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

2023-24

- Neurogenetics Core: NEPR 213 (Aut)

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)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Cindy Lin, Kathryn Wu

Postdoctoral Faculty Sponsor

Tetsuya Akiyama, Jacob Blum, Caiwei Guo, Chang Liu, Jay Ross, 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

- **HDGFL2 cryptic proteins report presence of TDP-43 pathology in neurodegenerative diseases.** *Molecular neurodegeneration*

  Calliari, A., Daughrity, L. M., Albagli, E. A., Castellanos Otero, P., Yue, M., Jansen-West, K., Islam, N. N., Caulfield, T., Rawlinson, B., DeTure, M., Cook, C., Graff-Radford, N. R., Day, et al

  2024; 19 (1): 29

- **A molecular atlas of adult C. elegans motor neurons reveals ancient diversity delineated by conserved transcription factor codes.** *Cell reports*

  Smith, J. J., Taylor, S. R., Blum, J. A., Feng, W., Collings, R., Gitler, A. D., Miller, D. M., Kratsios, P.

  2024; 43 (3): 113857

- **TDP-43 nuclear loss in FTD/ALS causes widespread alternative polyadenylation changes.** *bioRxiv : the preprint server for biology*

  Zeng, Y., Lovchykova, A., Akiyama, T., Liu, C., Guo, C., Jawahar, V. M., Sianto, O., Calliari, A., Prudencio, M., Dickson, D. W., Petrucelli, L., Gitler, A. D.

  2024

- **It's me, hi, I solved the problem, it's TF-seqFISH.** *Cell research*

  Gautier, O., Gitler, A. D.

  2024

- **Challenges of profiling motor neuron transcriptomes from human spinal cord.** *Neuron*
  Gautier, O., Blum, J. A., Maksymetz, J., Chen, D., Schweingruber, C., Mei, I., Hermann, A., Hackos, D. H., Hedlund, E., Ravits, J., Gitler, A. D.
  2023; 111 (23): 3739-3741

- **FTLD targets brain regions expressing recently evolved genes.** *medRxiv : the preprint server for health sciences*
  Pasquini, L., Pereira, F. L., Seddighi, S., Zeng, Y., Wei, Y., Illán-Gala, I., Vatsavayai, S. C., Friedberg, A., Lee, A. J., Brown, J. A., Spina, S., Grinberg, L. T., Sirkis, et al
  2023

- **A molecular atlas of adult C. elegans motor neurons reveals ancient diversity delineated by conserved transcription factor codes.** *bioRxiv : the preprint server for biology*
  Smith, J. J., Taylor, S. R., Blum, J. A., Gitler, A. D., Miller, D. M., Kratsios, P.
  2023

- **A 3' UTR Deletion Is a Leading Candidate Causal Variant at the TMEM106B Locus Reducing Risk for FTLD-TDP.** *medRxiv : the preprint server for health sciences*
  Chemparathy, A., Le Guen, Y., Zeng, Y., Gorzynski, J., Jensen, T., Kasireddy, N., Talozzi, L., Belloy, M. E., Stewart, I., Gitler, A. D., Wagner, A. D., Mormino, E., Henderson, et al
  2023

- **Drugging "undruggable" neurodegenerative disease targets with small molecules.** *Science bulletin*
  Lu, J., Li, Z., Gitler, A. D., Lu, B.
  2023

- **Poly(A)-binding protein is an ataxin-2 chaperone that regulates biomolecular condensates.** *Molecular cell*
  Boeynaems, S., Dorone, Y., Zhuang, Y., Shabardina, V., Huang, G., Marian, A., Kim, G., Sanyal, A., Sen, N., Griffith, D., Docampo, R., Lasker, K., Ruiz-Trillo, et al
  2023

- **Aberrant phase separation is a common killing strategy of positively charged peptides in biology and human disease.** *bioRxiv : the preprint server for biology*
  Boeynaems, S., Ma, X. R., Yeong, V., Ginell, G. M., Chen, J. H., Blum, J. A., Nakayama, L., Sanyal, A., Briner, A., Haver, D. V., Pauwels, J., Ekman, A., Schmidt, et al
  2023

- **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

- **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

- **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

- **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

- **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

- **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

- **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. V., 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., 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
- **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
- **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
- **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*
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