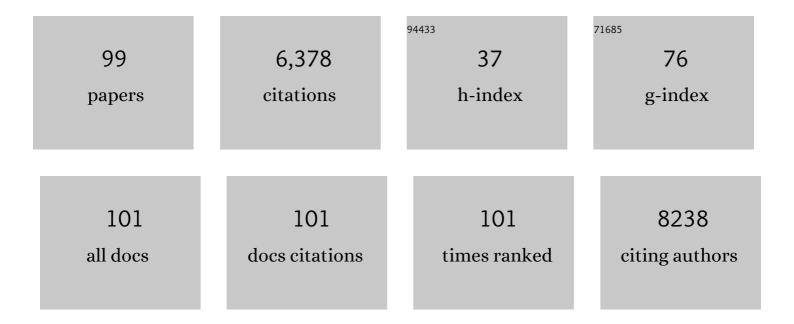
## Alfredo Ciccodicola

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	The DNA sequence of the human X chromosome. Nature, 2005, 434, 325-337.	27.8	985
2	Genomic rearrangement in NEMO impairs NF-κB activation and is a cause of incontinentia pigmenti. Nature, 2000, 405, 466-472.	27.8	709
3	A gene (RPGR) with homology to the RCC1 guanine nucleotide exchange factor is mutated in X–linked retinitis pigmentosa (RP3). Nature Genetics, 1996, 13, 35-42.	21.4	453
4	Mutational hot spot within a new RPGR exon in X-linked retinitis pigmentosa. Nature Genetics, 2000, 25, 462-466.	21.4	392
5	Uncovering the Complexity of Transcriptomes with RNA-Seq. Journal of Biomedicine and Biotechnology, 2010, 2010, 1-19.	3.0	315
6	RNA-Seq and human complex diseases: recent accomplishments and future perspectives. European Journal of Human Genetics, 2013, 21, 134-142.	2.8	216
7	Transcriptome Profiling in Human Diseases: New Advances and Perspectives. International Journal of Molecular Sciences, 2017, 18, 1652.	4.1	193
8	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
9	Isolation, physical mapping, and Northern analysis of the X-linked human gene encoding methyl CpG-binding protein, MECP2. Mammalian Genome, 1996, 7, 533-535.	2.2	116
10	Sequence of human glucose-6-phosphate dehydrogenase cloned in plasmids and a yeast artificial chromosome. Genomics, 1991, 10, 792-800.	2.9	110
11	Mapping human chromosomes by walking with sequence-tagged sites from end fragments of yeast artificial chromosome inserts. Genomics, 1992, 14, 241-248.	2.9	110
12	Filamin A Is Mutated in X-Linked Chronic Idiopathic Intestinal Pseudo-Obstruction with Central Nervous System Involvement. American Journal of Human Genetics, 2007, 80, 751-758.	6.2	106
13	Identification of new mutations in the Emery-Dreifuss muscular dystrophy gene and evidence for genetic heterogeneity of the disease. Human Molecular Genetics, 1995, 4, 1859-1863.	2.9	93
14	A synaptobrevin–like gene in the Xq28 pseudoautosomal region undergoes X inactivation. Nature Genetics, 1996, 13, 227-229.	21.4	78
15	Klinefelter's syndrome as a model of anomalous cerebral laterality: Testing gene dosage in the X chromosome pseudoautosomal region using a DNA microarray. Genesis, 1998, 23, 215-229.	2.1	77
16	Molecular genetics of autosomal dominant retinitis pigmentosa (ADRP): a comprehensive study of 43 Italian families. Journal of Medical Genetics, 2005, 42, e47-e47.	3.2	74
17	Yeast artificial chromosome-based genome mapping: Some lessons from Xq24–q28. Genomics, 1991, 11, 783-793.	2.9	71
18	Long-range sequence analysis in Xq28: thirteen known and six candidate genes in 219.4 kb of high GC DNA between the RCP/GCP and G6PD loci. Human Molecular Genetics, 1996, 5, 659-668.	2.9	69

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19	Complex Events in the Evolution of the Human Pseudoautosomal Region 2 (PAR2). Genome Research, 2003, 13, 281-286.	5.5	63
20	Locations and contexts of sequences that hybridize to poly(dG-dT).(dC-dA) in mammalian ribosomal DNAs and two X-linked genes. Nucleic Acids Research, 1988, 16, 865-881.	14.5	62
21	A muscle-specific DNase I-like gene in human Xq28. Human Molecular Genetics, 1995, 4, 1557-1564.	2.9	62
22	Massive-Scale RNA-Seq Analysis of Non Ribosomal Transcriptome in Human Trisomy 21. PLoS ONE, 2011, 6, e18493.	2.5	62
23	Molecular and Clinical Characterization of Albinism in a Large Cohort of Italian Patients. , 2011, 52, 1281.		58
24	PPARγΔ5, a Naturally Occurring Dominant-Negative Splice Isoform, Impairs PPARγ Function and Adipocyte Differentiation. Cell Reports, 2018, 25, 1577-1592.e6.	6.4	58
25	A Novel Peroxisome Proliferator-activated Receptor Î <sup>3</sup> Isoform with Dominant Negative Activity Generated by Alternative Splicing. Journal of Biological Chemistry, 2005, 280, 26517-26525.	3.4	55
26	Autosomal recessive familial exudative vitreoretinopathy: evidence for genetic heterogeneity. Clinical Genetics, 1998, 54, 315-320.	2.0	54
27	Experimental colitis: decreased Octn2 and Atb0+ expression in rat colonocytes induces carnitine depletion that is reversible by carnitineâ€oaded liposomes. FASEB Journal, 2006, 20, 2544-2546.	0.5	54
28	Heart failure: Pilot transcriptomic analysis of cardiac tissue by RNA-sequencing. Cardiology Journal, 2017, 24, 539-553.	1.2	54
29	PPARG: Gene Expression Regulation and Next-Generation Sequencing for Unsolved Issues. PPAR Research, 2010, 2010, 1-17.	2.4	52
30	New somatic mutations and <i>WNK1-B4GALNT3</i> gene fusion in papillary thyroid carcinoma. Oncotarget, 2015, 6, 11242-11251.	1.8	51
31	SSCP detection of novel mutations in patients with Emery-Dreifuss muscular dystrophy: definition of a small C-terminal region required for emerin function. Human Molecular Genetics, 1995, 4, 2003-2004.	2.9	48
32	YAC Contig Organization and CpG Island Analysis in Xq28. Genomics, 1994, 24, 149-158.	2.9	44
33	Clinical features of X linked juvenile retinoschisis associated with new mutations in the XLRS1 gene in Italian families. British Journal of Ophthalmology, 2003, 87, 1130-1134.	3.9	43
34	Pan-Cancer Mutational and Transcriptional Analysis of the Integrator Complex. International Journal of Molecular Sciences, 2017, 18, 936.	4.1	41
35	Non-coding RNA and pseudogenes in neurodegenerative diseases: "The (un)Usual Suspectsâ€; Frontiers in Genetics, 2012, 3, 231.	2.3	40
36	Hoxa5 undergoes dynamic DNA methylation and transcriptional repression in the adipose tissue of mice exposed to high-fat diet. International Journal of Obesity, 2016, 40, 929-937.	3.4	40

ALFREDO CICCODICOLA

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37	Identification of novel RP2 mutations in a subset of X-linked retinitis pigmentosa families and prediction of new domains. Human Mutation, 2001, 18, 109-119.	2.5	39
38	Identification and characterisation of the retinitis pigmentosa 1-like1 gene (RP1L1): a novel candidate for retinal degenerations. European Journal of Human Genetics, 2003, 11, 155-162.	2.8	39
39	Human glucose-6-phosphate dehydrogenase gene carried on a yeast artificial chromosome encodes active enzyme in monkey cells. Genomics, 1990, 7, 531-534.	2.9	38
40	Sequence-Based Exon Prediction around the Synaptophysin Locus Reveals a Gene-Rich Area Containing Novel Genes in Human Proximal Xp. Genomics, 1997, 45, 340-347.	2.9	38
41	Antigen delivery by filamentous bacteriophage fd displaying an anti― <scp>DEC</scp> â€205 singleâ€chain variable fragment confers adjuvanticity by triggering a <scp>TLR</scp> 9â€mediated immune response. EMBO Molecular Medicine, 2015, 7, 973-988.	6.9	38
42	A novel pseudoautosomal gene encoding a putative GTP-binding protein resides in the vicinity of the Xp/Yp telomere. Human Molecular Genetics, 1998, 7, 407-414.	2.9	37
43	Smith-Lemli-Opitz syndrome: evidence of T93M as a common mutation of Δ7-sterol reductase in Italy and report of three novel mutations. European Journal of Human Genetics, 1999, 7, 937-940.	2.8	37
44	Characterization of a Novel Polymorphism in PPARG Regulatory Region Associated with Type 2 Diabetes and Diabetic Retinopathy in Italy. Journal of Biomedicine and Biotechnology, 2009, 2009, 1-7.	3.0	36
45	Impairment of circulating endothelial progenitors in Down syndrome. BMC Medical Genomics, 2010, 3, 40.	1.5	36
46	Genetic and epigenetic alterations of RB2/p130 tumor suppressor gene in human sporadic retinoblastoma: implications for pathogenesis and therapeutic approach. Oncogene, 2005, 24, 5827-5836.	5.9	34
47	A novel pseudoautosomal human gene encodes a putative protein similar to Ac-like transposases. Human Molecular Genetics, 1999, 8, 61-67.	2.9	31
48	Glucose impairs tamoxifen responsiveness modulating connective tissue growth factor in breast cancer cells. Oncotarget, 2017, 8, 109000-109017.	1.8	31
49	Mutation analysis of the RPGR gene reveals novel mutations in south European patients with X-linked retinitis pigmentosa. European Journal of Human Genetics, 1999, 7, 687-694.	2.8	30
50	ZPLD1 gene is disrupted in a patient with balanced translocation that exhibits cerebral cavernous malformations. Neuroscience, 2008, 155, 345-349.	2.3	30
51	Transcriptional Regulation: Molecules, Involved Mechanisms, and Misregulation. International Journal of Molecular Sciences, 2019, 20, 1281.	4.1	30
52	Detrimental effects of <i>Bartonella henselae</i> are counteracted by <scp> </scp> -arginine and nitric oxide in human endothelial progenitor cells. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 9427-9432.	7.1	29
53	PR/SET Domain Family and Cancer: Novel Insights from the Cancer Genome Atlas. International Journal of Molecular Sciences, 2018, 19, 3250.	4.1	29
54	Nutritional genomics era: opportunities toward a genome-tailored nutritional regimen. Journal of Nutritional Biochemistry, 2010, 21, 457-467.	4.2	28

ALFREDO CICCODICOLA

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55	Differential Expression Pattern of XqPAR-Linked Genes SYBL1 and IL9R Correlates with the Structure and Evolution of the Region. Human Molecular Genetics, 1997, 6, 1917-1923.	2.9	26
56	Identification of a Novel Mutation in the Myosin VIIA Motor Domain in a Family with Autosomal Dominant Hearing Loss (DFNA11). Audiology and Neuro-Otology, 2006, 11, 157-164.	1.3	25
57	Conserved sequence-tagged sites: a phylogenetic approach to genome mapping Proceedings of the National Academy of Sciences of the United States of America, 1992, 89, 3681-3685.	7.1	24
58	PPARÎ <sup>3</sup> and Diabetes: Beyond the Genome and Towards Personalized Medicine. Current Diabetes Reports, 2021, 21, 18.	4.2	23
59	New evidence for the correlation of the p.G130V mutation in the <i>GJB2</i> gene and syndromic hearing loss with palmoplantar keratoderma. American Journal of Medical Genetics, Part A, 2009, 149A, 685-688.	1.2	22
60	Oncogenic Properties of the Antisense lncRNA <i>COMET</i> in <i>BRAF</i> and <i>RET</i> -Driven Papillary Thyroid Carcinomas. Cancer Research, 2019, 79, 2124-2135.	0.9	22
61	Investigation of Gamma-aminobutyric acid (GABA) A receptors genes and migraine susceptibility. BMC Medical Genetics, 2008, 9, 109.	2.1	21
62	Pharmacogenomics of Drug Response in Type 2 Diabetes: Toward the Definition of Tailored Therapies?. PPAR Research, 2015, 2015, 1-10.	2.4	18
63	Alternative Splicing in Adhesion- and Motility-Related Genes in Breast Cancer. International Journal of Molecular Sciences, 2016, 17, 121.	4.1	18
64	Escape from X Inactivation of Two New Genes Associated with DXS6974E and DXS7020E. Genomics, 1997, 43, 183-190.	2.9	17
65	Evidence of Bacteroides fragilis Protection from Bartonella henselae-Induced Damage. PLoS ONE, 2012, 7, e49653.	2.5	17
66	Analysis of SEMA6B gene expression in breast cancer: Identification of a new isoform. Biochimica Et Biophysica Acta - General Subjects, 2013, 1830, 4543-4553.	2.4	16
67	Identification and expression analysis of novel Jakmip1 transcripts. Gene, 2007, 402, 1-8.	2.2	15
68	Cloning and gene structure of the rod cGMP phosphodiesterase delta subunit gene (PDED) in man and mouse. European Journal of Human Genetics, 1998, 6, 283-290.	2.8	14
69	Characterization of MPP4, a gene highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. Gene, 2002, 297, 33-38.	2.2	14
70	Is <i>PPARG</i> the key gene in diabetic retinopathy?. British Journal of Pharmacology, 2012, 165, 1-3.	5.4	14
71	In Vitro-Generated Hypertrophic-Like Adipocytes Displaying PPARG Isoforms Unbalance Recapitulate Adipocyte Dysfunctions In Vivo. Cells, 2020, 9, 1284.	4.1	14
72	Identification and characterization of C1orf36, a transcript highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. Biochemical and Biophysical Research Communications, 2003, 308, 414-421.	2.1	13

ALFREDO CICCODICOLA

#	Article	IF	CITATIONS
73	DDX11L: a novel transcript family emerging from human subtelomeric regions. BMC Genomics, 2009, 10, 250.	2.8	13
74	The "next-generation―knowledge of papillary thyroid carcinoma. Cell Cycle, 2015, 14, 2018-2021.	2.6	13
75	Hepatic Insulin Resistance in Hyperthyroid Rat Liver: Vitamin E Supplementation Highlights a Possible Role of ROS. Antioxidants, 2022, 11, 1295.	5.1	13
76	AnaLysis of Expression on human chromosome 21, ALE-HSA21: a pilot integrated web resource. Database: the Journal of Biological Databases and Curation, 2014, 2014, bau009.	3.0	12
77	SFMetaDB: a comprehensive annotation of mouse RNA splicing factor RNA-Seq datasets. Database: the Journal of Biological Databases and Curation, 2017, 2017, .	3.0	12
78	Mapping of MRX81 in Xp11.2-Xq12 suggests the presence of a new gene involved in nonspecific X-linked mental retardation. American Journal of Medical Genetics Part A, 2003, 118A, 217-222.	2.4	10
79	RNA-Seq for the identification of novel Mediator transcripts in endothelial progenitor cells. Gene, 2014, 547, 98-105.	2.2	10
80	Integrated Network Pharmacology Approach for Drug Combination Discovery: A Multi-Cancer Case Study. Cancers, 2022, 14, 2043.	3.7	10
81	Human protein kinase C iota gene (PRKCI) is closely linked to the BTK gene in Xq21.3. Genomics, 1995, 26, 629-631.	2.9	9
82	Functional expression of human glucose-6-phosphate dehydrogenase in Escherichia coli. Gene, 1989, 78, 365-370.	2.2	8
83	Complete congenital stationary night blindness maps on Xp11.4 in a Sardinian family. European Journal of Human Genetics, 1999, 7, 574-578.	2.8	8
84	Human and mouse SYBL1 gene structure and expression. Gene, 1999, 240, 233-238.	2.2	8
85	Identification and characterization of a novel human brain-specific gene, homologous to S. scrofa tmp83.5 , in the chromosome 10q24 critical region for temporal lobe epilepsy and spastic paraplegia. Gene, 2002, 282, 87-94.	2.2	8
86	Screening forGJB2andGJB6gene mutations in patients from Campania region with sensorineural hearing loss. International Journal of Audiology, 2010, 49, 326-331.	1.7	8
87	Novel Transcription Factor Variants through RNA-Sequencing: The Importance of Being "Alternativeâ€. International Journal of Molecular Sciences, 2015, 16, 1755-1771.	4.1	8
88	Computational Analysis of Single Nucleotide Polymorphisms Associated with Altered Drug Responsiveness in Type 2 Diabetes. International Journal of Molecular Sciences, 2016, 17, 1008.	4.1	8
89	E2 multimeric scaffold for vaccine formulation: immune response by intranasal delivery and transcriptome profile of E2-pulsed dendritic cells. BMC Microbiology, 2016, 16, 152.	3.3	8
90	TNFα Mediates Inflammation-Induced Effects on PPARG Splicing in Adipose Tissue and Mesenchymal Precursor Cells. Cells, 2022, 11, 42.	4.1	6

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91	Filamin (FLN1),plexin (SEX), major palmitoylated proteinp55 (MPP1), and von-Hippel Lindau binding protein (VBP1) are not involved in incontinentia pigmenti type 2. American Journal of Medical Genetics Part A, 2000, 94, 79-84.	2.4	5
92	A new de novo mutation of the connexin-32 gene in a patient with X-linked Charcot-Marie-Tooth type 1 disease. Neurological Sciences, 2000, 21, 109-112.	1.9	4
93	RBPMetaDB: a comprehensive annotation of mouse RNA-Seq datasets with perturbations of RNA-binding proteins. Database: the Journal of Biological Databases and Curation, 2018, 2018, .	3.0	4
94	Human homologue of the murinebare patches/striated gene is not mutated in incontinentia pigmenti type 2. , 2000, 91, 241-244.		3
95	Human dbl proto-oncogene in 85 kb of Xq26, and determination of the transcription initiation site. Gene, 2000, 253, 107-115.	2.2	3
96	Sequence-tagged sites (STSs) from YAC insert-ends and X-specific flow-sorted chromosomes. Mammalian Genome, 1994, 5, 511-514.	2.2	2
97	Expressed STSs and transcription of human Xq28. Gene, 1997, 187, 185-191.	2.2	2
98	Non-coding RNA in Neurodegeneration. Current Geriatrics Reports, 2012, 1, 219-228.	1.1	0
99	High-Throughput Analysis of Noncoding RNAs. , 2016, , 215-238.		Ο