Stanford

Jacinda Sampson

Clinical Professor, Adult Neurology

Curriculum Vitae available Online

CLINICAL OFFICE (PRIMARY)

- **Stanford Neuroscience Health Center**
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ACADEMIC CONTACT INFORMATION

- **Alternate Contact**
  Gayla Weng - Neuromuscular Program Coordinator
  Email gweng@stanford.edu
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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 in the clinical application of next-generation genomic sequencing to genetic testing.

CLINICAL FOCUS

- Neurogenetics
- Neuromuscular Medicine

ACADEMIC APPOINTMENTS

- Clinical Professor, Adult Neurology
- Member, Cardiovascular Institute
- Member, Wu Tsai Human Performance Alliance
- Member, Wu Tsai Neurosciences Institute

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
- 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

- Board Certification: Neurology, American Board of Psychiatry and Neurology (2005)
- 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

Research & Scholarship

CLINICAL TRIALS

- Calyx MT-3-01, Recruiting
- ION373-CS1, Recruiting
- MT-2-01, Not Recruiting

Publications

PUBLICATIONS

- **Longitudinal Psychometric Properties of the Myotonic Dystrophy Health Index in a Large Multicenter Cohort of People Living With Myotonic Dystrophy Type 1.** *Muscle & nerve*
  Sansone, V. A., Lizio, A., Ferrari Aggradi, C. R., Greco, L. C., Gagnon, C., Subramony, S., Roxburgh, R. H., Mul, K., Hamel, J., Eichinger, K., Hung, M., Statland, J. M., Turner, et al
  2026

- **Prospective Study of Video Hand Opening Time as a Quantitative Measurement of Myotonia in Patients With Myotonic Dystrophy Type 1.** *Neurology*
  Leeuwenberg, K. E., Sansone, V. A., Hamel, J., Hung, M., Dekdebrun, J. M., Lizio, A., Eichinger, K., Gagnon, C., Roxburgh, R. H., Subramony, S. H., Statland, J. M., Elsheikh, B. H., Turner, et al
  2026; 106 (7): e214747

- **Generation of induced pluripotent stem cell lines from patients with Emery-Dreifuss muscular dystrophy.** *Stem cell research*
  Chorsi, M. S., Mui, B. J., Tan, R., Fernandez, H. M., Xu, L., Chao, J. T., Sampson, J. B., Wheeler, M. T., Wu, J. C.
  2026; 91: 103926

- **Biallelic LAMP3 Variants in Five Families with Interstitial Lung Disease: Evidence of a Disease-Gene Association.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Keehan, L. A., Ono-Minagi, H., Hadhud, M., Rips, J., Hinds, D. M., Fischer, A. J., Bartlett, J. A., McCray, P. B., Qawasmi, N., Nathan, N., Louvrier, C., Desroziers, T., Damme, et al
  2026: 102531

- **Establishing biomarkers and clinical endpoints in myotonic dystrophy type 1 (END-DM1): Protocol of an international natural history study.** *PloS one*
  Mul, K., Eichinger, K., Hung, M., Sansone, V. A., Gagnon, C., Subramony, S., Roxburgh, R. H., Hamel, J., Statland, J. M., Elsheikh, B., Turner, C., Sampson, J., Ragole, et al
  2025; 20 (12): e0331163

- **Cerebrospinal fluid proteomic profiling reveals potential biomarkers and altered pathways in myotonic dystrophy type 1** *FRONTIERS IN NEUROSCIENCE*
  Zafarullah, M., Kamali, T., Hagerman, K. A., Ghiglieri, L., Duong, T., Wang, E., Sampson, J. B., Day, J. W.
  2025; 19: 1709678

- **Stanford Center for Undiagnosed Diseases: a decade of advancing genetic diagnosis and discovery**
  Bonner, D., Carter, J., Reuter, C., Marwaha, S., Kravets, E., Hom, J., Sampson, J., Fisher, P., Ashley, E., Tabor, H., Halley, M., Wheeler, M., Bernstein, et al
  SPRINGERNATURE.2025: 877

- **An optimized variant prioritization process for rare disease diagnostics: recommendations for Exomiser and Genomiser.** *Genome medicine*
  Cooperstein, I. B., Marwaha, S., Ward, A., Kobren, S. N., Carter, J. N., Wheeler, M. T., Marth, G. T.
  2025; 17 (1): 127

- **RAPSN-Associated Congenital Myasthenic Syndrome due to Biallelic Single Nucleotide Variants at the Same Position.** *Case reports in genetics*
  Keehan, L., Carter, J. N., Kravets, E., Wheeler, M. T., Bernstein, J. A., Maselli, R. A., Sampson, J. B., Bachir, S.
  2025; 2025: 1882021

- **Integrating cognition and neuroimaging: a study of cerebral regions in myotonic dystrophy**
  Duong, T., Karman, L., Yao, M., Ismail, S., Rogers, M., Deutsch, G., Sampson, J., Day, J., Hageman, N.
  PERGAMON-ELSEVIER SCIENCE LTD.2025

- **Results from 15 mg/kg single dose PGN-EDODM1 cohort of FREEDOM-DM1-a phase 1 study in people with myotonic dystrophy type 1 (DM1)**
  Hamel, J., Brisson, J., Lochmuller, H., Wheeler, T., Sampson, J., Goyal, N., Johnson, N., Statland, J., Lilleker, J., Turner, C., Pfeffer, G., Shoskes, J., Holland, et al
  PERGAMON-ELSEVIER SCIENCE LTD.2025

- **Joint, multifaceted genomic analysis enables diagnosis of diverse, ultra-rare monogenic presentations** *NATURE COMMUNICATIONS*
  Kobren, S., Moldovan, M. A., Reimers, R., Traviglia, D., Li, X., Barnum, D., Veit, A., Corona, R. I., Neto, G., Willett, J., Berselli, M., Ronchetti, W., Nelson, et al
  2025; 16 (1): 7267

- **Motoneuron Involvement in CANVAS: A Case of ACAGG Expansions in a Guamanian Man**
  Dufort, G., Bonner, D., Anguiano, B., Bernstein, J., Wheeler, M., Sampson, J.
  LIPPINCOTT WILLIAMS & WILKINS.2025

- **Multimodal Investigation of Brain Structural Changes, Neurocognitive Function, and Sleep Disruptions in Myotonic Dystrophy Type 1**
  Kamali, T., Hagerman, K., Sampson, J., Day, J.
  LIPPINCOTT WILLIAMS & WILKINS.2025

- **CSF Proteomic Profiling Reveals Altered Pathways in Myotonic Dystrophy Type 1**
  Zafarullah, M., Kamali, T., Hagerman, K., Ghiglieri, L., Sampson, J., Day, J.
  LIPPINCOTT WILLIAMS & WILKINS.2025

- **Hyoscyamine as a Novel Treatment for Intestinal Pseudo-obstruction in Myotonic Dystrophy Type 1**
  Roses, S., Sampson, J., Nguyen, L., Day, J.
  LIPPINCOTT WILLIAMS & WILKINS.2025

- **Detailed tandem repeat allele profiling in 1,027 long-read genomes reveals genome-wide patterns of pathogenicity.** *bioRxiv : the preprint server for biology*
  Danzi, M. C., Xu, I. R., Fazal, S., Dolzhenko, E., Pellerin, D., Weisburd, B., Reuter, C., Sampson, J., Folland, C., Wheeler, M., O'Donnell-Luria, A., Wuchty, S., Ravenscroft, et al
  2025

- **A multi-omic approach characterizes a rare repeat expansion in RFC1 in an Asian-Pacific family with motor neuronopathy**
  Bonner, D., Sampson, J., Anguiano, B., Kravets, E., Marwaha, S., Montgomery, S., Wheeler, M., Bernstein, J.
  SAGE PUBLICATIONS LTD.2025: 607

- **Telehealth Is Effective in the Evaluation of Individuals With Undiagnosed Rare Disorders: An Undiagnosed Diseases Network Study.** *American journal of medical genetics. Part A*
  Tan, Q. K., McConkie-Rosell, A., Mahoney, R., Spillmann, R. C., Schoch, K., Chanprasert, S., Acosta, M. T., Toro, C., Rosenfeld, J. A., Orengo, J. P., Scott, D. A., Granadillo, J. L., Sisco, et al
  2024: e63956

- **Practical Approach to Longitudinal Neurologic Care of Adults With X-Linked Adrenoleukodystrophy and Adrenomyeloneuropathy.** *Neurology. Genetics*
  Kornbluh, A. B., Baldwin, A., Fatemi, A., Vanderver, A., Adang, L. A., Van Haren, K., Sampson, J., Eichler, F. S., Sadjadi, R., Engelen, M., Orthmann-Murphy, J. L.
  2024; 10 (5): e200192

- **Development and implementation of enhanced AI-derived DTI features for precision mapping of neural tract damage in myotonic dystrophy**
  Kamali, T., Hageman, N., Yazdavar, T., Piccolli, C., Day, J., Sampson, J., Wozniak, J.
  PERGAMON-ELSEVIER SCIENCE LTD.2024

- **Evaluating EEG as an outcome measure for CNS Symptoms in myotonic dystrophy type 1: a clinical trial analysis**
  Allen, S., Kamali, T., Parker, D., Seto, A., Ehrich, E., Wang, E., Sampson, J.
  PERGAMON-ELSEVIER SCIENCE LTD.2024

- **Large-scale mutational analysis identifies UNC93B1 variants that drive TLR-mediated autoimmunity in mice and humans.** *The Journal of experimental medicine*
  Rael, V. E., Yano, J. A., Huizar, J. P., Slayden, L. C., Weiss, M. A., Turcotte, E. A., Terry, J. M., Zuo, W., Thiffault, I., Pastinen, T., Farrow, E. G., Jenkins, J. L., Becker, et al
  2024; 221 (8)

- **The Undiagnosed Diseases Network: Characteristics of solvable applicants and diagnostic suggestions for non-accepted ones.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Mulvihill, J. J., Findley, L., Ni, W., Sinsheimer, J. S., Cole, F. S., Esteves, C., Bernstein, J. A., Newman, J. H., Wheeler, M. T., Mokry, J. R.
  2024: 101203

- **Loss of function of FAM177A1, a Golgi complex localized protein, causes a novel neurodevelopmental disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Kohler, J. N., Legro, N. R., Baldridge, D., Shin, J., Bowman, A., Ugur, B., Jackstadt, M. M., Shriver, L. P., Patti, G. J., Zhang, B., Feng, W., McAdow, A. R., Goddard, et al
  2024: 101166

- **Immunological and hematological findings as major features in a patient with a new germline pathogenic CBL variant.** *American journal of medical genetics. Part A*
  Stellacci, E., Carter, J. N., Pannone, L., Stevenson, D., Moslehi, D., Venanzi, S., Bernstein, J. A., Tartaglia, M., Martinelli, S.
  2024: e63627

- **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*
  Pucel, J., Briere, L. C., Reuter, C., Gochyyev, P., LeBlanc, K.
  2024: 101115

- **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

- **A multi-omics approach to the characterization of a novel repeat expansion in FAM193B in a family with oculopharyngodistal myopathy**
  Reuter, C., Fazal, S., Jensen, T., Gorzynski, J., Marwaha, S., Bonner, D., Kohler, J., Mendez, R., Voutos, Emami, S., Kravets, E., Smith, K., Goddard, P., et al
  SAGE PUBLICATIONS LTD.2024: 400-401

- **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, Ł. 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.
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