Adamo Pio d'Adamo

List of Publications by Year
in descending order

104	10,715	66343	33894
papers	citations	42 h-index	99 g-index

105	105	105	21464
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	27.8	1,328
2	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. Nature Genetics, 2013, 45, 145-154.	21.4	675
3	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. Nature, 2014, 514, 92-97.	27.8	548
4	Somatic mosaic IDH1 and IDH2 mutations are associated with enchondroma and spindle cell hemangioma in Ollier disease and Maffucci syndrome. Nature Genetics, 2011, 43, 1256-1261.	21.4	488
5	Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies. Nature Genetics, 2010, 42, 1077-1085.	21.4	445
6	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. Nature Communications, 2016, 7, 10023.	12.8	412
7	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	21.4	403
8	New gene functions in megakaryopoiesis and platelet formation. Nature, 2011, 480, 201-208.	27.8	401
9	MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. Nature Genetics, 2006, 38, 570-575.	21.4	380
10	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	27.8	320
11	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. Nature Genetics, 2012, 44, 260-268.	21.4	303
12	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	21.4	281
13	Genetic Structure of Europeans: A View from the Northâ€“East. PLoS ONE, 2009, 4, e5472.	2.5	279
14	Meta-analysis identifies multiple loci associated with kidney functionâ€“related traits in east Asian populations. Nature Genetics, 2012, 44, 904-909.	21.4	254
15	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. Nature Genetics, 2009, 41, 654-656.	21.4	233
16	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	7.6	229
17	Severe X-Linked Mitochondrial Encephalomyopathy Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2010, 86, 639-649.	6.2	199
18	Ethylmalonic Encephalopathy Is Caused by Mutations in ETHE1, a Gene Encoding a Mitochondrial Matrix Protein. American Journal of Human Genetics, 2004, 74, 239-252.	6.2	192

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19	Genome-wide association and longitudinal analyses reveal genetic loci linking pubertal height growth, pubertal timing and childhood adiposity. Human Molecular Genetics, 2013, 22, 2735-2747.	2.9	188
20	Severe Infantile Encephalomyopathy Caused by a Mutation in COX6B1, a Nucleus-Encoded Subunit of Cytochrome C Oxidase. American Journal of Human Genetics, 2008, 82, 1281-1289.	6.2	165
21	Variation in the Bitterâ€taste Receptor Gene TAS2R38, and Adiposity in a Genetically Isolated Population in Southern Italy. Obesity, 2008, 16, 2289-2295.	3.0	165
22	Mutations in TTC19 cause mitochondrial complex III deficiency and neurological impairment in humans and flies. Nature Genetics, 2011, 43, 259-263.	21.4	148
23	Meta-Analysis of Genome-Wide Association Studies Identifies Six New Loci for Serum Calcium Concentrations. PLoS Genetics, 2013, 9, e1003796.	3.5	142
24	Functional analysis of mutations in SLC7A9, and genotypeâ€“phenotype correlation in non-Type I cystinuria. Human Molecular Genetics, 2001, 10, 305-316.	2.9	125
25	Genetic Variation in Taste Sensitivity to 6â€nâ€Propylthiouracil and Its Relationship to Taste Perception and Food Selection. Annals of the New York Academy of Sciences, 2009, 1170, 126-139.	3.8	112
26	FASTKD2 Nonsense Mutation in an Infantile Mitochondrial Encephalomyopathy Associated with Cytochrome C Oxidase Deficiency. American Journal of Human Genetics, 2008, 83, 415-423.	6.2	107
27	Cigarette toxicity triggers Leberâ€™s hereditary optic neuropathy by affecting mtDNA copy number, oxidative phosphorylation and ROS detoxification pathways. Cell Death and Disease, 2015, 6, e2021-e2021.	6.3	107
28	Heterogeneity in Circulating Tumor Cells: The Relevance of the Stem-Cell Subset. Cancers, 2019, 11, 483.	3.7	107
29	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. Scientific Reports, 2017, 7, 45040.	3.3	98
30	Behavioral Disorder, Dementia, Ataxia, and Rigidity in a Large Family With TATA Box-Binding Protein Mutation. Archives of Neurology, 2004, 61, 1314-20.	4.5	82
31	Genome-wide association study of sexual maturation in males and females highlights a role for body mass and menarche loci in male puberty. Human Molecular Genetics, 2014, 23, 4452-4464.	2.9	82
32	Evidence of Inbreeding Depression on Human Height. PLoS Genetics, 2012, 8, e1002655.	3.5	79
33	Hearing function and thresholds: a genome-wide association study in European isolated populations identifies new loci and pathways. Journal of Medical Genetics, 2011, 48, 369-374.	3.2	71
34	Dominant inheritance of a novel integrin Â3 mutation associated with a hereditary macrothrombocytopenia and platelet dysfunction in two Italian families. Haematologica, 2009, 94, 663-669.	3.5	64
35	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. European Journal of Human Genetics, 2013, 21, 659-665.	2.8	64
36	Modulation of Genetic Associations with Serum Urate Levels by Body-Mass-Index in Humans. PLoS ONE, 2015, 10, e0119752.	2.5	64

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37	Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index. Molecular Psychiatry, 2017, 22, 192-201.	7.9	63
38	DNA mismatch repair gene MSH6 implicated in determining age at natural menopause. Human Molecular Genetics, 2014, 23, 2490-2497.	2.9	56
39	A novel GJA1 mutation causes oculodentodigital dysplasia without syndactyly. American Journal of Medical Genetics, Part A, 2005, 133A, 58-60.	1.2	55
40	Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. Brain, 2016, 139, e17-e17.	7.6	51
41	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. Gene, 2014, 534, 236-239.	2.2	50
42	Autosomal recessive stickler syndrome due to a loss of function mutation in the COL9A3 gene. American Journal of Medical Genetics, Part A, 2014, 164, 42-47.	1.2	49
43	A GWAS sequence variant for platelet volume marks an alternative DNM3 promoter in megakaryocytes near a MEIS1 binding site. Blood, 2012, 120, 4859-4868.	1.4	44
44	Association Analysis of Bitter Receptor Genes in Five Isolated Populations Identifies a Significant Correlation between TAS2R43 Variants and Coffee Liking. PLoS ONE, 2014, 9, e92065.	2.5	41
45	Does epidermal thickening explain GJB2 high carrier frequency and heterozygote advantage?. European Journal of Human Genetics, 2009, 17, 284-286.	2.8	35
46	Phospholipase C-Î²3 Is a Key Modulator of IL-8 Expression in Cystic Fibrosis Bronchial Epithelial Cells. Journal of Immunology, 2011, 186, 4946-4958.	0.8	34
47	Maps of open chromatin highlight cell typeâ€“restricted patterns of regulatory sequence variation at hematological trait loci. Genome Research, 2013, 23, 1130-1141.	5.5	34
48	Next generation sequencing in nonsyndromic intellectual disability: From a negative molecular karyotype to a possible causative mutation detection. American Journal of Medical Genetics, Part A, 2014, 164, 170-176.	1.2	34
49	Genome-wide meta-analysis of common variant differences between men and women. Human Molecular Genetics, 2012, 21, 4805-4815.	2.9	33
50	Influence of age, sex and ethnicity on platelet count in five Italian geographic isolates: mild thrombocytopenia may be physiological. British Journal of Haematology, 2012, 157, 384-387.	2.5	33
51	Charcotâ€“Marieâ€“Tooth disease type 2C: a distinct genetic entity. Clinical and molecular characterization of the first European family. Neuromuscular Disorders, 2002, 12, 399-404.	0.6	31
52	Genetic heterogeneity of FG syndrome: a fourth locus (FGS4) maps to Xp11.4-p11.3 in an Italian family. Human Genetics, 2003, 112, 124-130.	3.8	30
53	Juvenile hemochromatosis locus maps to chromosome 1q in a French Canadian population. European Journal of Human Genetics, 2003, 11, 585-589.	2.8	28
54	Genomic profiling by wholeâ€genome single nucleotide polymorphism arrays in Wilms tumor and association with relapse. Genes Chromosomes and Cancer, 2012, 51, 644-653.	2.8	28

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55	Pharmacogenetics and induction/consolidation therapy toxicities in acute lymphoblastic leukemia patients treated with AIEOP-BFM ALL 2000 protocol. Pharmacogenomics Journal, 2017, 17, 4-10.	2.0	28
56	Cohen syndrome resulting from a novel large intragenic COH1 deletion segregating in an isolated Greek island population. American Journal of Medical Genetics, Part A, 2008, 146A, 2221-2226.	1.2	26
57	A second locus mapping to 2q35â€“36 for familial pseudohyperkalaemia. European Journal of Human Genetics, 2004, 12, 1073-1076.	2.8	25
58	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
59	Variation of hemoglobin levels in normal Italian populations from genetic isolates. Haematologica, 2008, 93, 1372-1375.	3.5	25
60	Cystinuria type I: Identification of eight new mutations in SLC3A1. Kidney International, 2001, 59, 1250-1256.	5.2	24
61	Genetic landscape of populations along the Silk Road: admixture and migration patterns. BMC Genetics, 2014, 15, 131.	2.7	24
62	A nonsense mutation of human <scp>XRCC</scp> 4 is associated with adultâ€onset progressive encephalocardiomyopathy. EMBO Molecular Medicine, 2015, 7, 918-929.	6.9	24
63	Detection of Epidermal Thickening inGJB2Carriers with Epidermal US. Radiology, 2009, 251, 280-286.	7.3	23
64	Narrowing of the critical region in autosomal recessive spastic paraplegia linked to the SPG5 locus. Neurogenetics, 2004, 5, 49-54.	1.4	21
65	Does the 1.5â€‰Mb microduplication in chromosome band Xp22.31 have a pathogenetic role? New contribution and a review of the literature. American Journal of Medical Genetics, Part A, 2012, 158A, 461-464.	1.2	21
66	Adult-onset Alexander disease, associated with a mutation in an alternative GFAP transcript, may be phenotypically modulated by a non-neutral HDAC6 variant. Orphanet Journal of Rare Diseases, 2013, 8, 66.	2.7	21
67	MRX87 family with Aristaless Xdup24bp mutation and implication for polyAlanine expansions. BMC Medical Genetics, 2007, 8, 25.	2.1	20
68	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. Genetics in Medicine, 2015, 17, 396-399.	2.4	19
69	Phenotypic and genetic characterization of a family carrying two Xq21.1-21.3 interstitial deletions associated with syndromic hearing loss. Molecular Cytogenetics, 2015, 8, 18.	0.9	19
70	Systematic analysis of factors that improve homologous direct repair (HDR) efficiency in CRISPR/Cas9 technique. PLoS ONE, 2021, 16, e0247603.	2.5	19
71	Metabonomics and population studies: age-related amino acids excretion and inferring networks through the study of urine samples in two Italian isolated populations. Amino Acids, 2010, 38, 65-73.	2.7	18
72	Brain-derived neurotrophic factor serum levels in genetically isolated populations: gender-specific association with anxiety disorder subtypes but not with anxiety levels or Val66Met polymorphism. PeerJ, 2015, 3, e1252.	2.0	18

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73	Age-related hearing loss in four Italian genetic isolates: An epidemiological study. International Journal of Audiology, 2009, 48, 465-472.	1.7	17
74	TBL1Y: a new gene involved in syndromic hearing loss. European Journal of Human Genetics, 2019, 27, 466-474.	2.8	17
75	A novel autosomal dominant non-syndromic deafness locus (DFNA48) maps to 12q13-q14 in a large Italian family. Human Genetics, 2003, 112, 319-320.	3.8	15
76	Frequency of hearing loss in a series of rural communities of five developing countries located along the Silk Road. Audiological Medicine, 2011, 9, 135-140.	0.4	15
77	Exome analysis of HIV patients submitted to dendritic cells therapeutic vaccine reveals an association of CNOT1 gene with response to the treatment. Journal of the International AIDS Society, 2014, 17, 18938.	3.0	15
78	Association of a variant in the CHRNA5-A3-B4 gene cluster region to heavy smoking in the Italian population. European Journal of Human Genetics, 2011, 19, 593-596.	2.8	13
79	Genetic determinants for methotrexate response in juvenile idiopathic arthritis. Frontiers in Pharmacology, 2015, 6, 52.	3.5	13
80	GNE-related thrombocytopenia: evidence for a mutational hotspot in the ADP/substrate domain of the GNE bifunctional enzyme. Haematologica, 2022, 107, 750-754.	3.5	11
81	A Novel CRYBB2 Missense Mutation Causing Congenital Autosomal Dominant Cataract in an Italian Family. Ophthalmic Genetics, 2013, 34, 115-117.	1.2	10
82	ACASQ1founder mutation in three Italian families with protein aggregate myopathy and hyperCKaemia. Journal of Medical Genetics, 2015, 52, 617-626.	3.2	10
83	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2015, 781, 32-36.	1.0	10
84	Notch Signaling Regulation in Autoinflammatory Diseases. International Journal of Molecular Sciences, 2020, 21, 8847.	4.1	10
85	Benign hereditary chorea and deletions outside NKX2-1: What's the role of MBIP?. European Journal of Medical Genetics, 2018, 61, 581-584.	1.3	9
86	High prevalence of rare FBLIM1 gene variants in an Italian cohort of patients with Chronic Non-bacterial Osteomyelitis (CNO). Pediatric Rheumatology, 2020, 18, 55.	2.1	9
87	Impact of Methylmercury and Other Heavy Metals Exposure on Neurocognitive Function in Children Aged 7 Years: Study Protocol of the Follow-up. Journal of Epidemiology, 2021, 31, 157-163.	2.4	9
88	Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. Journal of Neurology, 2005, 252, 897-900.	3.6	8
89	The type 2 diabetes associated rs7903146 T allele within TCF7L2 is significantly under-represented in Hereditary Multiple Exostoses: Insights into pathogenesis. Bone, 2015, 72, 123-127.	2.9	8
90	A new locus (DFNA47) for autosomal dominant non-syndromic inherited hearing loss maps to 9p21-22 in a large Italian family. European Journal of Human Genetics, 2003, 11, 121-124.	2.8	7

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91	The Genetic Diagnosis of Ultrarare DEEs: An Ongoing Challenge. Genes, 2022, 13, 500.	2.4	7
92	Dental anomalies as a possible clue of 1p36 deletion syndrome due to germline mosaicism: a case report. BMC Pediatrics, 2020, 20, 201.	1.7	6
93	Otosclerosis: exclusion of linkage to the OTSC1 and OTSC2 loci in four Italian families: Otoesclerosis: exclusiÃ³n de enlaces entre los loci OTSC1 y OTSC2 en cuatro familias italianas. International Journal of Audiology, 2003, 42, 475-480.	1.7	5
94	Plant Antimicrobial Peptides as Potential Tool for Topic Treatment of Hidradenitis Suppurativa. Frontiers in Microbiology, 2021, 12, 795217.	3.5	5
95	Could the <scp>MED13</scp> mutations manifest as a <scp>Kabuki</scp>â€like syndrome?. American Journal of Medical Genetics, Part A, 2021, 185, 584-590.	1.2	3
96	Incidence of Congenital Clubfoot: Preliminary Data from Italian CeDAP Registry. International Journal of Environmental Research and Public Health, 2022, 19, 5406.	2.6	3
97	Juvenile stroke in combined syndrome of hereditary hemorrhagic telangiectasia and juvenile polyposis. Neurological Sciences, 2014, 35, 1315-1318.	1.9	2
98	Carbamazepine-induced thrombocytopenic purpura in a child: Insights from a genomic analysis. Blood Cells, Molecules, and Diseases, 2016, 59, 97-99.	1.4	2
99	Things come in threes: A new complex allele and a novel deletion within the CFTR gene complicate an accurate diagnosis of cystic fibrosis. Molecular Genetics & Genomic Medicine, 2022, 10, e1926.	1.2	2
100	Opioid Resistance Associated with CYP3A4 Hyperactivity and COMT Polymorphism in an Oncological Patient. Pain Medicine, 2018, 19, 638-640.	1.9	1
101	New Tools for Congenital Hyperinsulinism. Clinical Pediatrics, 2021, 60, 336-340.	0.8	1
102	Whole-Genome Methylation Study of Congenital Lung Malformations in Children. Frontiers in Oncology, 2021, 11, 689833.	2.8	1
103	Identification of a New Mutation in RSK2, the Gene for Coffinâ€“Lowry Syndrome (CLS), in Two Related Patients with Mild and Atypical Phenotypes. Brain Sciences, 2021, 11, 1105.	2.3	1
104	Searching for genetic modifiers of Leber's hereditary optic neuropathy penetrance. Mitochondrion, 2012, 12, 572.	3.4	0