

## Curriculum Vitae

### Personal Information<sup>1</sup>

**Jacinda Sampson MD PhD**  
**Dept. Of Neurology and Neurological Sciences**  
**Division of Neuromuscular Medicine**  
**Stanford Neuroscience Health Center**  
**213 Quarry Rd, M/C 5979**  
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**jacindas@stanford.edu**

### Education

Year	Degree	Institution	Field/Discipline	Honor/Award
7/1991 – 6/2000	MD	University of Alabama School of Medicine	MD/PhD program	
7/1991 – 6/1999	PhD	University of Alabama	Biochemistry	
8/1985 – 5/1991	BA	St. Mary's College of Maryland	Biology, Music	Magna cum laude, Brent-Calvert Scholarship

### Residency and Fellowship Training<sup>2</sup>

Year(s)	Title	Specialty/Discipline	Place of Training
7/2004 – 6/2006	Fellowship	Neurogenetics	University of Utah School of Medicine
7/2001 – 6/2004	Residency	Neurology	University of Utah School of Medicine
7/2000 – 6/2001	Internship	Internal Medicine	University of Utah School of Medicine

<sup>1</sup> Exclude personal contact information unless it is used for business purposes. Exclude personal identifying information such as date of birth, birthplace, gender, race/ethnicity, citizenship, marital status, hobbies and interests.

<sup>2</sup> List here all internships, residencies, clinical and research fellowships.

**Licensure and Certification**

Year	Type of License or Certification	Registration #	State Issuing
2015-present	Medical License	C135776	California
2012 - 2017	Medical License	264588-1	New York
2001 – 2014	Medical License	4978808-1205, -8905	Utah
2001 - 2014	DEA Certificate	BS7645762	United States
2005-2015 MOC renewal 2015	Neurology	53426	American Board of Psychiatry and Neurology

**Academic Appointments**

Year(s)	Academic Title	Institution
8/1/2015	Associate Professor of Neurology	Stanford University Medical Center
07/2012 – 7/31/2015	Assistant Professor of Neurology, Neurogenetics Clinic Director, Adjunct Assistant Professor Pediatrics (3/2014)	Columbia University Medical Center, Department of Neurology
07/2010- 07/2012	Assistant Professor, Clinical Track	University of Utah School of Medicine
07/2006- 07/2010	Assistant Professor, Tenure Track	University of Utah School of Medicine

**Clinical Teaching Responsibilities**

Year(s)	Role	Institution/Organization
2013-2014	Instructor, small group teaching, Advanced Neurology exam	CUMC

**Other Teaching Experience<sup>3</sup>**

Date(s)	Course/Lecture Name	Institution
3/7/2014	Ethics of Human Subjects Research: “Genetic testing: diagnostics, clinical research, and the space between	CUMC
9/25/2013	“The Body: In health and disease II: Neuroscience”	CUMC
5/22/2013	“Amyotrophic Lateral Sclerosis: Clinical	CUMC

<sup>3</sup> List here presentations to students and trainees. Limit these to the 6 most significant or recent.

	care”	
3/7/2013	“How to Draw a Pedigree”	CUMC
2010	Nervous & Musculoskeletal Exams, Nervous System & Exam Demo, Intro to Cranial Nerve Exam & Demonstration, HEENT Practice	University of Utah School of Medicine

#### **Students / Trainees<sup>4</sup>**

Name	Relationship	Year Trained	Current Title	Current Institution
David Sant	Advisor/Mentor (undergrad)	2011 – 2013	Graduate Student	University of Miami
Missy Whipple	Mentor (undergrad)	2011 – 2013	Medical Student	University of Utah
Jackie Whitesell	Advisor, research project (neuromuscular fellow)	2008-2009	Neurologist	Saint Alphonsus Medical Group Neurology Clinic

#### **Hospital or Affiliated Institution Appointments**

Year(s)	Title	Hospital/Affiliated Institution
8/1/2015-present	Clinical Associate Professor of Neurology- Consulting	Lucile Packard Children’s Hospital
8/1/2015-present	Clinical Associate Professor of Neurology	Stanford Hospitals and Clinics
07/2012 – 07/2015	Assistant Professor of Neurology, Neurogenetics Clinic Director, Adjunct Assistant Professor Pediatrics (3/2014)	New York Presbyterian Hospital, Department of Neurology

#### **Hospital and Health Care Organization Service Responsibilities**

Year(s)	Role	Institution/Organization
07/2012 – 7/2015	Neurogenetics Clinic Director	Columbia University Medical Center, Department of Neurology

<sup>4</sup> List three to five students/trainees. Provide a mix of current and former, if possible.

**Major Administrative Responsibilities or Committee Assignments<sup>5</sup>**

Year(s)	Title	Institution (local, national, government, foundation)
2006 - 2011	Neurology & Neurosurgery Grand Rounds Organizer	University of Utah Department of Neurology

**Other Professional Positions<sup>6</sup>**

Year(s)	Position/Title	Institution/Company
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**Community Service Related to Professional Work**

Year(s)	Title	Organization
2008	Panelist, Utah Professional Chaplains Association, Annual Conference of the Utah Professional Chaplains Association, St. Mark's Hospital	University of Utah School of Medicine
2010 - 2012	Member, Utah Physicians for a Healthy Environment	University of Utah School of Medicine
2011	Preceptor, Junior League of Salt Lake Community Care Fair, Annual Salt Lake C.A.R.E Fair event offering free healthcareservices to underserved members of the community	University of Utah School of Medicine
2010 - 2012	Panel Member, Institutional Review Board, University of Utah	University of Utah School of Medicine
2014	MDA Summer Camp physician (Long Island, NY)	Muscular Dystrophy Association

**Professional Societies<sup>7</sup>**

Year(s)	Role <sup>8</sup>	Society
2013 - present	Member	American Society of Human Genetics
2005 - Present	Member	World Muscle Society
2005 - Present	Member	International Restless Legs Syndrome Study Group
2001 – Present	Member	American Academy of Neurology

<sup>5</sup> Indicate if you held a leadership role.

<sup>6</sup> List only those positions that are related to your professional field.

<sup>7</sup> Societies to your professional field. Indicate if you held a leadership role.

<sup>8</sup> E.g., member; officer; committee assignment

### **Editorial Boards**

Year(s)	Role <sup>9</sup>	Journal
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### **Post-Degree Honors and Awards**

Year(s)	Honor/Award	Grantor
2014	Stephen Q. Shafer Award for Humanism	Neurology Residents, CUMC

### **Other Study and Research Opportunities<sup>10</sup>**

Year(s)	Title/Role	Institution/Funder	Description
2014-2015	Principal Investigator	Sarepta	Promovi: An open-label, multi-center, 48-week study with a concurrent untreated control arm to evaluate the efficacy and safety of Eteplirsen in Duchenne Muscular Dystrophy
2014-2015	Principal Investigator	PTC Therapeutics	ACT DMD: A phase 3 efficacy and safety study of ataluren (PTC124) in patients with nonsense mutation dystrophinopathy
2013	Principal Investigator	GlaxoSmithKline	An Exploratory study to assess two doses of GSK2402968 in the treatment of ambulant boys with Duchenne Muscular Dystrophy
2011-2015	Principal Investigator	PTC Therapeutics	An open label study of low dose ataluren in patients with nonsense mutation dystrophinopathy
2008	Principal Investigator	PTC Therapeutics	A Phase 2a Extension Study of PTC124 in Subjects with Nonsense-Mutation-Mediated Duchenne Muscular Dystrophy
2008	Principal Investigator	PTC Therapeutics	A Phase 2b Extension Study of PTC124 in Subjects with Nonsense-Mutation Mediated Duchenne and Becker Muscular Dystrophy
2007	Co-investigator	Insmmed	A Placebo-Controlled, Randomized, Double-Blind Phase II Clinical Trial to

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<sup>10</sup> If you have sponsored research to list include in the description your role (e.g. PI, Co-Investigator), the granting agency, the award type (e.g. K-08, R-01). Indicate whether the funding is current, pending approval, future or past. List all patents awarded. Exclude dollar amount of the sponsored research.

			Evaluate Tolerability, Safety, and Efficacy Endpoints after Administration of Recombinant Human Insulin-like Growth Factor -1/Recombinant Human Insulin-like Growth Factor Binding Protein 3
2006 - 2007	Co-investigator	Wyeth	A Double-Blind, Placebo Controlled, Randomized, Multiple Ascending Dose, Safety Study of MYO-029 Administered to Adult Patients with Becker, Fascioscapulohumeral and Limb-Girdle Muscular Dystrophy
2006	Co-investigator	PTC Therapeutics	A Phase 2 Study of PTC 124 as an Oral Treatment for Nonsense-Mutation-Mediated Duchenne Muscular Dystrophy

### Bibliography<sup>11</sup>

#### Peer-Reviewed Journal Articles (original work)

Author(s)	"Article Title"	<i>Journal</i>	Volume (Year):	Page(s)
Bushby K, Finkel R, Wong B, Barohn R, Campbell C, Comi GP, Goemans N, Jones K, Mercuri E, Ryan MM, Tulinius M, Voit T, Moore SA, Sweeney HL, Abresch RT, Coleman KL, Eagle M, Florence J, Gappmaier E, Glanzman AM, Henricson E, Barth J, Elfring G, Reha A, Spiegel RJ, O'Donnell, MW, Peltz SW, McDonald CM, <b>PTC-124-GD-007-DMD Study Group.</b>	Ataluren treatment of patients with nonsense mutation dystrophinopathy	<i>Muscle Nerve</i>	50(4): (2014)	477-87
Kariya S, <b>Sampson JB</b> , Northrup LE, Luccarelli CM, Naini AB, Re DB, Hirano M, Mitsumoto H.	Nuclear localization of SMN and FUS is not altered in fibroblasts from patients with sporadic ALS.	<i>Amyotrophic Lateral Sclerosis and</i>	15 (7-8): (2014)	581-7.

<sup>11</sup> Format author name "Last, First M." If there are multiple authors bold your name. Include current submissions designated as "(in press)." Distinguish between peer-reviewed (intended to delineate original work) and non-peer-reviewed publications. References to large collaborative projects may include annotations explaining your individual contributions.

	Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration	<i>Frontotemporal Degeneration</i>		
Tazen S, Figueroa K, Kwan JY, Goldman J, Hunt A, Sampson J, Gutmann L, Pulst SM, Mitsumoto H, Kuo SH	Amyotrophic lateral sclerosis and spinocerebellar ataxia 2 in a family with full CAG repeat expansions of ATN2	<i>JAMA Neurol</i>	Oct 70(10): 2013	1302-4
Churko JM, Kelly JJ, Macdonald A, Lee J, <b>Sampson J</b> , Bai D, Laird DW	The G60S Cx43 mutant enhances keratinocyte proliferation and differentiation	<i>Exp Dermatol</i>	Aug 21(8): (2012)	612-8
Walker KA, <b>Sampson JB</b> , Skalabrin EJ, Majersik J.	Clinical Characteristics and Thrombolytic Outcomes of Infective Endocarditis-Associated Stroke.	<i>The Neurohospitalist</i>	2(3): (2012)	87-91
Flanigan KM, Dunn DM, von Niederhausern A, Soltanzadeh P, Howard MT, <b>Sampson JB</b> , Swoboda KJ, Bromberg MB, Mendell JR, Taylor L, Anderson CB, Pestronk A, Florence J, Connolly AM, Mathews KD, Wong B, Finkel RS, Bonnemann CG, Day JW, McDonald C, Weiss RB.	Nonsense mutation-associated Becker muscular dystrophy: interplay between exon definition and splicing regulatory elements within the DMD gene.	<i>Hum Mutat.</i>	Mar 32(3): (2011)	299-308.
Soltanzadeh, P, Friez MJ, Dunn DM, von Niederhausern A, Gurvich OL, Swoboda KJ, <b>Sampson JB</b> , Pestronk A, Connolly A, Florence J, Finkel R, Bonnemann CG, Medne L, Mendell JR, MATHews KD, Wong B, Sussman MD, Zonana J, Kovak K, Gappmaier E, Taylor LE, Howard MT, Weiss RB, Flanigan KM.	Clinical and Genetic Characterization of Manifesting Carriers of DMD Mutations.	<i>Neuromuscul Disord,</i>	Aug 20(8): (2010)	499-504.
Zhang F, Potocki L, <b>Sampson JB</b> , Liu P, Sanchez-Valle A, Robbins-Furman P, Deli Navarro AD, Wheeler PG, Spence JE, Brasington CK,	Identification of the predicted 5-Mb uncommon recurrent Potocki-Lupski syndrome associated duplication and the	<i>Am J Hum Genet.</i>	Mar 12, 86(3): (2010)	462-70

Withers MA, Lupski JR.	distribution of rearrangement types and mechanisms in PTLs.			
Churko J, Shao Q, Gong G, Swoboda K, <b>Sampson JB</b> , Laird D	Human Dermal Fibroblasts Derived from Oculodentodigital Dysplasia patients suggest that patients may have wound healing defects.	<i>Hum Mutat</i>	Apr, 32(4): (2011)	456-66
Flanigan KM, Dunn DM, von Niederhausern A, Soltanzadeh P, Gappmaier E, Howard MT, <b>Sampson JB</b> , Mendell JR, Wall C, King WM, Pestronk A, Florence JM, Connolly AM, Mathews KD, Stephan CM, Laubenthal KS, Wong BL, Morehart PJ, Meyer A, Finkel RS, Bonnemann CG, Medne L, Day JW, Dalton JC, Margolis MK, Hinton VJ, Weiss RB.	Mutational spectrum of DMD mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort.	<i>Hum Mutat</i>	30(12): (2009)	1657-66
<b>Sampson JB</b> , Smith SM, Smith AG, Singleton JR, Chin S, Pestronk A, Flanigan KM.	Paraneoplastic myopathy: response to intravenous immunoglobulin.	<i>Neuromuscul Disord</i>	17(5): (2007)	404-8
Davis I, Zhu CS, <b>Sampson JB</b> , Crow JP, Matalon S.	Inhibition of human surfactant protein A function by oxidation intermediates of nitrite.	<i>Free Radic Biol Med</i> ,	33(12): (2002)	1703-13
<b>Sampson JB</b> , Beckman JS.	Hydrogen peroxide damages the zinc-binding site of zinc-deficient Cu, Zn superoxide dismutase.	<i>Arch Biochem Biophys</i>	392(1): (2001)	8-13
Estevez AG, <b>Sampson JB</b> , Zhuang YX, Spear N, Richardson GJ, Crow JP, Tarpey MM, Barbeito L, Beckman JS.	Liposome-delivered superoxide dismutase prevents nitric oxide-dependent motor neuron death induced by trophic factor withdrawal	<i>Free Radic Biol Med</i>	28(3): (2000)	437-46
Estevez AG, Crow JP, <b>Sampson JB</b> , Reiter C, Zhuang Y, Richardson GJ, Tarpey MM, Barbeito L, Beckman JS	Induction of nitric oxide-dependent apoptosis in motor neurons by zinc-deficient superoxide	<i>Science</i>	286(5449): (1999)	2498-500



	dismutase			
<b>Sampson JB</b> , Ye, H Rosen, Beckman JS.	Myeloperoxidase and horseradish peroxidase catalyze tyrosine nitration in proteins from nitrite and hydrogen peroxide	<i>Arch Biochem Biophys</i> ,	356(2): (1998)	207-13
Crow JP, <b>Sampson JB</b> , Zhuang Y, Thompson JA, Beckman JS.	Decreased zinc affinity of amyotrophic lateral sclerosis-associated superoxide dismutase mutants leads to enhanced catalysis of tyrosine nitration by peroxynitrite.	<i>J Neurochem</i> ,	69(5): (1997)	1936-44
Benveniste EN, Kwon J, Chung WJ, <b>Sampson J</b> , Pandya K, Tang LP.	Differential modulation of astrocyte cytokine gene expression by TGF- $\beta$ .	<i>J Immunol</i> ,	153(11): (1994)	5210-21

#### Non Peer-Reviewed Journal Articles

Author(s)	"Article Title"	Journal	Volume (Year):	Page(s)
Patel A, <b>Sampson J</b> .	Cognitive Profile of C9orf72 in Frontotemporal Dementia and Amyotrophic Lateral Sclerosis	<i>Curr Neurol Neurosci Rep</i>	15 (2015)	582
Ulane C, Teed S, <b>Sampson J</b> .	Recent Advances in Myotonic Dystrophy Type 2.	<i>Current Neurological and Neuroscience Reports</i>	Feb 14(2): (2014)	429

#### Book Chapters

Author(s)	Chapter	Book	City Published:	Publisher	Year
Goldman J, <b>Sampson JB</b> .	Chapter 34: Genetic testing and DNA diagnosis.	Merritt's Neurology, 13 <sup>th</sup> edition.	Philadelphia PA	Lippencott, Williams and Wilkins	<i>In press</i>
Dalton J, Goldman J (ed), <b>Sampson JB</b>	"18: Overview of the Adult Muscular Dystrophies; 19: The Myopathies; 20: The Muscular Dystrophies; 21: The Myotonic Dystrophies"	<i>Genetic Counseling for Adult Neurogenetic Disease</i> .	New York, NY	Springer	2015

	“25: The Neurological Examination and Testing”				
Crow JP, <b>Sampson JB</b> , Beckman JS.	Peroxynitrite as a Mediator of Injury in Brain Ischemia, Atherosclerosis, and Amotrophic Lateral Sclerosis.	<i>Frontiers in Cerebrovascular Disease: Mechanism, Diagnosis, and Treatment</i>	Armonk, NY	Futura Publishing Co.	1998
<b>Sampson JB</b> , Rosen H, Beckman JS.	Peroxynitrite-dependent tyrosine nitration catalyzed by superoxide dismutase, myeloperoxidase, and horseradish peroxidase	<i>Methods in Enzymology</i>	New York, NY	Academic Press.	1996
<b>Sampson JB</b> , Crow JP, Strong M, Beckman JS.	Copper/Zinc Superoxide Dismutases in CNS	<i>Mineral and Metal Neurotoxicology</i>	Boca Raton, FL	CRC Press Inc.	1996

### Books

Author(s)	Title	City Published	Publisher	Year
none				

### Papers and Posters Presented at Meetings<sup>12</sup>

Author(s)	Paper/Poster Title	presented at Conference Name and Title	Conference Number	Date	Place
<b>Sampson JB</b> , Baird L, Stevens J, Matsunami N, Leppert M	Exome Sequencing of Two Individuals with Early Onset Familial Willis Ekbom Syndrome.	World Association of Sleep Medicine/Canadian Sleep Society Congress		2011	Quebec City, Canada.
<b>Sampson J</b> , Otterud B, Baird L, Leppert M	Self Report of Restlessness is Not Enough: Exclusion of Secondary Causes of RLS in Utah Kindreds and Search for Genetic	International Restless Legs Study Group Scientific Meeting,		2008	Baltimore, MD

<sup>12</sup> List here talks, lectures, invited presentations and addresses to regional, national and international meetings. Limit these to the 6 most significant or recent.

	Linkage to Published Loci				
<b>Sampson J,</b> Beckman J	Clues to peroxidase plus hydrogen peroxide plus nitrite catalysis of protein tyrosine nitration	Peroxynitrite Conference		1999	Crete, Greece
Crow J, <b>Sampson J,</b> Beckman J	Acquired function for SOD mutants in amyotrophic lateral sclerosis (ALS): formation of zinc-deficient SOD enhances nitration of neurofilaments of peroxynitrite	Oxygen Society Meeting		1995	Pasadena, CA

### Special Materials (editorials, book reviews, letters, epitomes)

Author(s)	"Title"	<i>Journal</i>	Volume (Year):	Page(s)
none				

### Digital Publications (software, on-line writings, other scholarly digital creations)

Author(s)	"Website Article"	<u>Website Name</u>	Day Month Year Electronically Published	<URL>
<b>Sampson JB,</b> Teed S, Goldman J.	Neurogenetics and Genetic and Genomic Testing	<i>MedLink</i>	2014	<a href="http://www.medlink.com">www.medlink.com</a>
<b>Sampson JB,</b> Kerr L (ed)	Myotonic dystrophy and Duchenne muscular dystrophy modules	<i>Medical Home Portal</i>	2010	www.medicalhomeportal.org

### Abstracts Not Published in Other Forms<sup>13</sup>

Author(s)	Paper/Poster Title	presented at Conference Name and Title	Conference Number	Date	Place
Churko J, Shao Q,	Mapping Oculodentodigital	American Society of Cell Biology		2008	San Francisco,

<sup>13</sup> Limit these to the 6 most significant or recent.

<b>Sampson J,</b> Flanigan K, Swoboda K, Laird D	Dysplasia-Linked Genetic Mutations in Cx43 to an Epidermal Phenotype Using Cell, Mouse, and Human Reference Models.				CA
J. Whitesell, <b>J. Sampson,</b> K. Whitehead.	Holter Monitoring versus Electrocardiogram (ECG) for the Detection of Cardiac Arrhythmia in Myotonic Dystrophy.	American Academy of Neurology	61st Annual Meeting	2009	Seattle, WA.
Walker K, <b>Sampson J,</b> Olsen J, Skalabrin E	Diagnosing Infective Endocarditis in the Thrombolytic Window of Acute Ischemic Stroke: A Retrospective Case Series from the University of Utah Stroke Database.	American Academy of Neurology	61st Annual Meeting	2009	Seattle, WA.
<b>Sampson JB,</b> Brimley C, Baird L, Leppert M.	Application of Actigraphy to Phenotypic Characterization of Familial Restless Legs Syndrome: An Objective Measure for a Subjective Disorder?	American Neurological Association	135 <sup>th</sup> annual meeting	2010	San Francisco, CA.