitative Interventions for FXTAS. The 20th NFXF International Fragile X Conference; Louisville, KY, July 16-19, 2026
2. **O’Keefe, JA.** Cortical activation patterns and white matter integrity in cerebello-cortical pathways predict gait performance in FXTAS. The 20th NFXF International Fragile X Conference; Louisville, KY, July 16-19, 2026
3. **O’Keefe, JA.** White matter integrity in cerebello-cortical pathways and the cerebellum is associated with cortical activation patterns during gait and digital gait metrics in FXTAS. Proceedings of the Sixth International Conference on the FMR1 Premutation; Polignano A Mare, Italy, September 08- 11, 2025.
4. **O’Keefe, JA.** Lower Verbal Fluency Negatively Impacts Balance, and Gait Variability and Rhythm are Predictive of Fallers in FXTAS. Proceedings of the Sixth International Conference on the FMR1 Premutation; Polignano A Mare, Italy, September 08- 11, 2025.
5. **O’Keefe JA.** “Characterization of Balance and Gait Deficits Following Unilateral MRgFUS Thalamotomy to Treat Essential Tremor”; 6th Annual Rush University Parkinson Disease and Movement Disorders Research Day; May 23, 2025.
6. **O’Keefe JA.** “Impact of executive function on balance, gait and falls in FXTAS”. Midwest Fragile X Research Conference 2024; Chicago, IL, October 4, 2024.
7. **O’Keefe JA.** “Cortical correlates of gait and balance deficits in Huntington’s Disease”. 5th Annual Rush University Parkinson Disease and Movement Disorders Research Day; May 13, 2024.
8. **O’Keefe JA.** “Neural Underpinnings of Balance and Gait Deficits in Huntington’s Disease”; Meet the Expert/Mentor Seminar Series, Rush University, May 29, 2023.
9. **O’Keefe JA.** “Trunk Instability during Single Task, Dual Task and Fast Paced Gait in Huntington’s Disease”; 4th Annual Rush University Parkinson Disease and Movement Disorders Research Day; May 2023
10. **O’Keefe JA,** “The Effects of Dual Task Cognitive Interference and Fast Paced Walking On Gait and Turns in FXTAS”; The 17th International Fragile X Virtual Conference Series (due to COVID-19); 07/22/2020.

11. **O’Keefe JA**, Early Detection of FXTAS motor symptoms in *FMRI* premutation Carriers. This paper entitled "Prodromal markers of upper limb deficits in FMR1 premutation carriers and quantitative outcome measures for future clinical trials in FXTAS" ; The 17th International Fragile X Virtual Conference Series (due to COVID-19); 07/22/2020.
12. **O’Keefe, JA**, Dual task cognitive motor interference exacerbates turn deficits in men with FXTAS; invited speaker at the *Proceedings of the Fourth International Conference on the FMRI Premutation*; 2019, September 25-27, 2019, Rotterdam, The Netherlands
13. **O’Keefe JA**. “Cognitive function impacts gait, functional mobility and falls in Fragile X-Associated Tremor/Ataxia Syndrome”, 16th International Fragile X Conference, Cincinnati Ohio, July 11-15, 2018.
14. **O’Keefe JA**, Ilg W, Mirelman A, Hausdorff J. 2017 Invited Key Symposium speaker “Balance and gait changes in preclinical stages of neurodegenerative movement disorders - Commonalities, specificities, open questions and future steps” at the International Society for Posture and Gait Research World Congress, Fort Lauderdale, Florida, June 25-29, 2017.
15. **O’Keefe JA** Grand Rounds speaker invitation, Department of Rehabilitative Medicine, NIH, Bethesda, MA; “Cognition, Balance, Gait and Falling in Movement Disorders”; June 21, 2017.
16. **O’Keefe, JA** *Rush University, Neurology Grand Rounds*; “Relationship between Cognition and Balance and Gait in Movement Disorders”; October 11, 2016.
17. **O’Keefe, JA**. “*FMRI* premutation carriers exhibit worse balance and gait performance under dual task and environmentally challenging conditions”; The 15th International Fragile X Conference; San Antonio, TX, July 2016
18. **O’Keefe, JA**. “Evidence-based practice for rehabilitative interventions in cerebellar ataxias”; The 15th International Fragile X Conference; San Antonio, TX, July 2016.
19. **O’Keefe, JA**. "Inertial sensors and Dual task balance and gait measures in Movement Disorders" Rush University Movement Disorders Research Conference, Chicago, IL, May 9, 2016.
20. **O’Keefe, JA**. "Ataxia and Evidence-based Practice for Rehabilitative Interventions", University of Illinois, Physical Therapy Program, Chicago, IL. March 30, 2016.
21. **O’Keefe, JA**. “Synopsis of Balance and Gait research and Cognitive interaction in Fragile X gene carriers”, Midwest Chapter of the National Fragile X Foundation Research Meeting, Chicago, IL; April, 23, 2016.
22. **O’Keefe JA**. “Lower Executive function is associated with worse balance and gait performance in *FMRI* premutation carriers.” Midwest Fragile X Research Group, Chicago, IL; Jan 25th 2016.

23. **O'Keefe JA.** 2015. "Executive function is associated with balance and gait dysfunction in *FMRI* premutation carriers." Proceedings of the Second International Conference on the FMRI Premutation: Basic Mechanisms and Clinical Involvement, Barcelona, Spain.
24. **O'Keefe, JA.** 2014. "Gait, Balance, and Mobility impairments in carriers of the Fragile X gene." September 2014. University of Illinois-Chicago, Department of Physical Therapy.
25. **O'Keefe, JA.** July, 2014. "Gait and Mobility Deficits in Fragile X-associated tremor/ataxia syndrome (FXTAS) using the instrumented Timed Up and Go test", The 14th International Fragile X Conference; San Diego, CA.
26. **O'Keefe, JA,** July 2014, "Reliability and validity of a markerless motion capture system to quantify hyperactivity in Fragile X Syndrome." 14th International Fragile X Conference; San Diego, CA.
27. **O'Keefe, JA.** June 2014. "Gait and Mobility Deficits in Fragile X-associated tremor/ataxia syndrome (FXTAS) using the instrumented Timed Up and Go test", The Clinical Gait and Movement Disorder Society; Vermont, USA.
28. **O'Keefe, JA.** 2013. "Characterization of balance deficits in *FMRI* premutation carriers"; Presented at the 1st International Conference on the FMRI Premutation: Basic Mechanisms and Clinical Involvement. June 23-26, 2013.
29. **O'Keefe, JA.** 2013. "Quantification of hyperactivity and stereotypies in Fragile X Syndrome using a novel markerless motion analyses system", The Clinical Gait and Movement Disorder Society, Cincinnati, Ohio.
30. **O'Keefe, JA.** 2012. "Characterization of balance deficits in *FMRI* premutation carriers with and without FXTAS reveals a unique profile of impaired vestibular control and postural instability"; 13th International Fragile X Research Conference, Miami, Florida, 2012.
31. **O'Keefe, JA.** 2012. "Development of a Markerless Motion Analysis Method to Quantify Hyperkinesia and Stereotypies in Males with Fragile X Syndrome"; 13th International Fragile X Research Conference, Miami, Florida, 2012.
32. **O'Keefe, JA.** 2001. "Efficacy of Neurodevelopmental Treatment", Chicago Chapter, Illinois Physical Therapy Association.
33. **O'Keefe, JA.** 1990 "Estrogen Receptor Development in the Rodent Hippocampus." Society for Neuroscience (Chicago Chapter) Graduate Student Symposium; University of Chicago, March 1990.
34. **O'Keefe, JA.** 1985. "Rotational changes in the lower limb following distal medial hamstring surgery in children with cerebral palsy", Marquette University, 1985.

35. O'Keefe, JA 1984. "Orthotic casting using the CASCADE DAFO system", *Physical Therapy Department, Cook County Hospital*, Chicago, 1984
36. O'Keefe, JA 1983. "Physical Therapy Management of the Burn Patient", *University of Illinois, Physical Therapy Program*, Chicago, 1983.

Funding History of Peer-Reviewed Grants (Federal, Foundations, Internal)

Current

Federal

Department of Defense PR230363 - Neural Underpinnings of the Relationship Between Cognitive and Gait Dysfunction in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). The primary goals of this project are to understand the structural and functional neural underpinnings of the relationship between cognitive and gait dysfunction in FXTAS, to inform future interventional studies.

Role: PI

Total Costs: \$ 315,958

28% salary effort

Dates: 09/30/2024 - 10/01/2026

NIH-NINDS U01 14234988 Clinical Outcome Assessment and Treatment Monitoring Markers for Clinical Trial Readiness in FXTAS. This multi-site longitudinal project aims to establish valid, reliable, and sensitive clinical outcome measures, including digital tremor and balance measurements and brain imaging markers, for use in future clinical trials of patients with FXTAS to monitor treatment response. These measures will be optimized across multiple clinic sites and guided by an Advisory Committee of patient advocates, clinicians, researchers, and drug industry representatives.

PI: David Hessel, PhD (MPI Deborah A. Hall, Peter Todd)

Role: Co-Investigator

Total costs: \$5,640,572.00

10% salary effort

Dates: 07/01/2025 – 6/30/2030

NIH-NINDS 1 U01 NS113851; "Study in Parkinson's Disease of Exercise Phase 3 Clinical Trial: SPARX3"

This project aims to test whether the progression of the signs of PD is attenuated at 12 months in non-medicated people with PD when they perform high-intensity endurance treadmill exercise.

PI: Daniel Corcos, PhD

Role: Rush Site Principal Investigator

Total costs: \$17,381,229

10% salary effort

Dates: 3/19/2021 - 7/31/2026

Note: Administrative Supplement submitted March, 2026 (for 6-month extension from August 1st, 2026 to January 31st 2027); see below for competitive renewal application slated to begin 02/01/2027.

1I21RX005133-01A1 “Stimulating the Vagus nerve to improve gait in Veterans with Parkinson's disease”; Department of Veterans Affairs: VA Rehabilitation Research and Development SPiRE Program

The goal is to develop a non-invasive, neuromodulatory transcutaneous cervical Vagal Nerve Stimulation as an at-home intervention to improve gait and balance in veterans with PD
Clinicaltrials.gov # NCT06562569

PI: Kletzel, Sandie

Role: Co-I

Total Costs: \$230,000

% effort: 7.5%/year

Dates: 01/01/2026 - 12/31/2027

Foundations/Philanthropy

STXBP1 Foundation: A Phase 0 Non-interventional, Multi-center, Natural History Study in Patients with Syntaxin Binding Protein 1 (STXBP1) Related Disorder

PI: Elizabeth Berry-Kravis, MD, PhD

Role: Co-I

Total Direct Costs: \$302,324

10% salary effort

Dates: 03/01/2026 – 02/28/2028

DDX3X Foundation: Characterizing the Natural History of DDX3X Syndrome to Inform the Development of Intervention Outcome Measures.

PI: Tamar Caceres, MD

Role: Co-I

Total Direct Costs: \$180,000

2% effort

Dates: 01/01/2024 – 12/31/2027

Cures Within Reach (NOA)

“Using a Wearable Leg Stimulation Device to Improve Mobility of Veterans with Parkinson's Disease.” This open-label, crossover pilot trial is evaluating whether the Cionic Neural Sleeve electrical stimulation device improves walking and freezing episodes in veterans with PD.

PI: Coletta K

Role: Co-Investigator

Total Direct Costs: \$60,000

Salary: \$6,000 consulting fees over two years

Dates: 04/01/2026 – 03/31/2028

Treatment of Neurological Symptoms of CLN1 Neuronal Ceroid Lipofuscinosis with Intrathecal TSHA-118 (scAAV9.CBh.hCLN1)

PI: Elizabeth Berry-Kravis
Role: Clinical Outcome Assessment Rater
Sponsor: Philanthropy
Salary: in-kind service
Dates: 10/4/2024 – 01/12/2032

An open-label single center study of an experimental antisense oligonucleotide treatment of three participants with Schuurs-Hoeijmakers syndrome due to PACS1 genetic mutation

PI: Elizabeth Berry-Kravis
Role: Co-Investigator
Sponsor: unfunded compassionate use
Salary: in-kind service
Dates: 03/06/2025 – 12/2030

Intrathecal 2-Hydroxypropyl- β -cyclodextrin for Neurological Decline in Patients with Niemann-Pick Disease Type C10

Sponsor: University of Notre Dame Philanthropy
PI: Elizabeth Berry-Kravis
Role: Co-Investigator
3-5% effort annually
Dates: 05/01/2014 – present (expected FDA approval August, 2026)

Faculty Mentor For Student Research Awards

Student's Name and Degree Program: Emily C. Timm, IBS PhD Program
Project Title: The Neural Mechanisms Underlying Cortical Control of Gait and Cognition in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS)
Funding Mechanism: Graduate Student Predoctoral Fellowship Award (\$25,000)
Agency: National Ataxia Foundation (an international foundation)
Faculty Sponsor: Joan A. O'Keefe, PhD
Dates of Project Period: February 01, 2025- Jan. 31, 2027 (One Year NCE)

Pending

Federal

FDA: Characterizing the Natural History of DDX3X Syndrome to Inform the Development of Intervention Outcome Measures and Explore Neurophysiologic Biomarkers. This is an observational multicenter investigation into the natural history and co-morbidities of DDX3X syndrome. We will collect detailed longitudinal data on individuals with a molecular diagnosis of DDX3X syndrome (pathogenic variant or deletion affecting the DDX3X gene) to increase our understanding of DDX3X syndrome and develop robust outcomes that will be useful in clinical trials.

PI: Tamar Caceres, MD
Role: Co-I
3% effort
Total Direct Costs: \$1,176,831
Dates: 9/1/2026-8/31/2030

NIH U01 Competitive Renewal Study in Parkinson's Disease of Exercise Phase 3 Clinical Trial: SPARX3" (5U01NS113851-05) Competitive Renewal. This is a Phase 3 clinical trial on the efficacy of aerobic exercise for slowing the progression of Parkinson's disease (PD). We are requesting four years of support to finish the exercise intervention on 144 participants, lock the database, publish the primary paper, and publish at least 22 planned manuscripts.

PI: Daniel Corcos, PhD

Role: Rush Site Principal Investigator

Total costs: \$7,464,956

10% salary effort

Submission Date: March 5, 2026

Dates: 02/01/2027 - 01/31/2031

VA pilot grant: Characterizing the neuromechanical features of continuous walking and freezing-of-gait in Veterans with Parkinson's disease (PD) using an instrumented leg sleeve. The research objective is to examine feasibility, acceptability and acute therapeutic gait effects (gait kinematics and freezing episodes) of electrical stimulation delivered by the Neural Sleeve in PD

PI: Matty Major, PhD

Role: Co-investigator

Total direct costs: \$200,000

% Effort: 5% salary/year

Dates: 10/1/2026 – 9/30/2028

Foundations

Aligning Research to Impact Autism (ARIA): Long-term, high support network to study and treat causes of severe Autism Spectrum Disorders (ASD).

PI: Elizabeth Berry-Kravis, MD, PhD

Role: Co-I

Total Costs: \$5.2 million

10% effort

Dates: July 01, 2026 - June 31, 2031 (NOA received with grant now slated to be funded in "Wave 2" from 2027/2028 through 2032/ 2033)

Parkinson Foundation: Powering up Gait and Mobility Rehabilitation in Parkinson's: Advancing a Smart Wearable Leg Sleeve. The major goals of this project are to examine feasibility, acceptability and acute therapeutic effects of electrical stimulation delivered by a neural sleeve on gait in atypical parkinsonism.

PI: Colletta, K

Total Direct Cost: \$180,000

Role: Co-investigator

5% salary/year

Dates: 5/1/26 – 4/30/28

Completed

Huntington's Disease Society of America Human Biology Project Award; "Neural underpinnings of cognitive, balance and gait deficits in Huntington's disease". The major goal of this project is to examine cortical activity via portable fNIRS technology and MRI metrics of GMV in cortical regions of interest and the striatum to examine structure- function relationships mediating cognitive, gait and balance impairments in Huntington's disease.

Role: PI (*best score*)

Total direct costs: \$ 150,000

Dates: 11/01/2021 to 10/31/2023 (NCE to 10/31/2025)

Rush Imaging Research Core Pilot Grant; "Structural and functional brain imaging to identify the neural underpinnings of the relationship between cognitive and gait dysfunction in Fragile X-associated tremor/ataxia syndrome (FXTAS)".

Role: PI

Total Direct costs: \$50,000

Dates: 06/01/2023 – 06/31/2025 (One Year NCE)

"Digital Assessment of Gait and Speech in Parkinson's Disease. Major goals are to assess the ability of a wearable insole to accurately measure gait in Parkinson's disease and detect clinically meaningful change in gait in Parkinson's disease."

Sponsor: Regeneron

PI: Deborah A. Hall, MD, PhD

Role: Co-I

Total Direct costs: \$ 126,000

10% effort

Dates: 01/01/2022- 12/31/2024

NIH-NICHD K01HD088762; "Impact of Cognition on Balance and Gait Markers for FXTAS"

Role: PI

The major goal of this project is to provide an early detection model for Fragile X-associated tremor/ataxia syndrome (FXTAS) using balance and gait markers and interacting cognitive and molecular factors.

Total costs: \$ 826,758; includes 75% salary support for 5.5 years

Dates: 7/1/2016- 06/30/2022 (funded extension due to COVID-19)

Rush Schweppe Translational Science Consortium Pilot grant: "Balance and Gait Biomarkers for Fragile X Tremor/Ataxia Syndrome (FXTAS)"

Role : PI

Costs: \$50,000

11/1/2015- 5/31/2017

Rush University Cohn Research Fellowship

This fellowship allowed the candidate protected time for research for her studies on gait and balance dysfunction in FXTAS and the use of markerless technology to quantify hyperactivity and stereotyped behaviors in FXS and autism. This award directly impacted her success in receiving NIH K01 funding.

Total Costs: \$20,000
Role: PI
20% salary effort
7/1/2014 – 6/30/2015

Private Philanthropic Donor

“Validity and test-retest reliability of a Markerless Motion Analysis System to Quantify Hyperkinesia in Fragile X Syndrome and Autism”

The goal is to evaluate the reliability and validity of a markerless motion analysis method to quantify stereotypies and other hyperactive motions in Fragile X Syndrome and autism for future use as outcome measures in clinical research trials.

Other PI: Markus A. Wimmer

Role: Co-PI

Total Costs: \$20,000
9/01/2013 - 8/31/14

Fragile X Research Foundation “Development of a Markerless Motion Analysis System to Quantify Hyperkinesia as an Outcome Measure for Clinical Trials in Fragile X Syndrome”

The goal of this project is to determine the feasibility of a markerless technique to quantify excessive motion in persons with fragile X syndrome

Role: Co-PI

PI: Elizabeth Berry-Kravis, MD, PhD

Total costs: \$43,824
2011-2012

NIH Academic Research Enhancement Award (AREA) R15 grant for proposal “Trk B Receptor Regulation by Cellular Signals”

Role: PI

Direct costs: \$111,438
1994-1997

“Regulation of NMDA Receptor Subunit Expression by Tyrosine Kinase pathways: Relevance to Alzheimer’s Disease (AD)” National Institute of Aging sponsored Alzheimer’s Disease Research Center Project pilot grant at the University of Kentucky.

PI: Joan O’Keefe

Total costs: \$20,000
1994-1995

Maternal Child Health (MCH) Core Faculty Affiliate for University Affiliated Program Grant “Interdisciplinary Training and Leadership Development”

Role: Co-Investigator

15% Salary support
1992-1995

University of Kentucky Postdoctoral Fellowship Award for Women in Science; grant entitled “FGF and other growth factor modulation of hippocampal neuron growth and survival”.

Role: PI

Costs: \$ 30,000
1992-1993

Current Industry Support

An open-label study of an experimental antisense oligonucleotide treatment for participants with a TUBB4A mutation.

Sponsor: n-Lorem Foundation

PI: Elizabeth Berry-Kravis

Role: Sub-Investigator/Clinical outcome assessment Rater

% effort: 2%

Dates: 07/28/2025 - present

A Multicenter, Open Label, Randomized, Dose-Escalation and Dose-Expansion Study of the Safety, Tolerability, and Efficacy of a Single Intrathecal Administration of TSHA-102, an AAV9-Delivered Gene Therapy, for the Treatment of Pediatric Females with Rett Syndrome
Sponsor: Taysha Gene Therapies, Inc.

PI: Elizabeth Berry-Kravis

Role: Sub-Investigator/Clinical outcome assessment Rater

% effort: 2%

Dates: start 10/29/2023 – 06/30/2030

RTT-200: A Baseline-Controlled, Open-Label, Multicenter, Single-Arm, Pivotal Study to Evaluate the Efficacy, Safety, and Tolerability of NGN-401 in Subjects with Rett Syndrome (Embolden)

Sponsor: Neurogene Inc.

PI: Elizabeth Berry-Kravis

Role: Sub-Investigator/Clinical outcome assessment Rater

% effort: 2%

Dates: 10/25/2024 – 09/30/2030

Past Industry or Philanthropic Support

Rett Syndrome Utility Methods Evaluation (RESUME): A Methods Study on Standardizing Clinical and At-Home Video Assessments

Sponsor: Red Nucleus Solutions, LLC

PI: RUMC site, Elizabeth Berry-Kravis

Role: Sub-Investigator/Clinical outcome assessment Rater

% effort: 1%

Dates: 08/19/2024 – 03/15/2026

Clinical Trial Protocol VTS301, A Phase 2b/3 Prospective, randomized, Double-Blind, Sham Controlled Trial of VTS270 (2-hydroxypropyl- β -cyclodextrin) in Subjects with Neurological Manifestations of Niemann-Pick Type C1 (NPC1) Disease

My role on this project is to supervise and administer sensitive tests of gait and balance outcome measures every 2 months.

Sponsor: Vtesse Pharmaceuticals
Co PIs: Berry-Kravis, Porter
Role: Sub-Investigator for Rush University
Total costs: \$455,242
5% Effort
Dates: 09/1/15-12/31/20

Intermediate size patient population IND for 2-hydroxypropyl-B-cyclodextrin (HP-B-CD) treatment of Niemann-Pick type C1 (NP-C1); conducted bimonthly gait, balance and neurodevelopmental assessments in patients in this IND.

Sponsors: Hope for Hayley Foundation, Samantha's Search for the Cure Foundation

PI: Berry-Kravis

Role: Co-Investigator

Total Direct Costs: \$75,000

5% effort

Dates: 2013- 2018

Efficacy, Safety, and Tolerability of Fosmetpantotenate (RE-024), a Phosphopantothenate Replacement Therapy, in Patients with Pantothenate Kinase-associated Neurodegeneration (PKAN): A Randomized, Double-blind, Placebo-controlled Study with an Open-label Extension
The goal is to determine if RE-024 is effective for reducing progression of neurological deterioration in PKAN.

Sponsor: Retrophin, Inc

Rush PI: Berry-Kravis

Role: Co-Investigator

Dates: 01/01/2017 - 04/20/2020

PUBLICATIONS

Submitted

Hall D, Gonzalez D, Klusek J, Todd P, **O'Keefe JA**, Maltman N, Monttanaro FAM, Protic D, Scorr, L, Malik R, Turcano P, Meyers, Seritan A. "Management of Fragile X-Associated Tremor Ataxia Syndrome: A Guide for Clinicians"; Frontiers in Neurology, section Movement Disorders, May 01, 2026 (under revision review)

Peer Reviewed Journals

*** Indicates student/trainee author**

**** indicates top 5 recent research articles**

Öz G, Coccozza S, Rezende TJR, Henry PG, Faber J, Harding IH; Ataxia Global Initiative Working Group on MRI Biomarkers. MRI end-points for clinical trials in ataxias: recommendations from the **Ataxia Global Initiative MRI Biomarkers Working Group**. Nat Rev Neurol. 2026 Jun 3. doi: 10.1038/s41582-026-01218-7. Epub ahead of print. PMID: 42236987.

Hall DA, Kwei K, Acosta RJ, Koyani B, Robertson R, Rai P, Barge R, Timm EC*, Purcell N*, Desai N, Patel D, Visoiu-Knapp AM, Stuart S, Alaj R, Wipperman MF, Levy O, **O'Keefe JA**. Validity of a wearable digital insole for assessing gait ON and OFF in Parkinson's disease; Annals of Clinical and Translational Neurology, 2026; 0:1-11.

Purcell, N.L.*, Stuart, S., Vitorio, R., Timm, E.C.,* Hall, D.A., Cooper, C., & **O'Keefe, J.A. Cortical Correlates of Balance During Single and Dual Tasks in Huntington's Disease: a preliminary fNIRS study. Neurorehabilitation & Neural Repair, (2026) 20(1) 24-36.

Mancini M, Afshari M, Almeida Q, Amundsen-Huffmaster S, Balfany K, Camicioli R, Christiansen C, Dale ML, Dibble L, Earhart G, Ellis T, Griffith G, Hackney M, Hopkins J, Horak F, Jones K, Ling L, **O'Keefe JA**, Kwei K, Olivier G, Rao A, Sivaramakrishnan A, and Corcos D. Digital gait biomarkers in Parkinson's disease: susceptibility/risk, progression, response to exercise, and prognosis"; npj Parkinson's Disease (2025) 11:51, <https://doi.org/10.1038/s41531-025-00897-1>

Tait, P.; Graham, L.; Vitorio, R.; Watermeyer T; Timm EC*; **O'Keefe J**; Stuart S, Morris R. Neuroimaging and cognitive correlates of postural control in Parkinson's Disease: a systematic review. J Neuroeng Rehabil 2025 Feb 8;22(1):24. doi: 10.1186/s12984-024-01539-y.

Timm, E.C.*; Purcell, N.L.*; Ouyang, B.; Berry-Kravis, E.; Hall, D.A.; **O'Keefe, J.A. Potential Prodromal Digital Postural Sway Markers for Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS) Detected via Dual-Tasking and Sensory Manipulation. Sensors 2024, 24, 2586. <https://doi.org/10.3390/s24082586>

Robertson-Dick EE*, Timm EC*, Pal G, Ouyang B, Liu Y, Berry-Kravis E, Hall DA, **O'Keefe JA. Digital gait markers to potentially distinguish fragile X-associated tremor/ataxia syndrome, Parkinson's disease, and essential tremor. Front Neurol. 2023 Dec 7;14:1308698. doi: 10.3389/fneur.2023.1308698. PMID: 38162443; PMCID: PMC10755476

Cramer LA*, Wimmer MA, Malloy P, **O'Keefe JA**, Knowlton CB, Ferrigno C. Validity and Reliability of the Insole3 Instrumented Shoe Insole for Ground Reaction Force Measurement during Walking and Running, Sensors, 2022, 22, 2203. <https://doi.org/10.3390/s22062203>.

Voss S*, Zampieri C, Biskis A*, Armajo N*, Purcell N*, Ouyang B, Liu Y, Berry-Kravis E, **O'Keefe JA**. Inertial sensor based normative postural sway parameters in typically developing children and young adults, Gait & Posture, 2021. 90: 112-119.

****O'Keefe, JA**, Guan J.*, Robertson E*, Biskis A*, Joyce J*, Ouyang B, Li, Y, Carnes D*, Purcell E*, Berry-Kravis E, Hall DA. The Effects of Dual Task Cognitive Interference and Fast Paced Walking on Gait and Turns in Men and Women with FXTAS, Cerebellum, 2021. 20: 212–221; PMID: 33118140

O'Keefe JA, Bang D*, Robertson E*, Biskis A*, Ouyang B, Liu Y, Pal G, Hall DA and Berry-Kravis E. Prodromal markers of upper limb deficits in *FMRI* premutation carriers and

quantitative outcome measures for future clinical trials in FXTAS, Movement Disorders Clinical Practice, 2020; 7(7): 810–819. doi: 10.1002/mdc3.13045. PMID: 33043077

Robertson EE*, Hall DA, Pal G, Bichun Ouyang B, Liu Y, Joyce JM*, Berry-Kravis E, **O'Keefe JA**. Tremorography in fragile X-associated tremor/ataxia syndrome, Parkinson's disease and essential tremor;. Clin Park Relat Disord. 2020 Jan 23;3:100040. doi: 10.1016/j.prdoa.2020.100040. eCollection 2020.PMID: 34316626.

Voss S*, Joyce JA*, Biskis, A*, Armajo N*, Tracy R*, Palmer A*, Ouyang B, Liu Y, Zampieri C, Parulekar M*, Fefferman M*, Berry-Kravis E, **O'Keefe JA. Inertial sensor-based normative spatiotemporal gait parameters in typically developing children and young adults, Gait & Posture, 2020; Jul;80:206-213. PMID: 32531757

Gera A* , **O'Keefe JA**, Ouyang B, Liu Y, Ruehl S, Buder B, Joyce J*, Purcell N*, Pal G. Gait asymmetry in glucocerebrosidase mutation carriers with Parkinson's disease. PLoS One. 2020 Jan 24;15(1):e0226494. doi: 10.1371/journal.pone.0226494. eCollection 2020. PMID: 31978134

Purcell N*, Goldman JG, Bernard B, **O'Keefe JA**. The effects of dual-task cognitive interference on gait and turning in Huntington's disease; PLoS One. 2020 Jan 7;15(1):e0226827. doi: 10.1371/journal.pone.0226827. eCollection 2020. PMID: 31910203

Hall DA, Robertson E.* , Leehey M, McAsey A*, Ouyang B, Berry-Kravis E, **O'Keefe JA**. Open-label pilot trial of citicoline for fragile X-associated tremor/ataxia syndrome (FXTAS); PLoS One. 2020 Feb 13;15(2):e0225191. doi: 10.1371/journal.pone.0225191. eCollection 2020. PMID: 32053612

Purcell N*, Goldman J, Bernard B, **O'Keefe JA**. The Effects of Dual-Task Cognitive Interference on Balance in Huntington's disease, Movement Disorders Clinical Practice, 2019; 6(3):202-212; PMID:30949551; PMCID:PMC6417749

O'Keefe JA, Robertson E., Ouyang B, Carns D*, McAsey, A*, Liu Y, Swanson M*, Bernard B, Berry-Kravis E, Hall DA. Cognitive function impacts gait, functional mobility and falls in Fragile X-Associated Tremor/Ataxia Syndrome; Gait and Posture. 2018;66:288-293; PMID: 30243213

Berry-Kravis, E, Chin, J Hoffmann, A, Winston A, Stoner R, Lagorio L, Friedmann K, **O'Keefe, JA**. Long-Term Treatment of Niemann-Pick Type C1 Disease with Hydroxypropyl-Beta-Cyclodextrin; Pediatric Neurology. 2018 Mar;80:24-34. PMID: 29429782

Pal G, **O'Keefe JA**, Robertson E*, Bernard B, Anderson S, Hall DA. Global cognitive function and processing speed are associated with gait and balance dysfunction in Parkinson's disease; J NeuroEng and Rehab. 2016. 13(1): 94-102.

Robertson E*, Hall DA, McAsey AR*, **O'Keefe JA**. Fragile X-associated Tremor/Ataxia Syndrome: Phenotypic comparisons with other Movement Disorders, Clin Neuropsychol. 2016; 30(6):849-900.

Hall DA, Robertson E*, Shelton AL, Losh MC, Mila M, Moreno EG, Gomez-Anson B, Martínez-Cerdeño V, Grigsby J, Lozano R, Hagerman R, Santa Maria L, Berry-Kravis E, **O’Keefe JA**. Update on the Clinical, Radiographic, and Neurobehavioral Manifestations in FXTAS and *FMR1* Premutation Carriers, Cerebellum; 2016;15:578-586.

O’Keefe JA, Robertson E*, Hall DA and Berry-Kravis E. Gait and Functional Mobility Deficits in Fragile X-Associated Tremor/Ataxia Syndrome; Cerebellum; 2016;15(4):475-82.

Hall, DA, Robertson-Dick, E*, **O’Keefe JA**, Hadd AG, Zhou L, Berry-Kravis E. Repeat Size and X-inactivation in the Clinical Phenotype of Fragile X Premutation Carrier Sisters; Neurol Genet. 2016; 2(1):e45. PMID: 27066582

Pal G, Robertson E*, **O’Keefe JA**, Hall DA. The Cognitive and Motor Profile of GBA-associated Parkinson Disease: A Review; Movement Disorders: Clinical Practice, 2015; 3: 4–8.

O’Keefe JA, Robertson E, Dunn E*, Li Y, Deng Y, Fiutko A*, Berry-Kravis E, Hall DA. Characterization and Early Detection of Balance Deficits in Fragile X Premutation Carriers With and Without Fragile X Associated Tremor/Ataxia Syndrome (FXTAS). Cerebellum, 2015; 14(6):650-62.

Hall DA, Birch R, Anheim M, Jonch A, Pintado E, **O’Keefe, JA**, Troller J, Hagermann RJ, Fahn S, Berry-Kravis, E, Leehey MA. Emerging topics in FXTAS. J. Neurodev Dis. 2014; 6: 31-41.

O’Keefe JA, Espinosa-Orias A , Khan H, Hall DA, Berry-Kravis E, Wimmer MA,. Implementation of a Novel Markerless Motion Analyses System to Quantify Hyperactivity in males with Fragile X Syndrome, Gait and Posture, 2014; 39 (2):827-30.

Hall DA, **O’Keefe JA**. Clinical Neurogenetics: Fragile X-associated tremor/ataxia syndrome. Neurol Clin. 2013; 31(4):1073-84.

Hall DA, **O’Keefe JA**. Fragile X-Associated Tremor Ataxia Syndrome: The Expanding Clinical Picture, Pathophysiology, Epidemiology, and Update of Treatment. Tremor Hyperkin Mov;2. Epub 2012; pii: tre-02-56-352-1. PMID: 23439567

Geiger RA*, Allen, **O’Keefe JA** and Hicks M. Balance and mobility following stroke: effects of physical therapy intervention with and without biofeedback/ forceplate training. Phys Ther.; 2001; 81:995-1005.

Mattson MP, **O’Keefe JA** and Smith-Swintosky VL. Similarity between ALS and Alzheimer’s Disease: the role of intracellular calcium? In: S. Appel (Ed.) ALS 1: Research in Therapy. 1996.

Cheng B, Furukawa K, **O’Keefe JA**, Goodman Y, Kohiko M, Fabian T and Mattson, MP. Basic FGF selectivity increases levels of AMPA receptor subunit GluR1 and differentially modulates Ca²⁺ responses to AMPA and NMDA in hippocampal neurons, J. Neurochem. 1995. 65:2525-2536.

O’Keefe JA, Li Y, Burgess LA and Handa RJ 1995. Estrogen receptor mRNA alterations in the developing rat hippocampus. Mol Brain Res. 30:115-124.

Handa RJ, Burgess LH, Kerr JE and **O’Keefe JA**. 1994. Gonadal steroid hormone receptors and sex differences in the hypothalamo-pituitary-adrenal axis. Hormones and Behav. 28:464-476.

O’Keefe JA, Pedersen EB, Castro AJ and Handa RJ. 1993. Ontogeny of estrogen receptors in heterochronic hippocampal and neocortical transplants demonstrates an intrinsic developmental program, Dev. Brain Res. 75:105-112.

Pederson EB, **O’Keefe JA**, Handa RJ and Castro AJ. 1992. Estrogen receptors are present in neocortical transplants, J. Neurotranspl. Plast. 3:135-138.

O’Keefe JA and RJ Handa. 1990. Transient elevation of estrogen receptors in the neonatal rat hippocampus. Dev Brain Res., 57: 119-127.

Other publications

Hall DA, **O’Keefe J**. Fragile X-Associated Tremor Ataxia Syndrome: The Expanding Clinical Picture, Pathophysiology, Epidemiology, and Update of Treatment. 2013. Rush Neuroscience Review, 47-51.

Book Chapters

Tait, PJ, Timm, EC*, **O’Keefe, JA**, Watermeyer, T, Vitorio, ., Morris, R, *et al.* (2024). MoBI: Mobile Brain/Body Imaging to understand walking and balance. In F. A. Barbieri, R. Vitorio, & P. C. R. dos Santos (Eds.), In *Locomotion and posture in older adults: The role of aging and movement disorders* (pp. 19–38). Springer Nature Switzerland. https://doi.org/10.1007/978-3-031-74123-4_2

Tait PJ, Morris R, Watermeyer T, Timm EC, **O’Keefe, JA**, Vitorio R, Stuart S. Chapter 15 - Balance assessment in neurological conditions, Editor(s): Samuel Stuart, Rosie Morris; In *Gait, Balance, and Mobility Analysis*, Academic Press, 2025, Pages 405-434, ISBN 9780443214844, <https://doi.org/10.1016/B978-0-443-21484-4.00015-6>

Robertson-Dick, E, **O’Keefe JA**, Hall DA. Clinical Trials in Fragile X-Associated Tremor/Ataxia Syndrome; *Contemporary Clinical Neuroscience*, 2023, Part F6, pp. 649–661.

O’Keefe, JA. Genomics and Genetic Syndromes Affecting Movement. In *Physical Therapy for Children 4th Edition* (SK Campbell, RJ Palisano and Orlin, MN Eds), St. Louis, MO, Elsevier, E Chapter 2, 2011. ISBN 9780323290944

O’Keefe, JA. Genomics and Genetic Syndromes Affecting Movement. In *Physical Therapy for Children 3rd Edition* (SK Campbell, DW Vander Linden and RJ Palisano, Eds), St. Louis, MO, Saunders, 2006, pp. 217- 255.ISBN13: 9780721603780

Complete List of Published Work in [My Bibliography - NCBI](#)

Complete List of Published Work in [Scopus](#)

Peer-Reviewed Abstracts, Published Conference Proceedings, and Poster Presentations

* Indicates student author

1. **O’Keefe, JA**, Timm, E*, Sica C, Niaz MR, Ouyang B, Hall DA. White matter integrity in cerebello-cortical pathways and the cerebellum is associated with cortical activation patterns during gait and digital gait metrics in FXTAS. *Proceedings of the Sixth International Conference on the FMR1 Premutation*; Polignano A Mare, Italy, September 08- 11, 2025. (selected for platform presentation).
2. **O’Keefe, JA**, Zaidi A*, Timm E, Ouyang B, Hall DA. Lower Verbal Fluency Negatively Impacts Balance, and Gait Variability and Rhythm are Predictive of Fallers in FXTAS. *Proceedings of the Sixth International Conference on the FMR1 Premutation*; Polignano A Mare, Italy, September 08- 11, 2025 (selected for platform presentation).
3. Timm EC*, Stuart S, Hall DA, **O’Keefe, JA**. The Neural Correlates of Gait and Executive Function Deficits in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS): A Preliminary Study. *Proceedings of the Sixth International Conference on the FMR1 Premutation*; Polignano A Mare, Italy, September 08- 11, 2025. (selected for platform presentation by EC Timm, Dr. O’Keefe’s PhD student)
4. **O’Keefe, JA**; Zaidi, A, Timm, E.; Hall DA. Lower Verbal Fluency Negatively Impacts Balance and Gait Variability and Rhythm are Predictive of Fallers in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). Poster presentation at the *Proceedings of the 2025 International Society for Posture and Gait Research World Congress*; Maastricht, The Netherlands; June 29- Jul 3, 2025.
5. Timm EC, Alyassir K, Purcell NL, Garcia L, **O’Keefe JA**, Trunk Instability during Single Task, Fast Paced, and Dual Task Gait in Huntington’s Disease. Poster presentation at the *Proceedings of the 2025 International Society for Posture and Gait Research World Congress*; Maastricht, The Netherlands; June 29- Jul 3, 2025.
6. Timm, E.C.; Sica C, Niaz MR, Stuart S, Hall DA, **O’Keefe, J.A** The Neural Correlates of Gait and Executive Function Deficits in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS): A Preliminary Study. Poster presentation at the *Proceedings of the 2025 International Society for Posture and Gait Research World Congress*; Maastricht, The Netherlands; June 29- Jul 3, 2025.

7. Hall DA, Kwei K, Purcell N*, Timm EC*, Acosta RJ, Koyani B, Robertson R, Wipperman MF, Levy O, **O’Keefe JA**. Initial validity and clinical utility of the Moticon insole system for assessing gait in Parkinson’s disease. Poster presentation at the *2025 American Academy of Neurology Conference*, San Diego, CA, April 5-9, 2025
8. Timm EC, Paras J, Hall DA, **O’Keefe JA**. The Neural Mechanisms Underlying Cortical Control of Gait and Cognition in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS); Poster presentation at the *International Congress for Ataxia Research 2024*, London, EN, Nov. 12-15, 2024.
9. **O’Keefe JA**, Zaidi A*, Ouyang B, Timm EC*, Hall DA. Impact of executive function on balance, gait and falls in FXTAS. *Midwest Fragile X Research Conference 2024* (conference organizer and platform presenter). Chicago, IL, October 4, 2024.
10. Timm EC*, Sica C, Niaz MR, Stuart S, Hall DA, **O’Keefe JA**. The Neural Mechanisms Underlying Cortical Control of Gait and Cognition in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). *Midwest Fragile X Research Conference 2024* (selected for platform presentation by EC Timm, Dr. O’Keefe’s PhD student), Chicago, IL, October 4, 2024.
11. **O’Keefe JA**, Purcell N*, Timm EC*, Acosta RJ, Koyani B, Robertson R, Wipperman MF, Levy O, Kwei K, Hall DA. Initial validity and clinical utility of the Moticon insole system for assessing gait in Parkinson’s disease. Poster presentation at the *2024 International Parkinson Study Group Conference*, Nashville, TN; Dec. 2024.
12. **O’Keefe, J.A**, Timm, E.C.*; Purcell, N.L.*; Ouyang, B.; Berry-Kravis, E.; Hall, D.A. Potential Prodromal Digital Postural Sway Markers for FXTAS Detected via Dual Tasking and Sensory Manipulation. Poster presentation at the *19th NFXF International Fragile X Conference*, Orlando, FL, 2024
13. Timm EC, Paras J, Hall DA, **O’Keefe JA**. The Cortical Control of Gait and Cognition in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). Poster presentation at the *19th NFXF International Fragile X Conference*, Orlando, FL, 2024
14. **O’Keefe JA**, Purcell NL*, Stuart S, Vitorio R, Ouyang, B, Hall DA. Cortical Control of Gait and Balance During Single and Dual Tasks in Huntington’s Disease: an fNIRS imaging study. Poster presentation at the *2023 International Huntington’s Study Group Conference*, Phoenix, AZ, 2023
15. Alyassir, K, Purcell NL*, Timm EC, Goldman J, **O’Keefe JA**. Trunk Instability during Single Task, Dual Task and Fast Paced Gait in Huntington’s Disease. Poster presentation at the *2023 International Huntington’s Study Group Conference*, Phoenix, AZ, 2023.
16. Tosin M, Hessel D, **O’Keefe, JA**, Hall DA. Multiple methods to improve recruitment, enrollment, and retention of study participants for validation of the Fragile X-Associated Tremor Ataxia Syndrome Rating Scale (FXTAS-RS). Poster presentation at the *2023 International Congress of Parkinson's Disease and Movement Disorders*, Copenhagen, Denmark, 2023.

17. Timm EC*, Cao N, Purcell NL*, Ouyang B, Liu Y, Hall DA, **O'Keefe JA**. Impact of dual tasking and sensory manipulation on balance in Fragile X-associated tremor/ataxia syndrome (FXTAS) and potential prodromal postural sway deficits in asymptomatic *FMR1* premutation carriers. Poster presentation at the *2023 International Congress of Parkinson's Disease and Movement Disorders, Copenhagen, Denmark, 2023*.
18. **O'Keefe JA**, Purcell NL*, Stuart S, Vitorio R, Ouyang, B, Hall DA. Cortical Control of Gait and Balance During Single and Dual Tasks in Huntington's Disease: an fNIRS study.
Poster presentation at the *2023 International Congress of Parkinson's Disease and Movement Disorders, Copenhagen, Denmark, 2023*
19. Fleisher JE, Woo K, Sennott BJ, **O'Keefe J**, Gill C, Anderson S, Purcell N, Ouyang B, Chodosh J. Randomized, waitlist-controlled trial of Karate Intervention to Change Kinematic Outcomes in PD (KICK OUT PD). Poster presentation at the *2023 International Congress of Parkinson's Disease and Movement Disorders, Copenhagen, Denmark, 2023*.
20. **O'Keefe JA**, Bang D* , Joyce J*, Armijo N*, Purcell N*, Biskis A, Berry-Kravis E, Hall DA. Feasibility Study of Dual-Task Treadmill Training to Improve Gait and Balance in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). Poster presentation at the *2022 International Congress for Ataxia Research, Dallas TX, Nov.01-05, 2022*.
21. Purcell N, **O'Keefe J**. Cortical control of balance and gait in Huntington's disease. *Journal of Huntington's disease* 11, S35 (2022); N Purcell, Dr. O'Keefe's PhD student, poster presentation at the 2023 International Huntington's Study Group Conference.
22. Robertson-Dick, E, Hall, DA and **O'Keefe, JA** Unique balance impairments distinguish Fragile X-associated Tremor/Ataxia Syndrome from Parkinson disease and essential tremor. Poster presentation at the *International Congress for Ataxia Research 2022, Dallas TX, Nov.01-05, 2022*
23. Timm EC*, Cao N, Purcell NL*, Ouyang B, Liu Y, Hall DA, **O'Keefe JA**. Impact of dual tasking and sensory manipulation on balance in Fragile X-associated tremor/ataxia syndrome (FXTAS) and potential prodromal postural sway deficits in asymptomatic *FMR1* premutation carriers. Poster presentation at the *International Congress for Ataxia Research 2022, Dallas TX, Nov.01-05, 2022*
24. Dual tasking causes increases postural jerk in FXTAS and uncovers prodromal postural sway deficits in Fragile X carriers at risk for FXTAS. Poster presentation at the *Proceedings of the International Society for Posture and Gait Research, 2022, Montreal, CA*.

25. Purcell, N and **O'Keefe JA**. The Cortical Control of Cognition, Balance, and Gait in Huntington's Disease. Poster presentation at the *Proceedings of the International Society for Posture and Gait Research, 2022*, Montreal, CA.
26. **O'Keefe JA**, "The Effects of Dual Task Cognitive Interference and Fast Paced Walking On Gait and Turns in FXTAS"; *The 17th International Fragile X Virtual Conference*; 07/22/2020.
27. **O'Keefe JA**, Early Detection of FXTAS motor symptoms in *FMR1* premutation Carriers. This paper entitled "Prodromal markers of upper limb deficits in *FMR1* premutation carriers and quantitative outcome measures for future clinical trials in FXTAS" ; *The 17th International Fragile X Virtual Conference*; 07/22/2020.
28. **O'Keefe JA**, Joyce J, Purcell N, Wrobel K, Ouyang B, Liu Y, Berry-Kravis E. Efficacy of long term intrathecal 2-hydroxypropyl- β -cyclodextrin treatment on balance and gait deficits in Niemann-Pick Type C1; Poster presentation at the *2019 International Society for Neuroscience Conference*; Oct. 19-23, Chicago, IL.
29. Purcell, N, Goldman, J, Bernard B, **O'Keefe JA**. The Negative Effects of Cognitive Interference on Mobility in Huntington's disease; Poster presentation at the *2019 International Society for Neuroscience Conference*; Oct. 19-23, Chicago, IL.
30. Bang D, Robertson E, Biskis A, Ouyang B, Liu Y, Pal G, Hall DA, Berry-Kravis E. **O'Keefe JA**. Tremor and Bradykinesia Quantification by Inertial Sensor-based Tremorography and Potential for Early Disease Identification in Fragile X- Associated Tremor /Ataxia Syndrome; Poster presentation at the *2019 International Society for Neuroscience Conference*; Oct. 19-23, Chicago, IL.
31. Guan J. Robertson E, Biskis A, Joyce J, Ouyang B, Li, Y, Carnes D, Purcell E, Berry-Kravis E, Hall DA and **O'Keefe, JA**. Dual-task cognitive motor interference exacerbates turn deficits in fragile X-associated tremor/ataxia syndrome (FXTAS), Poster presentation at the *2019 International Society for Neuroscience Conference*; Oct. 19-23, Chicago, IL.
32. **O'Keefe, JA**, Guan J. Robertson E, Biskis A, Joyce J, Ouyang B, Li, Y, Carnes D, Purcell E, Berry-Kravis E, Hall DA and. Dual task cognitive motor interference exacerbates turn deficits in men with FXTAS; *Proceedings of the Fourth International Conference on the FMR1 Premutation*; 2019, September 25-27, 2019, Rotterdam, The Netherlands
33. Bang D, Biskis A, Hall DA, Berry-Kravis E. **O'Keefe JA**. Pilot feasibility study of dual task treadmill training in FXTAS. *Proceedings of the Fourth International Conference on the FMR1 Premutation*; September 25-27, 2019, Rotterdam, The Netherlands
34. **O'Keefe JA**, Bang D, Robertson E, Biskis A, Ouyang B, Liu Y, Pal G, Hall DA, Berry-Kravis E.. Tremor and Bradykinesia Quantification by Inertial Sensor-based

Tremorography and Potential for Early Disease Identification in FXTAS; *Proceedings of the Fourth International Conference on the FMR1 Premutation*; September 25-27, 2019, Rotterdam, The Netherlands

35. **O’Keefe JA**, Voss S, Tracy R, Palmer A, Parulekar M, Purcell N, Fefferman M, Berry-Kravis E. Inertial sensor based normative spatiotemporal gait and Time Up and Go parameters in typically developing children and young adults; Poster presentation at the *Proceedings of the 2019 International Society for Posture and Gait Research*, June 30 –July 5, 2019; Edinburg, Scotland.
36. **O’Keefe, JA**, Palmer A, Voss S, Tracy R, Parulekar M, Purcell N, Fefferman M, Elizabeth Berry-Kravis. 2019 Inertial sensor based normative postural sway parameters in typically developing children and young adults. Poster presentation at the *Proceedings of the 2019 International Society for Posture and Gait Research*, June 30 –July 5, 2019; Edinburg, Scotland.
37. **O’Keefe JA**, Robertson E, McAsey A, Chin J, Palukar M, Berry-Kravis E. Objective inertial sensor based gait outcome measures for efficacy of cyclodextrin treatment in Niemann-Pick Type C1 (NPC); Poster presentation at the *Proceedings of the 2019 International Society for Posture and Gait Research*, June 30 –July 5, 2019; Edinburg, Scotland.
38. Purcell, N, Goldman, J, Bernard B, **O’Keefe JA**. The Effects of Cognitive Interference on Gait and Turning in Huntington’s disease; Poster presentation at the *Proceedings of the 2019 International Society for Posture and Gait Research*, June 30 –July 5, 2019; Edinburg, Scotland.
39. **O’Keefe JA**, Joyce J, Nicollette Purcell, Wrobel K, Parulekar M, Berry-Kravis E. Objective gait and balance outcome measures for efficacy of cyclodextrin treatment in Niemann-Pick Type C1 (NPC1): a case series; Poster presentation at the *Proceedings of the 2019 International Society for Posture and Gait Research*, June 30 –July 5, 2019; Edinburg, Scotland.
40. **O’Keefe, JA** , Carnes D, Joseph Guan, Erin Robertson, BS¹ , Timothy Young, BS³ , Nicollette Purcell, BS¹ , Elizabeth Berry-Kravis, MD, PhD^{2,4,5} , Deborah A. Hall, MD, PhD² Fast Paced Gait May Be More Discriminating Than Dual Tasking for Detecting Severity of Gait and Turn Deficits In Fragile X-associated tremor/ataxia syndrome (FXTAS); Poster presentation at the *Proceedings of the 2019 International Society for Posture and Gait Research*, June 30 –July 5, 2019; Edinburg, Scotland.
41. **O’Keefe, JA**^{a,b}, Erin E. Robertson,^a , Danielle Carnes^a, Timothy Yung^c, Nicollette Purcell^a , Andrew McAsey^a , Maija Swanson^c, Elizabeth Berry-Kravis^{b,d,e}, and Deborah A Hall^b Fast Paced Gait May Be More Discriminating Than Dual Tasking for Detection of the Severity of Gait and Turn Deficits In FXTAS. Poster presentation at the *16th International Fragile X Conference*, Cincinnati Ohio, July 11-15, 2018

42. **O'Keefe, JA**^{1,2}, Erin E. Robertson¹, Bichun Ouyang², Danielle Carnes¹, Andrew McAsey¹, Yuanqing Liu², Maija Swanson³, Bryan Bernard², Elizabeth Berry-Kravis^{2,4,5}, and Deborah A Hall² Cognitive function impacts gait, functional mobility and falls in Fragile X-Associated Tremor/Ataxia Syndrome, *16th International Fragile X Conference*, Cincinnati Ohio, July 11-15, 2018. (Platform presentation).
43. **O'Keefe, JA**, Chin J, McAsey, A, Parulekar M, Berry-Kravis, E. Objective gait and balance outcome measures for efficacy of cyclodextrin treatment in Niemann-Pick Type C1 (NPC1): a case series; Poster presentation at the *2018 Michael, Marcia & Christa Parseghian Scientific Conference*, Tucson, AZ, June 2018.
44. Berry-Kravis E, **O'Keefe J**, Chin J, Friedmann K. Treatment of very young children with intrathecal 2-hydroxy-beta-cyclodextrin (vts-270): experience with 6 NPC1 patients; Poster presentation at the *2018 Michael, Marcia & Christa Parseghian Scientific Conference*, Tucson, AZ, June 2018.
45. **O'Keefe, JA**, Robertson E, McAsey, A, Chin J, Parulekar M, Berry-Kravis, E. Objective inertial sensor based gait outcome measures for efficacy of cyclodextrin treatment in Niemann-Pick Type C1 (NPC1); Poster presentation at the *Clinical Gait and Movement Analysis Society*; Indianapolis, IN, May 2018
46. Purcell, N, Goldman, J, Bernard B, **O'Keefe, JA**; The Effects of Dual-Task Cognitive Interference on Balance in Huntington's disease; *Clinical Gait and Movement Analysis Society*; Indianapolis, IN, May 2018. (N Purcell invited as Platform Presenter)
47. **O'Keefe, JA**, Carnes D, Robertson E, Young T, Purcell, N, Berry-Kravis, E, Hall, DA. Dual-task cognitive motor interference and fast paced walking exacerbates gait deficits in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS); Poster presentation at the *2018 National Ataxia Foundation Investigator's meeting*, Philadelphia PA, April, 2018
48. Hall DA, Robertson-Dick E, Leehey M, McAsey A, Berry-Kravis E, **O'Keefe JA**. Open-label pilot trial of citicoline for fragile X-associated tremor/ataxia syndrome (FXTAS); Poster presentation at the *2018 National Ataxia Foundation Investigator's meeting*, Philadelphia PA, April, 2018
49. **O'Keefe JA**, Robertson E., McAsey, A, Swanson, M, Bernard, B, Berry-Kravis E, Hall DA. Executive function impacts balance and gait in Fragile X premutation carriers with and without Fragile X-Associated Tremor/Ataxia Syndrome; Poster presentation at the *Proceedings of the Third International Conference on the FMR1 Premutation: Basic Mechanisms and Clinical Involvement*, Israel, 2017.
50. **O'Keefe JA**, Parulekar M, Berry A, Huml C, Kravis K, Arca CD, McAsey A, DeSonia A, Berry-Kravis E. 2017. Inertial sensor based normative spatiotemporal gait and postural sway parameters in typically developing children and young adults, Poster presentation at the *Fifth International Conference on Ambulatory Monitoring of Physical Activity and Movement*, Bethesda, MA, June, 2017.

51. **O'Keefe JA**, Robertson E, McAsey A, Chin J, Palukar M, Berry-Kravis E. Objective inertial sensor based gait outcome measures for efficacy of cyclodextrin treatment in Niemann-Pick Type C1 (NPC): Preliminary analysis; Poster presentation at the *Fifth International Conference on Ambulatory Monitoring of Physical Activity and Movement*, Bethesda, MA, June, 2017.
52. Robertson E, McAsey A, Swanson M, Berry A, Huml C, Berry-Kravis E, Hall DA, **O'Keefe JA**. Fragile X-associated tremor/ataxia syndrome, Parkinson disease, and essential tremor subjects demonstrate distinct gait and balance deficits under normal, environmentally challenging, and dual-task conditions. Poster presentation at the *Proceedings of the International Society for Posture and Gait Research*, June 2017, Fort Lauderdale, FL
53. Robertson E, McAsey A, Swanson M, Berry A, Huml C, Berry-Kravis E, Hall DA, **O'Keefe JA**. Fragile X-associated tremor/ataxia syndrome, Parkinson disease, and essential tremor subjects demonstrate distinct gait and balance deficits under normal, environmentally challenging, and dual-task conditions. Poster presentation at the *18th International Congress on Parkinson's Disease and Movement Disorders*, Vancouver, BC, Canada, June 4-8, 2017.
54. **O'Keefe JA**, Potential preclinical gait and balance markers for developing Fragile X-Associated Tremor/ Ataxia Syndrome (FXTAS); Poster presentation at the *2017 Proceedings of the International Society for Posture and Gait Research*, June 2017, Fort Lauderdale, FL
55. **O'Keefe JA**, Potential preclinical gait and balance markers for developing Fragile X-Associated Tremor/ Ataxia Syndrome (FXTAS); Poster presentation at the *2017 International Congress on Parkinson's Disease and Movement Disorders*, Vancouver, BC, Canada, June, 2017.
56. Robertson E, McAsey, A, Swanson, M, Berry-Kravis, E, Hall, DA., **O'Keefe, JA**. Fragile X-associated tremor/ataxia syndrome, Parkinson disease, and essential tremor subjects demonstrate distinct gait and balance deficits under normal, environmentally challenging, and dual-task conditions; Poster presentation at the *International Society for Posture and Gait Research World Congress*; Fort Lauderdale, Florida, June 2017.
57. Berry-Kravis E, **O'Keefe J**, Hoffmann A, Winston A, Stoner R, LaGorio L, Robertson E, Chin J, Friedmann K. Performance on objective clinical efficacy outcome measures during long-term cyclodextrin (VTS-270) treatment in NPC. Poster presentation at the *Michael, Marcia & Christa Parseghian Scientific Conference*, Tucson, AZ, June 2016.
58. Hall DA, Robertson-Dick EE, **O'Keefe JA**, Hadd AL, Zhou L, Berry-Kravis E. Repeat Size and X Inactivation in the clinical phenotype of fragile X premutation carrier sisters: a familial case series. Poster presentation at *The International Parkinson and Movement Disorder Society Congress*, Berlin, 2016.

59. Berry-Kravis E, **O'Keefe J**, Hoffmann A, Winston A, Stoner R, LaGorio L, Robertson E, Chin J, Friedmann K. Performance on objective clinical efficacy outcome measures during long-term cyclodextrin (VTS-270) treatment in NPC. Poster presentation at the *National Niemann-Pick Disease Foundation Family Conference*, Boston, August 2016.
60. **O'Keefe, JA**, McAsey, A, Swanson, M, Robertson E, Berry-Kravis, E, Hall, DA. FMR1 premutation carriers demonstrate worse gait and balance performance at fast paced speeds and under dual task and environmentally challenging conditions; Invited speaker at the *15th International Fragile X Conference*, San Antonio, TX, 2016
61. Robertson E, McAsey, A, Swanson, M, Berry-Kravis, E, Hall, DA., **O'Keefe, JA**. Cognitive dysfunction and balance and gait deficits in FXTAS; invited speaker at the *15th International Fragile X Conference*, San Antonio, TX, 2016
62. Swanson, M., Robertson E., Bernard, B, Berry-Kravis E, Hall DA, **O'Keefe, JA**. Lower executive function negatively impacts gait and balance in Fragile X premutation carriers with and without fragile X-associated tremor/ataxia syndrome (FXTAS), *Poster presentation at the American Academy of Neurology 2016 Annual meeting*.
63. Berry-Kravis E, **O'Keefe J**, Hoffmann A, Winston A, LaGorio L, Robertson E, Chin J, Leurgans S. Objective clinical efficacy outcome measures for cyclodextrin treatment in Niemann-Pick type C (NP-C): a five domain approach. *Poster presentation at Ann Neurol 2015;78:S219*.
64. **O'Keefe J**, Robertson E., Berry-Kravis E, Hall DA. 2015 Executive function is associated with balance and gait in *FMR1* premutation carriers. *Proceedings of the Second International Conference on the FMR1 Premutation: Basic Mechanisms and Clinical Involvement*, Barcelona, Spain.
65. **O'Keefe J**, Robertson E., Hall DA and Berry-Kravis E. 2015 Gait and Functional Mobility Deficits in FXTAS measured with an instrumented Timed Up and Go, *Proceedings of the Second International Conference on the FMR1 Premutation: Basic Mechanisms and Clinical Involvement*, Barcelona, Spain.
66. Robertson E, **O'Keefe J**, Hadd A, Zhou L, Berry-Kravis E., Hall, DA 2015. Repeat Size And X-Inactivation In The Clinical Phenotype Of Fragile X Premutation Carrier Sisters: A Familial Case Series. *Proceedings of the Second International Conference on the FMR1 Premutation: Basic Mechanisms and Clinical Involvement*, Barcelona, Spain.
67. Berry-Kravis E, **O'Keefe J**, Hoffmann A, Winston A, LaGorio L, Robertson E, Chin J, Leurgans S. Objective Clinical Efficacy Outcome Measures for Cyclodextrin Treatment in Niemann-Pick Type C: A Five Domain Approach. *Parseghian Conference on Niemann Pick Type C*, University of Notre Dame, June 2015.

68. Robertson E, Hall, DA, Berry-Kravis E., **O'Keefe J.** 2015 Characterization and early detection of balance and gait deficits in Fragile X premutation carriers with and without fragile X-associated tremor/ataxia syndrome (FXTAS), *Proceedings of the International Society for Posture and Gait Research*, Seville, Spain.
69. Berry-Kravis, E, **O'Keefe,JA**, Pounardjian, T ,Chin, J, Forbes, Porter, D, Ory, D,Alam, Kasturi Haldar 2014. Intermediate Size Patient Population Ind For 2-Hydroxypropyl- β -Cyclodextrin (Hp- β -Cd) Outpatient Treatment of Siblings with Neimann-Pick Type C1 (Np-C1) And Disparate Rates Of Progression: Exploration of Outcome Measures And Biomarkers; *Academy of Child Neurology and Proceedings 2015 and the 2015 Michael, Marcia, and Christa Parseghian Scientific Conference for Niemann-Pick Type C Research*.
70. Berry-Kravis E, **O'Keefe J**, Hoffmann A, Chin J, Pounardjian T, Porter FD, Alam S, Haldar K, Ory D. Intermediate size patient population IND for 2-hydroxypropyl-B-cyclodextrin (HP-B-CD) outpatient treatment of Niemann-Pick type C1 (NP-C1) and disparate rates of progression: exploration of outcome measures and biomarkers. *Ann Neurol.* 2014;76:S215-216.
71. **O'Keefe, JA**, Kohjima JW, Walsh, ME, Berry-Kravis, E, Hall, DA. 2014. Gait and Mobility Deficits in FXTAS using the instrumented Timed Up and Go test; *17th International Congress on Parkinson's Disease and Movement Disorders*, Stockholm, Sweden.
72. **O'Keefe, JA**, Kuo, B, and Hall, DA, and Berry-Kravis, E July 2014, Reliability and validity of a markerless motion capture system to quantify hyperactivity in Fragile X Syndrome. *Proceedings of the 14th International Fragile X Conference*, San Diego, CA.
73. **O'Keefe, JA**, Kohjima JW, Walsh, ME, Berry-Kravis, E, Hall, DA. 2014 Gait and Mobility Deficits in FXTAS using an instrumented Timed Up and Go test, *Proceedings of the 14th International Fragile X Conference*, San Diego, CA.
74. **O'Keefe, JA**, Kohjima JW, Walsh, ME, Berry-Kravis, E, Hall, DA. 2014 Gait and Mobility Deficits in Fragile X-associated tremor/ataxia syndrome (FXTAS) using the instrumented Timed Up and Go test, *Proceedings of The Clinical Gait and Movement Disorder Society*.
75. **O'Keefe, JA**, Fiutko, A, Dunn, EJ, Berry-Kravis,E, Ouyang, B., Hall, DA. 2013 Characterization of balance deficits in *FMRI* premutation carriers. *Proceedings of the 1st International Conference on the FMRI Premutation: Basic Mechanisms and Clinical Involvement*, Perugia Italy.
76. Hall DA, Berry-Kravis E, Goetz CG, Leehey MA, Hagerman R, **O'Keefe JA**, Kehinde T, Stebbins G. Clinimetrics of the FXTAS Rating Scale. 2013. *Proceedings of the 1st International Conference on the FMRI Premutation: Basic Mechanisms and Clinical Involvement*, Perugia Italy.

77. Hall DA, Kehinde T, Berry-Kravis E, Goetz C, **O'Keefe JA**, Stebbins G. Responsivity of the fragile X-associated tremor ataxia syndrome. 2013. *16th International Congress on Parkinson's Disease and Movement Disorders*, Sydney, Australia.
78. **O'Keefe, JA**, Khan H, Trombley, R, Hall, DA, Espinoza Orías, AA, Wimmer, MA, Berry-Kravis, E. 2013. Quantification of hyperactivity and stereotypies in Fragile X Syndrome using a novel markerless motion analyses system; *Proceedings of the Clinical Gait and Movement Disorder Society*, Cincinnati, Ohio.
79. Kehinde T, **O'Keefe, JA**, Berry-Kravis, E and Hall, DA, 2013. "Responsivity of the Fragile X-Associated Tremor Ataxia Syndrome Rating Scale, *American Academy of Neurology*, San Diego, March, 2013.
80. Dunn EJ, Hall, DA, Berry-Kravis, E., Ouyang, B and **O'Keefe JA**. Impaired vestibular control mechanisms underlie balance deficits in Fragile X-associated tremor/ataxia syndrome, *American Academy of Neurology, New Orleans, LA. Neurology; 2012; 78(Meeting Abstracts 1): P06.051*
81. **O'Keefe, JA**, Dunn, EJ, Berry-Kravis,E, Ouyang, B., Hall, DA. Characterization of balance deficits in FMR1 premutation carriers with and without FXTAS reveals a unique profile of impaired vestibular control and postural instability; The Movement Disorder Society, Dublin , Ireland. *Movement Disorders 2012; 27, Suppl 1, pg. S549, # 589*
82. **O'Keefe, JA**, Dunn, EJ, Berry-Kravis,E, Ouyang, B., Hall, DA. 2012 Characterization of balance deficits in FMR1 premutation carriers with and without FXTAS reveals a unique profile of impaired vestibular control and postural instability, *Proceedings of the International Fragile X Research Foundation*, Miami, FL.
83. **O'Keefe, JA**, Khan H,Trombley, R, Hall, DA, Espinoza Orías, AA, Wimmer, MA, Berry-Kravis, E, 2012. Development of a Markerless Motion Analysis Method to Quantify Hyperkinesia and Stereotypies in Males with Fragile X Syndrome; *Proceedings of the International Fragile X Research Foundation*, Miami, FL.
84. **O'Keefe JA** and Kihiko, M. 1996 Protein kinase modulation of NMDA receptors in the neonatal and aged rat hippocampus and cerebral cortex: implications for spatial learning, II Step Conference, *Proceedings of the American Physical Therapy Association*, New Hampshire.
85. Wilson, ME, **O'Keefe, JA**, Price R and Handa RJ. 1995. Androgen receptor (AR) and AR mRNA in primary cultures of rat hippocampus. *Soc. Neurosci Abst., 21:429.*
86. English TE, **O'Keefe, JA** and Kasarkis EJ. 1995 Correlation of the peak torque output of the biceps muscle with the forced vital capacity in people with amyotrophic lateral sclerosis, *Physical Therapy Abstract.*

87. Cheng, B., **O'Keefe, JA**, Goodman, Y and Mattson, MP. 1994 Basic FGF increases AMPA receptor subunit GluR1 expression in hippocampal neurons, *Soc. Neurosci Abst.*, 20:482.
88. **O'Keefe, JA**, Kihiko, M, Goodman, T and Mattson MP. 1994. Evidence that protein kinase activity modulates NMDAR1 subunit expression in the hippocampus and cerebral cortex in vivo. *Soc. Neurosci Abst.*, 20:742.
89. **O'Keefe, JA**, Kihiko, M, Goodman, T and Mattson MP. 1993. Depolarizing stimuli increase trk b protein in embryonic rat hippocampal neurons, *Soc. Neurosci Abst.*, 19:1295.
90. **O'Keefe, JA**, and RJ Handa. 1992. Estrogen receptor mRNA are transiently elevated in the rat hippocampus during the early postnatal period. *Soc. Neurosci Abst.*, 18:232.
91. McGivern, RF, Bollnow, MR, **O'Keefe JA**, and Handa RJ. 1992. Increases in estrogen receptor mRNA in the medial preoptic area of the male rat following long term castration, *Soc. Neurosci Abst.*, 18:893.
92. **O'Keefe, JA**, EB Pedersen, AJ Castro and RJ Handa. 1990. Estrogen receptors in hippocampal and neocortical transplants during development, *Soc. Neurosci Abst.*, 16:327.
93. Pederson, EB., **JA O'Keefe**, RJ Handa and AJ Castro. 1990. Intrinsic regulation of estrogen receptor concentration in neocortical transplants during postnatal development. *Europ. Neurosci. Assoc. Abst. Europ I Neurosci.*, Suppl. 3:80.
94. **O'Keefe, JA** and RJ Handa. 1989. Transient elevation of estrogen receptors in the neonatal rat hippocampus. *Soc. Neurosci Abst.*, 15: 89
95. Sullivan, RC, **O'Keefe, JA**, Harris, GF. 1985. Rotational changes in the lower limb following distal medial hamstring surgery in cerebral palsied children., *Proc Amer. Acad. Cer. Pal. Dev. Med. 39th Ann Mtg*, Seattle, WA.