

# Emily G Spencer

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8449943/publications.pdf>

Version: 2024-02-01

22  
papers

2,133  
citations

516561

16  
h-index

642610

23  
g-index

25  
all docs

25  
docs citations

25  
times ranked

6181  
citing authors

#	ARTICLE	IF	CITATIONS
1	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. <i>Science</i> , 2014, 343, 506-511.	6.0	466
2	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. <i>Nature Genetics</i> , 2016, 48, 1071-1076.	9.4	314
3	Emergence and rapid transmission of SARS-CoV-2 B.1.1.7 in the United States. <i>Cell</i> , 2021, 184, 2587-2594.e7.	13.5	285
4	Genomic surveillance reveals multiple introductions of SARS-CoV-2 into Northern California. <i>Science</i> , 2020, 369, 582-587.	6.0	253
5	Clinical and Mutational Spectrum of Neurofibromatosis Type 1-like Syndrome. <i>JAMA - Journal of the American Medical Association</i> , 2009, 302, 2111.	3.8	160
6	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. <i>Neuron</i> , 2014, 84, 1226-1239.	3.8	95
7	AMPD2 Regulates GTP Synthesis and Is Mutated in a Potentially Treatable Neurodegenerative Brainstem Disorder. <i>Cell</i> , 2013, 154, 505-517.	13.5	94
8	Mutations in CSPP1 Lead to Classical Joubert Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 80-86.	2.6	75
9	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 1074-1078.	1.4	64
10	Re-analysis of whole-exome sequencing data uncovers novel diagnostic variants and improves molecular diagnostic yields for sudden death and idiopathic diseases. <i>Genome Medicine</i> , 2019, 11, 83.	3.6	54
11	Molecular Autopsy for Sudden Unexpected Death. <i>JAMA - Journal of the American Medical Association</i> , 2016, 316, 1492.	3.8	42
12	Emergence of an early SARS-CoV-2 epidemic in the United States. <i>Cell</i> , 2021, 184, 4939-4952.e15.	13.5	31
13	Mutations of the mitochondrial carrier translocase channel subunit TIM22 cause early-onset mitochondrial myopathy. <i>Human Molecular Genetics</i> , 2018, 27, 4135-4144.	1.4	30
14	Whole-Exome Sequencing Identifies Mutated C12orf57 in Recessive Corpus Callosum Hypoplasia. <i>American Journal of Human Genetics</i> , 2013, 92, 392-400.	2.6	28
15	Validation of a genetic risk score for atrial fibrillation: A prospective multicenter cohort study. <i>PLoS Medicine</i> , 2018, 15, e1002525.	3.9	24
16	A feasibility study of colorectal cancer diagnosis via circulating tumor DNA derived CNV detection. <i>PLoS ONE</i> , 2018, 13, e0196826.	1.1	22
17	Identification of <i>SPRED1</i> deletions using RT-PCR, multiplex ligation-dependent probe amplification and quantitative PCR. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1352-1359.	0.7	15
18	Combined accelerometer and genetic analysis to differentiate essential tremor from Parkinson's disease. <i>PeerJ</i> , 2018, 6, e5308.	0.9	14

#	ARTICLE	IF	CITATIONS
19	Novel <i>STAMBP</i> mutation and additional findings in an Arabic family. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 805-809.	0.7	11
20	Mutation of <i>WIF1</i> : a potential novel cause of a Nail-Patella-like disorder. <i>Genetics in Medicine</i> , 2017, 19, 1179-1183.	1.1	7
21	Returning Results to Family Members: Professional Duties in Genomics Research in the United States. <i>Journal of Legal Medicine</i> , 2018, 38, 201-219.	0.4	7
22	Direct to Consumer Fitness DNA Testing. <i>Clinical Chemistry</i> , 2019, 65, 45-47.	1.5	6