Clinical Professor, Neurology & Neurological Sciences

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 in the clinical application of next-generation genomic sequencing to genetic testing.

CLINICAL FOCUS
• Neurogenetics
• Neuromuscular Medicine

ACADEMIC APPOINTMENTS
• Clinical Professor, Neurology & Neurological Sciences
• Member, Cardiovascular Institute
• Member, Wu Tsai Human Performance Alliance

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 (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
• Stanford Neuromuscular Biobank: https://med.stanford.edu/day-lab/biobank.html

Publications

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• Exome and genome sequencing in a heterogeneous population of patients with rare disease: Identifying predictors of a diagnosis. *Genetics in medicine : official journal of the American College of Medical Genetics*
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• Cerebrospinal Fluid Proteomic Changes after Nusinersen in Patients with Spinal Muscular Atrophy. *Journal of clinical medicine*
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  2023; 12 (20)

• Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care. *JOURNAL OF PEDIATRICS*
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• Correction: Distinct germline genetic susceptibility profiles identified for common non-Hodgkin lymphoma subtypes. *Leukemia*
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Fatemi, A., Koehler, W., Eichler, F., Moehel, F., Sadjadi, R., Lund, T., Sampson, J., Shuhaimer, H., Amartino, H., Sgobbi, P., Kappler, M., Kay, R., Pina, et al
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Kamali, T., Day, J., Sampson, J., Murad, A., Chaufy, J.
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Koehler, W., Engelen, M., Eichler, F., Lachmann, R., Fatemi, A., Sampson, J., Salsano, E., Gamez, J., Molnar, M. J., Pascual, S., Rovira, M., Vila, A., Pina, et al
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Evaluating 2-3 year responses to disease modifying treatment in adults with spinal muscular atrophy
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• Mesial Temporal Enlargement in Adult-Onset Myotonic Dystrophy Type 1
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• Characterization of HNRNPA1 mutations defines diversity in pathogenic mechanisms and clinical presentation.
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• A variant of uncertain significance in SDHAF1, the succinate dehydrogenase chaperone protein, in an adult patient with spastic paraparesis and leukoencephalopathy.
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• Advances in the therapy of Spinal Muscular Atrophy.
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• Nusinersen Treatment in Adults With Spinal Muscular Atrophy.
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• A resource of lipidomics and metabolomics data from individuals with undiagnosed diseases
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  Kyle, J. E., Stratton, K. G., Zink, E. M., Kim, Y., Bloodsworth, K. J., Monroe, M. E., Bacino, C. A., Bacino, C. A., Hanchard, N. A., Lewis, R. A., Rosenfeld, J. A., Scott, D. A., Tran, et al
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Toward Developing Robust Myotonic Dystrophy Brain Biomarkers using White Matter Tract Profiles Sub-Band Energy and A Framework of Ensemble Predictive Learning.
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Diagnosis of Myotonic Dystrophy Based on Resting State fMRI Using Convolutional Neural Networks.
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2020

Diagnosis of Myotonic Dystrophy Based on Resting State fMRI Using Convolutional Neural Networks
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• Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review. *American journal of medical genetics. Part A*

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• A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing. *Journal of genetic counseling*
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• A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis. *Journal of general internal medicine*
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• Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students. *Journal of genetic counseling*
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• Nusinersen Efficacy in Adults with Spinal Muscular Atrophy
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