

# Elaine T Lim

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8928956/publications.pdf>

Version: 2024-02-01

26  
papers

4,752  
citations

377584

21  
h-index

620720

26  
g-index

30  
all docs

30  
docs citations

30  
times ranked

11443  
citing authors

#	ARTICLE	IF	CITATIONS
1	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.	13.5	1,422
2	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. <i>Neuron</i> , 2013, 77, 259-273.	3.8	383
3	An enhanced CRISPR repressor for targeted mammalian gene regulation. <i>Nature Methods</i> , 2018, 15, 611-616.	9.0	361
4	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. <i>PLoS Genetics</i> , 2014, 10, e1004494.	1.5	351
5	Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. <i>PLoS Genetics</i> , 2013, 9, e1003671.	1.5	253
6	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. <i>Neuron</i> , 2013, 77, 235-242.	3.8	242
7	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. <i>Nature Neuroscience</i> , 2017, 20, 1217-1224.	7.1	212
8	Ataxia, Dementia, and Hypogonadotropism Caused by Disordered Ubiquitination. <i>New England Journal of Medicine</i> , 2013, 368, 1992-2003.	13.9	208
9	The Genetic Landscape of Diamond-Blackfan Anemia. <i>American Journal of Human Genetics</i> , 2018, 103, 930-947.	2.6	184
10	Whole-Exome Sequencing and Homozygosity Analysis Implicate Depolarization-Regulated Neuronal Genes in Autism. <i>PLoS Genetics</i> , 2012, 8, e1002635.	1.5	164
11	REST and Neural Gene Network Dysregulation in iPSC Models of Alzheimer's Disease. <i>Cell Reports</i> , 2019, 26, 1112-1127.e9.	2.9	150
12	Recessive gene disruptions in autism spectrum disorder. <i>Nature Genetics</i> , 2019, 51, 1092-1098.	9.4	109
13	Engineering adeno-associated viral vectors to evade innate immune and inflammatory responses. <i>Science Translational Medicine</i> , 2021, 13, .	5.8	99
14	A Homozygous Mutation in KCTD7 Links Neuronal Ceroid Lipofuscinosis to the Ubiquitin-Proteasome System. <i>American Journal of Human Genetics</i> , 2012, 91, 202-208.	2.6	97
15	Germline Mutations Affecting $Ca^{2+}$ in Hypoparathyroidism. <i>New England Journal of Medicine</i> , 2013, 368, 2532-2534.	13.9	85
16	The ESCRT-III Protein CHMP1A Mediates Secretion of Sonic Hedgehog on a Distinctive Subtype of Extracellular Vesicles. <i>Cell Reports</i> , 2018, 24, 973-986.e8.	2.9	79
17	The landscape of somatic mutation in cerebral cortex of autistic and neurotypical individuals revealed by ultra-deep whole-genome sequencing. <i>Nature Neuroscience</i> , 2021, 24, 176-185.	7.1	73
18	Genome-wide Analysis of Body Proportion Classifies Height-Associated Variants by Mechanism of Action and Implicates Genes Important for Skeletal Development. <i>American Journal of Human Genetics</i> , 2015, 96, 695-708.	2.6	67

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19	An Excess of Risk-Increasing Low-Frequency Variants Can Be a Signal of Polygenic Inheritance in Complex Diseases. <i>American Journal of Human Genetics</i> , 2014, 94, 437-452.	2.6	55
20	A pharmacogenetic study implicates <i>SLC9A9</i> in multiple sclerosis disease activity. <i>Annals of Neurology</i> , 2015, 78, 115-127.	2.8	39
21	A Novel Test for Recessive Contributions to Complex Diseases Implicates Bardet-Biedl Syndrome Gene BBS10 in Idiopathic Type 2 Diabetes and Obesity. <i>American Journal of Human Genetics</i> , 2014, 95, 509-520.	2.6	29
22	No evidence for rare recessive and compound heterozygous disruptive variants in schizophrenia. <i>European Journal of Human Genetics</i> , 2015, 23, 555-557.	1.4	21
23	Orgo-Seq integrates single-cell and bulk transcriptomic data to identify cell type specific-driver genes associated with autism spectrum disorder. <i>Nature Communications</i> , 2022, 13, .	5.8	11
24	Enabling multiplexed testing of pooled donor cells through whole-genome sequencing. <i>Genome Medicine</i> , 2018, 10, 31.	3.6	10
25	Whole exome sequencing uncovered highly penetrant recessive mutations for a spectrum of rare genetic pediatric diseases in Bangladesh. <i>Npj Genomic Medicine</i> , 2021, 6, 14.	1.7	8
26	An unbiased index to quantify participant's phenotypic contribution to an open-access cohort. <i>Scientific Reports</i> , 2017, 7, 46148.	1.6	6