Stanford

Jacinda Sampson

Clinical Professor, Neurology & Neurological Sciences

Curriculum Vitae available Online

CLINICAL OFFICES

- **Stanford Neuroscience Health Center**

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ACADEMIC CONTACT INFORMATION

- **Alternate Contact**

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Bio

BIO

Dr. Jacinda Sampson received her MD and a PhD in biochemistry from University of Alabama at Birmingham, and completed her neurology residency and neurogenetics fellowship at the University of Utah. She served at Columbia University Medical Center prior to joining Stanford University Medical Center in 2015. Her areas of interest include myotonic dystrophies, Duchenne muscular dystrophy, and neurogenetic disorders such as neurofibromatosis, hereditary spastic paraparesis, spinocerebellar ataxia, among others. She is interested in clinical trials for treatment of neurogenetic disorders, and is the clinical application of next-generation genomic sequencing to genetic testing.

CLINICAL FOCUS

- Neurology
- Neurogenetics

ACADEMIC APPOINTMENTS

- Clinical Professor, Neurology & Neurological Sciences

HONORS AND AWARDS

- Stephen Q. Shafer Award for Humanism in Neurology, Columbia University Neurology Residents (2014)

PROFESSIONAL EDUCATION

- Fellowship: University of Utah School of Medicine Registrar (2006) UT
- Board Certification: Neurology, American Board of Psychiatry and Neurology (2005)
- Residency: University of Utah School of Medicine (2004) UT
- Internship: University of Utah School of Medicine (2001) UT
- Medical Education: University of Alabama at Birmingham (2000) AL
- Fellowship, University of Utah School of Medicine , Neurogenetics (2006)
- Residency, University of Utah School of Medicine , Neurology (2004)

- Internship, University of Utah School of Medicine , Internal Medicine (2001)
- PhD, University of Alabama , Biochemistry (1999)
- MD, University of Alabama School of Medicine , Medical Degree (2000)

LINKS

- Muscular Dystrophy Association (MDA): www.mda.org
- Myotonic Dystrophy Foundation: http://www.myotonic.org
- Parent Project Muscular Dystrophy: http://www.parentprojectmd.org
- Get a Second Opinion: https://stanfordhealthcare.org/second-opinion/overview.html

Research & Scholarship

CLINICAL TRIALS

- Safety, Tolerability and Pharmacokinetics of ERX-963 in Adults With Myotonic Dystrophy Type 1, Not Recruiting

Publications

PUBLICATIONS

- **Consensus-based care recommendations for adults with myotonic dystrophy type 1** *NEUROLOGY-CLINICAL PRACTICE*
  Ashizawa, T., Gagnon, C., Groh, W. J., Gutmann, L., Johnson, N. E., Meola, G., Moxley, R., Pandya, S., Rogers, M. T., Simpson, E., Angeard, N., Bassez, G., Berggren, et al
  2018; 8 (6): 507–20

- **Experience using Spinraza to treat adults with spinal muscular atrophy**
  Day, J., Wolford, C., Macpherson, C., Hagerman, K., Paulose, S., Zeineh, M., Martens, W., McDermott, M., Darras, B., De Vivo, D., Cunningham, Z., Finkel, R., Sampson, et al
  PERGAMON-ELSEVIER SCIENCE LTD.2018: S81

- **FGFR3 Antibodies in Neuropathy: What to Do With Them?** *Journal of clinical neuromuscular disease*
  Samara, V., Sampson, J., Muppidi, S.
  2018; 20 (1): 35–40

- **Minimally Invasive Thymectomy and Lung Volume Reduction in a Patient with Myasthenia Gravis.** *The Annals of thoracic surgery*
  Salna, M., Kidambi, S., Sampson, J., Shrager, J. B.
  2018

- **FGFR3 Antibodies in Neuropathy. What to do with them?**
  Samara, V., Sampson, J., Muppidi, S.
  LIPPINCOTT WILLIAMS & WILKINS.2018

- **Nusinersen Efficacy in Adults with Spinal Muscular Atrophy**
  Day, J., Wolford, C., MacPherson, C., Martens, W., McDermott, M., Darras, B., De Vivo, D., Cunningham, Z., Finkel, R., Sampson, J., Duong, T.
  LIPPINCOTT WILLIAMS & WILKINS.2018

- **Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy** *NEW ENGLAND JOURNAL OF MEDICINE*
  Mercuri, E., Darras, B. T., Chiriboga, C. A., Day, J. W., Campbell, C., Connolly, A. M., Iannaccone, S. T., Kirschner, J., Kuntz, N. L., Saito, K., Shieh, P. B., Tulinius, M., Mazzone, et al
  2018; 378 (7): 625–35

- **Increased EEG Theta Spectral Power in Sleep in Myotonic Dystrophy Type 1.** *Journal of clinical sleep medicine : JCSM : official publication of the American Academy of Sleep Medicine*
  Cheung, J., Ruoff, C., Moore, H., Hagerman, K. A., Perez, J., Sakamuri, S., Warby, S. C., Mignot, E., Day, J., Sampson, J.
  2018; 14 (2): 229–35

- **Loss-of-Function Mutations in LGI4, a Secreted Ligand Involved in Schwann Cell Myelination, Are Responsible for Arthrogryposis Multiplex Congenita** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Xue, S., Maluenda, J., Marguet, F., Shboul, M., Quevarec, L., Bonnard, C., Ng, A. Y., Tohari, S., Thong Teck Tan, T. T., Kong, M. K., Monaghan, K. G., Cho, M. T., Siskind, et al
  2017; 100 (4): 659-665

- **A 31-Year-Old Man With a Ring-Enhancing Brain Lesion.** *Journal of neuro-ophthalmology*
  Riley, C. S., Roth, L. A., Sampson, J. B., Radhakrishnan, J., Herlitz, L. C., Blitz, A. M., Moazami, G.
  2017

- **Specific functional pathologies of Cx43 mutations associated with oculodentodigital dysplasia.** *Molecular biology of the cell*
  Kelly, J. J., Esseltine, J. L., Shao, Q., Jabs, E. W., Sampson, J., Auranen, M., Bai, D., Laird, D. W.
  2016; 27 (14): 2172-2185

- **Manipulating Cx43 expression triggers gene reprogramming events in dermal fibroblasts from oculodentodigital dysplasia patients.** *Biochemical journal*
  Esseltine, J. L., Shao, Q., Huang, T., Kelly, J. J., Sampson, J., Laird, D. W.
  2015; 472 (1): 55-69

- **Nuclear localization of SMN and FUS is not altered in fibroblasts from patients with sporadic ALS** *AMYOTROPHIC LATERAL SCLEROSIS AND FRONTOTEMPORAL DEGENERATION*
  Kariya, S., Sampson, J. B., Northrop, L. E., Lucarelli, C. M., Naini, A. B., Re, D. B., Hirano, M., Mitsumoto, H.
  2014; 15 (7-8): 581-587

- **ATALUREN TREATMENT OF PATIENTS WITH NONSENSE MUTATION DYSTROPHINOPATHY** *MUSCLE & NERVE*
  Bushby, K., Finkel, R., Wong, B., Barohn, R., Campbell, C., Comi, G. P., Connolly, A. M., Day, J. W., Flanigan, K. M., Goemans, N., Jones, K. J., Mercuri, E., Quinlivan, et al
  2014; 50 (4): 477-487

- **The G60S Cx43 mutant enhances keratinocyte proliferation and differentiation** *EXPERIMENTAL DERMATOLOGY*
  Churko, J. M., Kelly, J. J., Macdonald, A., Lee, J., Sampson, J., Bai, D., Laird, D. W.
  2012; 21 (8): 612-618

- **Human Dermal Fibroblasts Derived from Oculodentodigital Dysplasia Patients Suggest That Patients May Have Wound-Healing Defects** *HUMAN MUTATION*
  Churko, J. M., Shao, Q., Gong, X., Swoboda, K. J., Bai, D., Sampson, J., Laird, D. W.
  2011; 32 (4): 456-466

- **Nonsense Mutation-Associated Becker Muscular Dystrophy: Interplay Between Exon Definition and Splicing Regulatory Elements within the DMD Gene** *HUMAN MUTATION*
  Flanigan, K. M., Dunn, D. M., von Niederhausern, A., Soltanzadeh, P., Howard, M. T., Sampson, J. B., Swoboda, K. J., Bromberg, M. B., Mendell, J. R., Taylor, L. E., Anderson, C. B., Pestronk, A., Florence, et al
  2011; 32 (3): 299-308

- **Clinical and genetic characterization of manifesting carriers of DMD mutations** *NEUROMUSCULAR DISORDERS*
  Soltanzadeh, P., Friez, M. J., Dunn, D., von Niederhausern, A., Gurvich, O. L., Swoboda, K. J., Sampson, J. B., Pestronk, A., Connolly, A. M., Florence, J. M., Finkel, R. S., Boennemann, C. G., Medne, et al
  2010; 20 (8): 499-504

- **Identification of Uncommon Recurrent Potocki-Lupski Syndrome-Associated Duplications and the Distribution of Rearrangement Types and Mechanisms in PTLS** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Zhang, F., Potocki, L., Sampson, J. B., Liu, P., Sanchez-Valle, A., Robbins-Furman, P., Delicado Navarro, A., Wheeler, P. G., Spence, J. E., Brasington, C. K., Withers, M. A., Lupski, J. R.
  2010; 86 (3): 462-470

- **Mutational Spectrum of DMD Mutations in Dystrophinopathy Patients: Application of Modern Diagnostic Techniques to a Large Cohort** *HUMAN MUTATION*
  Flanigan, K. M., Dunn, D. M., von Niederhausern, A., Soltanzadeh, P., Gappmaier, E., Howard, M. T., Sampson, J. B., Mendell, J. R., Wall, C., King, W. M., Pestronk, A., Florence, J. M., Connolly, et al
  2009; 30 (12): 1657-1666

- **Paraneoplastic myopathy: response to intravenous immunoglobulin** *NEUROMUSCULAR DISORDERS*

Sampson, J. B., Smith, S. M., Smith, A. G., Singleton, J. R., Chin, S., Pestronk, A., Flanigan, K. M.
2007; 17 (5): 404-408

- **Inhibition of human surfactant protein a function by oxidation intermediates of nitrite** *FREE RADICAL BIOLOGY AND MEDICINE*
  Davis, I. C., Zhu, S., Sampson, J. B., Crow, J. P., Matalon, S.
  2002; 33 (12): 1703-1713

- **Liposome-delivered superoxide dismutase prevents nitric oxide-dependent motor neuron death induced by trophic factor withdrawal** *FREE RADICAL BIOLOGY AND MEDICINE*
  Estevez, A. G., Sampson, J. B., Zhuang, Y. X., Spear, N., Richardson, G. J., Crow, J. P., Tarpey, M. M., Barbeito, L., Beckman, J. S.
  2000; 28 (3): 437-446

- **Myeloperoxidase and horseradish peroxidase catalyze tyrosine nitration in proteins from nitrite and hydrogen peroxide** *ARCHIVES OF BIOCHEMISTRY AND BIOPHYSICS*
  Sampson, J. B., Ye, Y. Z., Rosen, H., Beckman, J. S.
  1998; 356 (2): 207-213

- **DIFFERENTIAL MODULATION OF ASTROCYTE CYTOKINE GENE-EXPRESSION BY TGF-BETA** *JOURNAL OF IMMUNOLOGY*
  Benveniste, E. N., Kwon, J. B., Chung, W. J., Sampson, J., Pandya, K., Tang, L. P.
  1994; 153 (11): 5210-5221