Vamsi K Mootha

List of Publications by Year
in descending order

103	70,352	15504 65	29157 104
papers	citations	h-index	g-index

118	118	118	101520
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Gene set enrichment analysis: A knowledge-based approach for interpreting genome-wide expression profiles. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 15545-15550.	7.1	38,922
2	PGC-1Î±-responsive genes involved in oxidative phosphorylation are coordinately downregulated in human diabetes. Nature Genetics, 2003, 34, 267-273.	21.4	8,185
3	A Mitochondrial Protein Compendium Elucidates Complex I Disease Biology. Cell, 2008, 134, 112-123.	28.9	1,766
4	Integrative genomics identifies MCU as an essential component of the mitochondrial calcium uniporter. Nature, 2011, 476, 341-345.	27.8	1,596
5	MitoCarta2.0: an updated inventory of mammalian mitochondrial proteins. Nucleic Acids Research, 2016, 44, D1251-D1257.	14.5	1,170
6	Proteomic Mapping of Mitochondria in Living Cells via Spatially Restricted Enzymatic Tagging. Science, 2013, 339, 1328-1331.	12.6	1,023
7	Directed evolution of APEX2 for electron microscopy and proximity labeling. Nature Methods, 2015, 12, 51-54.	19.0	1,014
8	Integrated Analysis of Protein Composition, Tissue Diversity, and Gene Regulation in Mouse Mitochondria. Cell, 2003, 115, 629-640.	28.9	815
9	MitoCarta3.0: an updated mitochondrial proteome now with sub-organelle localization and pathway annotations. Nucleic Acids Research, 2021, 49, D1541-D1547.	14.5	760
10	MICU1 encodes a mitochondrial EF hand protein required for Ca2+ uptake. Nature, 2010, 467, 291-296.	27.8	747
11	Mitochondrial disorders as windows into an ancient organelle. Nature, 2012, 491, 374-383.	27.8	633
12	ErrÂ and Gabpa/b specify PGC-1Â-dependent oxidative phosphorylation gene expression that is altered in diabetic muscle. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 6570-6575.	7.1	627
13	Engineered ascorbate peroxidase as a genetically encoded reporter for electron microscopy. Nature Biotechnology, 2012, 30, 1143-1148.	17.5	584
14	How many human proteoforms are there?. Nature Chemical Biology, 2018, 14, 206-214.	8.0	580
15	EMRE Is an Essential Component of the Mitochondrial Calcium Uniporter Complex. Science, 2013, 342, 1379-1382.	12.6	537
16	The Mitochondrial Proteome and Human Disease. Annual Review of Genomics and Human Genetics, 2010, 11, 25-44.	6.2	497
17	Metabolic enzyme expression highlights a key role for MTHFD2 and the mitochondrial folate pathway in cancer. Nature Communications, 2014, 5, 3128.	12.8	438
18	Common Inherited Variation in Mitochondrial Genes Is Not Enriched for Associations with Type 2 Diabetes or Related Glycemic Traits. PLoS Genetics, 2010, 6, e1001058.	3.5	429

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19	Proteomic Mapping of the Human Mitochondrial Intermembrane Space in Live Cells via Ratiometric APEX Tagging. Molecular Cell, 2014, 55, 332-341.	9.7	414
20	A bacterial cytidine deaminase toxin enables CRISPR-free mitochondrial base editing. Nature, 2020, 583, 631-637.	27.8	409
21	Molecular Diagnosis of Infantile Mitochondrial Disease with Targeted Next-Generation Sequencing. Science Translational Medicine, 2012, 4, 118ra10.	12.4	406
22	MICU1 Controls Both the Threshold and Cooperative Activation of the Mitochondrial Ca2+ Uniporter. Cell Metabolism, 2013, 17, 976-987.	16.2	397
23	MICU2, a Paralog of MICU1, Resides within the Mitochondrial Uniporter Complex to Regulate Calcium Handling. PLoS ONE, 2013, 8, e55785.	2.5	387
24	Hypoxia as a therapy for mitochondrial disease. Science, 2016, 352, 54-61.	12.6	339
25	High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. Nature Genetics, 2010, 42, 851-858.	21.4	332
26	Mitochondrial dysfunction remodels one-carbon metabolism in human cells. ELife, 2016, 5, .	6.0	332
27	Complementation of mitochondrial electron transport chain by manipulation of the NAD ⁺ /NADH ratio. Science, 2016, 352, 231-235.	12.6	314
28	Nutrient-sensitized screening for drugs that shift energy metabolism from mitochondrial respiration to glycolysis. Nature Biotechnology, 2010, 28, 249-255.	17.5	290
29	The molecular era of the mitochondrial calcium uniporter. Nature Reviews Molecular Cell Biology, 2015, 16, 545-553.	37.0	280
30	Proteomic mapping of cytosol-facing outer mitochondrial and ER membranes in living human cells by proximity biotinylation. ELife, 2017, 6, .	6.0	276
31	Architecture of the mitochondrial calcium uniporter. Nature, 2016, 533, 269-273.	27.8	256
32	A Genome-wide CRISPR Death Screen Identifies Genes Essential for Oxidative Phosphorylation. Cell Metabolism, 2016, 24, 875-885.	16.2	244
33	Mitochondrial Reprogramming Underlies Resistance to BCL-2 Inhibition in Lymphoid Malignancies. Cancer Cell, 2019, 36, 369-384.e13.	16.8	224
34	<scp>MICU</scp> 1 and <scp>MICU</scp> 2 play nonredundant roles in the regulation of the mitochondrial calcium uniporter. EMBO Reports, 2014, 15, 299-307.	4.5	193
35	Widespread Chromosomal Losses and Mitochondrial DNA Alterations as Genetic Drivers in HÃ¼rthle Cell Carcinoma. Cancer Cell, 2018, 34, 242-255.e5.	16.8	185
36	Discovery of Genes Essential for Heme Biosynthesis through Large-Scale Gene Expression Analysis. Cell Metabolism, 2009, 10, 119-130.	16.2	178

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37	MCU encodes the pore conducting mitochondrial calcium currents. ELife, 2013, 2, e00704.	6.0	156
38	Targeted exome sequencing of suspected mitochondrial disorders. Neurology, 2013, 80, 1762-1770.	1.1	155
39	IGF2BP2/IMP2-Deficient Mice Resist Obesity through Enhanced Translation of Ucp1 mRNA and Other mRNAs Encoding Mitochondrial Proteins. Cell Metabolism, 2015, 21, 609-621.	16.2	148
40	Evolutionary Diversity of the Mitochondrial Calcium Uniporter. Science, 2012, 336, 886-886.	12.6	146
41	Reconstitution of the mitochondrial calcium uniporter in yeast. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 8985-8990.	7.1	136
42	Distinct mitochondrial defects trigger the integrated stress response depending on the metabolic state of the cell. ELife, 2020, 9, .	6.0	133
43	A plasma signature of human mitochondrial disease revealed through metabolic profiling of spent media from cultured muscle cells. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 1571-1575.	7.1	126
44	Genetic Screen for Cell Fitness in High or Low Oxygen Highlights Mitochondrial and Lipid Metabolism. Cell, 2020, 181, 716-727.e11.	28.9	126
45	Cryo-EM structure of a fungal mitochondrial calcium uniporter. Nature, 2018, 559, 570-574.	27.8	125
46	Epstein-Barr-Virus-Induced One-Carbon Metabolism Drives B Cell Transformation. Cell Metabolism, 2019, 30, 539-555.e11.	16.2	119
47	Hypoxia treatment reverses neurodegenerative disease in a mouse model of Leigh syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E4241-E4250.	7.1	117
48	A Metabolic Signature of Mitochondrial Dysfunction Revealed through a Monogenic Form of Leigh Syndrome. Cell Reports, 2015, 13, 981-989.	6.4	113
49	Highâ€affinity cooperative Ca²⁺ binding by <scp>MICU</scp>1â€“<scp>MICU</scp>2 serves as an onâ€“off switch for theÂuniporter. EMBO Reports, 2017, 18, 1397-1411.	4.5	111
50	A Compendium of Genetic Modifiers of Mitochondrial Dysfunction Reveals Intra-organelle Buffering. Cell, 2019, 179, 1222-1238.e17.	28.9	109
51	Hepatic NADH reductive stress underlies common variation in metabolic traits. Nature, 2020, 583, 122-126.	27.8	108
52	Expansion of Biological Pathways Based on Evolutionary Inference. Cell, 2014, 158, 213-225.	28.9	107
53	The Human Knockout Gene CLYBL Connects Itaconate to Vitamin B12. Cell, 2017, 171, 771-782.e11.	28.9	102
54	SARS-CoV-2 hijacks folate and one-carbon metabolism for viral replication. Nature Communications, 2021, 12, 1676.	12.8	102

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55	Circulating markers of NADH-reductive stress correlate with mitochondrial disease severity. Journal of Clinical Investigation, 2021, 131, .	8.2	95
56	Effects of sodium benzoate, a widely used food preservative, on glucose homeostasis and metabolic profiles in humans. Molecular Genetics and Metabolism, 2015, 114, 73-79.	1.1	93
57	Functional Genomic Analysis of Human Mitochondrial RNA Processing. Cell Reports, 2014, 7, 918-931.	6.4	86
58	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59.	1.9	86
59	An engineered enzyme that targets circulating lactate to alleviate intracellular NADH:NAD+ imbalance. Nature Biotechnology, 2020, 38, 309-313.	17.5	86
60	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. American Journal of Human Genetics, 2017, 101, 239-254.	6.2	83
61	Leigh Syndrome Mouse Model Can Be Rescued by Interventions that Normalize Brain Hyperoxia, but Not HIF Activation. Cell Metabolism, 2019, 30, 824-832.e3.	16.2	83
62	Spatiotemporal compartmentalization of hepatic NADH and NADPH metabolism. Journal of Biological Chemistry, 2018, 293, 7508-7516.	3.4	81
63	Oxygen and mammalian cell culture: are we repeating the experiment of Dr. Ox?. Nature Metabolism, 2019, 1, 858-860.	11.9	81
64	CRISPR-free base editors with enhanced activity and expanded targeting scope in mitochondrial and nuclear DNA. Nature Biotechnology, 2022, 40, 1378-1387.	17.5	81
65	Hypoxia Rescues Frataxin Loss by Restoring Iron Sulfur Cluster Biogenesis. Cell, 2019, 177, 1507-1521.e16.	28.9	80
66	A genetically encoded tool for manipulation of NADP+/NADPH in living cells. Nature Chemical Biology, 2017, 13, 1088-1095.	8.0	77
67	Natural underlying mt<scp>DNA</scp> heteroplasmy as a potential source of intraâ€person hi<scp>PSC</scp> variability. EMBO Journal, 2016, 35, 1979-1990.	7.8	71
68	Comparative Analysis of Mitochondrial N-Termini from Mouse, Human, and Yeast. Molecular and Cellular Proteomics, 2017, 16, 512-523.	3.8	71
69	Itaconyl-CoA forms a stable biradical in methylmalonyl-CoA mutase and derails its activity and repair. Science, 2019, 366, 589-593.	12.6	71
70	Early loss of mitochondrial complex I and rewiring of glutathione metabolism in renal oncocytoma. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E6283-E6290.	7.1	70
71	Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. Human Molecular Genetics, 2017, 26, 4257-4266.	2.9	63
72	An essential role for cardiolipin in the stability and function of the mitochondrial calcium uniporter. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 16383-16390.	7.1	63

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73	Purifying Selection against Pathogenic Mitochondrial DNA in Human T Cells. New England Journal of Medicine, 2020, 383, 1556-1563.	27.0	62
74	MICU1 imparts the mitochondrial uniporter with the ability to discriminate between Ca ²⁺ and Mn ²⁺. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E7960-E7969.	7.1	59
75	Mitochondrial DNA variation across 56,434 individuals in gnomAD. Genome Research, 2022, 32, 569-582.	5.5	59
76	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	6.2	58
77	Airway stem cells sense hypoxia and differentiate into protective solitary neuroendocrine cells. Science, 2021, 371, 52-57.	12.6	52
78	Cardiovascular homeostasis dependence on MICU2, a regulatory subunit of the mitochondrial calcium uniporter. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E9096-E9104.	7.1	48
79	A Middle Eastern Founder Mutation Expands the Genotypic and Phenotypic Spectrum of Mitochondrial MICU1 Deficiency: A Report of 13 Patients. JIMD Reports, 2018, 43, 79-83.	1.5	46
80	Crystal structure of MICU2 and comparison with MICU1 reveal insights into the uniporter gating mechanism. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 3546-3555.	7.1	39
81	Metabolic profiles of exercise in patients with McArdle disease or mitochondrial myopathy. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 8402-8407.	7.1	37
82	Structural insights into the Ca2+-dependent gating of the human mitochondrial calcium uniporter. ELife, 2020, 9, .	6.0	34
83	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
84	Loss of LUC7L2 and U1 snRNP subunits shifts energy metabolism from glycolysis to OXPHOS. Molecular Cell, 2021, 81, 1905-1919.e12.	9.7	33
85	Meclizine Preconditioning Protects the Kidney Against Ischemiaâ€“Reperfusion Injury. EBioMedicine, 2015, 2, 1090-1101.	6.1	32
86	Exploring the InÂVivo Role of the Mitochondrial Calcium Uniporter in Brown Fat Bioenergetics. Cell Reports, 2019, 27, 1364-1375.e5.	6.4	31
87	Combinatorial GxGxE CRISPR screen identifies SLC25A39 in mitochondrial glutathione transport linking iron homeostasis to OXPHOS. Nature Communications, 2022, 13, 2483.	12.8	31
88	CLIC, a tool for expanding biological pathways based on co-expression across thousands of datasets. PLoS Computational Biology, 2017, 13, e1005653.	3.2	30
89	CLYBL is a polymorphic human enzyme with malate synthase and Î²-methylmalate synthase activity. Human Molecular Genetics, 2014, 23, 2313-2323.	2.9	29
90	The uniporter: From newly identified parts to function. Biochemical and Biophysical Research Communications, 2014, 449, 370-372.	2.1	26

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91	Natural Product Screening Reveals Naphthoquinone Complex I Bypass Factors. PLoS ONE, 2016, 11, e0162686.	2.5	22
92	CYB561A3 is the key lysosomal iron reductase required for Burkitt B-cell growth and survival. Blood, 2021, 138, 2216-2230.	1.4	20
93	Human genetic analyses of organelles highlight the nucleus in age-related trait heritability. ELife, 2021, 10, .	6.0	20
94	Pheochromocytoma-Induced Cardiomyopathy. Circulation, 2000, 102, E11-3.	1.6	19
95	Hypoxia ameliorates brain hyperoxia and NAD+ deficiency in a murine model of Leigh syndrome. Molecular Genetics and Metabolism, 2021, 133, 83-93.	1.1	16
96	Biochemical Characterization of Pathogenic Mutations in Human Mitochondrial Methionyl-tRNA Formyltransferase. Journal of Biological Chemistry, 2014, 289, 32729-32741.	3.4	11
97	Oxygen in mitochondrial disease: can there be too much of a good thing?. Journal of Inherited Metabolic Disease, 2018, 41, 761-763.	3.6	11
98	A patient with homozygous nonsense variants in two Leigh syndrome disease genes: Distinguishing a dual diagnosis from a hypomorphic proteinâ€truncating variant. Human Mutation, 2019, 40, 893-898.	2.5	8
99	On the dynamic and even reversible nature of Leigh syndrome: Lessons from human imaging and mouse models. Current Opinion in Neurobiology, 2022, 72, 80-90.	4.2	8
100	Evolutionary divergence reveals the molecular basis of EMRE dependence of the human MCU. Life Science Alliance, 2020, 3, e202000718.	2.8	5
101	Editorial: Mitochondrial medicine special issue. Journal of Inherited Metabolic Disease, 2021, 44, 289-291.	3.6	3
102	Megaloblastic Anemia and Mitochondriopathy Caused by a Homozygous Mutation in Sideroflexin-4.. Blood, 2012, 120, 79-79.	1.4	0
103	Ironâ€dependent regulation of mitochondrial form and function. FASEB Journal, 2013, 27, lb65.	0.5	0