Nadav Ahituv

List of Publications by Year
in descending order

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118 papers	10,081 citations	61984 43 h-index	45317 90 g-index
148 all docs	148 docs citations	148 times ranked	14981 citing authors

#	Article	IF	Citations
1	In vivo enhancer analysis of human conserved non-coding sequences. Nature, 2006, 444, 499-502.	27.8	1,072
2	Massively parallel functional dissection of mammalian enhancers in vivo. Nature Biotechnology, 2012, 30, 265-270.	17.5	468
3	A distal enhancer and an ultraconserved exon are derived from a novel retroposon. Nature, 2006, 441, 87-90.	27.8	452
4	Chromatin connectivity maps reveal dynamic promoterâ€“enhancer long-range associations. Nature, 2013, 504, 306-310.	27.8	405
5	A Genome-wide Framework for Mapping Gene Regulation via Cellular Genetic Screens. Cell, 2019, 176, 377-390.e19.	28.9	379
6	Mutations in DMRT3 affect locomotion in horses and spinal circuit function in mice. Nature, 2012, 488, 642-646.	27.8	364
7	Mutation in Transcription Factor POU4F3 Associated with Inherited Progressive Hearing Loss in Humans. Science, 1998, 279, 1950-1954.	12.6	322
8	Variant Interpretation: Functional Assays to the Rescue. American Journal of Human Genetics, 2017, 101, 315-325.	6.2	275
9	Deletion of Ultraconserved Elements Yields Viable Mice. PLoS Biology, 2007, 5, e234.	5.6	255
10	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. Genome Research, 2017, 27, 38-52.	5.5	244
11	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	12.6	234
12	CRISPR-mediated activation of a promoter or enhancer rescues obesity caused by haploinsufficiency. Science, 2019, 363, .	12.6	230
13	Massively parallel decoding of mammalian regulatory sequences supports a flexible organizational model. Nature Genetics, 2013, 45, 1021-1028.	21.4	226
14	MYO6, the Human Homologue of the Gene Responsible for Deafness in Snellâ€™s Waltzer Mice, Is Mutated in Autosomal Dominant Nonsyndromic Hearing Loss. American Journal of Human Genetics, 2001, 69, 635-640.	6.2	212
15	Decoding enhancers using massively parallel reporter assays. Genomics, 2015, 106, 159-164.	2.9	208
16	The Notch ligand Jagged1 is required for inner ear sensory development. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 3873-3878.	7.1	206
17	Coding exons function as tissue-specific enhancers of nearby genes. Genome Research, 2012, 22, 1059-1068.	5.5	202
18	Medical Sequencing at the Extremes of Human Body Mass. American Journal of Human Genetics, 2007, 80, 779-791.	6.2	199

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19	Enhancer Interaction Networks as a Means for Singular Olfactory Receptor Expression. Cell, 2014, 159, 543-557.	28.9	173
20	Single-cell epigenomics reveals mechanisms of human cortical development. Nature, 2021, 598, 205-213.	27.8	154
21	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. Nature Communications, 2019, 10, 3583.	12.8	152
22	Integrating Diverse Datasets Improves Developmental Enhancer Prediction. PLoS Computational Biology, 2014, 10, e1003677.	3.2	149
23	Function and Regulation of AUTS2, a Gene Implicated in Autism and Human Evolution. PLoS Genetics, 2013, 9, e1003221.	3.5	145
24	Limb development: a paradigm of gene regulation. Nature Reviews Genetics, 2017, 18, 245-258.	16.3	131
25	A PAX1 enhancer locus is associated with susceptibility to idiopathic scoliosis in females. Nature Communications, 2015, 6, 6452.	12.8	122
26	The role of AUTS2 in neurodevelopment and human evolution. Trends in Genetics, 2013, 29, 600-608.	6.7	120
27	Gene Regulatory Elements, Major Drivers of Human Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 45-63.	6.2	115
28	Association of functionally significant Melanocortin-4 but not Melanocortin-3 receptor mutations with severe adult obesity in a large North American case-control study. Human Molecular Genetics, 2009, 18, 1140-1147.	2.9	112
29	A systematic evaluation of the design and context dependencies of massively parallel reporter assays. Nature Methods, 2020, 17, 1083-1091.	19.0	111
30	Whole-Genome Sequencing of Pharmacogenetic Drug Response in Racially Diverse Children with Asthma. American Journal of Respiratory and Critical Care Medicine, 2018, 197, 1552-1564.	5.6	102
31	Gain-of-Function R225W Mutation in Human AMPKÎ³3 Causing Increased Glycogen and Decreased Triglyceride in Skeletal Muscle. PLoS ONE, 2007, 2, e903.	2.5	80
32	Identification and Massively Parallel Characterization of Regulatory Elements Driving Neural Induction. Cell Stem Cell, 2019, 25, 713-727.e10.	11.1	76
33	Mapping cis-regulatory domains in the human genome using multi-species conservation of synteny. Human Molecular Genetics, 2005, 14, 3057-3063.	2.9	70
34	cisâ€regulatory mutations are a genetic cause of human limb malformations. Developmental Dynamics, 2011, 240, 920-930.	1.8	68
35	A genome-wide association study identifies four novel susceptibility loci underlying inguinal hernia. Nature Communications, 2015, 6, 10130.	12.8	68
36	Use antibiotics in cell culture with caution: genome-wide identification of antibiotic-induced changes in gene expression and regulation. Scientific Reports, 2017, 7, 7533.	3.3	67

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37	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. Nature Protocols, 2020, 15, 2387-2412.	12.0	65
38	Transcriptomic and epigenomic characterization of the developing bat wing. Nature Genetics, 2016, 48, 528-536.	21.4	64
39	Oestrogen engages brain MC4R signalling to drive physical activity in female mice. Nature, 2021, 599, 131-135.	27.8	59
40	MPRAnalyze: statistical framework for massively parallel reporter assays. Genome Biology, 2019, 20, 183.	8.8	58
41	Minor Loops in Major Folds: Enhancerâ€“Promoter Looping, Chromatin Restructuring, and Their Association with Transcriptional Regulation and Disease. PLoS Genetics, 2015, 11, e1005640.	3.5	57
42	Identification and characterization of novel polymorphisms in the basal promoter of the human transporter, MATE1. Pharmacogenetics and Genomics, 2009, 19, 770-780.	1.5	56
43	Human Developmental Enhancers Conserved between Deuterostomes and Protostomes. PLoS Genetics, 2012, 8, e1002852.	3.5	55
44	Functional characterization of tissue-specific enhancers in the DLX5/6 locus. Human Molecular Genetics, 2012, 21, 4930-4938.	2.9	54
45	COVID-19 mRNA Vaccination in Lactation: Assessment of Adverse Events and Vaccine Related Antibodies in Mother-Infant Dyads. Frontiers in Immunology, 2021, 12, 777103.	4.8	53
46	Genome-wide distribution of Auts2 binding localizes with active neurodevelopmental genes. Translational Psychiatry, 2014, 4, e431-e431.	4.8	51
47	Bat Accelerated Regions Identify a Bat Forelimb Specific Enhancer in the HoxD Locus. PLoS Genetics, 2016, 12, e1005738.	3.5	51
48	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. Human Mutation, 2019, 40, 1280-1291.	2.5	46
49	Humanâ€“chimpanzee fused cells reveal cis-regulatory divergence underlying skeletal evolution. Nature Genetics, 2021, 53, 467-476.	21.4	46
50	Exploiting human-fish genome comparisons for deciphering gene regulation. Human Molecular Genetics, 2004, 13, R261-R266.	2.9	43
51	Modulating gene regulation to treat genetic disorders. Nature Reviews Drug Discovery, 2020, 19, 757-775.	46.4	41
52	Genomic Characterization of Metformin Hepatic Response. PLoS Genetics, 2016, 12, e1006449.	3.5	41
53	Evaluation of Messenger RNA From COVID-19 BTN162b2 and mRNA-1273 Vaccines in Human Milk. JAMA Pediatrics, 2021, 175, 1069.	6.2	40
54	CRISPRâ€“Cas9-mediated functional dissection of 3â€²-UTRs. Nucleic Acids Research, 2017, 45, 10800-10810.	14.5	39

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55	Deletion of CTCF sites in the SHH locus alters enhancerâ€“promoter interactions and leads to acheiropodia. Nature Communications, 2021, 12, 2282.	12.8	37
56	Systematic Dissection of Coding Exons at Single Nucleotide Resolution Supports an Additional Role in Cell-Specific Transcriptional Regulation. PLoS Genetics, 2014, 10, e1004592.	3.5	36
57	Genome-Wide Discovery of Drug-Dependent Human Liver Regulatory Elements. PLoS Genetics, 2014, 10, e1004648.	3.5	36
58	The cis-regulatory effects of modern human-specific variants. ELife, 2021, 10, .	6.0	36
59	Genome-wide meta-analysis and replication studies in multiple ethnicities identify novel adolescent idiopathic scoliosis susceptibility loci. Human Molecular Genetics, 2018, 27, 3986-3998.	2.9	34
60	Sequence signatures extracted from proximal promoters can be used to predict distal enhancers. Genome Biology, 2013, 14, R117.	9.6	32
61	The Hydrodynamic Tail Vein Assay as a Tool for the Study of Liver Promoters and Enhancers. Methods in Molecular Biology, 2013, 1015, 279-289.	0.9	32
62	The cartilage matrisome in adolescent idiopathic scoliosis. Bone Research, 2020, 8, 13.	11.4	31
63	Mouse models for human deafness: current tools for new fashions. Trends in Molecular Medicine, 2002, 8, 447-451.	6.7	30
64	Identification and Characterization of Proximal Promoter Polymorphisms in the Human Concentrative Nucleoside Transporter 2 (SLC28A2). Journal of Pharmacology and Experimental Therapeutics, 2009, 328, 699-707.	2.5	30
65	A novel 13 base pair insertion in the sonic hedgehog ZRS limb enhancer (ZRS/LMBR1) causes preaxial polydactyly with triphalangeal thumb. Human Mutation, 2012, 33, 1063-1066.	2.5	29
66	A Novel ZRS Mutation Leads to Preaxial Polydactyly Type 2 in a Heterozygous Form and Werner Mesomelic Syndrome in a Homozygous Form. Human Mutation, 2014, 35, 945-948.	2.5	29
67	A PYY Q62P variant linked to human obesity. Human Molecular Genetics, 2006, 15, 387-391.	2.9	28
68	In vivo characterization of human APOA5 haplotypes. Genomics, 2007, 90, 674-679.	2.9	25
69	Functional Characterization of Liver Enhancers That Regulate Drug-Associated Transporters. Clinical Pharmacology and Therapeutics, 2011, 89, 571-578.	4.7	24
70	Dysregulation of STAT3 signaling is associated with endplate-oriented herniations of the intervertebral disc in Adgrg6 mutant mice. PLoS Genetics, 2019, 15, e1008096.	3.5	24
71	Genome-wide identification of signaling center enhancers in the developing limb. Development (Cambridge), 2014, 141, 4194-4198.	2.5	21
72	A point mutation in the pre-ZRS disrupts sonic hedgehog expression in the limb bud and results in triphalangeal thumbâ€“polysyndactyly syndrome. Genetics in Medicine, 2018, 20, 1405-1413.	2.4	21

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73	An ENU-induced mutation in AP-2Î± leads to middle earand ocular defects in Doarad mice. Mammalian Genome, 2004, 15, 424-432.	2.2	20
74	Genetic variation in the SIM1 locus is associated with erectile dysfunction. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 11018-11023.	7.1	20
75	Genetic Association Study of Adiposity and Melanocortin-4 Receptor (MC4R) Common Variants: Replication and Functional Characterization of Non-Coding Regions. PLoS ONE, 2014, 9, e96805.	2.5	20
76	Genomic structure of the human unconventional myosin VI gene. Gene, 2000, 261, 269-275.	2.2	19
77	A compact, in vivo screen of all 6-mers reveals drivers of tissue-specific expression and guides synthetic regulatory element design. Genome Biology, 2013, 14, R72.	9.6	19
78	Uncovering drug-responsive regulatory elements. Pharmacogenomics, 2015, 16, 1829-1841.	1.3	19
79	Cellular and transcriptional diversity over the course of human lactation. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2121720119.	7.1	19
80	Pharmacogene regulatory elements: from discovery to applications. Genome Medicine, 2012, 4, 45.	8.2	18
81	Functionally conserved enhancers with divergent sequences in distant vertebrates. BMC Genomics, 2015, 16, 882.	2.8	18
82	The human ARF tumor suppressor senses blastema activity and suppresses epimorphic tissue regeneration. ELife, 2015, 4, .	6.0	18
83	Identification of three novelFGF16mutations in Xâ€linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. Molecular Genetics & Genomic Medicine, 2014, 2, 402-411.	1.2	17
84	Mutations in the fourth Î²-propeller domain of LRP4 are associated with isolated syndactyly with fusion of the third and fourth fingers. Human Mutation, 2018, 39, 811-815.	2.5	17
85	Genomic and epigenomic mapping of leptin-responsive neuronal populations involved in body weight regulation. Nature Metabolism, 2019, 1, 475-484.	11.9	17
86	High-throughput characterization of the role of non-B DNA motifs on promoter function. Cell Genomics, 2022, 2, 100111.	6.5	17
87	Comparative genomic analysis reveals a distant liver enhancer upstream of the COUP-TFII gene. Mammalian Genome, 2005, 16, 91-95.	2.2	16
88	Exonic enhancers: proceed with caution in exome and genome sequencing studies. Genome Medicine, 2016, 8, 14.	8.2	16
89	A novel ZRS variant causes preaxial polydactyly type I by increased sonic hedgehog expression in the developing limb bud. Genetics in Medicine, 2020, 22, 189-198.	2.4	16
90	Co-option of the lineage-specific LAVA retrotransposon in the gibbon genome. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 19328-19338.	7.1	16

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91	Massively parallel reporter perturbation assays uncover temporal regulatory architecture during neural differentiation. Nature Communications, 2022, 13, 1504.	12.8	16
92	Metaâ€analysis of massively parallel reporter assays enables prediction of regulatory function across cell types. Human Mutation, 2019, 40, 1299-1313.	2.5	15
93	A multidisciplinary review of triphalangeal thumb. Journal of Hand Surgery: European Volume, 2019, 44, 59-68.	1.0	15
94	A new mouse mutant for the LDL receptor identified using ENU mutagenesis. Journal of Lipid Research, 2008, 49, 2452-2462.	4.2	13
95	Alternative approach to a heavy weight problem. Genome Research, 2008, 18, 214-220.	5.5	13
96	A novel ZRS mutation in a Balochi tribal family with triphalangeal thumb, preâ€axial polydactyly, postâ€axial polydactyly, and syndactyly. American Journal of Medical Genetics, Part A, 2012, 158A, 2031-2035.	1.2	13
97	Absent from DNA and protein: genomic characterization of nullomers and nullpeptides across functional categories and evolution. Genome Biology, 2021, 22, 245.	8.8	13
98	Functional characterization of SIM1-associated enhancers. Human Molecular Genetics, 2014, 23, 1700-1708.	2.9	12
99	Genomic characterization of the adolescent idiopathic scoliosis-associated transcriptome and regulome. Human Molecular Genetics, 2021, 29, 3606-3615.	2.9	12
100	Identification of novel Fgf enhancers and their role in dental evolution. Evolution & Development, 2016, 18, 31-40.	2.0	10
101	Functional analysis of limb enhancers in the developing fin. Development Genes and Evolution, 2013, 223, 395-399.	0.9	7
102	Rare Variants in theABCG2Promoter Modulate In Vivo Activity. Drug Metabolism and Disposition, 2018, 46, 636-642.	3.3	7
103	Characterization of functional transposable element enhancers in acute myeloid leukemia. Science China Life Sciences, 2020, 63, 675-687.	4.9	7
104	High-throughput techniques enable advances in the roles of DNA and RNA secondary structures in transcriptional and post-transcriptional gene regulation. Genome Biology, 2022, 23, .	8.8	7
105	Comparative Genomic Characterization of the Multimammate Mouse Mastomys coucha. Molecular Biology and Evolution, 2019, 36, 2805-2812.	8.9	6
106	Noncoding SNPs associated with increased GDF15 levels located in a metformin-activated enhancer region upstream of GDF15. Pharmacogenomics, 2020, 21, 509-520.	1.3	6
107	Ancestry- and sex-specific effects underlying inguinal hernia susceptibility identified in a multiethnic genome-wide association study meta-analysis. Human Molecular Genetics, 2022, 31, 2279-2293.	2.9	6
108	In Vivo Hepatic Enhancer Elements in the HumanABCG2Locus. Drug Metabolism and Disposition, 2017, 45, 208-215.	3.3	5

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109	ABCG2 regulatory single-nucleotide polymorphisms alter in vivo enhancer activity and expression. Pharmacogenetics and Genomics, 2017, 27, 454-463.	1.5	5
110	Asymmetron: a toolkit for the identification of strand asymmetry patterns in biological sequences. Nucleic Acids Research, 2021, 49, e4-e4.	14.5	5
111	Institutional Profile: The University of California Pharmacogenomics Center: at the interface of genomics, biological mechanisms and drug therapy. Pharmacogenomics, 2009, 10, 1569-1576.	1.3	3
112	Classification of topological domains based on gene expression and regulation. Genome, 2013, 56, 415-423.	2.0	2
113	Gene Regulatory Elements. , 2012, , 1-17.		1
114	Myosin VI. , 2003, , .		1
115	Location, Location,Cis-mutation. Human Mutation, 2011, 32, iv-iv.	2.5	0
116	Reply to Liu et al.: Tissue specificity of SIM1 gene expression and erectile dysfunction. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 3349-3350.	7.1	0
117	Cis-Regulatory Enhancer Mutations are a Cause of Human Limb Malformations. , 2012, , 73-93.		0
118	Functional Characterization of Gene Regulatory Elements. FASEB Journal, 2018, 32, 20.2.	0.5	0