Stanford

Jacinda Sampson

Clinical Professor, Neurology & Neurological Sciences

Curriculum Vitae available Online

CLINICAL OFFICE (PRIMARY)

- **Stanford Neuroscience Health Center**

  213 Quarry Rd

  MC 5957 Fl 2

  Palo Alto, CA 94304

  Tel (650) 723-6469 **Fax** (650) 725-0390

ACADEMIC CONTACT INFORMATION

- **Alternate Contact**

  Gayla Weng - Neuromuscular Program Coordinator

  Email gweng@stanford.edu

  Tel 650-725-7623

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

CLINICAL FOCUS

- Neurology
- Neurogenetics

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

PUBLICATIONS

- **Recurring homozygous ACTN2 variant (p.Arg506Gly) causes a recessive myopathy.** *Annals of clinical and translational neurology*
  Donkervoort, S., Mohassel, P., O'Leary, M., Bonner, D. E., Hartley, T., Acquaye, N., Brull, A., Mozaffar, T., Saporta, M. A., Dyment, D. A., Sampson, J. B., Pajusalu, S., Austin-Tse, et al
  2024

- **Cerebrospinal Fluid Proteomic Changes after Nusinersen in Patients with Spinal Muscular Atrophy.** *Journal of clinical medicine*
  Beaudin, M., Kamali, T., Tang, W., Hagerman, K. A., Dunaway Young, S., Ghiglieri, L., Parker, D. M., Lehallier, B., Tesi-Rocha, C., Sampson, J. B., Duong, T., Day, J. W.
  2023; 12 (20)

- **Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care** *JOURNAL OF PEDIATRICS*
  Halley, M. C., Young, J. L., Tang, C., Mintz, K. T., Lucas-Griffin, S., Maghiro, A., Ashley, E. A., Tabor, H. K., Undiagnosed Diseases Network
  2023; 261

- **Correction: Distinct germline genetic susceptibility profiles identified for common non-Hodgkin lymphoma subtypes.** *Leukemia*
  Berndt, S. I., Vijai, J., Benavente, Y., Camp, N. J., Nieters, A., Wang, Z., Smedby, K. E., Kleinstern, G., Hjalgrim, H., Besson, C., Skibola, C. F., Morton, L. M., Brooks-Wilson, et al
  2023

- **A Phase 3 Clinical Trial of Leriglitazone with Adaptive Placebo-Controlled Treatment Duration in Adults with Cerebral Adrenoleukodystrophy**
  Fatemi, A., Koehler, W., Eichler, F., Mochel, F., Sadjadi, R., Lund, T., Sampson, J., Shuhaiber, H., Amartino, H., Sgobbi, P., Kappler, M., Kay, R., Pina, et al
  WILEY.2023: S138-S139

- **Learning Spectral Fractional Anisotropy and Mean Diffusivity Features as Neuroimaging Biomarkers for Tracking White Matter Integrity Changes in Myotonic Dystrophy Type 1 Patients using Deep Convolutional Neural Networks.** *Annual International Conference of the IEEE Engineering in Medicine and Biology Society. IEEE Engineering in Medicine and Biology Society. Annual International Conference*
  Kamali, T., Day, J. W., Deutsch, G. K., Sampson, J. B., Murad, A., Chaufty, J., Parker, D., Wozniak, J. R.
  2023; 2023: 1-4

- **Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care.** *The Journal of pediatrics*
  Halley, M. C., Young, J. L., Tang, C., Mintz, K. T., Lucas-Griffin, S., Maghiro, A. S., Ashley, E. A., Tabor, H. K.
  2023: 113537

- **Choroid plexus mis-splicing and altered cerebrospinal fluid composition in myotonic dystrophy type 1.** *Brain : a journal of neurology*
  Nutter, C. A., Kidd, B. M., Carter, H. A., Hamel, J. I., Mackie, P. M., Kumbkarni, N., Davenport, M. L., Tuyn, D. M., Gopinath, A., Creigh, P. D., Sznajder, #. J., Wang, E. T., Ranum, et al
  2023

- **A Multimodal Neuroimaging Feature Extraction Framework for Biomarker Discovery in Myotonic Dystrophies**
  Kamali, T., Day, J., Sampson, J., Murad, A., Chaufty, J.
  LIPPINCOTT WILLIAMS & WILKINS.2023

- **Participation in a national diagnostic research study: assessing the patient experience.** *Orphanet journal of rare diseases*
  Rosenfeld, L. E., LeBlanc, K., Nagy, A., Ego, B. K., Undiagnosed Diseases Network, McCray, A. T., Acosta, M. T., Adam, M., Adams, D. R., Alvarez, R. L., Alvey, J., Amendola, L., Andrews, A., et al
  2023; 18 (1): 73

- **Safety and efficacy of leriglitazone for preventing disease progression in men with adrenomyeloneuropathy (ADVANCE): a randomised, double-blind, multi-centre, placebo-controlled phase 2-3 trial.** *The Lancet. Neurology*
  Kohler, 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
  2023; 22 (2): 127-136

- **Safety and efficacy of leriglitazone for preventing disease progression in men with adrenomyeloneuropathy (ADVANCE): a randomised, double-blind, multi-centre, placebo-controlled phase 2-3 trial** *LANCET NEUROLOGY*
  Koehler, W., Engelen, M., Eichler, F., Lachmann, R., Fatemi, A., Sampson, J., Salsano, E., Gamez, J., Molnar, M., Pascual, S., Rovira, M., Vila, A., Pina, et al
  2023; 22 (2): 127-136

- **A concurrent dual analysis of genomic data augments diagnoses: experiences of two clinical sites in the Undiagnosed Diseases Network.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Spillmann, R. C., Tan, Q. K., Reuter, C., Schoch, K., Kohler, J., Bonner, D., Zastrow, D., Alkelai, A., Baugh, E., Cope, H., Marwaha, S., Wheeler, M. T., Bernstein, et al
  2022

- **Multimodal fusion of neuroimaging and neuropsych data: A machine learning approach to study brain alterations linked with cognitive domains in DM1**
  Kamali, T., Parker, D., Deutsch, G., Sampson, J., Day, J., Wozniak, J.
  PERGAMON-ELSEVIER SCIENCE LTD.2022: S132

- **Evaluating 2-3 year responses to disease modifying treatment in adults with spinal muscular atrophy**
  Duong, T., Tang, W., Young, S., Parker, D., Wolford, C., Sampson, J., Day, J.
  PERGAMON-ELSEVIER SCIENCE LTD.2022: S90

- **Correction: Healthcare resource utilization, total costs, and comorbidities among patients with myotonic dystrophy using U.S. insurance claims data from 2012 to 2019.** *Orphanet journal of rare diseases*
  Howe, S. J., Lapidus, D., Hull, M., Yeaw, J., Stevenson, T., Sampson, J. B.
  2022; 17 (1): 260

- **Cognitive Impairment Analysis of Myotonic Dystrophy via Weakly Supervised Classification of Neuropsychological Features.** *Annual International Conference of the IEEE Engineering in Medicine and Biology Society. IEEE Engineering in Medicine and Biology Society. Annual International Conference*
  Kamali, T., Deutsch, G. K., Hagerman, K. A., Parker, D., Day, J. W., Sampson, J. B., Wozniak, J. R.
  2022; 2022: 4377-4382

- **Mesial Temporal Enlargement in Adult-Onset Myotonic Dystrophy Type 1**
  Fecto, F., Parker, D., Sampson, J., Mueller, B., Lim, K., Wozniak, J., Hagerman, K., Day, J.
  LIPPINCOTT WILLIAMS & WILKINS.2022

- **Mesial Temporal Enlargement in Adult-Onset Myotonic Dystrophy Type 1**
  Fecto, F., Parker, D., Sampson, J., Mueller, B., Lim, K., Wozniak, J., Hagerman, K., Day, J.
  LIPPINCOTT WILLIAMS & WILKINS.2022

- **Exploring Protein Changes in Cerebrospinal Fluid of Spinal Muscular Atrophy Patients Pre-Nusinersen vs. Post-Nusinersen Treatment using Bayesian Machine Learning Algorithms**
  Kamali, T., Hagerman, K., Duong, T., Parker, D., Young, S., Tang, W., Sampson, J., Day, J.
  LIPPINCOTT WILLIAMS & WILKINS.2022

- **Healthcare resource utilization, total costs, and comorbidities among patients with myotonic dystrophy using U.S. insurance claims data from 2012 to 2019.** *Orphanet journal of rare diseases*
  Howe, S. J., Ladipus, D., Hull, M., Yeaw, J., Stevenson, T., Sampson, J. B.
  2022; 17 (1): 79

- **Brief assessment of cognitive function in myotonic dystrophy: multicenter longitudinal study using computer-assisted evaluation.** *Muscle & nerve*
  Deutsch, G. K., Hagerman, K. A., Sampson, J., Dent, G., Dekdebrun, J., Parker, D. M., Thornton, C. A., Heatwole, C. R., Subramony, S. H., Mankodi, A. K., Ashizawa, T., Statland, J. M., Arnold, et al
  2022

- **Leriglitazone Reduces Cerebral Lesions and Improves Biomarkers Related to Axonal Degeneration, Inflammation and Compromised Blood-Brain-Barrier in Patients with Adrenomyeloneuropathy**
  Mochel, F., Eichler, F., Engelen, M., Lachman, R., Fatemi, A., Sampson, J., Salsano, E., Gamez, J., Judith Molnar, M., Vilalta, A., Rodriguez-Pascau, L., Pizcueta, P., Pascual, et al
  WILEY.2021: S129

- **Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation.** *Journal of genetic counseling*
  Kohler, J. N., Kelley, E. G., Boyd, B. M., Sillari, C. H., Marwaha, S., Undiagnosed Diseases Network, Wheeler, M. T., Acosta, M. T., Adam, M., Adams, D. R., Agrawal, P. B., Alejandro, M. E., Alvey, J., et al
  2021

- **Characterization of HNRNPA1 mutations defines diversity in pathogenic mechanisms and clinical presentation.** *JCI insight*
  Beijer, D., Kim, H. J., Guo, L., O'Donovan, K., Mademan, I., Deconinck, T., Van Schil, K., Fare, C. M., Drake, L. E., Ford, A. F., Kochanski, A., Kabzinska, D., Dubuisson, et al
  2021; 6 (14)

- **A variant of uncertain significance in SDHAF1, the succinate dehydrogenase chaperone protein, in an adult patient with spastic paraparesis and leukoencephalopathy.** *Multiple sclerosis and related disorders*
  Vlahovic, L., Lock, C. B., Han, M. H., Van Haren, K., Sampson, J. B.
  2021; 54: 103132

- **Advances in the therapy of Spinal Muscular Atrophy.** *The Journal of pediatrics*
  Klotz, J., Rocha, C. T., Young, S. D., Duong, T., Buu, M., Sampson, J., Day, J. W.
  2021

- **Nusinersen Treatment in Adults With Spinal Muscular Atrophy.** *Neurology. Clinical practice*
  Duong, T., Wolford, C., McDermott, M. P., Macpherson, C. E., Pasternak, A., Glanzman, A. M., Martens, W. B., Kichula, E., Darras, B. T., De Vivo, D. C., Zolkipli-Cunningham, Z., Finkel, R. S., Zeineh, et al
  2021; 11 (3): e317-e327

- **Dominant and Recessive Congenital Myasthenic Syndromes Caused by SYT2 Mutations.** *Muscle & nerve*
  Maselli, R. A., Wei, D. T., Hodgson, T. S., Sampson, J., Vazquez, J., Smith, H. L., Pytel, P., Ferns, M.
  2021

- **A resource of lipidomics and metabolomics data from individuals with undiagnosed diseases** *SCIENTIFIC DATA*
  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
  2021; 8 (1): 114

- **Results of Double-blind, Placebo-controlled, Dose Range Finding, Crossover Study of Single Day Administration of ERX-963 (IV Flumazenil) in Adults with Myotonic Dystrophy Type 1**
  Sampson, J., Wang, E., Day, J., Gutmann, L., Mezerhane, E., Seto, A., Ehrich, E.
  LIPPINCOTT WILLIAMS & WILKINS.2021

- **Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain** *GENETICS IN MEDICINE*
  Marbach, F., Stoyanov, G., Erger, F., Stratakis, C. A., Settas, N., London, E., Rosenfeld, J. A., Torti, E., Haldeman-Englert, C., Sklirou, E., Kessler, E., Ceulemans, S., Nelson, et al
  2021

- **Exome testing most useful for people with recessive CMT**
  Siskind, C., Sampson, J., Goyal, N., Rocha, A., Day, J.
  WILEY.2021: 141–42

- **Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases.** *Genetics in medicine : official journal of the American College of Medical Genetics*

Kobren, S. N., Baldridge, D., Velinder, M., Krier, J. B., LeBlanc, K., Esteves, C., Pusey, B. N., Zuchner, S., Blue, E., Lee, H., Huang, A., Bastarache, L., Bican, et al

2021

- **Transcriptome alterations in myotonic dystrophy frontal cortex.** *Cell reports*

  Otero, B. A., Poukalov, K. n., Hildebrandt, R. P., Thornton, C. A., Jinnai, K. n., Fujimura, H. n., Kimura, T. n., Hagerman, K. A., Sampson, J. B., Day, J. W., Wang, E. T.

  2021; 34 (3): 108634

- **Toward Developing Robust Myotonic Dystrophy Brain Biomarkers using White Matter Tract Profiles Sub-Band Energy and A Framework of Ensemble Predictive Learning.** *Annual International Conference of the IEEE Engineering in Medicine and Biology Society. IEEE Engineering in Medicine and Biology Society. Annual International Conference*

  Kamali, T., Parker, D., Day, J. W., Sampson, J., Deutsch, G. K., Wozniak, J. R.

  2021; 2021: 3838-3841

- **Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy.** *Journal of comparative effectiveness research*

  Campbell, C., Barohn, R. J., Bertini, E., Chabrol, B., Comi, G. P., Darras, B. T., Finkel, R. S., Flanigan, K. M., Goemans, N., Iannaccone, S. T., Jones, K. J., Kirschner, J., Mah, et al

  2020

- **Diagnosis of Myotonic Dystrophy Based on Resting State fMRI Using Convolutional Neural Networks.** *Annual International Conference of the IEEE Engineering in Medicine and Biology Society. IEEE Engineering in Medicine and Biology Society. Annual International Conference*

  Kamali, T., Hagerman, K. A., Day, J. W., Sampson, J., Lim, K. O., Mueller, B. A., Wozniak, J.

  2020; 2020: 1714–17

- **Revised Recommendations for the Treatment of Infants Diagnosed with Spinal Muscular Atrophy Via Newborn Screening Who Have 4 Copies of SMN2.** *Journal of neuromuscular diseases*

  Glascock, J., Sampson, J., Connolly, A. M., Darras, B. T., Day, J. W., Finkel, R., Howell, R. R., Klinger, K. W., Kuntz, N., Prior, T., Shieh, P. B., Crawford, T. O., Kerr, et al

  2020

- **Diagnosis of Myotonic Dystrophy Based on Resting State fMRI Using Convolutional Neural Networks**

  Kamali, T., Hagerman, K. A., Day, J. W., Sampson, J., Lim, K. O., Mueller, B. A., Wozniak, J., IEEE

  IEEE.2020: 1714–17

- **Successful liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE).** *Molecular genetics and metabolism*

  Kripps, K. n., Nakayuenyongsuk, W. n., Shayota, B. J., Berquist, W. n., Gomez-Ospina, N. n., Esquivel, C. O., Concepcion, W. n., Sampson, J. B., Cristin, D. J., Jackson, W. E., Gilliland, S. n., Pomfret, E. A., Kueht, et al

  2020

- **Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Schoch, K. n., Esteves, C. n., Bican, A. n., Spillmann, R. n., Cope, H. n., McConkie-Rosell, A. n., Walley, N. n., Fernandez, L. n., Kohler, J. N., Bonner, D. n., Reuter, C. n., Stong, N. n., Mulvihill, et al

  2020

- **De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation.** *American journal of human genetics*

  Mao, D. n., Reuter, C. M., Ruzhnikov, M. R., Beck, A. E., Farrow, E. G., Emrick, L. T., Rosenfeld, J. A., Mackenzie, K. M., Robak, L. n., Wheeler, M. T., Burrage, L. C., Jain, M. n., Liu, et al

  2020

- **Multiomics Approach to Diagnosing Undiagnosed Patients**

  Wheeler, M. T., Kohler, J. N., Bonner, D. E., Zastrow, D. B., Reuter, C., Majcherska, M., Fernandez, L., McCormack, C., Marwaha, S., Curnin, C., Dries, A., Ruzhnikov, M., Hom, et al

  NATURE PUBLISHING GROUP.2019: 1163–64

- **Connexin43 is Dispensable for Early Stage Human Mesenchymal Stem Cell Adipogenic Differentiation But is Protective against Cell Senescence.** *Biomolecules*

  Shao, Q., Esseltine, J. L., Huang, T., Novielli-Kuntz, N., Ching, J. E., Sampson, J., Laird, D. W.

  2019; 9 (9)

- **Explaining RLS families using risk SNPs from GWAS**
  Tilch, E., Zhao, C., Salminen, A., Antic, A., Schormair, B., Oexle, K., Sampson, J. B., Muller-Myhsok, B., Winkelmann, J., EU-RLS-Gene Consortium
  NATURE PUBLISHING GROUP.2019: 658–59

- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Kumar, A., Zastrow, D. B., Kravets, E. J., Beleford, D., Ruzhnikov, M. Z., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Mackenzie, K. M., Eng, et al
  2019; 179 (6): 966–77

- **A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis** *JOURNAL OF GENERAL INTERNAL MEDICINE*
  Hom, J., Marwaha, S., Postolova, A., Kittle, J., Vasquez, R., Davidson, J., Kohler, J., Dries, A., Fernandez-Betancourt, L., Majcherska, M., Dearlove, J., Raghavan, S., Vogel, et al
  2019; 34 (6): 1058–62

- **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., Zeineh, M., Sampson, J., Hagerman, K., Duong, et al
  LIPPINCOTT WILLIAMS & WILKINS.2019

- **Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy Through Newborn Screening**
  Glascock, J., Sampson, J., Haidet-Phillips, A., Connolly, A., Darras, B., Day, J., Finkel, R., Howell, R., Klinger, K., Kuntz, N., Prior, T., Shieh, P., Crawford, et al
  LIPPINCOTT WILLIAMS & WILKINS.2019

- **Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students** *JOURNAL OF GENETIC COUNSELING*
  Grove, M. E., White, S., Fisk, D. G., Rego, S., Dagan-Rosenfeld, O., Kohler, J. N., Reuter, C. M., Bonner, D., Wheeler, M. T., Bernstein, J. A., Ormond, K. E., Hanson-Kahn, A. K., Undiagnosed Dis Network
  2019; 28 (2): 466–76

- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing** *JOURNAL OF GENETIC COUNSELING*
  Zastrow, D. B., Kohler, J. N., Bonner, D., Reuter, C. M., Fernandez, L., Grove, M. E., Fisk, D. G., Yang, Y., Eng, C. M., Ward, P. A., Bick, D., Worthey, E. A., Fisher, et al
  2019; 28 (2): 213–28

- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review.** *American journal of medical genetics. Part A*
  Kumar, A., Zastrow, D. B., Kravets, E. J., Beleford, D., Ruzhnikov, M. R., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Undiagnosed Diseases Network, Mackenzie, K. M., et al
  2019

- **Dynamic regulation of connexins in stem cell pluripotency.** *Stem cells (Dayton, Ohio)*
  Esseltine, J. L., Brooks, C. R., Edwards, N. A., Subasri, M. n., Sampson, J. n., Séguin, C. n., Betts, D. H., Laird, D. W.
  2019

- **Genomics in medicine: a novel elective rotation for internal medicine residents.** *Postgraduate medical journal*
  Geng, L. N., Kohler, J. N., Levonian, P. n., Bernstein, J. A., Ford, J. M., Ahuja, N. n., Witteles, R. n., Hom, J. n., Wheeler, M. n.
  2019

- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing.** *Journal of genetic counseling*
  Zastrow, D. B., Kohler, J. N., Bonner, D. n., Reuter, C. M., Fernandez, L. n., Grove, M. E., Fisk, D. G., Yang, Y. n., Eng, C. M., Ward, P. A., Bick, D. n., Worthey, E. A., Fisher, et al
  2019; 28 (2): 213–28

- **A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis.** *Journal of general internal medicine*
  Hom, J. n., Marwaha, S. n., Postolova, A. n., Kittle, J. n., Vasquez, R. n., Davidson, J. n., Kohler, J. n., Dries, A. n., Fernandez-Betancourt, L. n., Majcherska, M. n., Dearlove, J. n., Raghavan, S. n., Vogel, et al
  2019

- **Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts.** *Nature medicine*

Frésard, L. n., Smail, C. n., Ferraro, N. M., Teran, N. A., Li, X. n., Smith, K. S., Bonner, D. n., Kernohan, K. D., Marwaha, S. n., Zappala, Z. n., Balliu, B. n., Davis, J. R., Liu, et al

2019

- **Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students.** *Journal of genetic counseling*

  Grove, M. E., White, S. n., Fisk, D. G., Rego, S. n., Dagan-Rosenfeld, O. n., Kohler, J. N., Reuter, C. M., Bonner, D. n., Wheeler, M. T., Bernstein, J. A., Ormond, K. E., Hanson-Kahn, A. K.

  2019

- **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

- **Minimally Invasive Thymectomy and Lung Volume Reduction in a Patient With Myasthenia Gravis** *ANNALS OF THORACIC SURGERY*

  Salna, M., Kidambi, S., Sampson, J., Shrager, J. B.

  2018; 106 (6): E313–E315

- **Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease** *NEW ENGLAND JOURNAL OF MEDICINE*

  Splinter, K., Adams, D. R., Bacino, C. A., Bellen, H. J., Bernstein, J. A., Cheatle-Jarvela, A. M., Eng, C. M., Esteves, C., Gahl, W. A., Hamid, R., Jacob, H. J., Kikani, B., Koeller, et al

  2018; 379 (22): 2131–39

- **C-terminal proline deletions in KCNC3 cause delayed channel inactivation and an adult-onset progressive SCA13 with spasticity** *CEREBELLUM*

  Khare, S., Galeano, K., Zhang, Y., Nick, J. A., Nick, H. S., Subramony, S. H., Sampson, J., Kaczmarek, L. K., Waters, M. F.

  2018; 17 (5): 692–97

- **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. n., Ruoff, C. n., Moore, H. n., Hagerman, K. A., Perez, J. n., Sakamuri, S. n., Warby, S. C., Mignot, E. n., Day, J. n., Sampson, J. n.

  2018; 14 (2): 229–35

- **Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening.** *Journal of neuromuscular diseases*

  Glascock, J., Sampson, J., Haidet-Phillips, A., Connolly, A., Darras, B., Day, J., Finkel, R., Howell, R. R., Klinger, K., Kuntz, N., Prior, T., Shieh, P. B., Crawford, et al

2018; 5 (2): 145–58

- **Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease.** *The New England journal of medicine*
  Splinter, K. n., Adams, D. R., Bacino, C. A., Bellen, H. J., Bernstein, J. A., Cheatle-Jarvela, A. M., Eng, C. M., Esteves, C. n., Gahl, W. A., Hamid, R. n., Jacob, H. J., Kikani, B. n., Koeller, et al
  2018

- **Increased EEG theta spectral power in polysomnography of myotonic dystrophy type 1 compared to matched controls.**
  Ruoff, C., Cheung, J., Perez, J., Sakamuri, S., Mignot, E., Day, J., Sampson, J.
  LIPPINCOTT WILLIAMS & WILKINS.2017

- **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

- **Basal Ganglia Gliosis in a Case of Rapid-Onset Dystonia-Parkinsonism (DYT12) with a Novel Mutation in ATPase 1A3 (ATP1A3)** *MOVEMENT DISORDERS CLINICAL PRACTICE*
  Sampson, J. B., Michaeli, T. H., Wright, B. A., Goldman, J. E., Vonsattel, J., Fahn, S.
  2016; 3 (6): 618–20

- **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

- **Basal Ganglia Gliosis in a Case of Rapid-Onset Dystonia-Parkinsonism (DYT12) with a Novel Mutation in ATPase 1A3 (ATP1A3).** *Movement disorders clinical practice*
  Sampson, J. B., Michaeli, T. H., Wright, B. A., Goldman, J. E., Vonsattel, J. P., Fahn, S.
  2016; 3 (6): 618-620

- **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

- **Modeling Cx43-linked pathologies in the human context**
  Esseltine, J. L., Shao, Q., Brooks, C., Sampson, J., Betts, D. H., Seguin, C.
  AMER SOC CELL BIOLOGY.2015

- **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