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List of Publications by Year
in descending order

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58 papers	2,911 citations	257101 24 h-index	174990 52 g-index
58 all docs	58 docs citations	58 times ranked	5388 citing authors

#	Article	IF	Citations
1	Long noncoding RNAs in mouse embryonic stem cell pluripotency and differentiation. Genome Research, 2008, 18, 1433-1445.	2.4	698
2	Survival and dementia in <scp>GBA</scp>â€associated Parkinson's disease: <scp>T</scp>he mutation matters. Annals of Neurology, 2016, 80, 662-673.	2.8	312
3	A transcriptional sketch of a primary human breast cancer by 454 deep sequencing. BMC Genomics, 2009, 10, 163.	1.2	205
4	Expression of distinct RNAs from 3â€² untranslated regions. Nucleic Acids Research, 2011, 39, 2393-2403.	6.5	185
5	Genetic Association and Altered Gene Expression of Mir-155 in Multiple Sclerosis Patients. International Journal of Molecular Sciences, 2011, 12, 8695-8712.	1.8	93
6	A novel mutation within the MIR96 gene causes non-syndromic inherited hearing loss in an Italian family by altering pre-miRNA processing. Human Molecular Genetics, 2012, 21, 577-585.	1.4	92
7	The Characterization of GSDMB Splicing and Backsplicing Profiles Identifies Novel Isoforms and a Circular RNA That Are Dysregulated in Multiple Sclerosis. International Journal of Molecular Sciences, 2017, 18, 576.	1.8	81
8	Tumor-Derived Prostaglandin E2 Promotes p50 NF-ÎºB-Dependent Differentiation of Monocytic MDSCs. Cancer Research, 2020, 80, 2874-2888.	0.4	81
9	Glucocerebrosidase mutations in primary parkinsonism. Parkinsonism and Related Disorders, 2014, 20, 1215-1220.	1.1	63
10	The GBAP1 pseudogene acts as a ceRNA for the glucocerebrosidase gene GBA by sponging miR-22-3p. Scientific Reports, 2017, 7, 12702.	1.6	62
11	DNAJC12 and dopaâ€responsive nonprogressive parkinsonism. Annals of Neurology, 2017, 82, 640-646.	2.8	60
12	Not only cancer: the long non-coding RNA MALAT1 affects the repertoire of alternatively spliced transcripts and circular RNAs in multiple sclerosis. Human Molecular Genetics, 2019, 28, 1414-1428.	1.4	56
13	Radiomics and gene expression profile to characterise the disease and predict outcome in patients with lung cancer. European Journal of Nuclear Medicine and Molecular Imaging, 2021, 48, 3643-3655.	3.3	53
14	SNCA and MAPT genes: Independent and joint effects in Parkinson disease in the Italian population. Parkinsonism and Related Disorders, 2012, 18, 257-262.	1.1	51
15	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	1.8	44
16	LINE1 are spliced in non-canonical transcript variants to regulate T cell quiescence and exhaustion. Nature Genetics, 2022, 54, 180-193.	9.4	39
17	Clinical relevance of clonal hematopoiesis in persons aged â‰¥80 years. Blood, 2021, 138, 2093-2105.	0.6	37
18	The SPID-GBA study. Neurology: Genetics, 2020, 6, e523.	0.9	37

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19	First independent replication of the involvement of LARS2 in Perrault syndrome by whole-exome sequencing of an Italian family. Journal of Human Genetics, 2016, 61, 295-300.	1.1	34
20	Clinical and molecular characterisation of 21 patients affected by quantitative fibrinogen deficiency. Thrombosis and Haemostasis, 2015, 113, 567-576.	1.8	33
21	Functional variations modulating PRKCA expression and alternative splicing predispose to multiple sclerosis. Human Molecular Genetics, 2014, 23, 6746-6761.	1.4	32
22	A lysosomeâ€plasma membraneâ€sphingolipid axis linking lysosomal storage to cell growth arrest. FASEB Journal, 2018, 32, 5685-5702.	0.2	32
23	Characterization of the genomic structure of the human neuronal nicotinic acetylcholine receptor CHRNA5/A3/B4 gene cluster and identification of novel intragenic polymorphisms. Journal of Human Genetics, 2001, 46, 640-648.	1.1	29
24	No association of GBA mutations and multiple system atrophy. European Journal of Neurology, 2013, 20, e61-2.	1.7	28
25	Non-random retention of protein-coding overlapping genes in Metazoa. BMC Genomics, 2008, 9, 174.	1.2	26
26	An Ariadne's thread to the identification and annotation of noncoding RNAs in eukaryotes. Briefings in Bioinformatics, 2009, 10, 475-489.	3.2	25
27	Interpreting Non-coding Genetic Variation in Multiple Sclerosis Genome-Wide Associated Regions. Frontiers in Genetics, 2018, 9, 647.	1.1	25
28	In vivo RNAâ€“RNA duplexes from human Î±3 and Î±5 nicotinic receptor subunit mRNAs. Gene, 2005, 345, 155-164.	1.0	24
29	Molecular characterization of two novel mutations causing factor XI deficiency: A splicing defect and a missense mutation responsible for a CRM+ defect. Thrombosis and Haemostasis, 2008, 99, 523-530.	1.8	24
30	Shedding Light on the Dark Side of the Genome: Overlapping Genes in Higher Eukaryotes. Current Genomics, 2004, 5, 509-524.	0.7	24
31	Molecular characterization of in-frame and out-of-frame alternative splicings in coagulation factor XI pre-mRNA. Blood, 2010, 115, 2065-2072.	0.6	23
32	Meta-Analysis of Multiple Sclerosis Microarray Data Reveals Dysregulation in RNA Splicing Regulatory Genes. International Journal of Molecular Sciences, 2015, 16, 23463-23481.	1.8	22
33	The expanding spectrum of PRPS1-associated phenotypes: three novel mutations segregating with X-linked hearing loss and mild peripheral neuropathy. European Journal of Human Genetics, 2015, 23, 766-773.	1.4	22
34	Side-by-side comparison of next-generation sequencing, cytology, and histology in diagnosing locally advanced pancreatic adenocarcinoma. Gastrointestinal Endoscopy, 2021, 93, 597-604.e5.	0.5	22
35	4q-D4Z4 chromatin architecture regulates the transcription of muscle atrophic genes in facioscapulohumeral muscular dystrophy. Genome Research, 2019, 29, 883-895.	2.4	18
36	Next-Generation Sequencing Analysis of MiRNA Expression in Control and FSHD Myogenesis. PLoS ONE, 2014, 9, e108411.	1.1	17

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37	Role of Lysosomal Gene Variants in Modulating <scp>GBA</scp>â€Associated Parkinson's Disease Risk. Movement Disorders, 2022, 37, 1202-1210.	2.2	17
38	Functional characterization of two novel splicing mutations in the OCA2 gene associated with oculocutaneous albinism type II. Gene, 2014, 537, 79-84.	1.0	16
39	Alport syndrome cold cases: Missing mutations identified by exome sequencing and functional analysis. PLoS ONE, 2017, 12, e0178630.	1.1	16
40	The Asp620asn mutation in VPS35 is not a common cause of familial Parkinson's disease. Movement Disorders, 2012, 27, 800-801.	2.2	15
41	Fine Characterization of the Recurrent c.1584+18672A>G Deep-Intronic Mutation in the Cystic Fibrosis Transmembrane Conductance Regulator Gene. American Journal of Respiratory Cell and Molecular Biology, 2013, 48, 619-625.	1.4	15
42	First Replication of the Involvement of OTUD6B in Intellectual Disability Syndrome With Seizures and Dysmorphic Features. Frontiers in Genetics, 2018, 9, 464.	1.1	14
43	X-Linked Alport Syndrome in Women: Genotype and Clinical Course in 24 Cases. Frontiers in Medicine, 2020, 7, 580376.	1.2	14
44	Whole-gene CFTR sequencing combined with digital RT-PCR improves genetic diagnosis of cystic fibrosis. Journal of Human Genetics, 2016, 61, 977-984.	1.1	12
45	Genetic Association and Altered Gene Expression of CYBB in Multiple Sclerosis Patients. Biomedicines, 2018, 6, 117.	1.4	10
46	In-depth characterization of breast cancer tumor-promoting cell transcriptome by RNA sequencing and microarrays. Oncotarget, 2016, 7, 976-994.	0.8	10
47	A type II mutation (Glu117stop), induction of allele-specific mRNA degradation and factor XI deficiency. Haematologica, 2005, 90, 1716-8.	1.7	9
48	miR-634 is a Pol III-dependent intronic microRNA regulating alternative-polyadenylated isoforms of its host gene PRKCA. Biochimica Et Biophysica Acta - General Subjects, 2017, 1861, 1046-1056.	1.1	7
49	Saposin D variants are not a common cause of familial Parkinsonâ€™s disease among Italians. Brain, 2020, 143, e71-e71.	3.7	7
50	SLC22A4 Gene in Hereditary Non-syndromic Hearing Loss: Recurrence and Incomplete Penetrance of the p.C113Y Mutation in Northwest Africa. Frontiers in Genetics, 2021, 12, 606630.	1.1	7
51	Post-Biopsy Cell-Free DNA From Blood: An Open Window on Primary Prostate Cancer Genetics and Biology. Frontiers in Oncology, 2021, 11, 654140.	1.3	6
52	Dual Role of G-runs and hnRNP F in the Regulation of a Mutation-Activated Pseudoexon in the Fibrinogen Gamma-Chain Transcript. PLoS ONE, 2013, 8, e59333.	1.1	5
53	Two novel splicing mutations in the SLC45A2 gene cause Oculocutaneous Albinism Type IV by unmasking cryptic splice sites. Journal of Human Genetics, 2015, 60, 467-471.	1.1	5
54	Massive Accumulation of Sphingomyelin Affects the Lysosomal and Mitochondria Compartments and Promotes Apoptosis in Niemann-Pick Disease Type A. Journal of Molecular Neuroscience, 2022, 72, 1482-1499.	1.1	5

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55	Improving mRNA 5â€² coding sequence determination in the mouse genome. Mammalian Genome, 2014, 25, 149-159.	1.0	4
56	Role of Cytoskeletal Diaphanous-Related Formins in Hearing Loss. Cells, 2022, 11, 1726.	1.8	4
57	Response: Further thoughts on the â€œphantomâ€•Î”6/7 FXI isoform. Blood, 2010, 116, 1186-1187.	0.6	3
58	How I faced my prostate cancer: a molecular biologistâ€™s perspective. Npj Precision Oncology, 2021, 5, 88.	2.3	1