



## Richard J. Reimer, MD

Professor of Neurology and Neurological Sciences (Adult Neurology)

### CLINICAL OFFICE (PRIMARY)

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### Bio

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#### BIO

Dr. Reimer specializes in treatment of lysosomal storage disorders that affect the nervous system. He has been practicing as a neurologist for over 20 years. He has a particular interest in Fabry disease and Gaucher disease.

#### CLINICAL FOCUS

- Neurology

#### ACADEMIC APPOINTMENTS

- Professor - University Medical Line, Adult Neurology
- Member, Bio-X
- Member, Maternal & Child Health Research Institute (MCHRI)
- Member, Wu Tsai Neurosciences Institute

#### HONORS AND AWARDS

- Basil O'Connor Award, March of Dimes (2003-05)
- Brain and Immuno imaging Grant, Dana Foundation (2007-09)

#### PROFESSIONAL EDUCATION

- Board Certification: Neurology, American Board of Psychiatry and Neurology (1998)
- Residency: University of California at San Francisco School of Medicine (1995) CA
- Internship: University of California at San Francisco School of Medicine (1992) CA
- MD, Emory University, Medicine (1991)
- BA, Yale University, Molecular Biophysics and Biochemistry (1985)

## LINKS

- Reimer Lab Site: <https://web.stanford.edu/~rjreimer>
- Get a Second Opinion: <https://stanfordhealthcare.org/second-opinion/overview.html>

## Research & Scholarship

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### CURRENT RESEARCH AND SCHOLARLY INTERESTS

Reimer Lab interests

A primary interest of our lab is to understand how nerve cells make and recycle neurotransmitters, the small molecules that they use to communicate with each other. In better defining these processes we hope to achieve our long-term goal of identifying novel sites for treatment of diseases such as epilepsy and Parkinson Disease. In our studies on neurotransmitter metabolism we have focused our efforts on transporters, a functional class of proteins that move neurotransmitters and other small molecules across membranes in cells. Transporters have many characteristics that make them excellent pharmacological targets, and not surprisingly some of the most effective treatments for neuropsychiatric disorders are directed at transporters. We are specifically focusing on two groups of transporters vesicular neurotransmitter transporters that package neurotransmitters into vesicles for release, and glutamine transporters that shuttle glutamine, a precursor for two major neurotransmitters glutamate and GABA, to neurons from glia, the supporting cells that surround them. We are pursuing these goals through molecular and biochemical studies, and, in collaboration with the Huguenard and Prince labs, through physiological and biosensor based imaging studies to better understand how pharmacological targeting of these molecules will influence neurological disorders.

A second interest of our lab is to define mechanism underlying the pathology of lysosomal storage disorders. Lysosomes are membrane bound acidic intracellular organelles filled with hydrolytic enzymes that normally function as recycling centers within cells by breaking down damaged cellular macromolecules. Several degenerative diseases designated as lysosomal storage disorders (LSDs) are associated with the accumulation of material within lysosomes. Tay-Sachs disease, Neimann-Pick disease and Gaucher disease are some of the more common LSDs. For reasons that remain incompletely understood, these diseases often affect the nervous system out of proportion to other organs. As a model for LSDs we are studying the lysosomal free sialic acid storage disorders. These diseases are the result of a defect in transport of sialic acid across lysosomal membranes and are associated with mutations in the gene encoding the sialic acid transporter sialin. We are using molecular, genetic and biochemical approaches to better define the normal function of sialin and to determine how loss of sialin function leads to neurodevelopmental defects and neurodegeneration associated with the lysosomal free sialic acid storage disorders.

## Teaching

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### GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Molecular and Cellular Physiology (Phd Program)
- Neurosciences (Phd Program)

## Publications

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### PUBLICATIONS

- **Symptom Burden and Care Satisfaction in US Military Veterans With ALS: Results of a National Survey.** *Muscle & nerve*  
Howard, I., Lyerly, M., Reimer, R., Patwa, H., Darling, L.  
2026
- **Cystinosis is Associated with Deficits in Muscle Mass, Strength and Hip Bone Density in Children and Young Adults.** *Kidney360*

- Sheldon, C. R., Grimm, P. C., Long, J., Whalen, J. R., Kent, K., Reimer, R. J., Strickland, A., Phelps, M., Leonard, M. B.  
2025
- **Collaborative research efforts drive therapeutic advancements for free sialic acid storage disorder (FSASD)**  
Malicdan, M., Adams, D. R., Brooks, P. J., Dobrenis, K., Gahl, W. A., Gasnier, B., Hackbarth, M. E., Hossain, M., Leoyklang, P., Anne-Longin, C., Paavola, L., Platt, F., Pollard, et al  
ACADEMIC PRESS INC ELSEVIER SCIENCE.2024
  - **An electronic health record cohort of Veterans with amyotrophic lateral sclerosis.** *Amyotrophic lateral sclerosis & frontotemporal degeneration*  
Reimer, R. J., Goncalves, A., Soper, B., Cadena, J., Wilson, J. L., Gryshuk, A. L., Suarez, P., Osborne, T. F., Grimes, K. V., Ray, P.  
2023: 1-7
  - **A concerted action to explore therapies for free sialic acid storage disease**  
Huizing, M., Walkley, S. U., Wasserstein, M., Anne-Longin, C., Wang, R. Y., Reimer, R., Pollard, L., Paavola, L., Malicdan, M., Sabir, M. S., Leoyklang, P., Adams, D. R., Gahl, et al  
ACADEMIC PRESS INC ELSEVIER SCIENCE.2023: 64-65
  - **Glial control of sphingolipid levels sculpts diurnal remodeling in a circadian circuit.** *Neuron*  
Vaughen, J. P., Theisen, E., Rivas-Serna, I. M., Berger, A. B., Kalakuntla, P., Anreiter, I., Mazurak, V. C., Rodriguez, T. P., Mast, J. D., Hartl, T., Perlstein, E. O., Reimer, R. J., Clandinin, et al  
2022
  - **Metalworking Fluid Exposure and Stroke Mortality Among U.S. Autoworkers.** *American journal of epidemiology*  
Elser, H., Chen, K. T., Arteaga, D., Reimer, R., Picciotto, S., Costello, S., Eisen, E. A.  
1800
  - **Acute on Chronic Neuromuscular Respiratory Failure in the Intensive Care Unit: Optimization of Triage, Ventilation Modes, and Extubation** *CUREUS*  
Murray, N. M., Reimer, R. J., Cao, M.  
2021; 13 (7)
  - **Acute on Chronic Neuromuscular Respiratory Failure in the Intensive Care Unit: Optimization of Triage, Ventilation Modes, and Extubation.** *Cureus*  
Murray, N. M., Reimer, R. J., Cao, M.  
2021; 13 (7): e16297
  - **Leigh Disease-Like Neuropathology in an Adult Following Covid-19 Infection and Global Anoxia**  
Sobel, R., Dulken, B., Reimer, R., Louie, C.  
OXFORD UNIV PRESS INC.2021: 582
  - **Correction to: Air pollution, methane super-emitters, and oil and gas wells in Northern California: the relationship with migraine headache prevalence and exacerbation.** *Environmental health : a global access science source*  
Elser, H., Morello-Frosch, R., Jacobson, A., Pressman, A., Kioumourtoglou, M., Reimer, R., Casey, J. A.  
2021; 20 (1): 57
  - **Air pollution, methane super-emitters, and oil and gas wells in Northern California: the relationship with migraine headache prevalence and exacerbation.** *Environmental health : a global access science source*  
Elser, H., Morello-Frosch, R., Jacobson, A., Pressman, A., Kioumourtoglou, M., Reimer, R., Casey, J. A.  
2021; 20 (1): 45
  - **Mini-Review Free Sialic Acid Storage Disorder: Progress and Promise.** *Neuroscience letters*  
Huizing, M., Hackbarth, M. E., Adams, D. R., Wasserstein, M., Patterson, M. C., Walkley, S. U., Gahl, W. A., FSASD Consortium, Adams, D. R., Dobrenis, K., Foglio, J., Gahl, W. A., Gasnier, B., et al  
2021: 135896
  - **Air pollution, methane super-emitters, and oil and gas wells in Northern California: the relationship with migraine headache prevalence and exacerbation**  
Elser, H., Morello-Frosch, R., Jacobson, A., Pressman, A., Kioumourtoglou, M., Reimer, R., Casey, J.  
LIPPINCOTT WILLIAMS & WILKINS.2021
  - **NEUROMUSCULAR RESPIRATORY FAILURE IN THE ICU: AN "UNWEANABLE" PATIENT AND A NOVEL TIERED PROTOCOL**

Murray, N., Reimer, R., Cao, M.  
LIPPINCOTT WILLIAMS & WILKINS.2020

- **Targeted genomic CRISPR-Cas9 screen identifies MAP4K4 as essential for glioblastoma invasion.** *Scientific reports*  
Prolo, L. M., Li, A. n., Owen, S. F., Parker, J. J., Foshay, K. n., Nitta, R. T., Morgens, D. W., Bolin, S. n., Wilson, C. M., Vega L, J. C., Luo, E. J., Nwagbo, G. n., Waziri, et al  
2019; 9 (1): 14020
- **Bassoon Controls Presynaptic Autophagy through Atg5.** *Neuron*  
Okerlund, N. D., Schneider, K., Leal-Ortiz, S., Montenegro-Venegas, C., Kim, S. A., Garner, L. C., Gundelfinger, E. D., Reimer, R. J., Garner, C. C.  
2017; 93 (4): 897-913 e7
- **Human Astrocyte Maturation Captured in 3D Cerebral Cortical Spheroids Derived from Pluripotent Stem Cells.** *Neuron*  
Sloan, S. A., Darmanis, S. n., Huber, N. n., Khan, T. A., Birey, F. n., Caneda, C. n., Reimer, R. n., Quake, S. R., Barres, B. A., Paşca, S. P.  
2017; 95 (4): 779–90.e6
- **Trio, a Rho Family GEF, Interacts with the Presynaptic Active Zone Proteins Piccolo and Bassoon** *PLOS ONE*  
Terry-Lorenzo, R. T., Torres, V. I., Wagh, D., Galaz, J., Swanson, S. K., Florens, L., Washburn, M. P., Waites, C. L., Gundelfinger, E. D., Reimer, R. J., Garner, C. C.  
2016; 11 (12)
- **Plasma taurine levels are not affected by vigabatrin in pediatric patients.** *Epilepsia*  
Spelbrink, E. M., Mabud, T. S., Reimer, R., Porter, B. E.  
2016; 57 (8): e168-72
- **Piccolo Directs Activity Dependent F-Actin Assembly from Presynaptic Active Zones via Daam1** *PLOS ONE*  
Wagh, D., Terry-Lorenzo, R., Waites, C. L., Leal-Ortiz, S. A., Maas, C., Reimer, R. J., Garner, C. C.  
2015; 10 (4)
- **Endozepines.** *Advances in pharmacology (San Diego, Calif.)*  
Farzampour, Z., Reimer, R. J., Huguenard, J.  
2015; 72: 147-164
- **Endozepines DIVERSITY AND FUNCTIONS OF GABA RECEPTORS: A TRIBUTE TO HANNS MOHLER, PT A**  
Farzampour, Z., Reimer, R. J., Huguenard, J.  
edited by Rudolph, U.  
2015; 72: 147-164
- **alpha 5-GABAA receptors negatively regulate MYC-amplified medulloblastoma growth** *ACTA NEUROPATHOLOGICA*  
Sengupta, S., Weeraratne, S. D., Sun, H., Phallen, J., Rallapalli, S. K., Teider, N., Kosaras, B., Amani, V., Pierre-Francois, J., Tang, Y., Brian Nguyen, B., Yu, F., Schubert, et al  
2014; 127 (4): 593-603
- **A local glutamate-glutamine cycle sustains synaptic excitatory transmitter release.** *Neuron*  
Tani, H., Dulla, C. G., Farzampour, Z., Taylor-Weiner, A., Huguenard, J. R., Reimer, R. J.  
2014; 81 (4): 888-900
- **SLC17: a functionally diverse family of organic anion transporters.** *Molecular aspects of medicine*  
Reimer, R. J.  
2013; 34 (2-3): 350-359
- **Vesicular uptake and exocytosis of L-aspartate is independent of sialin** *FASEB JOURNAL*  
Morland, C., Nordengen, K., Larsson, M., Prolo, L. M., Farzampour, Z., Reimer, R. J., Gundersen, V.  
2013; 27 (3): 1264-1274
- **Glutamate biosensor imaging reveals dysregulation of glutamatergic pathways in a model of developmental cortical malformation** *NEUROBIOLOGY OF DISEASE*  
Dulla, C. G., Tani, H., Brill, J., Reimer, R. J., Huguenard, J. R.  
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2012; 49C: 232-246
- **Biochemistry to the Rescue: A CIC-2 Auxiliary Subunit Provides a Tangible Link to Leukodystrophy** *NEURON*  
Maduke, M. C., Reimer, R. J.  
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- **Structure-Function Studies of the SLC17 Transporter Sialin Identify Crucial Residues and Substrate-induced Conformational Changes** *JOURNAL OF BIOLOGICAL CHEMISTRY*  
Courville, P., Quick, M., Reimer, R. J.  
2010; 285 (25): 19316-19323
- **Glutamine Is Required for Persistent Epileptiform Activity in the Disinhibited Neocortical Brain Slice** *JOURNAL OF NEUROSCIENCE*  
Tani, H., Dulla, C. G., Huguenard, J. R., Reimer, R. J.  
2010; 30 (4): 1288-1300
- **The Lysosomal Sialic Acid Transporter Sialin Is Required for Normal CNS Myelination** *JOURNAL OF NEUROSCIENCE*  
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- **GLUTAMINE POTENTIATES GLUTAMATE RELEASE DURING HIGH FREQUENCY STIMULATION OF ISOLATED LAYER 1 AXONS**  
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- **Synaptic Vesicle Protein NTT4/XT1 (SLC6A17) Catalyzes Na<sup>+</sup>-coupled Neutral Amino Acid Transport** *JOURNAL OF BIOLOGICAL CHEMISTRY*  
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- **Imaging of glutamate in brain slices using FRET sensors** *JOURNAL OF NEUROSCIENCE METHODS*  
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2008; 168 (2): 306-319
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Myall, N. J., Wreden, C. C., Wlizla, M., Reimer, R. J.  
2007; 92 (4): 371-374
- **Analysis of glutamate recycling in maintenance of epileptiform activity the acute slice**  
Reimer, R. J., Tani, H., Dulla, C. G., Huguenard, J. R.  
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- **Membrane topology of the Drosophila vesicular glutamate transporter** *JOURNAL OF NEUROCHEMISTRY*  
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- **Modulation of epileptiform activity by glutamine and system A transport in a model of post-traumatic epilepsy** *NEUROBIOLOGY OF DISEASE*  
Tani, H., Bandrowski, A. E., Parada, I., Wynn, M., Huguenard, J. R., Prince, D. A., Reimer, R. J.  
2007; 25 (2): 230-238
- **Biochemical and genetic analysis of ANK in arthritis and bone disease** *AMERICAN JOURNAL OF HUMAN GENETICS*  
Gurley, K. A., Reimer, R. J., Kingsley, D. M.  
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- **Detection of glutamate release from neurons by genetically encoded surface-displayed FRET nanosensors** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*  
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Wreden, C. C., Wlizla, M., Reimer, R. J.  
2005; 280 (2): 1408-1416
- **Organic anion transport is the primary function of the SLC17/type I phosphate transporter family** *PFLUGERS ARCHIV-EUROPEAN JOURNAL OF PHYSIOLOGY*  
Reimer, R. J., Edwards, R. H.  
2004; 447 (5): 629-635
- **Vesicular neurotransmitter transporter expression in developing postnatal rodent retina: GABA and glycine precede glutamate** *JOURNAL OF NEUROSCIENCE*  
Johnson, J., Tian, N., Caywood, M. S., Reimer, R. J., Edwards, R. H., Copenhagen, D. R.  
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