Avinash Abhyankar

List of Publications by Year
in descending order

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	OP035: Rapid Whole Genome Sequencing (rWGS) in the cardiac NICU. Genetics in Medicine, 2022, 24, S362-S363.	2.4	1
2	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
3	Embryonic lethal genetic variants and chromosomally normal pregnancy loss. Fertility and Sterility, 2021, 116, 1351-1358.	1.0	5
4	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
5	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
6	Detection of Copy Number Variants by Short Multiply Aggregated Sequence Homologies. Journal of Molecular Diagnostics, 2020, 22, 1476-1481.	2.8	0
7	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
8	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
9	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
10	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
11	Differential burden of rare protein truncating variants in Alzheimerâ€™s disease patients compared to centenarians. Human Molecular Genetics, 2016, 25, ddw150.	2.9	10
12	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
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	Disease variants in genomes of 44 centenarians. Molecular Genetics & Genomic Medicine, 2014, 2, 438-450.	1.2	58
15	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. Human Molecular Genetics, 2013, 22, 769-781.	2.9	58
16	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
17	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. Journal of Experimental Medicine, 2013, 210, 1743-1759.	8.5	119
18	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

#	Article	IF	Citations
19	Ribosomal Protein SA Haploinsufficiency in Humans with Isolated Congenital Asplenia. Science, 2013, 340, 976-978.	12.6	176
20	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
21	Heterozygous TBK1 mutations impair TLR3 immunity and underlie herpes simplex encephalitis of childhood. Journal of Experimental Medicine, 2012, 209, 1567-1582.	8.5	231
22	De novo gain-of-function KCNT1 channel mutations cause malignant migrating partial seizures of infancy. Nature Genetics, 2012, 44, 1255-1259.	21.4	436
23	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. Nature Immunology, 2012, 13, 1178-1186.	14.5	410
24	Impaired intrinsic immunity to HSV-1 in human iPSC-derived TLR3-deficient CNS cells. Nature, 2012, 491, 769-773.	27.8	288
25	Mycobacterial Disease and Impaired IFN-Î³ Immunity in Humans with Inherited ISG15 Deficiency. Science, 2012, 337, 1684-1688.	12.6	455
26	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
27	Dominant-negative STAT1 SH2 domain mutations in unrelated patients with mendelian susceptibility to mycobacterial disease. Human Mutation, 2012, 33, 1377-1387.	2.5	71
28	Exome Sequencing Identifies PDE4D Mutations as Another Cause of Acrodysostosis. American Journal of Human Genetics, 2012, 90, 740-745.	6.2	115
29	Human RHOH deficiency causes T cell defects and susceptibility to EV-HPV infections. Journal of Clinical Investigation, 2012, 122, 3239-3247.	8.2	134
30	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
31	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
32	Comparative Sequence Analysis of the Non-Protein-Coding Mitochondrial DNA of Inbred Rat Strains. PLoS ONE, 2009, 4, e8148.	2.5	11
33	Increased renal arterial resistance predicts the course of renal function in type 2 diabetes with microalbuminuria. Diabetes, 2006, 55, 234-9.	0.6	28
34	Altered Transcapillary Escape of Albumin and Microalbuminuria Reflects Two Different Pathogenetic Mechanisms. Diabetes, 2005, 54, 228-233.	0.6	23