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Education

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

Academic Appointments

- 2017- Associate Professor, Department of Anatomy & Cell Biology and Neurological Sciences, Rush University, Chicago, IL.
- 2018- Adjunct Associate Professor, Department of Occupational Therapy, College of Health Sciences, Rush University, Chicago, IL.
- 2009 - 2017 Assistant Professor, Department of Anatomy and Cell Biology, Rush University College of Medicine, Chicago, IL.
- 2015- 2018 Adjunct Assistant Professor, Department of Occupational Therapy, College of Health Sciences, Rush University, Chicago, IL.
- 2015- 2017 Adjunct Assistant Professor, Department of Neurological Sciences, Rush University, Chicago, IL.
- 2017 - Associate Professor, Graduate College, Rush University.
- 2014- 2017 Assistant Professor, Graduate College, Rush University.
- 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

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

Employment/Clinical Experience

- 1996–2012 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

- 2021-2023 Huntington’s Disease Society of America Human Biology Fellowship Award
- 2017 Rush Excellence Award in Education
- 2017 Member of the Mark Lepper Society of Teachers for Teaching Excellence
- 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
- 1977-1981 Dean’s List University of Illinois
- 1978 Alpha Lambda Delta Honor Society
- 1977 Illinois State Scholar

Society Memberships

- 2023- Parkinson’s Study Group, Professional Member

- 2023- Huntington's Study Group, Professional Member; Digital Outcomes Working Group member
- 2017- Executive Committee of the Chicago Chapter of the Society for Neuroscience
- 2017- The International Society for the Measurement of Physical Behaviour
- 2014- International Society for Posture and Gait Research
- 2010- Gait and Clinical Movement Analysis Society
- 2009- Movement Disorder Society
- 2009- American Association of Anatomists
- 2009- American Association of Clinical Anatomists
- 1991- American Physical Therapy Association: Neurology, Pediatric and Research section member
- 1998- Society for Neuroscience

TEACHING

Rush University

- 2017- 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. Lecture hours: 30; Laboratory hours: 10.
- 2015- 2018 Co-Block Administrator; M1 CNS/Head and Neck curriculum Block RMD 516. Responsibilities include 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 of gross anatomy practical exams.
- 2015- Course Director: Introduction to Neurobiology ANA 500; This course is for doctoral students in the Rush University College of Nursing's Anesthesia program as well as for graduate students in Rush's Department of Anatomy and Cell Biology. Lecture hours: 30; Laboratory hours: 10.
- 2017- 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 educator for occupational therapy students.
- 2016- Proseminar in Neurobiology; GCC 698-1; Rush Graduate College; 2 lecture hours" "Neurophysiology of gait"
- 2012- Discipline Director; M1 CNS/Head and Neck curriculum Block RMD 516 Lecture hours: 11; Gross anatomy lab hours: 27; 18 Neuro case application workshop hours; Small group i-human case session hours: 8

- 2011- 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- 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 Neurobiology Courses to First Year Medical students. 110 Laboratory hours; Rush Medical College.

Other Institutions

- 1996 Course Director/ Lecturer, “Mechanics of Motor Control”, 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, conducted 5 one week teaching sessions (all day sessions).

Continuing Education Workshops

- 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

MENTORSHIP TO STUDENTS / TRAINEES IN RESEARCH

Rush University

Sabrina Nguyen, MD candidate; Rush University Dean’s Fellowship Scholars program; 2023

Kyle Alyssir, MS; Rush University MS in Biotechnology Program, Primary Mentor; 2022-2023

Emily C Timm, PhD candidate; Primary PhD faculty advisor/mentor 2022-

Christopher Ly, MD candidate; Summer research 2022

Alex Weiss, Undergraduate summer research student 2002

Cassandra Cisneros; January, 2022- present; Primary advisor for research on MRI imaging in Huntington's disease and association with cortical activity; Occupational Therapy Doctorate Student | Class of 2024

Emma Sims; January, 2022- present; Primary advisor for research on MRI imaging in Huntington's disease and association with cortical activity; Occupational Therapy Doctorate Student | Class of 2024

Olyvia Gryzik; January, 2022- present; Primary advisor for research on MRI imaging in Huntington's disease and association with cortical activity; Occupational Therapy Doctorate Student | Class of 2024

Annam Zaidi, MD; Primary Faculty thesis advisor for MS in Clinical Research 2021-2024; "Impact of Cognition on Balance, Gait and Falls in FXTAS".

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.

Diana Abassi, MS; thesis committee member (2019-); 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.

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.

Leora Kramer, 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.

Elizabeth Pierobon, DPT, part-time PhD candidate in the CHS; part-time rotation in my lab began in September, 2019; worked on research training and potential PhD projects.

Sharon Arhin, Mentor for Capstone Research Project for MS in Biotechnology in Rush Graduate College; 2019-2020; applying for medical school.

Harlem Murray Summer 2019; Primary Mentor, Rush Summer Research Scholars Program; *Fine motor coordination outcome measures for efficacy of cyclodextrin treatment in Niemann-Pick Type C1.*

Nicholas Armijo, BS, Primary Faculty MS thesis advisor 2018 to 04/2020; *Effects of a Karate Intervention on Balance and Gait in Parkinson's Disease*. Author on two publications. Applying to medical school.

Anjali Gera, MD; MS in Clinical Research; 2018 to present; *Gait and Balance Analysis of Glucocerebrosidase Mutation Carriers with Parkinson's Disease*. Role: Co-Mentor Primary author on one publication.

Nicollette Purcell, MS, PhD candidate; Primary PhD faculty advisor 2017 to present: *Cortical activation patterns in Huntington's Disease during cognitive and motor tasks*. First author on 2 publications and author on 2 additional papers and 15 scientific abstracts.

Erin Robertson, PhD, Primary faculty advisor 2013- 2018, Department of Anatomy and Cell Biology, Thesis title: "*Balance, gait and cognitive dysfunction in FXTAS versus Essential Tremor and Parkinson's Disease*"; recipient of a 2015 National Fragile X Foundation Fellowship 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 May 2021 from Northwestern University. Presently a Scientist at Regeneron.

David J. Marmion, M.S. 2017- 08/2020; Ph.D. student in Kordower Laboratory; Thesis title: "*Establishment of Novel Viral Vector-Mediated Rat and Nonhuman Primate Models of Multiple System Atrophy*"; Role PhD thesis committee; starting his postdoctoral fellowship 9/2020.

Jessica Karl, MS in PA, 2016- 2020; received her PhD student in College of Health Sciences, Rush University; *Investigating how a Deep Brain Stimulation programming paradigm in Parkinson's disease patients affects gait and balance*; PhD Thesis committee member; junior faculty member in Neurological Sciences and the College of Health Science.

Nicollette Purcell, 2015- 2017; Master's student in Department of Anatomy; Thesis title: "*Dual cognitive task impact on balance and gait in Huntington Disease*"; Primary advisor; 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; Winner 2018 Rush Research Forum Graduate College poster award.

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*"; Caitlin is presently working here at Rush writing up 2 manuscripts from her MS project. Winner 2018 Rush Research Forum Graduate College poster award. First author on a manuscript under revision review in PLOS one.

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; PhD candidate in Cognitive Psychology at the University of Illinois-Champaign/Urbana

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.

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.

Kathryn Wrobel, 2018 M1/M2 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 will be immersed in a program for medical students with exceptional promise to be a clinician scientist. Won a first place award at Rush Research Forum 2018 for her poster presentation.

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”; entered Residency program

Jessica Joyce, BS; Research associate 2018-2019; Author on 3 manuscripts; admitted to Rush Medical School 2022.

Danielle Carnes, PhD; Research associate and PhD student in Neuropsychology 2017-2019; author on 2 publications; presently a practicing Neuropsychologist.

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*”.

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*”.

Colleen Huml, MS, 2016 - 2017; Research Associate; received her MD; in residency

Medha Parulekar, 2016; 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

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 submitted publications.

Andrew McAsey, 2015-16, Research Associate; author on 4 scientific abstracts and two publications; entered residency

Maija Swanson, 2015; Rush Dean’s Summer Research Fellowship Program; “*Effects of Cognitive Function and dual-task interference on balance and gait in preutation carriers of the FMR1 gene*”; entered residency; author on 3 scientific abstracts and one manuscript; entered residency

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.

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.

Joji W Kohjima, Rush Dean’s Summer Research Fellowship Program, 2013; “*Characterization of balance and gait deficits using novel inertial sensor technology in FMR1 preutation carriers*”.

Maura Walsh, Rush Summer Research Intern, 2013; “*Characterization of balance and gait deficits in FMR1 preutation carriers and molecular correlates of disease onset*”.

Brittany Witkiewicz, MS in Anatomy and Cell Biology, Master’s Thesis committee member, 2013

Amber Fuitko, Rush Dean’s Summer Research Fellowship Program, 2012; “*Characterization of balance deficits in FMR1 preutation carriers*”, entered residency; author on one publication.

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

Emily Dunn, Rush Dean’s Summer Research Fellowship Program, 2011; “*Characterization of balance deficits in FMR1 preutation carriers*”, recipient of an American Academy of Neurology travel award; Attending in Neurology; author on 2 scientific abstracts and 1 manuscript.

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

Rachel Neal, 1994-1996
Margo Van Eck, 1995- 1997
Ruth Anne Geiger, 1995-1997
Billie Schanen, 1995-1997; Chair
Steve Langermann, 1995 – 1997, Chair

Invited Grand Rounds and Lectures

O'Keefe JA. Meet the Expert/Mentor Seminar Series, “Cortical Control of Gait and Balance in Huntington's Disease: an fNIRS study”; Rush University, Spring 2023.

O'Keefe JA. “Trunk Instability during Single Task, Dual Task and Fast Paced Gait in Huntington's Disease”; Rush University Movement Disorders Research Day; May 2023

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.

O'Keefe, JA Rush University Neurological Grand Rounds; “Relationship between Cognition and Balance and Gait in Movement Disorders”; October 11, 2016

O'Keefe, JA. "Ataxia and Evidence based practice for Rehabilitative Interventions", University of Illinois, Physical Therapy Program, Chicago, IL. March 30, 2016.

O'Keefe, JA. “Rotational changes in the lower limb following distal medial hamstring surgery in children with cerebral palsy”, Marquette University, 1985.

O'Keefe, JA “Orthotic casting using the CASCADE DAFO system”, Cook County Hospital, Chicago, 1984

O'Keefe, JA “Physical Therapy Management of the Burn Patient”, University of Illinois, Physical Therapy Program, Chicago, 1983.

COMMITTEE AND ADMINISTRATIVE SERVICES

Rush University

2022- Advisory Board, Center for Academic Excellence
2019-2022 Academic Standards Committee, Rush Graduate College
2019-2022 Departmental Advisory Committee, Cell & Molecular Medicine, Rush University
2018- 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- 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 students for Rush University Dean's Fellowship summer research internships

SCIENTIFIC AND SCHOLARLY ACTIVITIES

Membership or Offices in Professional Societies (terms)

2019- 2022 President, Chicago Chapter of the Society for Neuroscience
 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.
 1997-99 Board of Directors, Director of Research, Neurodevelopmental treatment Organization.

Reviewer for Funding Agencies

2019- present Parkinson's Research Program (PRP); Monitoring and Early Detection peer review panel of the Department of Defense Congressionally Directed Medical Research Program (CDMRP)
 2016- 2017 Department of Defense (DOD) Duchenne Muscular Dystrophy Research Program, AdHoc Reviewer

Reviewer for Professional Journals

2021- Editorial Board, Frontiers in Neurology
 2019- Reviewer, Gait & Posture
 2016- Reviewer, Scientific Reports
 2016- Reviewer, The Clinical Neuropsychologist
 2016- Reviewer, The Cerebellum
 2016- Reviewer, Genes
 2016- Reviewer, Journal of Neurology and Neurosurgery
 2014- Reviewer, Clinical Genetics
 2014- Reviewer, Genes, Brain and Behavior
 2014- Reviewer, Geriatric Psychiatry
 2014- Reviewer, Neurology
 2009- Reviewer, Anatomical Sciences Education
 2009- Book reviewer, American Association of Anatomists
 1993- 96 Reviewer, Journal of Physical Therapy Education

NIH and other Scientific Committees

- 2023- Huntington's Study Group; digital outcomes working group member.
- 2020-2021 Orphazyme Treatment Team Steering Committee; advisory for treatment team member's knowledge regarding the drug Arimoclomol for the treatment of Niemann-Pick type C Disease.
- 2018-2019 Member, 2018 NIH Task Force for updating the NIH Research Plan on Fragile X Syndrome and FMR1-related conditions; served on the premutation/FXTAS workgroup; attended a Fragile X strategic planning meeting at the NIH in Bethesda regarding this on 3/8 and 3/9/2018.
- 2014-15 NIH Outcome measures Scientific Advisory Board committee for planning a Phase 2/3 clinical trial in Niemann Pick C type 1. Travelled 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

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

Pending (funded)

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

Role: PI

Total Costs: \$ 315,958

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

Current

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 – 05/31/2024

Huntington's Disease Society of America; "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 to examine cortical functional relationships mediating cognitive, gait and balance impairments in Huntington's disease.

Role: PI

Total direct costs: \$ 150,000

Dates: 11/01/2021 to 4/30/2024

NIH-NINDS 1 U01 NS113851; “Study in Parkinson Disease of Exercise Phase 3 Clinical Trial: SPARX3”

Role: Rush Site Co-Investigator

Costs: 10 % effort

The goal of this project is 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.

Dates: 3/19/2021 - 7/31/2025

Regeneron Pharmaceuticals: Digital Assessment of Gait and Speech in Parkinson’s Disease

Role: Co-Principal Investigator; 10% effort

Major goals are to assess the ability of wearable insole to accurately measure gait in Parkinson disease and detect clinically meaningful change in gait in Parkinson disease.

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

Completed

NIH-NICHHD K01HD088762; “Impact of Cognition on Balance and Gait Markers for FXTAS”

Role: PI; 75% effort

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: \$ 711,083

Dates: 7/1/2016- 06/30/2022 (funded extension)

Clinical Trial Protocol, Retrophin Pharmaceuticals

Rush PI: Berry-Kravis,

01/01/2017-12/31-2022

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.

Role: Co-Investigator: O’Keefe

Clinical Trial Protocol VTS301, Vtesse Pharmaceuticals

Berry-Kravis, Porter (Co-PIs)

09/1/15-12/31/20

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. The goal is to determine if VTS270 is effective for reducing progression of neurological deterioration in NPC1.

Role: Co-Investigator for Rush University

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

Role: PI; 20% effort

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

7/1/2014 – 6/30/2015

Private Philanthropic Donor

Role: Co-PI

Other PI: Markus A. Wimmer

“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.

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”

Role: Co-PI

PI: Elizabeth Berry-Kravis, MD, PhD

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

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

PI; pilot support grant for proposal “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 grant at the University of Kentucky.

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 for grant “FGF
and other growth factor modulation of hippocampal neuron growth and survival”.

Role: PI

Costs: \$ 30,000

1992-1993

Past Philanthropic Support and Service

Fujifilm

Hall (PI)

7/1/2019 - 8/1/2022

“A Phase I/IIa Trial to Evaluate the Safety and Tolerability of FCDI DAPC-1 (iPSC-derived dopamine neuron progenitor cells) in Subjects with Parkinson’s Disease”. This is a first in human study, currently in planning phase, with dopamine neuron transplantation for Parkinson disease.

Role: Sub-Investigator (5% salary support)

2013- 2018 Intermediate size patient population IND for 2-hydroxypropyl-B-cyclodextrin (HP-B-CD) treatment of Niemann-Pick type C1 (NP-C1) Hope for Hayley Foundation, Samantha’s Search for the Cure Foundation, \$75,000; conducted bimonthly gait, balance and neurodevelopmental assessments in patients in this IND.

Role: Sub-investigator; 5 % effort

PI: RUMC site, Elizabeth Berry-Kravis

Current Industry Support and Service

Regeneron

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

Role: Co-PI

% effort: 10

Total Direct costs: \$ 126,000

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

2015-2020 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

Vtesse Pharmaceuticals; \$455,242 directs

Role: Subinvestigator at Rush University Site; 5% effort

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

PI: RUMC site, Elizabeth Berry-Kravis

- 2017- 2019 Efficacy, Safety, And Tolerability Of Fosmetpantotate (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
Retrophin Protocol Number: 024PKAN15004
Role: Subinvestigator at Rush University Site; 2% consultant level effort
My role on this project is to administer a 2 minute walk test and the Functional Independence Measure (FIM) to adults and children (WeeFIM) in this clinical trial
PI at Rush University: Cynthia Comella
- 2020- present A Randomized, Double-Blind, Placebo-Controlled, Parallel-Group Study to Evaluate the Efficacy and Safety of Intravenously Administered BIIB092 in Participants with Progressive Supranuclear Palsy

RUSH UNIVERSITY SERVICE

- 2019- 2023 Graduate College Academic Standards Committee
2019- Committee on Academic Excellence; Graduate College Faculty representative
2018- 2020 Graduate College Council
2017- Rush Schwappe Translational Science Consortium grant reviewer
2017- Rush University Cohn Fellowship reviewer
2016-17 M1/M2 Curriculum block design committee
2015-18 Committee on Curriculum and Evaluation (CCE)
2014- Faculty advisor – Student AMA Association
2013-16 Committee on Senior Faculty Appointments and Promotions (COSFAP)
2013 Junior faculty committee for LCME site visit
2013 Department Liaison to Faculty Development Committee
2012-17 M1 working group committee
2011- Judge, Rush University Research Forum posters

ADDITIONAL RUSH AND GLOBAL SERVICE ACTIVITIES

2014- present Sub investigator in both an IND and industry led clinical trial on the effects of 2-hydroxypropyl-B-cyclodextrin (HP-B-CD) treatment of Niemann-Pick type C1 (NP-C1) for over 3 years. All activities result in a significant contribution to the lives of individuals with

NPC who travel to Rush from numerous states and other countries (Mexico, Costa Rica, and India) to receive these treatments and extensive motor outcome assessments at regular intervals.

COMMUNITY SERVICE

- 2016 - “Overview of Neuroanatomy”, Neuro Camp for Chicagoland High School students; Rush University Medical Center, 7/18/2016, July 2017
- 2009- Workshops/Guided tours in Anatomy Laboratory for numerous high school groups
- 2009- 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.

PUBLICATIONS

Peer Reviewed Journals

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>.

Purcell N, O’Keefe J. Cortical control of balance and gait in Huntington’s disease. *Journal of Huntington's disease* 11, S35 (2022).

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, **Joan A. 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 2016. 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

Robertson-Dick, E, **O'Keefe JA**, Hall DA. Clinical Trials in Fragile X-Associated Tremor/Ataxia Syndrome. In Trials for Cerebellar Ataxias; May 2023; DOI: [10.1007/978-3-031-24345-5_26](https://doi.org/10.1007/978-3-031-24345-5_26)

O'Keefe, JA. Genomics and Genetic syndromes Affecting Movement. In Physical Therapy for Children IV Edition (SK Campbell, RJ Palisano and Orlin, MN Eds), St. Louis, MO, Elsevier, E chapter 2, 2011.

O'Keefe, JA. Genomics and Genetic syndromes Affecting Movement. In Physical Therapy for Children III Edition (SK Campbell, DW Vander Linden and RJ Palisano, Eds), St. Louis, MO, Elsevier, 2006, pp. 217- 255.

Peer Reviewed Abstracts

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). *International Congress of Parkinson's Disease and Movement Disorders, Copenhagen, Denmark, 2023.*

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 *FMRI* premutation carriers. *International Congress of Parkinson's Disease and Movement Disorders, Copenhagen, Denmark, 2023*.

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. *International Congress of Parkinson's Disease and Movement Disorders, Copenhagen, Denmark, 2023*

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). *International Congress of Parkinson's Disease and Movement Disorders, Copenhagen, Denmark, 2023*.

Deborah Bang, MD, MS, MM, Deborah A. Hall, MD, PhD, Jessica Joyce, Nicholas Armijo, Nicolette Purcell, MS, Alexandras Biskis, Elizabeth Berry-Kravis, MD, PhD and, **Joan A. O'Keefe**, PhD, PT. A Feasibility Study of Dual-Task Treadmill Training to Improve Gait and Balance in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). International Congress for Ataxia Research 2022, Dallas TX, Nov.01-05, 2023

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. International Congress for Ataxia Research 2022, Dallas TX, Nov.01-05, 2023

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 *FMRI* premutation carriers. International Congress for Ataxia Research 2022, Dallas TX, Nov.01-05, 2023

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; 2019 International Society for Neuroscience Conference; Oct. 19-23, Chicago, IL.

Purcell, N, Goldman, J, Bernard B, **O'Keefe JA**. The Negative Effects of Cognitive Interference on Mobility in Huntington's disease; 2019 International Society for Neuroscience Conference; Oct. 19-23, Chicago, IL.

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; 2019 International Society for Neuroscience Conference; Oct. 19-23, Chicago, IL.

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), 2019 International Society for Neuroscience Conference; Oct. 19-23, Chicago, IL.

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 *FMRI* Premutation; 2019, September 25-27, 2019, Rotterdam, The Netherlands

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 *FMRI* Premutation; September 25-27, 2019, Rotterdam, The Netherlands

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 *FMRI* Premutation; September 25-27, 2019, Rotterdam, The Netherlands

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; Proceedings of the International Society for Posture and Gait Research, June 30 –July 5, 2019; Edinburg, Scotland.

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. Proceedings of the International Society for Posture and Gait Research, June 30 –July 5, 2019; Edinburg, Scotland.

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); Proceedings of the International Society for Posture and Gait Research, June 30 –July 5, 2019; Edinburg, Scotland.

Purcell, N, Goldman, J, Bernard B, **O’Keefe JA**. The Effects of Cognitive Interference on Gait and Turning in Huntington’s disease; Proceedings of the International Society for Posture and Gait Research, June 30 –July 5, 2019; Edinburg, Scotland.

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; Proceedings of the International Society for Posture and Gait Research, June 30 –July 5, 2019; Edinburg, Scotland.

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); Proceedings of the International Society for Posture and Gait Research, June 30 –July 5, 2019; Edinburg, Scotland.

Joan A. O’Keefe^{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. 16th International Fragile X Conference, Cincinnati Ohio, July 11-15, 2018

Joan A. O’Keefe^{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).

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; 2018 Michael, Marcia & Christa Parseghian Scientific Conference, Tucson, AZ, June 2018.

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; 2018 Michael, Marcia & Christa Parseghian Scientific Conference, Tucson, AZ, June 2018.

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); Clinical Gait and Movement Analysis Society; Indianapolis, IN, May 2018

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

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); 2018 National Ataxia Foundation Investigator’s meeting, Philadelphia PA, April, 2018

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); 2018 National Ataxia Foundation Investigator’s meeting, Philadelphia PA, April, 2018

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; Proceedings of the Third International Conference on the *FMR1* Premutation: Basic Mechanisms and Clinical Involvement, Israel, 2017.

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, Fifth International Conference on Ambulatory Monitoring of Physical Activity and Movement, Bethesda, MA, June, 2017.

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; Fifth International Conference on Ambulatory Monitoring of Physical Activity and Movement, Bethesda, MA, June, 2017.

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. Proceedings of the International Society for Posture and Gait Research, June 2017, Fort Lauderdale, FL and the 18th International Congress on Parkinson's Disease and Movement Disorders, Vancouver, BC, Canada, June 4-8, 2017.

O'Keefe JA, Potential preclinical gait and balance markers for developing Fragile X- Associated Tremor/ Ataxia Syndrome (FXTAS); Proceedings of the International Society for Posture and Gait Research, June 2017, Fort Lauderdale, FL and the 18th International Congress on Parkinson's Disease and Movement Disorders, Vancouver, BC, Canada, June, 2017

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; International Society for Posture and Gait Research World Congress; Fort Lauderdale, Florida, June 2017.

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. Michael, Marcia & Christa Parseghian Scientific Conference, Tucson, AZ, June 2016.

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. The International Parkinson and Movement Disorder Society Congress, Berlin, 2016.

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. National Niemann-Pick Disease Foundation Family Conference, Boston, August 2016.

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; the 15th International Fragile X Conference, San Antonio, TX, 2016

Robertson E, McAsey, A, Swanson, M, Berry-Kravis, E, Hall, DA., **O'Keefe, JA**. Cognitive dysfunction and balance and gait deficits in FXTAS; the 15th International Fragile X Conference, San Antonio, TX, 2016

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), American Academy of Neurology 2016 Annual meeting; in press.

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. *Ann Neurol* 2015;78:S219.

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.

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.

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.

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.

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.

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.

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.

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.

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.

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.

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.

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