Elaine T Lim

List of Publications by Year
in descending order

26 papers	4,752 citations	331670 21 h-index	552781 26 g-index
30 all docs	30 docs citations	30 times ranked	10401 citing authors

#	Article	IF	Citations
1	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
2	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. Neuron, 2013, 77, 259-273.	8.1	383
3	An enhanced CRISPR repressor for targeted mammalian gene regulation. Nature Methods, 2018, 15, 611-616.	19.0	361
4	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. PLoS Genetics, 2014, 10, e1004494.	3.5	351
5	Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. PLoS Genetics, 2013, 9, e1003671.	3.5	253
6	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	8.1	242
7	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. Nature Neuroscience, 2017, 20, 1217-1224.	14.8	212
8	Ataxia, Dementia, and Hypogonadotropism Caused by Disordered Ubiquitination. New England Journal of Medicine, 2013, 368, 1992-2003.	27.0	208
9	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	6.2	184
10	Whole-Exome Sequencing and Homozygosity Analysis Implicate Depolarization-Regulated Neuronal Genes in Autism. PLoS Genetics, 2012, 8, e1002635.	3.5	164
11	REST and Neural Gene Network Dysregulation in iPSC Models of Alzheimerâ€™s Disease. Cell Reports, 2019, 26, 1112-1127.e9.	6.4	150
12	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	21.4	109
13	Engineering adeno-associated viral vectors to evade innate immune and inflammatory responses. Science Translational Medicine, 2021, 13, .	12.4	99
14	A Homozygous Mutation in KCTD7 Links Neuronal Ceroid Lipofuscinosis to the Ubiquitin-Proteasome System. American Journal of Human Genetics, 2012, 91, 202-208.	6.2	97
15	Germline Mutations Affecting Gîą₁₁ in Hypoparathyroidism. New England Journal of Medicine, 2013, 368, 2532-2534.	27.0	85
16	The ESCRT-III Protein CHMP1A Mediates Secretion of Sonic Hedgehog on a Distinctive Subtype of Extracellular Vesicles. Cell Reports, 2018, 24, 973-986.e8.	6.4	79
17	The landscape of somatic mutation in cerebral cortex of autistic and neurotypical individuals revealed by ultra-deep whole-genome sequencing. Nature Neuroscience, 2021, 24, 176-185.	14.8	73
18	Genome-wide Analysis of Body Proportion Classifies Height-Associated Variants by Mechanism of Action and Implicates Genes Important for Skeletal Development. American Journal of Human Genetics, 2015, 96, 695-708.	6.2	67

#	Article	IF	Citations
19	An Excess of Risk-Increasing Low-Frequency Variants Can Be a Signal of Polygenic Inheritance in Complex Diseases. American Journal of Human Genetics, 2014, 94, 437-452.	6.2	55
20	A pharmacogenetic study implicates <scp>SLC9a9</scp> in multiple sclerosis disease activity. Annals of Neurology, 2015, 78, 115-127.	5.3	39
21	A Novel Test for Recessive Contributions to Complex Diseases Implicates Bardet-Biedl Syndrome Gene BBS10 in Idiopathic Type 2 Diabetes and Obesity. American Journal of Human Genetics, 2014, 95, 509-520.	6.2	29
22	No evidence for rare recessive and compound heterozygous disruptive variants in schizophrenia. European Journal of Human Genetics, 2015, 23, 555-557.	2.8	21
23	Orgo-Seq integrates single-cell and bulk transcriptomic data to identify cell type specific-driver genes associated with autism spectrum disorder. Nature Communications, 2022, 13, .	12.8	11
24	Enabling multiplexed testing of pooled donor cells through whole-genome sequencing. Genome Medicine, 2018, 10, 31.	8.2	10
25	Whole exome sequencing uncovered highly penetrant recessive mutations for a spectrum of rare genetic pediatric diseases in Bangladesh. Npj Genomic Medicine, 2021, 6, 14.	3.8	8
26	An unbiased index to quantify participantâ€™s phenotypic contribution to an open-access cohort. Scientific Reports, 2017, 7, 46148.	3.3	6