Stanford

Aaron D. Gitler

Stanford Medicine Basic Science Professor

Genetics

Bio

ACADEMIC APPOINTMENTS

- Professor, Genetics
- Member, Bio-X
- Member, Wu Tsai Neurosciences Institute

HONORS AND AWARDS

- Eberly College of Science Alumni Society's Outstanding Science Alumni Award, Penn State University (2020)
- Sheila Essey Award: An Award for ALS Research, American Academy of Neurology (2019)
- Friedrich Merz Guest Professorship Award, Merz Pharma (2017)
- R35 Research Program Award, NINDS (2016)
- Glenn Award for Research in Biological Mechanisms of Aging, Glenn Foundation for Medical Research (2015)
- Young Scientist Lectureship Award, International Society for Neurochemistry (2013)
- Addgene Innovation Award, Addgene (2011)
- Eppendorf and Science Prize for Neurobiology, Finalist, Eppendorf and Science Magazine (2011)
- Instituto Paulo Gontijo International Medicine PG Award, Instituto Paulo Gontijo (2011)
- Scientist to Watch, The Scientist (2010)
- NIH Director''s New Innovator Award, NIH (2008)
- Pew Scholar in the Biomedical Sciences, The Pew Charitable Trusts (2008)
- Rita Allen Foundation Scholar, The Rita Allen Foundation (2008)

PROFESSIONAL EDUCATION

- Postdoctoral Fellow, Whitehead Institute for Biomedical Research , Cell biology and genetics (2007)
- Ph.D., University of Pennsylvania , Cell and Molecular Biology (2004)
- B.S., Penn State University , Biochemistry and Molecular Biology (2000)

LINKS

- Gitler Lab: http://gitlerlab.org

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

We use the baker's yeast, Saccharomyces cerevisiae, as a model system to study the cell biology underpinning protein-misfolding diseases like Parkinson's disease and ALS. Since dealing with misfolded proteins is an ancient problem, we hypothesize that the mechanisms employed to cope with them are likely conserved from yeast to man. Our long-term goal is to identify the critical genes and cellular pathways affected by misfolded human disease proteins.

C9orf72 in ALS and FTD: Disease models and mechanisms

Mutations in the C9orf72 gene are the most common cause of ALS and frontotemporal dementia (FTD). The mutation is a massive hexanucleotide repeat (GGGGCC) expansion in the intron of C9orf72. The mechanism by which C9orf72 mutations cause disease has remained unclear and of intense interest. In collaboration with the Petrucelli laboratory we have recently identified a way to selectively inhibit the expression of both sense and antisense mutant C9orf72 transcripts, which could offer therapeutic potential (Kramer et al., Science 2016).

New yeast models of neurodegenerative diseases

Encouraged by the power of the yeast system to gain insight into #-synuclein biology, we are creating new yeast models to study additional protein-misfolding disorders, including Alzheimer's disease and ALS. We recently developed a yeast model to study the ALS disease protein TDP-43 (Johnson et al., Proc Natl Acad Sci USA 2008).

We have used yeast and in vitro biochemistry (in collaboration with Jim Shorter at PENN) to analyze the effects of ALS-linked TDP-43 mutations on aggregation and toxicity (Johnson et al., J Biol Chem 2009). We are now using these models to perform high-throughput genetic and small molecule screens to elucidate the molecular pathways that regulate the function of these disease proteins and control their conversion to a pathological conformation. We are currently analyzing hits from recent high-throughput screens that identified potent modifiers of TDP-43 toxicity. We are validating these hits in cell culture, animal models (mouse, fly, and zebrafish), and human patient samples.

These TDP-43 modifier screens are providing insight in two main ways:

1. The genes and pathways that are able to modify TDP-43 toxicity in yeast are now good candidates for evaluation as genetic contributors to ALS and related disorders in humans (e.g., see ataxin 2 below).

2. The yeast hits and their homologs are candidate therapeutic targets, especially gene deletions (Armakola et al., Nat Genet 2012; Kim et al., Nat Genet 2014).

Ataxin-2 and ALS

Interestingly, one of the hits from our yeast TDP-43 genetic modifier screen, PBP1, is the homolog of a human neurodegenerative disease protein, ataxin 2. We have validated this genetic interaction in the fly nervous system (in collaboration with Nancy Bonini at PENN), used biochemistry to show the proteins physically associate in an RNA-dependent manner.

We analyzed the ataxin 2 gene in 915 individuals with ALS and 980 healthy controls and found mutations in this gene as a common geneticrisk factor for ALS in humans. Long polyglutamine (polyQ) expansions (>34Q) in ataxin 2 cause spinocerebellar ataxia type 2 (SCA2). We found intermediate-length polyQ expansions in ataxin 2 (27-33Q) significantly associated with increased risk for ALS (Elden et al., Nature 2010). A role for polyQ expansions in ataxin 2 in ALS and related diseases is being evaluated by us and others in independent patient populations worldwide. Click here for an updated summary of these results.

We found that lowering levels of ataxin 2 in mouse, either by knockout or with antisense oligonucleotides (ASOs) can markedly extend survival and reduce pathology in TDP-43 transgenic mice (Becker et al., Nature 2017). We are extending these studies to additional mouse models and testing effects of ataxin 2 lowering in human cell models.

Teaching

COURSES

2022-23

- Neurogenetics Core: NEPR 213 (Win)
- Prions in Health & Disease: BIOS 277 (Aut)

2021-22

- Neurogenetics Core: NEPR 213 (Aut)

2020-21

- Neurogenetics Core: NEPR 213 (Aut)

2019-20

- Molecular Mechanisms of Neurodegenerative Disease: BIO 267 (Win)
- Neurogenetics Core: NEPR 213 (Aut)
- Prions in Health & Disease: BIOS 277 (Aut)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Cindy Lin, Kathryn Wu

Postdoctoral Faculty Sponsor

Tetsuya Akiyama, Jacob Blum, Caiwei Guo, Maya Maor Nof, Yi Zeng

Doctoral Dissertation Advisor (AC)

Olivia Gautier

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (Phd Program)
- Neurosciences (Phd Program)

Publications

PUBLICATIONS

- **TDP-43 represses cryptic exon inclusion in the FTD-ALS gene UNC13A.** *Nature*
  Ma, X. R., Prudencio, M., Koike, Y., Vatsavayai, S. C., Kim, G., Harbinski, F., Briner, A., Rodriguez, C. M., Guo, C., Akiyama, T., Schmidt, H. B., Cummings, B. B., Wyatt, et al
  2022

- **A prion-like protein regulator of seed germination undergoes hydration-dependent phase separation.** *Cell*
  Dorone, Y., Boeynaems, S., Flores, E., Jin, B., Hateley, S., Bossi, F., Lazarus, E., Pennington, J. G., Michiels, E., De Decker, M., Vints, K., Baatsen, P., Bassel, et al
  2021

- **Single-cell transcriptomic analysis of the adult mouse spinal cord reveals molecular diversity of autonomic and skeletal motor neurons.** *Nature neuroscience*
  Blum, J. A., Klemm, S., Shadrach, J. L., Guttenplan, K. A., Nakayama, L., Kathiria, A., Hoang, P. T., Gautier, O., Kaltschmidt, J. A., Greenleaf, W. J., Gitler, A. D.
  2021

- **p53 is a central regulator driving neurodegeneration caused by C9orf72 poly(PR).** *Cell*
  Maor-Nof, M. n., Shipony, Z. n., Lopez-Gonzalez, R. n., Nakayama, L. n., Zhang, Y. J., Couthouis, J. n., Blum, J. A., Castruita, P. A., Linares, G. R., Ruan, K. n., Ramaswami, G. n., Simon, D. J., Nof, et al
  2021

- **RPS25 is required for efficient RAN translation of C9orf72 and other neurodegenerative disease-associated nucleotide repeats.** *Nature neuroscience*
  Yamada, S. B., Gendron, T. F., Niccoli, T. n., Genuth, N. R., Grosely, R. n., Shi, Y. n., Glaria, I. n., Kramer, N. J., Nakayama, L. n., Fang, S. n., Dinger, T. J., Thoeng, A. n., Rocha, et al
  2019

- **CRISPR-Cas9 screens in human cells and primary neurons identify modifiers of C9ORF72 dipeptide-repeat-protein toxicity.** *Nature genetics*
  Kramer, N. J., Haney, M. S., Morgens, D. W., Jovi#i#, A. n., Couthouis, J. n., Li, A. n., Ousey, J. n., Ma, R. n., Bieri, G. n., Tsui, C. K., Shi, Y. n., Hertz, N. T., Tessier-Lavigne, et al
  2018

- **Therapeutic reduction of ataxin-2 extends lifespan and reduces pathology in TDP-43 mice** *NATURE*
  Becker, L. A., Huang, B., Bieri, G., Ma, R., Knowles, D. A., Jafar-Nejad, P., Messing, J., Kim, H. J., Soriano, A., Auburger, G., Pulst, S. M., Taylor, J. P., Rigo, et al
  2017; 544 (7650): 367-?

- **Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts.** *Science*
  Kramer, N. J., Carlomagno, Y., Zhang, Y., Almeida, S., Cook, C. N., Gendron, T. F., Prudencio, M., van Blitterswijk, M., Belzil, V., Couthouis, J., Paul, J. W., Goodman, L. D., Daughrity, et al
  2016; 353 (6300): 708-712

- **Modifiers of C9orf72 dipeptide repeat toxicity connect nucleocytoplasmic transport defects to FTD/ALS.** *Nature neuroscience*
  Jovicic, A., Mertens, J., Boeynaems, S., Bogaert, E., Chai, N., Yamada, S. B., Paul, J. W., Sun, S., Herdy, J. R., Bieri, G., Kramer, N. J., Gage, F. H., Van Den Bosch, et al
  2015; 18 (9): 1226-1229

- **Ataxin-2 intermediate-length polyglutamine expansions are associated with increased risk for ALS** *NATURE*
  Elden, A. C., Kim, H., Hart, M. P., Chen-Plotkin, A. S., Johnson, B. S., Fang, X., Armakola, M., Geser, F., Greene, R., Lu, M. M., Padmanabhan, A., Clay-Falcone, D., McCluskey, et al
  2010; 466 (7310): 1069-U77

- **Radiogenomics of C9orf72 expansion carriers reveals global transposable element de-repression and enables prediction of thalamic atrophy and clinical impairment.** *The Journal of neuroscience : the official journal of the Society for Neuroscience*
  Bonham, L. W., Geier, E. G., Sirkis, D. W., Leong, J. K., Ramos, E. M., Wang, Q., Karydas, A., Lee, S. E., Sturm, V. E., Sawyer, R. P., Friedberg, A., Ichida, J. K., Gitler, et al
  2022

- **Rnq1! You are still dangerous, but you can be my wingman anytime.** *Molecular cell*
  Guo, C., Gitler, A. D.
  2022; 82 (22): 4194-4196

- **Mesoscale connections and gene expression empower whole-brain modeling of alpha-synuclein spread, aggregation, and decay dynamics.** *Cell reports*
  Dadgar-Kiani, E., Bieri, G., Melki, R., Gitler, A. D., Lee, J. H.
  2022; 41 (6): 111631

- **Targeting RTN4/NoGo-Receptor reduces levels of ALS protein ataxin-2.** *Cell reports*
  Rodriguez, C. M., Bechek, S. C., Jones, G. L., Nakayama, L., Akiyama, T., Kim, G., Solow-Cordero, D. E., Strittmatter, S. M., Gitler, A. D.

2022; 41 (4): 111505

- **Genome-wide CRISPR screen reveals v-ATPase as a drug target to lower levels of ALS protein ataxin-2.** *Cell reports*
  Kim, G., Nakayama, L., Blum, J. A., Akiyama, T., Boeynaems, S., Chakraborty, M., Couthouis, J., Tassoni-Tsuchida, E., Rodriguez, C. M., Bassik, M. C., Gitler, A. D.
  2022; 41 (4): 111508

- **The material properties of a bacterial-derived biomolecular condensate tune biological function in natural and synthetic systems.** *Nature communications*
  Lasker, K., Boeynaems, S., Lam, V., Scholl, D., Stainton, E., Briner, A., Jacquemyn, M., Daelemans, D., Deniz, A., Villa, E., Holehouse, A. S., Gitler, A. D., Shapiro, et al
  2022; 13 (1): 5643

- **APOE told me put my fat in the bag and nobody gets hurt.** *Cell*
  Kim, G., Gitler, A. D.
  2022; 185 (13): 2201-2203

- **Why you always in a mood? Pumpin' polyP, actin' brand new.** *Neuron*
  Guttenplan, K. A., Gitler, A. D.
  2022; 110 (10): 1603-1605

- **Cracking the cryptic code in amyotrophic lateral sclerosis and frontotemporal dementia: Towards therapeutic targets and biomarkers.** *Clinical and translational medicine*
  Akiyama, T., Koike, Y., Petrucelli, L., Gitler, A. D.
  2022; 12 (5): e818

- **Singling out motor neurons in the age of single-cell transcriptomics.** *Trends in genetics : TIG*
  Blum, J. A., Gitler, A. D.
  2022

- **Confirming Pathogenicity of the F386L PSEN1 Variant in a South Asian Family With Early-Onset Alzheimer Disease.** *Neurology. Genetics*
  Eger, S. J., Le Guen, Y., Khan, R. R., Hall, J. N., Kennedy, G., Zaharchuk, G., Couthouis, J., Brooks, W. S., Velakoulis, D., Napolioni, V., Belloy, M. E., Dalgard, C. L., Mormino, et al
  1800; 8 (1): e647

- **Phenotypic Heterogeneity among GBA p.R202X Carriers in Lewy Body Spectrum Disorders.** *Biomedicines*
  Napolioni, V., Fredericks, C. A., Kim, Y., Channappa, D., Khan, R. R., Kim, L. H., Zafar, F., Couthouis, J., Davidzon, G. A., Mormino, E. C., Gitler, A. D., Montine, T. J., Schule, et al
  1800; 10 (1)

- **An optimized ATAC-seq protocol for genome-wide mapping of active regulatory elements in primary mouse cortical neurons.** *STAR protocols*
  Maor-Nof, M., Shipony, Z., Marinov, G. K., Greenleaf, W. J., Gitler, A. D.
  2021; 2 (4): 100854

- **Aaron Gitler** *NEURON*
  Gitler, A.
  2021; 109 (21): 3352-3354

- **You come at the misfolded proteins, you best not miss.** *Trends in biochemical sciences*
  Boeynaems, S., Gitler, A. D.
  2021

- **Neurotoxic reactive astrocytes induce cell death via saturated lipids.** *Nature*
  Guttenplan, K. A., Weigel, M. K., Prakash, P., Wijewardhane, P. R., Hasel, P., Rufen-Blanchette, U., Munch, A. E., Blum, J. A., Fine, J., Neal, M. C., Bruce, K. D., Gitler, A. D., Chopra, et al
  2021

- **C9orf72-derived arginine-containing dipeptide repeats associate with axonal transport machinery and impede microtubule-based motility.** *Science advances*
  Fumagalli, L., Young, F. L., Boeynaems, S., De Decker, M., Mehta, A. R., Swijsen, A., Fazal, R., Guo, W., Moisse, M., Beckers, J., Dedeene, L., Selvaraj, B. T., Vandoorne, et al
  2021; 7 (15)

- **A versatile system to record cell-cell interactions.** *eLife*
  Tang, R., Murray, C. W., Linde, I. L., Kramer, N. J., Lyu, Z., Tsai, M. K., Chen, L. C., Cai, H., Gitler, A. D., Engleman, E., Lee, W., Winslow, M. M.
  2020; 9

- **It's not all about those bases** *NATURE*
  Gautier, O., Gitler, A. D.
  2020; 585 (7823): 34–35

- **Just Took a DNA Test, Turns Out 100% Not That Phase.** *Molecular cell*
  Gitler, A. D., Shorter, J., Ha, T., Myong, S.
  2020; 78 (2): 193–94

- **A memory of eS25 loss drives resistance phenotypes.** *Nucleic acids research*
  Johnson, A. G., Flynn, R. A., Lapointe, C. P., Ooi, Y. S., Zhao, M. L., Richards, C. M., Qiao, W. n., Yamada, S. B., Couthouis, J. n., Gitler, A. D., Carette, J. E., Puglisi, J. D.
  2020

- **BraInMap Elucidates the Macromolecular Connectivity Landscape of Mammalian Brain.** *Cell systems*
  Pourhaghighi, R. n., Ash, P. E., Phanse, S. n., Goebels, F. n., Hu, L. Z., Chen, S. n., Zhang, Y. n., Wierbowski, S. D., Boudeau, S. n., Moutaoufik, M. T., Malty, R. H., Malolepsza, E. n., Tsafou, et al
  2020; 10 (4): 333–50.e14

- **Evolution of a Human-Specific Tandem Repeat Associated with ALS.** *American journal of human genetics*
  Course, M. M., Gudsnuk, K. n., Smukowski, S. N., Winston, K. n., Desai, N. n., Ross, J. P., Sulovari, A. n., Bourassa, C. V., Spiegelman, D. n., Couthouis, J. n., Yu, C. E., Tsuang, D. W., Jayadev, et al
  2020

- **ALS Genetics: Gains, Losses, and Implications for Future Therapies.** *Neuron*
  Kim, G. n., Gautier, O. n., Tassoni-Tsuchida, E. n., Ma, X. R., Gitler, A. D.
  2020

- **Knockout of reactive astrocyte activating factors slows disease progression in an ALS mouse model.** *Nature communications*
  Guttenplan, K. A., Weigel, M. K., Adler, D. I., Couthouis, J. n., Liddelow, S. A., Gitler, A. D., Barres, B. A.
  2020; 11 (1): 3753

- **Genome-wide synthetic lethal CRISPR screen identifies FIS1 as a genetic interactor of ALS-linked C9ORF72.** *Brain research*
  Chai, N., Haney, M. S., Couthouis, J., Morgens, D. W., Benjamin, A., Wu, K., Ousey, J., Fang, S., Finer, S., Bassik, M. C., Gitler, A. D.
  2019: 146601

- **Symmetric dimethylation of poly-GR correlates with disease duration in C9orf72 FTLD and ALS and reduces poly-GR phase separation and toxicity.** *Acta neuropathologica*
  Gittings, L. M., Boeynaems, S., Lightwood, D., Clargo, A., Topia, S., Nakayama, L., Troakes, C., Mann, D. M., Gitler, A. D., Lashley, T., Isaacs, A. M.
  2019

- **Genetic Spectrum and Variability in Chinese Patients with Amyotrophic Lateral Sclerosis.** *Aging and disease*
  Liu, Z., Lin, H., Wei, Q., Zhang, Q., Chen, C., Tao, Q., Liu, G., Ni, W., Gitler, A. D., Li, H., Wu, Z.
  2019; 10 (6): 1199–1206

- **Neuroinflammatory reactive astrocytes in acute injury and neurodegenerative disease**
  Guttenplan, K., Weigel, M., Munch, A., Bennett, M., Liddelow, S., Gitler, A., Barres, B.
  WILEY.2019: E520

- **LRRK2 modifies alpha-syn pathology and spread in mouse models and human neurons** *ACTA NEUROPATHOLOGICA*
  Bieri, G., Brahic, M., Bousset, L., Couthouis, J., Kramer, N. J., Ma, R., Nakayama, L., Monbureau, M., Defensor, E., Schuele, B., Shamloo, M., Melki, R., Gitler, et al
  2019; 137 (6): 961–80

- **Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly** *HUMAN GENETICS*
  Ullah, I., Kakar, N., Schrauwen, I., Hussain, S., Chakchouk, I., Liaqat, K., Acharya, A., Wasif, N., Santos-Cortez, R. P., Khan, S., Aziz, A., Lee, K., Couthouis, et al
  2019; 138 (6): 593–600

- **Toxic expanded GGGGCC repeat transcription is mediated by the PAF1 complex in C9orf72-associated FTD.** *Nature neuroscience*
  Goodman, L. D., Prudencio, M., Kramer, N. J., Martinez-Ramirez, L. F., Srinivasan, A. R., Lan, M., Parisi, M. J., Zhu, Y., Chew, J., Cook, C. N., Berson, A., Gitler, A. D., Petrucelli, et al
  2019

- **A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS** *GENOME RESEARCH*
  Gelfman, S., Dugger, S., Moreno, C., Ren, Z., Wolock, C. J., Shneider, N. A., Phatnani, H., Cirulli, E. T., Lasseigne, B. N., Harris, T., Maniatis, T., Rouleau, G. A., Brown, et al
  2019; 29 (5): 809–18

- **Spontaneous driving forces give rise to protein-RNA condensates with coexisting phases and complex material properties** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Boeynaems, S., Holehouse, A. S., Weinhardt, V., Kovacs, D., Van Lindt, J., Larabell, C., Van Den Bosch, L., Das, R., Tompa, P. S., Pappu, R., Gitler, A. D.
  2019; 116 (16): 7889–98

- **Spontaneous driving forces give rise to protein-RNA condensates with coexisting phases and complex material properties.** *Proceedings of the National Academy of Sciences of the United States of America*
  Boeynaems, S., Holehouse, A. S., Weinhardt, V., Kovacs, D., Van Lindt, J., Larabell, C., Van Den Bosch, L., Das, R., Tompa, P. S., Pappu, R. V., Gitler, A. D.
  2019; 116 (16): 7889–98

- **Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly.** *Human genetics*
  Ullah, I., Kakar, N., Schrauwen, I., Hussain, S., Chakchouk, I., Liaqat, K., Acharya, A., Wasif, N., Santos-Cortez, R. L., Khan, S., Aziz, A., Lee, K., Couthouis, et al
  2019

- **Loss of CREST leads to neuroinflammatory responses and ALS-like motor defects in mice** *TRANSLATIONAL NEURODEGENERATION*
  Cheng, C., Yang, K., Wu, X., Zhang, Y., Shan, S., Gitler, A., Ghosh, A., Qiu, Z.
  2019; 8: 13

- **A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS.** *Genome research*
  Gelfman, S., Dugger, S. A., Araujo Martins Moreno, C., Ren, Z., Wolock, C. J., Shneider, N., Phatnani, H., Cirulli, E. T., Lasseigne, B. N., Harris, T., Maniatis, T., Rouleau, G., Brown, et al
  2019

- **LRRK2 modifies alpha-syn pathology and spread in mouse models and human neurons.** *Acta neuropathologica*
  Bieri, G., Brahic, M., Bousset, L., Couthouis, J., Kramer, N. J., Ma, R., Nakayama, L., Monbureau, M., Defensor, E., Schule, B., Shamloo, M., Melki, R., Gitler, et al
  2019

- **Identification and functional analysis of novel mutations in the SOD1 gene in Chinese patients with amyotrophic lateral sclerosis.** *Amyotrophic lateral sclerosis & frontotemporal degeneration*
  Lin, H., Tao, Q., Wei, Q., Chen, C., Chen, Y., Li, H., Gitler, A. D., Wu, Z.
  2019: 1–7

- **Identification and functional analysis of novel mutations in the SOD1 gene in Chinese patients with amyotrophic lateral sclerosis** *AMYOTROPHIC LATERAL SCLEROSIS AND FRONTOTEMPORAL DEGENERATION*
  Lin, H., Tao, Q., Wei, Q., Chen, C., Chen, Y., Li, H., Gitler, A. D., Wu, Z.
  2019; 20 (3-4): 222–28

- **Axons Gonna Ride 'til They Can't No More.** *Neuron*
  Boeynaems, S. n., Gitler, A. D.
  2019; 104 (2): 179–81

- **In search of lost trafficking.** *Brain : a journal of neurology*
  Bechek, S. C., Gitler, A. D.
  2018; 141 (12): 3282–85

- **Disease protein muscles out of the nucleus** *NATURE*
  Becker, L. A., Gitler, A. D.
  2018; 563 (7732): 477–78

- **Pour Some Sugar on TDP(-43).** *Molecular cell*
  Boeynaems, S., Gitler, A. D.
  2018; 71 (5): 649–51
- **A matter of balance.** *eLife*
  Gitler, A. D., Fryer, J. D.
  2018; 7
- **These violent repeats have violent extends.** *Neurology. Genetics*
  Couthouis, J., Gitler, A. D.
  2018; 4 (4): e247
- **Poly(GR) impairs protein translation and stress granule dynamics in C9orf72-associated frontotemporal dementia and amyotrophic lateral sclerosis** *NATURE MEDICINE*
  Zhang, Y., Gendron, T. F., Ebbert, M. W., O'Raw, A. D., Yue, M., Jansen-West, K., Zhang, X., Prudencio, M., Chew, J., Cook, C. N., Daughrity, L. M., Tong, J., Song, et al
  2018; 24 (8): 1136-+
- **Targeted next-generation sequencing improves diagnosis of hereditary spastic paraplegia in Chinese patients** *JOURNAL OF MOLECULAR MEDICINE-JMM*
  Lu, C., Li, L., Dong, H., Wei, Q., Liu, Z., Ni, W., Gitler, A. D., Wu, Z.
  2018; 96 (7): 701–12
- **Hunting the G-unit in Huntington's.** *Brain : a journal of neurology*
  Huang, B., Gitler, A. D.
  2018; 141 (6): 1586–89
- **Ataxin-2 Is Droppin' Some Knowledge** *NEURON*
  Becker, L. A., Gitler, A. D.
  2018; 98 (4): 673–75
- **Phosphorylation Leads the Way for Protein Aggregate Disassembly** *DEVELOPMENTAL CELL*
  Boeynaems, S., Gitler, A. D.
  2018; 45 (3): 279–81
- **Stress Granule Assembly Disrupts Nucleocytoplasmic Transport** *CELL*
  Zhang, K., Daigle, J., Cunningham, K. M., Coyne, A. N., Ruan, K., Grima, J. C., Bowen, K. E., Wadhwa, H., Yang, P., Rigo, F., Taylor, J., Gitler, A. D., Rothstein, et al
  2018; 173 (4): 958-+
- **Nuclear-Import Receptors Reverse Aberrant Phase Transitions of RNA-Binding Proteins with Prion-like Domains** *CELL*
  Guo, L., Kim, H., Wang, H., Monaghan, J., Freyermuth, F., Sung, J. C., O'Donovan, K., Fare, C. M., Diaz, Z., Singh, N., Zhang, Z., Coughlin, M., Sweeny, et al
  2018; 173 (3): 677-+
- **Genome-wide Analyses Identify KIF5A as a Novel ALS Gene.** *Neuron*
  Nicolas, A. n., Kenna, K. P., Renton, A. E., Ticozzi, N. n., Faghri, F. n., Chia, R. n., Dominov, J. A., Kenna, B. J., Nalls, M. A., Keagle, P. n., Rivera, A. M., van Rheenen, W. n., Murphy, et al
  2018; 97 (6): 1268–83.e6
- **The epidemiology and genetics of Amyotrophic lateral sclerosis in China.** *Brain research*
  Liu, X. n., He, J. n., Gao, F. B., Gitler, A. D., Fan, D. n.
  2018
- **Yeast screen for modifiers of C9orf72 poly(Glycine-Arginine) dipeptide repeat toxicity.** *FEMS yeast research*
  Chai, N. n., Gitler, A. D.
  2018
- **Efficient Prevention of Neurodegenerative Diseases by Depletion of Starvation Response Factor Ataxin-2.** *Trends in neurosciences*
  Auburger, G., Sen, N. E., Meierhofer, D., Ba#ak, A. N., Gitler, A. D.
  2017; 40 (8): 507-516

- **ERAD defects and the HFE-H63D variant are associated with increased risk of liver damages in Alpha 1-Antitrypsin Deficiency.** *PloS one*
  Joly, P., Vignaud, H., Di Martino, J., Ruiz, M., Garin, R., Restier, L., Belmalih, A., Marchal, C., Cullin, C., Arveiler, B., Fergelot, P., Gitler, A. D., Lachaux, et al
  2017; 12 (6): e0179369

- **Unlocking the Mystery of ALS.** *Scientific American*
  Petrucelli, L., Gitler, A. D.
  2017; 316 (6): 46-51

- **Neurodegenerative disease: models, mechanisms, and a new hope** *DISEASE MODELS & MECHANISMS*
  Gitler, A. D., Dhillon, P., Shorter, J.
  2017; 10 (5): 499-502

- **Old moms say, no Sir.** *Science (New York, N.Y.)*
  Gitler, A. D., Jarosz, D. F.
  2017; 355 (6330): 1126-1127

- **Internalization, axonal transport and release of fibrillar forms of alpha-synuclein.** *Neurobiology of disease*
  Bieri, G., Gitler, A. D., Brahic, M.
  2017

- **ATXN2 trinucleotide repeat length correlates with risk of ALS** *NEUROBIOLOGY OF AGING*
  Sproviero, W., Shatunov, A., Stahl, D., Shoai, M., van Rheenen, W., Jones, A. R., Al-Sarraj, S., Andersen, P. M., Bonini, N. M., Conforti, F. L., Van Damme, P., Daoud, H., Amador, et al
  2017; 51

- **Raise the Roof: Boosting the Efficacy of a Spinal Muscular Atrophy Therapy.** *Neuron*
  Kramer, N. J., Gitler, A. D.
  2017; 93 (1): 3-5

- **Distinct repertoires of microRNAs present in mouse astrocytes compared to astrocyte-secreted exosomes.** *PloS one*
  Jovicic, A., Gitler, A. D.
  2017; 12 (2)

- **Glycolytic Enzymes Coalesce in G Bodies under Hypoxic Stress.** *Cell reports*
  Jin, M. n., Fuller, G. G., Han, T. n., Yao, Y. n., Alessi, A. F., Freeberg, M. A., Roach, N. P., Moresco, J. J., Karnovsky, A. n., Baba, M. n., Yates, J. R., Gitler, A. D., Inoki, et al
  2017; 20 (4): 895–908

- **Susan Lee Lindquist (1949-2016).** *Nature*
  Shorter, J., Gitler, A. D.
  2016; 540 (7631): 40

- **Defects in trafficking bridge Parkinson's disease pathology and genetics** *NATURE*
  Abeliovich, A., Gitler, A. D.
  2016; 539 (7628): 207-216

- **Semisynthetic and in Vitro Phosphorylation of Alpha-Synuclein at Y39 Promotes Functional Partly Helical Membrane-Bound States Resembling Those Induced by PD Mutations.** *ACS chemical biology*
  Dikiy, I., Fauvet, B., Jovicic, A., Mahul-Mellier, A., Desobry, C., El-Turk, F., Gitler, A. D., Lashuel, H. A., Eliezer, D.
  2016; 11 (9): 2428-2437

- **There has been an awakening: Emerging mechanisms of C9orf72 mutations in FTD/ALS** *BRAIN RESEARCH*
  Gitler, A. D., Tsuiji, H.
  2016; 1647: 19-29

- **Nuclear transport dysfunction: a common theme in amyotrophic lateral sclerosis and frontotemporal dementia.** *Journal of neurochemistry*
  Jovicic, A., Paul, J. W., Gitler, A. D.
  2016; 138: 134-144

- **Activation of HIPK2 Promotes ER Stress-Mediated Neurodegeneration in Amyotrophic Lateral Sclerosis.** *Neuron*

Lee, S., Shang, Y., Redmond, S. A., Urisman, A., Tang, A. A., Li, K. H., Burlingame, A. L., Pak, R. A., Jovicic, A., Gitler, A. D., Wang, J., Gray, N. S., Seeley, et al
2016; 91 (1): 41-55

- **Axonal transport and secretion of fibrillar forms of a-synuclein, Aß42 peptide and HTTExon 1.** *Acta neuropathologica*
  Brahic, M., Bousset, L., Bieri, G., Melki, R., Gitler, A. D.
  2016; 131 (4): 539-548

- **Analysis of COPII Vesicles Indicates a Role for the Emp47-Ssp120 Complex in Transport of Cell Surface Glycoproteins.** *Traffic*
  Margulis, N. G., Wilson, J. D., Bentivoglio, C. M., Dhungel, N., Gitler, A. D., Barlowe, C.
  2016; 17 (3): 191-210

- **Drosophila screen connects nuclear transport genes to DPR pathology in c9ALS/FTD** *SCIENTIFIC REPORTS*
  Boeynaems, S., Bogaertl, E., Michiels, E., Gijselinck, I., Sieben, A., Jovicic, A., De Baets, G., Scheveneels, W., Steyaert, J., Cuijt, I., Verstrepen, K. J., Callaerts, P., Rousseau, et al
  2016; 6

- **Drosophila screen connects nuclear transport genes to DPR pathology in c9ALS/FTD.** *Scientific reports*
  Boeynaems, S., Bogaert, E., Michiels, E., Gijselinck, I., Sieben, A., Jovicic, A., De Baets, G., Scheveneels, W., Steyaert, J., Cuijt, I., Verstrepen, K. J., Callaerts, P., Rousseau, et al
  2016; 6: 20877-?

- **Regrowing axons with alternative splicing.** *eLife*
  Kramer, N. J., Gitler, A. D.
  2016; 5

- **CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia.** *Nature communications*
  Williams, K. L., Topp, S., Yang, S., Smith, B., Fifita, J. A., Warraich, S. T., Zhang, K. Y., Farrawell, N., Vance, C., Hu, X., Chesi, A., Leblond, C. S., Lee, et al
  2016; 7: 11253-?

- **Fragile X protein mitigates TDP-43 toxicity by remodeling RNA granules and restoring translation** *HUMAN MOLECULAR GENETICS*
  Coyne, A. N., Yamada, S. B., Siddegowda, B. B., Estes, P. S., Zaepfel, B. L., Johannesmeyer, J. S., Lockwood, D. B., Pham, L. T., Hart, M. P., Cassel, J. A., Freibaum, B., Boehringer, A. V., Taylor, et al
  2015; 24 (24): 6886-6898

- **Fragile X protein mitigates TDP-43 toxicity by remodeling RNA granules and restoring translation.** *Human molecular genetics*
  Coyne, A. N., Yamada, S. B., Siddegowda, B. B., Estes, P. S., Zaepfel, B. L., Johannesmeyer, J. S., Lockwood, D. B., Pham, L. T., Hart, M. P., Cassel, J. A., Freibaum, B., Boehringer, A. V., Taylor, et al
  2015; 24 (24): 6886-98

- **Neurodegeneration: A Leg Up on TDP-43.** *Current biology : CB*
  Figley, M. D., Gitler, A. D.
  2015; 25 (16): R728-31

- **Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways** *SCIENCE*
  Cirulli, E. T., Lasseigne, B. N., Petrovski, S., Sapp, P. C., Dion, P. A., Leblond, C. S., Couthouis, J., Lu, Y., Wang, Q., Krueger, B. J., Ren, Z., Keebler, J., Han, et al
  2015; 347 (6229): 1436-1441

- **Parkinson's Disease Genes VPS35 and EIF4G1 Interact Genetically and Converge on a-Synuclein.** *Neuron*
  Dhungel, N., Eleuteri, S., Li, L., Kramer, N. J., Chartron, J. W., Spencer, B., Kosberg, K., Fields, J. A., Stafa, K., Adame, A., Lashuel, H., Frydman, J., Shen, et al
  2015; 85 (1): 76-87

- **It's all starting to come together.** *eLife*
  Becker, L. A., Gitler, A. D.
  2015; 4

- **Targeted Exon Capture and Sequencing in Sporadic Amyotrophic Lateral Sclerosis** *PLOS GENETICS*
  Couthouis, J., Raphael, A. R., Daneshjou, R., Gitler, A. D.
  2014; 10 (10)

- **Targeted exon capture and sequencing in sporadic amyotrophic lateral sclerosis.** *PLoS genetics*
  Couthouis, J., Raphael, A. R., Daneshjou, R., Gitler, A. D.
  2014; 10 (10)

- **Cell Biology. Clogging information flow in ALS.** *Science (New York, N.Y.)*
  Paul, J. W., Gitler, A. D.
  2014; 345 (6201): 1118-9

- **The novel Parkinson's disease linked mutation G51D attenuates in vitro aggregation and membrane binding of a-synuclein, and enhances its secretion and nuclear localization in cells.** *Human molecular genetics*
  Fares, M., Ait-Bouziad, N., Dikiy, I., Mbefo, M. K., Jovicic, A., Kiely, A., Holton, J. L., Lee, S., Gitler, A. D., Eliezer, D., Lashuel, H. A.
  2014; 23 (17): 4491-4509

- **Congenital muscular dystrophy and generalized epilepsy caused by GMPPB mutations.** *Brain research*
  Raphael, A. R., Couthouis, J., Sakamuri, S., Siskind, C., Vogel, H., Day, J. W., Gitler, A. D.
  2014; 1575: 66-71

- **A Cellular System that Degrades Misfolded Proteins and Protects against Neurodegeneration** *MOLECULAR CELL*
  Guo, L., Giasson, B. I., Glavis-Bloom, A., Brewer, M. D., Shorter, J., Gitler, A. D., Yang, X.
  2014; 55 (1): 15-30

- **Profilin 1 Associates with Stress Granules and ALS-Linked Mutations Alter Stress Granule Dynamics** *JOURNAL OF NEUROSCIENCE*
  Figley, M. D., Bieri, G., Kolaitis, R., Taylor, J. P., Gitler, A. D.
  2014; 34 (24): 8083-8097

- **Profilin 1 associates with stress granules and ALS-linked mutations alter stress granule dynamics.** *The Journal of neuroscience : the official journal of the Society for Neuroscience*
  Figley, M. D., Bieri, G., Kolaitis, R. M., Taylor, J. P., Gitler, A. D.
  2014; 34 (24): 8083-97

- **A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories.** *Journal of medical genetics*
  Akimoto, C., Volk, A. E., van Blitterswijk, M., Van den Broeck, M., Leblond, C. S., Lumbroso, S., Camu, W., Neitzel, B., Onodera, O., van Rheenen, W., Pinto, S., Weber, M., Smith, et al
  2014; 51 (6): 419-424

- **Exome sequencing identifies a DNAJB6 mutation in a family with dominantly-inherited limb-girdle muscular dystrophy.** *Neuromuscular disorders*
  Couthouis, J., Raphael, A. R., Siskind, C., Findlay, A. R., Buenrostro, J. D., Greenleaf, W. J., Vogel, H., Day, J. W., Flanigan, K. M., Gitler, A. D.
  2014; 24 (5): 431-435

- **Evaluating noncoding nucleotide repeat expansions in amyotrophic lateral sclerosis.** *Neurobiology of aging*
  Figley, M. D., Thomas, A., Gitler, A. D.
  2014; 35 (4): 936 e1-4

- **Evaluating noncoding nucleotide repeat expansions in amyotrophic lateral sclerosis** *NEUROBIOLOGY OF AGING*
  Figley, M. D., Thomas, A., Gitler, A. D.
  2014; 35 (4)

- **TDP-43 in ALS: stay on target…almost there.** *Neuron*
  Jovicic, A., Gitler, A. D.
  2014; 81 (3): 463-465

- **Therapeutic modulation of eIF2 alpha phosphorylation rescues TDP-43 toxicity in amyotrophic lateral sclerosis disease models** *NATURE GENETICS*
  Kim, H., Raphael, A. R., LaDow, E. S., McGurk, L., Weber, R. A., Trojanowski, J. Q., Lee, V. M., Finkbeiner, S., Gitler, A. D., Bonini, N. M.
  2014; 46 (2): 152-?

- **Exome sequencing to identify de novo mutations in sporadic ALS trios.** *Nature neuroscience*
  Chesi, A., Staahl, B. T., Jovicic, A., Couthouis, J., Fasolino, M., Raphael, A. R., Yamazaki, T., Elias, L., Polak, M., Kelly, C., Williams, K. L., Fifita, J. A., Maragakis, et al
  2013; 16 (7): 851-855

- **Kinetic Analysis of npBAF to nBAF Switching Reveals Exchange of SS18 with CREST and Integration with Neural Developmental Pathways.** *journal of neuroscience*
  Staahl, B. T., Tang, J., Wu, W., Sun, A., Gitler, A. D., Yoo, A. S., Crabtree, G. R.
  2013; 33 (25): 10348-10361

- **Exome sequencing to identify de novo mutations in sporadic ALS trios.** *Nature neuroscience*
  Chesi, A., Staahl, B. T., Jovicic, A., Couthouis, J., Fasolino, M., Raphael, A. R., Yamazaki, T., Elias, L., Polak, M., Kelly, C., Williams, K. L., Fifita, J. A., Maragakis, et al
  2013; 16 (7): 851-855

- **Stress granules as crucibles of ALS pathogenesis.** *journal of cell biology*
  Li, Y. R., King, O. D., Shorter, J., Gitler, A. D.
  2013; 201 (3): 361-372

- **Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS** *NATURE*
  Kim, H. J., Kim, N. C., Wang, Y., Scarborough, E. A., Moore, J., Diaz, Z., MacLea, K. S., Freibaum, B., Li, S., Molliex, A., Kanagaraj, A. P., Carter, R., Boylan, et al
  2013; 495 (7442): 467-?

- **Parallel PARKing: Parkinson's genes function in common pathway.** *Neuron*
  Chuang, R. S., Gitler, A. D.
  2013; 77 (3): 377-379

- **Yeast genetic screen reveals novel therapeutic strategy for ALS** *Rare Diseases*
  Figley, M., Gitler, AD
  2013; 1 (1): e24420

- **Yeast genetic screen reveals novel therapeutic strategy for ALS.** *Rare diseases (Austin, Tex.)*
  Figley, M. D., Gitler, A. D.
  2013; 1

- **A Template for New Drugs Against Alzheimer's Disease** *Cell*
  Aguzzi, A., Gitler, A. D.
  2013; 154 (6): 1182–1184

- **Inhibition of RNA lariat debranching enzyme suppresses TDP-43 toxicity in ALS disease models** *NATURE GENETICS*
  Armakola, M., Higgins, M. J., Figley, M. D., Barmada, S. J., Scarborough, E. A., Diaz, Z., Fang, X., Shorter, J., Krogan, N. J., Finkbeiner, S., Farese, R. V., Gitler, A. D.
  2012; 44 (12): 1302-1309

- **TDP-43 and FUS/TLS yield a target-rich haul in ALS** *NATURE NEUROSCIENCE*
  Gitler, A. D.
  2012; 15 (11): 1467-1469

- **Compartmentalization of superoxide dismutase 1 (SOD1G93A) aggregates determines their toxicity** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Weisberg, S. J., Lyakhovetsky, R., Werdiger, A., Gitler, A. D., Soen, Y., Kaganovich, D.
  2012; 109 (39): 15811-15816

- **Modeling Human Disease** *SCIENCE*
  Gitler, A. D., Lehmann, R.
  2012; 337 (6092): 269-269

- **ALS-Associated Ataxin 2 PolyQ Expansions Enhance Stress-Induced Caspase 3 Activation and Increase TDP-43 Pathological Modifications** *JOURNAL OF NEUROSCIENCE*
  Hart, M. P., Gitler, A. D.
  2012; 32 (27): 9133-9142

- **Evaluating the role of the FUS/TLS-related gene EWSR1 in amyotrophic lateral sclerosis** *HUMAN MOLECULAR GENETICS*
  Couthouis, J., Hart, M. P., Erion, R., King, O. D., Diaz, Z., Nakaya, T., Ibrahim, F., Kim, H., Mojsilovic-Petrovic, J., Panossian, S., Kim, C. E., Frackelton, E. C., Solski, et al

2012; 21 (13): 2899-2911

- **The tip of the iceberg: RNA-binding proteins with prion-like domains in neurodegenerative disease** *BRAIN RESEARCH*
  King, O. D., Gitler, A. D., Shorter, J.
  2012; 1462: 61-80

- **The Role of the Parkinson's Disease Gene PARK9 in Essential Cellular Pathways and the Manganese Homeostasis Network in Yeast** *PLOS ONE*
  Chesi, A., Kilaru, A., Fang, X., Cooper, A. A., Gitler, A. D.
  2012; 7 (3)

- **The modulation of Amyotrophic Lateral Sclerosis risk by Ataxin-2 intermediate polyglutamine expansions is a specific effect** *NEUROBIOLOGY OF DISEASE*
  Gispert, S., Kurz, A., Waibel, S., Bauer, P., Liepelt, I., Geisen, C., Gitler, A. D., Becker, T., Weber, M., Berg, D., Andersen, P. M., Krueger, R., Riess, et al
  2012; 45 (1): 356-361

- **Distinct TDP-43 pathology in ALS patients with ataxin 2 intermediate-length polyQ expansions** *Acta Neuropathol*
  Hart MP, Brettschneider J, Lee VM, Trojanowski JQ, Gitler AD
  2012; 124 (2): 221-230

- **A yeast functional screen predicts new candidate ALS disease genes** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Couthouis, J., Hart, M. P., Shorter, J., DeJesus-Hernandez, M., Erion, R., Oristano, R., Liu, A. X., Ramos, D., Jethava, N., Hosangadi, D., Epstein, J., Chiang, A., Diaz, et al
  2011; 108 (52): 20881-20890

- **Local RNA Translation at the Synapse and in Disease** *JOURNAL OF NEUROSCIENCE*
  Liu-Yesucevitz, L., Bassell, G. J., Gitler, A. D., Hart, A. C., Klann, E., Richter, J. D., Warren, S. T., Wolozin, B.
  2011; 31 (45): 16086-16093

- **Neuroscience. Another reason to exercise.** *Science*
  Gitler, A. D.
  2011; 334 (6056): 606-607

- **Model Organisms Reveal Insight into Human Neurodegenerative Disease: Ataxin-2 Intermediate-Length Polyglutamine Expansions Are a Risk Factor for ALS** *JOURNAL OF MOLECULAR NEUROSCIENCE*
  Bonini, N. M., Gitler, A. D.
  2011; 45 (3): 676-683

- **RNA-binding proteins with prion-like domains in ALS and FTLD-U** *PRION*
  Gitler, A. D., Shorter, J.
  2011; 5 (3): 179-187

- **High-throughput Yeast Plasmid Overexpression Screen** *JOVE-JOURNAL OF VISUALIZED EXPERIMENTS*
  Fleming, M. S., Gitler, A. D.
  2011

- **A yeast model for polyalanine-expansion aggregation and toxicity** *MOLECULAR BIOLOGY OF THE CELL*
  Konopka, C. A., Locke, M. N., Gallagher, P. S., Ngan Pham, N., Hart, M. P., Walker, C. J., Gitler, A. D., Gardner, R. G.
  2011; 22 (12): 1971-1984

- **Evaluating the prevalence of polyglutamine repeat expansions in amyotrophic lateral sclerosis** *NEUROLOGY*
  Lee, T., Li, Y. R., Chesi, A., Hart, M. P., Ramos, D., Jethava, N., Hosangadi, D., Epstein, J., Hodges, B., Bonini, N. M., Gitler, A. D.
  2011; 76 (24): 2062-2065

- **Ataxin-2 intermediate-length polyglutamine expansions in European ALS patients** *HUMAN MOLECULAR GENETICS*
  Lee, T., Li, Y. R., Ingre, C., Weber, M., Grehl, T., Gredal, O., De Carvalho, M., Meyer, T., Tysnes, O., Auburger, G., Gispert, S., Bonini, N. M., Andersen, et al
  2011; 20 (9): 1697-1700

- **Molecular Determinants and Genetic Modifiers of Aggregation and Toxicity for the ALS Disease Protein FUS/TLS** *PLOS BIOLOGY*
  Sun, Z., Diaz, Z., Fang, X., Hart, M. P., Chesi, A., Shorter, J., Gitler, A. D.
  2011; 9 (4)

- **PolyQ Repeat Expansions in ATXN2 Associated with ALS Are CAA Interrupted Repeats** *PLOS ONE*
  Yu, Z., Zhu, Y., Chen-Plotkin, A. S., Clay-Falcone, D., McCluskey, L., Elman, L., Kalb, R. G., Trojanowski, J. Q., Lee, V. M., Van Deerlin, V. M., Gitler, A. D., Bonini, N. M.
  2011; 6 (3)

- **TDP-43 toxicity in yeast** *METHODS*
  Armakola, M., Hart, M. P., Gitler, A. D.
  2011; 53 (3): 238-245

- **High-throughput yeast plasmid overexpression screen.** *Journal of visualized experiments : JoVE*
  Fleming, M. S., Gitler, A. D.
  2011

- **Prion-like disorders: blurring the divide between transmissibility and infectivity** *JOURNAL OF CELL SCIENCE*
  Cushman, M., Johnson, B. S., King, O. D., Gitler, A. D., Shorter, J.
  2010; 123 (8): 1191-1201

- **GTPase Activity Plays a Key Role in the Pathobiology of LRRK2** *PLOS GENETICS*
  Xiong, Y., Coombes, C. E., Kilaru, A., Li, X., Gitler, A. D., Bowers, W. J., Dawson, V. L., Dawson, T. M., Moore, D. J.
  2010; 6 (4)

- **TDP-43 Is Intrinsically Aggregation-prone, and Amyotrophic Lateral Sclerosis-linked Mutations Accelerate Aggregation and Increase Toxicity** *JOURNAL OF BIOLOGICAL CHEMISTRY*
  Johnson, B. S., Snead, D., Lee, J. J., McCaffery, J. M., Shorter, J., Gitler, A. D.
  2009; 284 (30): 20329-20339

- **Bridging high-throughput genetic and transcriptional data reveals cellular responses to alpha-synuclein toxicity** *NATURE GENETICS*
  Yeger-Lotem, E., Riva, L., Su, L. J., Gitler, A. D., Cashikar, A. G., King, O. D., Auluck, P. K., Geddie, M. L., Valastyan, J. S., Karger, D. R., Lindquist, S., Fraenkel, E.
  2009; 41 (3): 316-323

- **alpha-Synuclein is part of a diverse and highly conserved interaction network that includes PARK9 and manganese toxicity** *NATURE GENETICS*
  Gitler, A. D., Chesi, A., Geddie, M. L., Strathearn, K. E., Hamamichi, S., Hill, K. J., Caldwell, K. A., Caldwell, G. A., Cooper, A. A., Rochet, J., Lindquist, S.
  2009; 41 (3): 308-315

- **Disease models and mechanisms in the classroom** *DISEASE MODELS & MECHANISMS*
  Gitler, A. D.
  2009; 2 (3-4): 103-106

- **Evidence That alpha-Synuclein Does Not Inhibit Phospholipase D** *BIOCHEMISTRY*
  Rappley, I., Gitler, A. D., Selvy, P. E., LaVoie, M. J., Levy, B. D., Brown, H. A., Lindquist, S., Selkoe, D. J.
  2009; 48 (5): 1077-1083

- **Discovery and characterization of three novel synuclein genes in zebrafish** *DEVELOPMENTAL DYNAMICS*
  Sun, Z., Gitler, A. D.
  2008; 237 (9): 2490-2495

- **A yeast TDP-43 proteinopathy model: Exploring the molecular determinants of TDR-43 aggregation and cellular toxicity** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Johnson, B. S., McCaffery, J. M., Lindquist, S., Gitler, A. D.
  2008; 105 (17): 6439-6444

- **The Parkinson's disease protein alpha-synuclein disrupts cellular Rab homeostasis** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Gitler, A. D., Bevis, B. J., Shorter, J., Strathearn, K. E., Hamamichi, S., Su, L. J., Caldwell, K. A., Caldwell, G. A., Rochet, J., McCaffery, J. M., Barlowe, C., Lindquist, S.
  2008; 105 (1): 145-150

- **Beer and bread to brains and beyond: Can yeast cells teach us about neurodegenerative disease?** *NEUROSIGNALS*
  Gitler, A. D.

2008; 16 (1): 52-62

- **A suite of Gateway (R) cloning vectors for high-throughput genetic analysis in Saccharomyces cerevisiae** *YEAST*
  Alberti, S., Gitler, A. D., Lindquist, S.
  2007; 24 (10): 913-919
- **Prime time for alpha-synuclein** *JOURNAL OF NEUROSCIENCE*
  Gitler, A. D., Shorter, J.
  2007; 27 (10): 2433-2434
- **Kermit 2/XGIPC, an IGF1 receptor interacting protein, is required for IGF signaling in Xenopus eye development** *DEVELOPMENT*
  Wu, J., O'Donnell, M., Gitler, A. D., Klein, P. S.
  2006; 133 (18): 3651-3660
- **alpha-synuclein blocks ER-Golgi traffic and Rab1 rescues neuron loss in Parkinson's models** *SCIENCE*
  Cooper, A. A., Gitler, A. D., Cashikar, A., Haynes, C. M., Hill, K. J., Bhullar, B., Liu, K., Xu, K., Strathearn, K. E., Liu, F., Cao, S., Caldwell, K. A., Caldwell, et al
  2006; 313 (5785): 324-328
- **Insertion of Cre into the Pax3 locus creates a new allele of Splotch and identifies unexpected Pax3 derivatives** *DEVELOPMENTAL BIOLOGY*
  Engleka, K. A., Gitler, A. D., Zhang, M. Z., Zhou, D. D., High, F. A., Epstein, J. A.
  2005; 280 (2): 396-406
- **Yeast cells as a discovery platform for neurodegenerative disease** *9th Annual International Conference on Research in Computational Molecular Biology (RECOMB 2005)*
  Lindquist, S., Fraenkel, E., Outeiro, T., Gitler, A., Su, J., Cashikar, A., Jagadish, S.
  SPRINGER-VERLAG BERLIN.2005: 102–102
- **Semaphorin-plexin signaling guides patterning of the developing vasculature** *DEVELOPMENTAL CELL*
  Torres-Vazquez, J., Gitler, A. D., Fraser, S. D., Berk, J. D., Pham, V. N., Fishman, M. C., Childs, S., Epstein, J. A., Weinstein, B. M.
  2004; 7 (1): 117-123
- **PlexinD1 and semaphorin signaling are required in endothelial cells for cadiovascular development** *DEVELOPMENTAL CELL*
  Gitler, A. D., Lu, M. M., Epstein, J. A.
  2004; 7 (1): 107-116
- **Tie2-cre-induced inactivation of a conditional mutant Nf1 allele in mouse results in a myeloproliferative disorder that models juvenile myelomonocytic leukemia** *PEDIATRIC RESEARCH*
  Gitler, A. D., Kong, Y., Choi, J. K., Zhu, Y., Pear, W. S., Epstein, J. A.
  2004; 55 (4): 581-584
- **Molecular markers of cardiac endocardial cushion development** *DEVELOPMENTAL DYNAMICS*
  Gitler, A. D., Lu, M. M., Jiang, Y. Q., Epstein, J. A., Gruber, P. J.
  2003; 228 (4): 643-650
- **Cloning and characterization of zebrafish tbx1** *GENE EXPRESSION PATTERNS*
  Kochilas, L. K., Potluri, V., Gitler, A., Balasubramanian, K., Chin, A. J.
  2003; 3 (5): 645-651
- **Cardiac hypertrophy and histone deacetylase-dependent transcriptional repression mediated by the atypical homeodomain protein Hop** *JOURNAL OF CLINICAL INVESTIGATION*
  Kook, H., Lepore, J. J., Gitler, A. D., Lu, M. M., Yung, W. W., Mackay, J., Zhou, R., Ferrari, V., Gruber, P., Epstein, J. A.
  2003; 112 (6): 863-871
- **Regulating heart development: the role of Nf1.** *Cell cycle*
  Gitler, A. D., Epstein, J. A.
  2003; 2 (2): 96-98
- **Nf1 has an essential role in endothelial cells** *NATURE GENETICS*
  Gitler, A. D., Zhu, Y., Ismat, F. A., Lu, M. M., Yamauchi, Y., Parada, L. F., Epstein, J. A.
  2003; 33 (1): 75-79

- **Regulating Heart Development The Role of Nf1** *CELL CYCLE*
  Gitler, A. D., Epstein, J. A.
  2003; 2 (2): 96-98
- **Hop is an unusual homeobox gene that modulates cardiac development** *CELL*
  Chen, F., Kook, H., Milewski, R., Gitler, A. D., Lu, M. M., Li, J., Nazarian, R., Schnepp, R., Jen, K., Biben, C., Runke, G., MacKay, J. P., Novotny, et al
  2002; 110 (6): 713-723
- **Neural crest migration and mouse models of congenital heart disease** *Cold Spring Harbor Symposium on Quantitative Biology*
  Gitler, A. D., Brown, C. B., Kochilas, L., Li, J., Epstein, J. A.
  COLD SPRING HARBOR LAB PRESS, PUBLICATIONS DEPT.2002: 57–62
- **Cloning and expression analysis of murine lupin, a member of a novel gene family that is conserved through evolution and associated with Lupus inclusions** *DEVELOPMENT GENES AND EVOLUTION*
  Lu, M. M., Chen, F., Gitler, A., Li, J., Jin, F. Z., Ma, X. K., Epstein, J. A.
  2000; 210 (10): 512-517
- **Apakaochtodenes A and B: Two tetrahalogenated monoterpenes from the red marine alga Portieria hornemannii** *JOURNAL OF NATURAL PRODUCTS*
  Gunatilaka, A. A., Paul, V. J., Park, P. U., Puglisi, M. P., Gitler, A. D., Eggleston, D. S., Haltiwanger, R. C., Kingston, D. G.
  1999; 62 (10): 1376-1378