Elizabeth T Cirulli

List of Publications by Year
in descending order

64 papers	8,833 citations	81900 39 h-index	106344 65 g-index
78 all docs	78 docs citations	78 times ranked	19388 citing authors

#	Article	IF	Citations
1	Uncovering the roles of rare variants in common disease through whole-genome sequencing. Nature Reviews Genetics, 2010, 11, 415-425.	16.3	1,248
2	Genome-wide association meta-analysis in 269,867 individuals identifies new genetic and functional links to intelligence. Nature Genetics, 2018, 50, 912-919.	21.4	893
3	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
4	Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. Nature, 2012, 488, 499-503.	27.8	522
5	Study of 300,486 individuals identifies 148 independent genetic loci influencing general cognitive function. Nature Communications, 2018, 9, 2098.	12.8	484
6	Cold Urticaria, Immunodeficiency, and Autoimmunity Related toPLCG2Deletions. New England Journal of Medicine, 2012, 366, 330-338.	27.0	391
7	Common Genetic Variation and the Control of HIV-1 in Humans. PLoS Genetics, 2009, 5, e1000791.	3.5	377
8	Emergence and rapid transmission of SARS-CoV-2 B.1.1.7 in the United States. Cell, 2021, 184, 2587-2594.e7.	28.9	285
9	Profound Perturbation of the Metabolome in Obesity Is Associated with Health Risk. Cell Metabolism, 2019, 29, 488-500.e2.	16.2	235
10	GWAS meta-analysis reveals novel loci and genetic correlates for general cognitive function: a report from the COGENT consortium. Molecular Psychiatry, 2017, 22, 336-345.	7.9	194
11	Whole-Genome Sequencing of a Single Proband Together with Linkage Analysis Identifies a Mendelian Disease Gene. PLoS Genetics, 2010, 6, e1000991.	3.5	189
12	Association of Liver Injury From Specific Drugs, or Groups ofÂDrugs, With Polymorphisms in HLA and Other Genes in aÂGenome-Wide Association Study. Gastroenterology, 2017, 152, 1078-1089.	1.3	174
13	The Characterization of Twenty Sequenced Human Genomes. PLoS Genetics, 2010, 6, e1001111.	3.5	144
14	A genome-wide study of common SNPs and CNVs in cognitive performance in the CANTAB. Human Molecular Genetics, 2009, 18, 4650-4661.	2.9	131
15	Using ERDS to Infer Copy-Number Variants in High-Coverage Genomes. American Journal of Human Genetics, 2012, 91, 408-421.	6.2	127
16	A comparison of the Cambridge Automated Neuropsychological Test Battery (CANTAB) with â€œtraditionalâ€•neuropsychological testing instruments. Journal of Clinical and Experimental Neuropsychology, 2013, 35, 319-328.	1.3	117
17	Screening the human exome: a comparison of whole genome and whole transcriptome sequencing. Genome Biology, 2010, 11, R57.	9.6	115
18	Inosine Triphosphate Protects Against Ribavirin-Induced Adenosine Triphosphate Loss by Adenylosuccinate Synthase Function. Gastroenterology, 2011, 140, 1314-1321.	1.3	111

#	Article	IF	Citations
19	Large-Scale Cognitive GWAS Meta-Analysis Reveals Tissue-Specific Neural Expression and Potential Nootropic Drug Targets. Cell Reports, 2017, 21, 2597-2613.	6.4	103
20	Genome-wide rare variant analysis for thousands of phenotypes in over 70,000 exomes from two cohorts. Nature Communications, 2020, 11, 542.	12.8	101
21	Minocycline hepatotoxicity: Clinical characterization and identification of HLA-Bâˆ—35:02 as a risk factor. Journal of Hepatology, 2017, 67, 137-144.	3.7	100
22	A Missense Variant in PTPN22 is a Risk Factor for Drug-induced Liver Injury. Gastroenterology, 2019, 156, 1707-1716.e2.	1.3	97
23	Exome Sequencing Followed by Large-Scale Genotyping Fails to Identify Single Rare Variants of Large Effect in Idiopathic Generalized Epilepsy. American Journal of Human Genetics, 2012, 91, 293-302.	6.2	95
24	Common genetic variation and performance on standardized cognitive tests. European Journal of Human Genetics, 2010, 18, 815-820.	2.8	90
25	Pleiotropic Meta-Analysis of Cognition, Education, and Schizophrenia Differentiates Roles of Early Neurodevelopmental and Adult Synaptic Pathways. American Journal of Human Genetics, 2019, 105, 334-350.	6.2	86
26	Precision medicine integrating whole-genome sequencing, comprehensive metabolomics, and advanced imaging. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 3053-3062.	7.1	85
27	Exome Sequencing Followed by Large-Scale Genotyping Suggests a Limited Role for Moderately Rare Risk Factors of Strong Effect in Schizophrenia. American Journal of Human Genetics, 2012, 91, 303-312.	6.2	81
28	In vitro assays fail to predict in vivo effects of regulatory polymorphisms. Human Molecular Genetics, 2007, 16, 1931-1939.	2.9	78
29	Functional Genomic Analyses of Mendelian and Sporadic Disease Identify Impaired eIF2Î± Signaling as a Generalizable Mechanism for Dystonia. Neuron, 2016, 92, 1238-1251.	8.1	68
30	Failure to replicate effect of kibra on human memory in two large cohorts of European origin. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 667-668.	1.7	62
31	SVA: software for annotating and visualizing sequenced human genomes. Bioinformatics, 2011, 27, 1998-2000.	4.1	62
32	Novel mutation in VCP gene causes atypical amyotrophic lateral sclerosis. Neurology, 2012, 79, 2201-2208.	1.1	61
33	SARS-CoV-2 variant Delta rapidly displaced variant Alpha in the United States and led to higher viral loads. Cell Reports Medicine, 2022, 3, 100564.	6.5	61
34	A genome-wide genetic signature of Jewish ancestry perfectly separates individuals with and without full Jewish ancestry in a large random sample of European Americans. Genome Biology, 2009, 10, R7.	9.6	59
35	Position effect on FGF13 associated with X-linked congenital generalized hypertrichosis. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 7790-7795.	7.1	53
36	Fine-Scale Crossover Rate Heterogeneity in Drosophila pseudoobscura. Journal of Molecular Evolution, 2007, 64, 129-135.	1.8	50

#	Article	IF	Citations
37	Identification and Characterization of Fenofibrate-Induced Liver Injury. Digestive Diseases and Sciences, 2017, 62, 3596-3604.	2.3	49
38	COMT Val108/158 Met Genotype Affects Neural but not Cognitive Processing in Healthy Individuals. Cerebral Cortex, 2010, 20, 672-683.	2.9	48
39	Severe and protracted cholestasis in 44 young men taking bodybuilding supplements: assessment of genetic, clinical and chemical risk factors. Alimentary Pharmacology and Therapeutics, 2019, 49, 1195-1204.	3.7	43
40	Pathogenic variants in actionable MODY genes are associated with type 2 diabetes. Nature Metabolism, 2020, 2, 1126-1134.	11.9	43
41	Individual Variation in Contagious Yawning Susceptibility Is Highly Stable and Largely Unexplained by Empathy or Other Known Factors. PLoS ONE, 2014, 9, e91773.	2.5	41
42	Host Genetic Determinants of T Cell Responses to the MRKAd5 HIV-1 gag/pol/nef Vaccine in the Step Trial. Journal of Infectious Diseases, 2011, 203, 773-779.	4.0	40
43	The Fractionated Orthology of Bs2 and Rx/Gpa2 Supports Shared Synteny of Disease Resistance in the Solanaceae. Genetics, 2009, 182, 1351-1364.	2.9	38
44	Analysis of Genetic and Non-Genetic Factors Influencing Timing and Time Perception. PLoS ONE, 2015, 10, e0143873.	2.5	36
45	The role of HLA-A*33:01 in patients with cholestatic hepatitis attributed to terbinafine. Journal of Hepatology, 2018, 69, 1317-1325.	3.7	32
46	Acetaminophen (Paracetamol) Use Modifies the Sulfation of Sex Hormones. EBioMedicine, 2018, 28, 316-323.	6.1	28
47	An unsupervised learning approach to identify novel signatures of health and disease from multimodal data. Genome Medicine, 2020, 12, 7.	8.2	27
48	Systematic assessment of imputation performance using the 1000 Genomes reference panels. Briefings in Bioinformatics, 2015, 16, 549-562.	6.5	23
49	Sensitivity of whole exome sequencing in detecting infantile- and late-onset Pompe disease. Molecular Genetics and Metabolism, 2017, 122, 189-197.	1.1	21
50	A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS. Genome Research, 2019, 29, 809-818.	5.5	21
51	Antimicrobials and Antiepileptics Are the Leading Causes of Idiosyncratic Drugâ€induced Liver Injury in American Children. Journal of Pediatric Gastroenterology and Nutrition, 2019, 69, 152-159.	1.8	21
52	HLA-Aâˆ—03:01 is associated with increased risk of fever, chills, and stronger side effects from Pfizer-BioNTech COVID-19 vaccination. Human Genetics and Genomics Advances, 2022, 3, 100084.	1.7	21
53	Genetic and environmental correlates of topiramateâ€induced cognitive impairment. Epilepsia, 2012, 53, e5-8.	5.1	19
54	Individual Differences in Scotopic Visual Acuity and Contrast Sensitivity: Genetic and Non-Genetic Influences. PLoS ONE, 2016, 11, e0148192.	2.5	17

#	Article	IF	Citations
55	A whole-genome analysis of premature termination codons. Genomics, 2011, 98, 337-342.	2.9	14
56	Factors affecting pitch discrimination performance in a cohort of extensively phenotyped healthy volunteers. Scientific Reports, 2017, 7, 16480.	3.3	13
57	Positive predictive value highlights four novel candidates for actionable genetic screening from analysis of 220,000 clinicogenomic records. Genetics in Medicine, 2021, 23, 2300-2308.	2.4	13
58	The Increasing Importance of Gene-Based Analyses. PLoS Genetics, 2016, 12, e1005852.	3.5	13
59	Identifying nootropic drug targets via large-scale cognitive GWAS and transcriptomics. Neuropsychopharmacology, 2021, 46, 1788-1801.	5.4	12
60	Contribution of Pastimes and Testing Strategies to the Performance of Healthy Volunteers on Cognitive Tests. Clinical Neuropsychologist, 2011, 25, 778-798.	2.3	8
61	Localization and Characterization of X Chromosome Inversion Breakpoints Separating Drosophila mojavensis and Drosophila arizonae. Journal of Heredity, 2007, 98, 111-114.	2.4	7
62	Genome-Wide Identification of Rare and Common Variants Driving Triglyceride Levels in a Nevada Population. Frontiers in Genetics, 2021, 12, 639418.	2.3	7
63	Using symptom-based case predictions to identify host genetic factors that contribute to COVID-19 susceptibility. PLoS ONE, 2021, 16, e0255402.	2.5	6
64	Multi-Trait Analysis of GWAS and Biological Insights Into Cognition: A Response to Hill (2018). Twin Research and Human Genetics, 2018, 21, 394-397.	0.6	3