Aaron D Gitler

List of Publications by Year in Descending Order

140 papers	15,773 citations	58 h-index	125 g-index
162 ext. papers	19,069 ext. citations	15.2 avg, IF	6.57 L-index

#	Paper	IF	Citations
140	TDP-43 represses cryptic exon inclusion in the FTD-ALS gene UNC13A.. *Nature*, **2022**,	50.4	14
139	Confirming Pathogenicity of the F386L Variant in a South Asian Family With Early-Onset Alzheimer Disease.. *Neurology: Genetics*, **2022**, 8, e647	3.8	
138	Cracking the cryptic code in amyotrophic lateral sclerosis and frontotemporal dementia: Towards therapeutic targets and biomarkers.. *Clinical and Translational Medicine*, **2022**, 12, e818	5.7	0
137	Why you always in a mood? PumpinRpolyP, actinRbrand new.. *Neuron*, **2022**, 110, 1603-1605	13.9	
136	Neurotoxic reactive astrocytes induce cell death via saturated lipids. *Nature*, **2021**, 599, 102-107	50.4	45
135	-derived arginine-containing dipeptide repeats associate with axonal transport machinery and impede microtubule-based motility. *Science Advances*, **2021**, 7,	14.3	17
134	Single-cell transcriptomic analysis of the adult mouse spinal cord reveals molecular diversity of autonomic and skeletal motor neurons. *Nature Neuroscience*, **2021**, 24, 572-583	25.5	22
133	p53 is a central regulator driving neurodegeneration caused by C9orf72 poly(PR). *Cell*, **2021**, 184, 689-708.e20	56.2	26
132	A prion-like protein regulator of seed germination undergoes hydration-dependent phase separation. *Cell*, **2021**, 184, 4284-4298.e27	56.2	17
131	An optimized ATAC-seq protocol for genome-wide mapping of active regulatory elements in primary mouse cortical neurons. *STAR Protocols*, **2021**, 2, 100854	1.4	0
130	BraInMap Elucidates the Macromolecular Connectivity Landscape of Mammalian Brain. *Cell Systems* , **2020**, 10, 333-350.e14	10.6	13
129	A memory of eS25 loss drives resistance phenotypes. *Nucleic Acids Research*, **2020**, 48, 7279-7297	20.1	2
128	Symmetric dimethylation of poly-GR correlates with disease duration in C9orf72 FTLD and ALS and reduces poly-GR phase separation and toxicity. *Acta Neuropathologica*, **2020**, 139, 407-410	14.3	20
127	A versatile system to record cell-cell interactions. *ELife*, **2020**, 9,	8.9	8
126	Genome-wide synthetic lethal CRISPR screen identifies FIS1 as a genetic interactor of ALS-linked C9ORF72. *Brain Research*, **2020**, 1728, 146601	3.7	11
125	ALS Genetics: Gains, Losses, and Implications for Future Therapies. *Neuron*, **2020**, 108, 822-842	13.9	72
124	Evolution of a Human-Specific Tandem Repeat Associated with ALS. *American Journal of Human Genetics*, **2020**, 107, 445-460	11	15

123	Knockout of reactive astrocyte activating factors slows disease progression in an ALS mouse model. *Nature Communications*, **2020**, 11, 3753	17.4	62
122	Just Took a DNA Test, Turns Out 100% Not That Phase. *Molecular Cell*, **2020**, 78, 193-194	17.6	5
121	Toxic expanded GGGGCC repeat transcription is mediated by the PAF1 complex in C9orf72-associated FTD. *Nature Neuroscience*, **2019**, 22, 863-874	25.5	38
120	Identification and functional analysis of novel mutations in the gene in Chinese patients with amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2019**, 20, 222-228	3.6	2
119	Loss of CREST leads to neuroinflammatory responses and ALS-like motor defects in mice. *Translational Neurodegeneration*, **2019**, 8, 13	10.3	3
118	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. *Human Genetics*, **2019**, 138, 593-600	6.3	11
117	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*, **2019**, 116, 7889-7898	11.5	186
116	LRRK2 modifies □syn pathology and spread in mouse models and human neurons. *Acta Neuropathologica*, **2019**, 137, 961-980	14.3	78
115	A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS. *Genome Research*, **2019**, 29, 809-818	9.7	14
114	Genetic Spectrum and Variability in Chinese Patients with Amyotrophic Lateral Sclerosis **2019**, 10, 1199-1206		21
113	RPS25 is required for efficient RAN translation of C9orf72 and other neurodegenerative disease-associated nucleotide repeats. *Nature Neuroscience*, **2019**, 22, 1383-1388	25.5	54
112	Axons Gonna Ride R̈il They CanR̈ No More. *Neuron*, **2019**, 104, 179-181	13.9	1
111	Yeast screen for modifiers of C9orf72 poly(glycine-arginine) dipeptide repeat toxicity. *FEMS Yeast Research*, **2018**, 18,	3.1	19
110	The epidemiology and genetics of Amyotrophic lateral sclerosis in China. *Brain Research*, **2018**, 1693, 121-126	3.7	26
109	CRISPR-Cas9 screens in human cells and primary neurons identify modifiers of C9ORF72 dipeptide-repeat-protein toxicity. *Nature Genetics*, **2018**, 50, 603-612	36.3	110
108	Nuclear-Import Receptors Reverse Aberrant Phase Transitions of RNA-Binding Proteins with Prion-like Domains. *Cell*, **2018**, 173, 677-692.e20	56.2	246
107	Stress Granule Assembly Disrupts Nucleocytoplasmic Transport. *Cell*, **2018**, 173, 958-971.e17	56.2	195
106	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296

105	Internalization, axonal transport and release of fibrillar forms of alpha-synuclein. *Neurobiology of Disease*, **2018**, 109, 219-225	7.5	54
104	Phosphorylation Leads the Way for Protein Aggregate Disassembly. *Developmental Cell*, **2018**, 45, 279-281	10.2	3
103	Targeted next-generation sequencing improves diagnosis of hereditary spastic paraplegia in Chinese patients. *Journal of Molecular Medicine*, **2018**, 96, 701-712	5.5	19
102	A matter of balance. *ELife*, **2018**, 7,	8.9	4
101	P1-135: RARE MISSENSE VARIANTS ON ZNF679 AND CTD-3214H19.16 SEGREGATE IN A FAMILY WITH A HISTORY OF SYNUCLEINOPATHY **2018**, 14, P324-P324		
100	In search of lost trafficking. *Brain*, **2018**, 141, 3282-3285	11.2	
99	Pour Some Sugar on TDP(-43). *Molecular Cell*, **2018**, 71, 649-651	17.6	5
98	Hunting the G-unit in Huntington's. *Brain*, **2018**, 141, 1586-1589	11.2	1
97	Ataxin-2 Is DroppinR Some Knowledge. *Neuron*, **2018**, 98, 673-675	13.9	5
96	Poly(GR) impairs protein translation and stress granule dynamics in C9orf72-associated frontotemporal dementia and amyotrophic lateral sclerosis. *Nature Medicine*, **2018**, 24, 1136-1142	50.5	149
95	ATXN2 trinucleotide repeat length correlates with risk of ALS. *Neurobiology of Aging*, **2017**, 51, 178.e1-178.e9	5.6	55
94	Therapeutic reduction of ataxin-2 extends lifespan and reduces pathology in TDP-43 mice. *Nature*, **2017**, 544, 367-371	50.4	278
93	Old moms say, no Sir. *Science*, **2017**, 355, 1126-1127	33.3	2
92	Raise the Roof: Boosting the Efficacy of a Spinal Muscular Atrophy Therapy. *Neuron*, **2017**, 93, 3-5	13.9	4
91	ERAD defects and the HFE-H63D variant are associated with increased risk of liver damages in Alpha 1-Antitrypsin Deficiency. *PLoS ONE*, **2017**, 12, e0179369	3.7	16
90	Glycolytic Enzymes Coalesce in G Bodies under Hypoxic Stress. *Cell Reports*, **2017**, 20, 895-908	10.6	77
89	[O1.03.06]: IDENTIFICATION OF AN ITGA7 VARIANT ASSOCIATED WITH ALZHEIMER'S DISEASE AND MULTIPLE OTHER NEURODEGENERATIVE DISEASES **2017**, 13, P193-P194		
88	Efficient Prevention of Neurodegenerative Diseases by Depletion of Starvation Response Factor Ataxin-2. *Trends in Neurosciences*, **2017**, 40, 507-516	13.3	38

87	[P2096]: NOVEL MISSENSE VARIANT ON EPHA1 IN A PROTECTED APOE4 FAMILY **2017**, 13, P643-P644		
86	[PL-05-01-01]: CELLULAR MECHANISMS OF FRONTOTEMPORAL LOBAR DEGENERATION (FTLD) **2017**, 13, P1451		
85	Distinct repertoires of microRNAs present in mouse astrocytes compared to astrocyte-secreted exosomes. *PLoS ONE*, **2017**, 12, e0171418	3.7	51
84	Defects in trafficking bridge Parkinson's disease pathology and genetics. *Nature*, **2016**, 539, 207-216	50.4	271
83	Drosophila screen connects nuclear transport genes to DPR pathology in c9ALS/FTD. *Scientific Reports*, **2016**, 6, 20877	4.9	179
82	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
81	Activation of HIPK2 Promotes ER Stress-Mediated Neurodegeneration in Amyotrophic Lateral Sclerosis. *Neuron*, **2016**, 91, 41-55	13.9	57
80	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*, **2016**, 11, 2428-37	4.9	39
79	Axonal transport and secretion of fibrillar forms of □synuclein, A□42 peptide and HTTExon 1. *Acta Neuropathologica*, **2016**, 131, 539-48	14.3	98
78	Analysis of COPII Vesicles Indicates a Role for the Emp47-Ssp120 Complex in Transport of Cell Surface Glycoproteins. *Traffic*, **2016**, 17, 191-210	5.7	9
77	Susan Lee Lindquist (1949-2016). *Nature*, **2016**, 540, 40	50.4	1
76	There has been an awakening: Emerging mechanisms of C9orf72 mutations in FTD/ALS. *Brain Research*, **2016**, 1647, 19-29	3.7	103
75	Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts. *Science*, **2016**, 353, 708-12	33.3	92
74	Nuclear transport dysfunction: a common theme in amyotrophic lateral sclerosis and frontotemporal dementia. *Journal of Neurochemistry*, **2016**, 138 Suppl 1, 134-44	6	35
73	Parkinson's disease genes VPS35 and EIF4G1 interact genetically and converge on □synuclein. *Neuron*, **2015**, 85, 76-87	13.9	122
72	Fragile X protein mitigates TDP-43 toxicity by remodeling RNA granules and restoring translation. *Human Molecular Genetics*, **2015**, 24, 6886-98	5.6	59
71	Neurodegeneration: A Leg Up on TDP-43. *Current Biology*, **2015**, 25, R728-31	6.3	1
70	Modifiers of C9orf72 dipeptide repeat toxicity connect nucleocytoplasmic transport defects to FTD/ALS. *Nature Neuroscience*, **2015**, 18, 1226-9	25.5	411

69	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. *Science*, **2015**, 347, 1436-41	33.3	642
68	It's all starting to come together. *ELife*, **2015**, 4,	8.9	4
67	The novel Parkinson's disease linked mutation G51D attenuates in vitro aggregation and membrane binding of α-synuclein, and enhances its secretion and nuclear localization in cells. *Human Molecular Genetics*, **2014**, 23, 4491-509	5.6	153
66	Therapeutic modulation of eIF2α phosphorylation rescues TDP-43 toxicity in amyotrophic lateral sclerosis disease models. *Nature Genetics*, **2014**, 46, 152-60	36.3	256
65	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*, **2014**, 51, 419-24	5.8	96
64	Cell Biology. Clogging information flow in ALS. *Science*, **2014**, 345, 1118-9	33.3	12
63	Profilin 1 associates with stress granules and ALS-linked mutations alter stress granule dynamics. *Journal of Neuroscience*, **2014**, 34, 8083-97	6.6	102
62	Congenital muscular dystrophy and generalized epilepsy caused by GMPPB mutations. *Brain Research*, **2014**, 1575, 66-71	3.7	25
61	Evaluating noncoding nucleotide repeat expansions in amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2014**, 35, 936.e1-4	5.6	13
60	A cellular system that degrades misfolded proteins and protects against neurodegeneration. *Molecular Cell*, **2014**, 55, 15-30	17.6	117
59	TDP-43 in ALS: stay on target—almost there. *Neuron*, **2014**, 81, 463-5	13.9	9
58	Targeted exon capture and sequencing in sporadic amyotrophic lateral sclerosis. *PLoS Genetics*, **2014**, 10, e1004704	6	39
57	Exome sequencing identifies a DNAJB6 mutation in a family with dominantly-inherited limb-girdle muscular dystrophy. *Neuromuscular Disorders*, **2014**, 24, 431-5	2.9	29
56	Parallel PARKing: Parkinson's genes function in common pathway. *Neuron*, **2013**, 77, 377-9	13.9	3
55	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. *Nature*, **2013**, 495, 467-73	50.4	965
54	Kinetic analysis of npBAF to nBAF switching reveals exchange of SS18 with CREST and integration with neural developmental pathways. *Journal of Neuroscience*, **2013**, 33, 10348-61	6.6	68
53	Stress granules as crucibles of ALS pathogenesis. *Journal of Cell Biology*, **2013**, 201, 361-72	7.3	599
52	Yeast genetic screen reveals novel therapeutic strategy for ALS. *Rare Diseases (Austin, Tex.)*, **2013**, 1, e24420		17

51	Exome sequencing to identify de novo mutations in sporadic ALS trios. *Nature Neuroscience*, **2013**, 16, 851-5	25.5	112
50	The tip of the iceberg: RNA-binding proteins with prion-like domains in neurodegenerative disease. *Brain Research*, **2012**, 1462, 61-80	3.7	450
49	The modulation of Amyotrophic Lateral Sclerosis risk by ataxin-2 intermediate polyglutamine expansions is a specific effect. *Neurobiology of Disease*, **2012**, 45, 356-61	7.5	61
48	Compartmentalization of superoxide dismutase 1 (SOD1G93A) aggregates determines their toxicity. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 15811-6	11.5	87
47	Inhibition of RNA lariat debranching enzyme suppresses TDP-43 toxicity in ALS disease models. *Nature Genetics*, **2012**, 44, 1302-9	36.3	170
46	ALS-associated ataxin 2 polyQ expansions enhance stress-induced caspase 3 activation and increase TDP-43 pathological modifications. *Journal of Neuroscience*, **2012**, 32, 9133-42	6.6	62
45	Distinct TDP-43 pathology in ALS patients with ataxin 2 intermediate-length polyQ expansions. *Acta Neuropathologica*, **2012**, 124, 221-30	14.3	32
44	Evaluating the role of the FUS/TLS-related gene EWSR1 in amyotrophic lateral sclerosis. *Human Molecular Genetics*, **2012**, 21, 2899-911	5.6	207
43	The role of the Parkinson's disease gene PARK9 in essential cellular pathways and the manganese homeostasis network in yeast. *PLoS ONE*, **2012**, 7, e34178	3.7	35
42	High-throughput yeast plasmid overexpression screen. *Journal of Visualized Experiments*, **2011**,	1.6	4
41	TDP-43 toxicity in yeast. *Methods*, **2011**, 53, 238-45	4.6	25
40	Local RNA translation at the synapse and in disease. *Journal of Neuroscience*, **2011**, 31, 16086-93	6.6	221
39	Neuroscience. Another reason to exercise. *Science*, **2011**, 334, 606-7	33.3	5
38	Model organisms reveal insight into human neurodegenerative disease: ataxin-2 intermediate-length polyglutamine expansions are a risk factor for ALS. *Journal of Molecular Neuroscience*, **2011**, 45, 676-83	3.3	34
37	A yeast model for polyalanine-expansion aggregation and toxicity. *Molecular Biology of the Cell*, **2011**, 22, 1971-84	3.5	9
36	Ataxin-2 intermediate-length polyglutamine expansions in European ALS patients. *Human Molecular Genetics*, **2011**, 20, 1697-700	5.6	112
35	RNA-binding proteins with prion-like domains in ALS and FTLD-U. *Prion*, **2011**, 5, 179-87	2.3	114
34	A yeast functional screen predicts new candidate ALS disease genes. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 20881-90	11.5	302

33	PolyQ repeat expansions in ATXN2 associated with ALS are CAA interrupted repeats. *PLoS ONE*, **2011**, 6, e17951	3.7	64
32	Molecular determinants and genetic modifiers of aggregation and toxicity for the ALS disease protein FUS/TLS. *PLoS Biology*, **2011**, 9, e1000614	9.7	321
31	Ataxin-2 intermediate-length polyglutamine expansions are associated with increased risk for ALS. *Nature*, **2010**, 466, 1069-75	50.4	844
30	GTPase activity plays a key role in the pathobiology of LRRK2. *PLoS Genetics*, **2010**, 6, e1000902	6	163
29	Prion-like disorders: blurring the divide between transmissibility and infectivity. *Journal of Cell Science*, **2010**, 123, 1191-201	5.3	231
28	TDP-43 is intrinsically aggregation-prone, and amyotrophic lateral sclerosis-linked mutations accelerate aggregation and increase toxicity. *Journal of Biological Chemistry*, **2009**, 284, 20329-39	5.4	512
27	Alpha-synuclein is part of a diverse and highly conserved interaction network that includes PARK9 and manganese toxicity. *Nature Genetics*, **2009**, 41, 308-15	36.3	451
26	Bridging high-throughput genetic and transcriptional data reveals cellular responses to alpha-synuclein toxicity. *Nature Genetics*, **2009**, 41, 316-23	36.3	242
25	Evidence that alpha-synuclein does not inhibit phospholipase D. *Biochemistry*, **2009**, 48, 1077-83	3.2	25
24	The Parkinson's disease protein alpha-synuclein disrupts cellular Rab homeostasis. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 145-50	11.5	415
23	Beer and bread to brains and beyond: can yeast cells teach us about neurodegenerative disease?. *NeuroSignals*, **2008**, 16, 52-62	1.9	37
22	A yeast TDP-43 proteinopathy model: Exploring the molecular determinants of TDP-43 aggregation and cellular toxicity. *Proceedings of the National Academy of Sciences of the United States of America* , **2008**, 105, 6439-44	11.5	317
21	Discovery and characterization of three novel synuclein genes in zebrafish. *Developmental Dynamics*, **2008**, 237, 2490-5	2.9	41
20	A suite of Gateway cloning vectors for high-throughput genetic analysis in Saccharomyces cerevisiae. *Yeast*, **2007**, 24, 913-9	3.4	320
19	Prime time for alpha-synuclein. *Journal of Neuroscience*, **2007**, 27, 2433-4	6.6	46
18	Kermit 2/XGIPC, an IGF1 receptor interacting protein, is required for IGF signaling in Xenopus eye development. *Development (Cambridge)*, **2006**, 133, 3651-60	6.6	25
17	Alpha-synuclein blocks ER-Golgi traffic and Rab1 rescues neuron loss in Parkinson's models. *Science*, **2006**, 313, 324-8	33.3	1084
16	Insertion of Cre into the Pax3 locus creates a new allele of Splotch and identifies unexpected Pax3 derivatives. *Developmental Biology*, **2005**, 280, 396-406	3.1	187

15	Tie2-Cre-induced inactivation of a conditional mutant Nf1 allele in mouse results in a myeloproliferative disorder that models juvenile myelomonocytic leukemia. *Pediatric Research*, **2004**, 55, 581-4	3.2	37
14	PlexinD1 and semaphorin signaling are required in endothelial cells for cardiovascular development. *Developmental Cell*, **2004**, 7, 107-16	10.2	301
13	Semaphorin-plexin signaling guides patterning of the developing vasculature. *Developmental Cell*, **2004**, 7, 117-23	10.2	309
12	Regulating Heart Development: The Role of Nf1. *Cell Cycle*, **2003**, 2, 95-97	4.7	8
11	Cloning and characterization of zebrafish tbx1. *Gene Expression Patterns*, **2003**, 3, 645-51	1.5	17
10	Molecular markers of cardiac endocardial cushion development. *Developmental Dynamics*, **2003**, 228, 643-50	2.9	94
9	Nf1 has an essential role in endothelial cells. *Nature Genetics*, **2003**, 33, 75-9	36.3	143
8	Regulating heart development: the role of Nf1. *Cell Cycle*, **2003**, 2, 96-8	4.7	4
7	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. *SSRN Electronic Journal*,	1	2
6	CRISPR-Cas9 screens in human cells and primary neurons identify modifiers of C9orf72 dipeptide repeat protein toxicity		1
5	Single-cell transcriptomic analysis of the adult mouse spinal cord		2
4	Hydration-dependent phase separation of a prion-like protein regulates seed germination during water stress		2
3	Regional Collapsing of Rare Variation Implicates Specific Genic Regions in ALS		2
2	C9orf72-derived arginine-containing dipeptide repeats associate with axonal transport machinery and impede microtubule-based motility		7
1	A modular platform for engineering function of natural and synthetic biomolecular condensates		6