Avinash Abhyankar

List of Publications by Year
in descending order

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34	4,437	257450 24	414414 32
papers	citations	h-index	g-index

35	35	35	8198
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. Journal of Experimental Medicine, 2011, 208, 1635-1648.	8.5	739
2	Mycobacterial Disease and Impaired IFN-Î³ Immunity in Humans with Inherited ISG15 Deficiency. Science, 2012, 337, 1684-1688.	12.6	455
3	De novo gain-of-function KCNT1 channel mutations cause malignant migrating partial seizures of infancy. Nature Genetics, 2012, 44, 1255-1259.	21.4	436
4	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. Nature Immunology, 2012, 13, 1178-1186.	14.5	410
5	Impaired intrinsic immunity to HSV-1 in human iPSC-derived TLR3-deficient CNS cells. Nature, 2012, 491, 769-773.	27.8	288
6	Whole-exome sequencing-based discovery of STIM1 deficiency in a child with fatal classic Kaposi sarcoma. Journal of Experimental Medicine, 2010, 207, 2307-2312.	8.5	268
7	Heterozygous TBK1 mutations impair TLR3 immunity and underlie herpes simplex encephalitis of childhood. Journal of Experimental Medicine, 2012, 209, 1567-1582.	8.5	231
8	A Dominant Mutation in Human RAD51 Reveals Its Function in DNA Interstrand Crosslink Repair Independent of Homologous Recombination. Molecular Cell, 2015, 59, 478-490.	9.7	227
9	Ribosomal Protein SA Haploinsufficiency in Humans with Isolated Congenital Asplenia. Science, 2013, 340, 976-978.	12.6	176
10	Human RHOH deficiency causes T cell defects and susceptibility to EV-HPV infections. Journal of Clinical Investigation, 2012, 122, 3239-3247.	8.2	134
11	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. Journal of Experimental Medicine, 2013, 210, 1743-1759.	8.5	119
12	Exome Sequencing Identifies PDE4D Mutations as Another Cause of Acrodysostosis. American Journal of Human Genetics, 2012, 90, 740-745.	6.2	115
13	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	2.5	113
14	New and recurrent gain-of-functionSTAT1mutations in patients with chronic mucocutaneous candidiasis from Eastern and Central Europe. Journal of Medical Genetics, 2013, 50, 567-578.	3.2	105
15	The human gene connectome as a map of short cuts for morbid allele discovery. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 5558-5563.	7.1	79
16	Dominant-negative STAT1 SH2 domain mutations in unrelated patients with mendelian susceptibility to mycobacterial disease. Human Mutation, 2012, 33, 1377-1387.	2.5	71
17	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. Human Molecular Genetics, 2013, 22, 769-781.	2.9	58
18	Disease variants in genomes of 44 centenarians. Molecular Genetics & Genomic Medicine, 2014, 2, 438-450.	1.2	58

#	Article	IF	Citations
19	Whole-exome sequencing to analyze population structure, parental inbreeding, and familial linkage. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 6713-6718.	7.1	53
20	Variant Classification Concordance using the ACMG-AMP Variant Interpretation Guidelines across Nine Genomic Implementation Research Studies. American Journal of Human Genetics, 2020, 107, 932-941.	6.2	51
21	A Mild Form of SLC29A3 Disorder: A Frameshift Deletion Leads to the Paradoxical Translation of an Otherwise Noncoding mRNA Splice Variant. PLoS ONE, 2012, 7, e29708.	2.5	50
22	Distinct roles of BRCA2 in replication fork protection in response to hydroxyurea and DNA interstrand cross-links. Genes and Development, 2020, 34, 832-846.	5.9	48
23	A Novel Homozygous p.R1105X Mutation of the AP4E1 Gene in Twins with Hereditary Spastic Paraplegia and Mycobacterial Disease. PLoS ONE, 2013, 8, e58286.	2.5	31
24	Increased renal arterial resistance predicts the course of renal function in type 2 diabetes with microalbuminuria. Diabetes, 2006, 55, 234-9.	0.6	28
25	Altered Transcapillary Escape of Albumin and Microalbuminuria Reflects Two Different Pathogenetic Mechanisms. Diabetes, 2005, 54, 228-233.	0.6	23
26	Analytical Validation of Clinical Whole-Genome and Transcriptome Sequencing of Patient-Derived Tumors for Reporting Targetable Variants in Cancer. Journal of Molecular Diagnostics, 2018, 20, 822-835.	2.8	23
27	Clinical whole exome sequencing from dried blood spot identifies novel genetic defect underlying asparagine synthetase deficiency. Clinical Case Reports (discontinued), 2018, 6, 200-205.	0.5	15
28	Comparative Sequence Analysis of the Non-Protein-Coding Mitochondrial DNA of Inbred Rat Strains. PLoS ONE, 2009, 4, e8148.	2.5	11
29	Differential burden of rare protein truncating variants in Alzheimerâ€™s disease patients compared to centenarians. Human Molecular Genetics, 2016, 25, ddw150.	2.9	10
30	CDG: An Online Server for Detecting Biologically Closest Disease-Causing Genes and its Application to Primary Immunodeficiency. Frontiers in Immunology, 2018, 9, 1340.	4.8	6
31	Embryonic lethal genetic variants and chromosomally normal pregnancy loss. Fertility and Sterility, 2021, 116, 1351-1358.	1.0	5
32	OP035: Rapid Whole Genome Sequencing (rWGS) in the cardiac NICU. Genetics in Medicine, 2022, 24, S362-S363.	2.4	1
33	Detection of Copy Number Variants by Short Multiply Aggregated Sequence Homologies. Journal of Molecular Diagnostics, 2020, 22, 1476-1481.	2.8	0
34	eP067: Diagnostic yield of genome sequencing versus targeted gene panel testing in diverse pediatric patients in the NYCKidSeq study. Genetics in Medicine, 2022, 24, S45.	2.4	0