Curriculum Vitae

Joan A. O'Keefe, PhD, PT
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Rush University
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Education

1981	BS 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 Cell & Molecular Medicine and Neurological Sciences, Rush University, Chicago, IL.
2018-	Adjunct Associate Professor, Department of Occupational Therapy, College of Health Sciences, Rush University, Chicago, IL.
2009 -	Assistant Professor, Department of Anatomy and Cell Biology, Rush University College of Medicine, Chicago, IL.
2015-	Adjunct Assistant Professor, Department of Occupational Therapy, College of Health Sciences, Rush University, Chicago, IL.
2015-	Adjunct Assistant Professor, Department of Neurological Sciences, Rush University, Chicago, IL.
2014-	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

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

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: 23; Laboratory hours: 10.

2015- **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: 23; 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

Nicollette Purcell, MS, Primary PhD faculty advisor 2017 to present: Dual task Cognitive interference in Huntington's Disease.

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 6 publications. 2017 1st Place winner in the 34th Annual Rush Forum for Research and Clinical Investigation Poster Competition, Graduate College Category; entering Clinical Doctorate program in Audiology September 2018, Northwestern University.

David J. Marmion, M.S. 2017-present; 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.

Jessica Karl, MS in PA, 2016- present; 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.

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; 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.

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; writing a publication.

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 involved in one publication in preparation.

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 involved in one publication in preparation.

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".

Joseph Guan, 2018 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".

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; attends medical school.

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.

Alexandra Bery, 2016; Rush Dean's Summer Research Fellowship Program; "Impact of cognition on balance and gait markers for FXTAS"; M3; submitted 1 scientific abstract and author on 2 submitted publications

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

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.

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 Hyperkinesis 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 premutation carriers".

Maura Walsh, Rush Summer Research Intern, 2013; "Characterization of balance and gait deficits in FMR1 premutation 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 premutation carriers", entered residency; author on one publication.

Avram Fraint, 2011-2012, medical student, Project: "Development of a Markerless Motion Analysis System to Quantify Hyperkinesis 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 premutation carriers", recipient of an American Academy of Neurology travel award; resident in Neurology; author on 2 scientific abstracts and 1 manuscript.

University of Illinois-Chicago
Savitha Subramanium; Department of Physical Therapy; 2015 to present; PhD Committee member

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 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

2018-	Integrated Biomedical Sciences PhD program Council member; Rush Graduate College
2018-	Advisory Council, Center for Student Success, Rush University
2016-	Member, M1/M2 curriculum design task force
2015-	Member, Committee on Curriculum and Evaluation, Rush Medical College
2013-16	Member, Committee on Senior Faculty Academic Promotions (COSFAP)
2012-	Member, M1 Working Group Committee, Rush Medical College
2012-	Faculty Advisor, Rush University Student American Medical Association
2012-	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-	Member, International FXTAS Consortium (IFC)
2018-	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.
2015- 2018	Member, Scientific Content Committee, International Society for Posture and Gait Research
1997-99	Board of Directors, Director of Research, Neurodevelopmental treatment Organization.

Reviewer for Funding Agencies

2016-	Department of Defense (DOD) Duchenne Muscular Dystrophy Research Program, AdHoc Reviewer

Reviewer for Professional Journals

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

2018 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 – present Scientific Advisory Board, Vtesse Pharmaceuticals, clinical trial design and treatment of Niemann-Pick type C

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

Current

NIH-NICHD 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 tremor/ataxia syndrome (FXTAS) using balance and gait markers and interacting cognitive and molecular factors.
Total costs: $566,927
7/1/2016- 6/30/2021

Completed

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 Hyperkinesis 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 Hyperkinesis 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

Current Philanthropic Support and Service

2013- present Intermediate size patient population IND for 2-hydroxypropyl-B-cylcodextrin (HP-B-CD) treatment of Niemann-Pick type C1 (NP-C1) Hope for Hayley Foundation, Samantha's Search for the Cure Foundation, $75,000; Currently conducting bimonthly gait, balance and neurodevelopmental assessments in 9 patients in this IND; Thus far my team and I have conducted ~ 100 assessments accounting for ~ 200 hours of research time.
Role: Subinvestigator; 5 % unpaid effort
PI: RUMC site, Elizabeth Berry-Kravis

Current Industry Support and Service

2015-present 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. Thus far my team and I have conducted ~ 220 assessments on 15 patients accounting for ~ 400 hours of research time
PI: RUMC site, Elizabeth Berry-Kravis

2017- present 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 StudyWith An Open-Label Extension
Retrophin Protocol Number: 024PKAN15004
Role: Subinvestigator at Rush University Site; 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

RUSH UNIVERSITY SERVICE

2107-	Rush Schweppe Translational Science Consortium grant reviewer
2017-	Rush University Cohn Fellowship reviewer
2016-17	M1/M2 Curriculum block design committee
2015-	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-cylcodextrin (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 (which I and my team conduct). My work on these projects resulted being featured in a People Magazine video where a family (and other families) with NPC describe their experiences with the disease and testing at Rush University: http://people.com/pen/00000158-3295-d6e5-a15b-f79d34930000/0000015a-8250-d8c1-ad7a-837325c50000/

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

Submitted and under review

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); Cerebellum, 2019.

Robertson E., Hall DA, McAsey A, Ouyang B, Berry-Kravis E, **O'Keefe JA.** Different tremor profiles in Fragile X-associated Tremor/Ataxia Syndrome, Parkinson disease and essential tremor, 2019; under revision review.

Peer Reviewed Journals

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; doi:10.1002/mdc3.12720.

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

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

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. Robertson [1] , Bichun Ouyang [2] , Danielle Carnes [1] , Andrew McAsey [1] , Yuanqing Liu [2], Maija Swanson [3] , Bryan Bernard [2], Elizabeth Berry-Kravis[2,4,5], and Deborah A Hall [2] 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).

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