Stanford

Aaron D. Gitler

The Stanford Medicine Basic Science Professor

Genetics

Bio

ACADEMIC APPOINTMENTS

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

HONORS AND AWARDS

- 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 Site: http://gitlerlab.googlepages.com

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

2018-19

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

2017-18

- Molecular Mechanisms of Neurodegenerative Disease: BIO 267, GENE 267, NENS 267 (Win)
- Neurogenetics Core: NEPR 213 (Aut)

2016-17

- Neurogenetics Core: NEPR 213 (Aut)

2015-16

- Molecular Mechanisms of Neurodegenerative Disease: BIO 267, GENE 267, NENS 267 (Win)
- Neurogenetics Core: NEPR 213 (Win)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Mazen Asaad, Yanniv Dorone, Gerald Tiu

Postdoctoral Faculty Sponsor

Steven Boeynaems, Maya Maor Nof, Caitlin Rodriguez

Doctoral Dissertation Advisor (AC)

Nicholas Kramer, Eduardo Tassoni Tsuchida

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

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

Publications

PUBLICATIONS

- **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., Genuth, N. R., Grosely, R., Shi, Y., Glaria, I., Kramer, N. J., Nakayama, L., Fang, S., Dinger, T. J., Thoeng, A., 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., Couthouis, J., Li, A., Ousey, J., Ma, R., Bieri, G., Tsui, C. K., Shi, Y., 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

- **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

- **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., Kenna, K. P., Renton, A. E., Ticozzi, N., Faghri, F., Chia, R., Dominov, J. A., Kenna, B. J., Nalls, M. A., Keagle, P., Rivera, A. M., van Rheenen, W., Murphy, et al
  2018; 97 (6): 1268–83.e6
- **The epidemiology and genetics of Amyotrophic lateral sclerosis in China.** *Brain research*
  Liu, X., He, J., Gao, F. B., Gitler, A. D., Fan, D.
  2018
- **Yeast screen for modifiers of C9orf72 poly(Glycine-Arginine) dipeptide repeat toxicity.** *FEMS yeast research*
  Chai, N., Gitler, A. D.
  2018
- **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., Fuller, G. G., Han, T., Yao, Y., Alessi, A. F., Freeberg, M. A., Roach, N. P., Moresco, J. J., Karnovsky, A., Baba, M., Yates, J. R., Gitler, A. D., Inoki, et al
  2017; 20 (4): 895–908
- **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): 507–16

- **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

- **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-?

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

- **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

- **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

- **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

- **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)

- **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

- **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*
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