

# Adamo Pio d'Adamo

## List of Publications by Year in descending order

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104  
papers

10,715  
citations

66343

42  
h-index

33894

99  
g-index

105  
all docs

105  
docs citations

105  
times ranked

21464  
citing authors

#	ARTICLE	IF	CITATIONS
1	GNE-related thrombocytopenia: evidence for a mutational hotspot in the ADP/substrate domain of the GNE bifunctional enzyme. <i>Haematologica</i> , 2022, 107, 750-754.	3.5	11
2	The Genetic Diagnosis of Ultrarare DEEs: An Ongoing Challenge. <i>Genes</i> , 2022, 13, 500.	2.4	7
3	Things come in threes: A new complex allele and a novel deletion within the <i>CFTR</i> gene complicate an accurate diagnosis of cystic fibrosis. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2022, 10, e1926.	1.2	2
4	Incidence of Congenital Clubfoot: Preliminary Data from Italian CeDAP Registry. <i>International Journal of Environmental Research and Public Health</i> , 2022, 19, 5406.	2.6	3
5	Impact of Methylmercury and Other Heavy Metals Exposure on Neurocognitive Function in Children Aged 7 Years: Study Protocol of the Follow-up. <i>Journal of Epidemiology</i> , 2021, 31, 157-163.	2.4	9
6	Could the <i>MED13</i> mutations manifest as a Kabuki-like syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 584-590.	1.2	3
7	Systematic analysis of factors that improve homologous direct repair (HDR) efficiency in CRISPR/Cas9 technique. <i>PLoS ONE</i> , 2021, 16, e0247603.	2.5	19
8	New Tools for Congenital Hyperinsulinism. <i>Clinical Pediatrics</i> , 2021, 60, 336-340.	0.8	1
9	Whole-Genome Methylation Study of Congenital Lung Malformations in Children. <i>Frontiers in Oncology</i> , 2021, 11, 689833.	2.8	1
10	Identification of a New Mutation in <i>RSK2</i> , the Gene for Coffin-Lowry Syndrome (CLS), in Two Related Patients with Mild and Atypical Phenotypes. <i>Brain Sciences</i> , 2021, 11, 1105.	2.3	1
11	Plant Antimicrobial Peptides as Potential Tool for Topic Treatment of Hidradenitis Suppurativa. <i>Frontiers in Microbiology</i> , 2021, 12, 795217.	3.5	5
12	High prevalence of rare <i>FBLIM1</i> gene variants in an Italian cohort of patients with Chronic Non-bacterial Osteomyelitis (CNO). <i>Pediatric Rheumatology</i> , 2020, 18, 55.	2.1	9
13	Notch Signaling Regulation in Autoinflammatory Diseases. <i>International Journal of Molecular Sciences</i> , 2020, 21, 8847.	4.1	10
14	Dental anomalies as a possible clue of 1p36 deletion syndrome due to germline mosaicism: a case report. <i>BMC Pediatrics</i> , 2020, 20, 201.	1.7	6
15	Heterogeneity in Circulating Tumor Cells: The Relevance of the Stem-Cell Subset. <i>Cancers</i> , 2019, 11, 483.	3.7	107
16	<i>TBL1Y</i> : a new gene involved in syndromic hearing loss. <i>European Journal of Human Genetics</i> , 2019, 27, 466-474.	2.8	17
17	Benign hereditary chorea and deletions outside <i>NKX2-1</i> : What's the role of <i>MBIP</i> ?. <i>European Journal of Medical Genetics</i> , 2018, 61, 581-584.	1.3	9
18	Opioid Resistance Associated with <i>CYP3A4</i> Hyperactivity and <i>COMT</i> Polymorphism in an Oncological Patient. <i>Pain Medicine</i> , 2018, 19, 638-640.	1.9	1

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19	Pharmacogenetics and induction/consolidation therapy toxicities in acute lymphoblastic leukemia patients treated with AIEOP-BFM ALL 2000 protocol. <i>Pharmacogenomics Journal</i> , 2017, 17, 4-10.	2.0	28
20	Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index. <i>Molecular Psychiatry</i> , 2017, 22, 192-201.	7.9	63
21	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. <i>Scientific Reports</i> , 2017, 7, 45040.	3.3	98
22	Carbamazepine-induced thrombocytopenic purpura in a child: Insights from a genomic analysis. <i>Blood Cells, Molecules, and Diseases</i> , 2016, 59, 97-99.	1.4	2
23	Parsing the differences in affected with LHON: genetic versus environmental triggers of disease conversion. <i>Brain</i> , 2016, 139, e17-e17.	7.6	51
24	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. <i>Nature Communications</i> , 2016, 7, 10023.	12.8	412
25	Cigarette toxicity triggers Leber's hereditary optic neuropathy by affecting mtDNA copy number, oxidative phosphorylation and ROS detoxification pathways. <i>Cell Death and Disease</i> , 2015, 6, e2021-e2021.	6.3	107
26	A nonsense mutation of human <i>XRCC4</i> is associated with adult-onset progressive encephalomyopathy. <i>EMBO Molecular Medicine</i> , 2015, 7, 918-929.	6.9	24
27	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. <i>Genetics in Medicine</i> , 2015, 17, 396-399.	2.4	19
28	New genetic loci link adipose and insulin biology to body fat distribution. <i>Nature</i> , 2015, 518, 187-196.	27.8	1,328
29	A <i>CASQ1</i> founder mutation in three Italian families with protein aggregate myopathy and hyperCKaemia. <i>Journal of Medical Genetics</i> , 2015, 52, 617-626.	3.2	10
30	The type 2 diabetes associated rs7903146 T allele within TCF7L2 is significantly under-represented in Hereditary Multiple Exostoses: Insights into pathogenesis. <i>Bone</i> , 2015, 72, 123-127.	2.9	8
31	Genetic determinants for methotrexate response in juvenile idiopathic arthritis. <i>Frontiers in Pharmacology</i> , 2015, 6, 52.	3.5	13
32	Phenotypic and genetic characterization of a family carrying two Xq21.1-21.3 interstitial deletions associated with syndromic hearing loss. <i>Molecular Cytogenetics</i> , 2015, 8, 18.	0.9	19
33	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2015, 781, 32-36.	1.0	10
34	Modulation of Genetic Associations with Serum Urate Levels by Body-Mass-Index in Humans. <i>PLoS ONE</i> , 2015, 10, e0119752.	2.5	64
35	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. <i>PeerJ</i> , 2015, 3, e1252.	2.0	18
36	Association Analysis of Bitter Receptor Genes in Five Isolated Populations Identifies a Significant Correlation between TAS2R43 Variants and Coffee Liking. <i>PLoS ONE</i> , 2014, 9, e92065.	2.5	41

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37	Exome analysis of HIV patients submitted to dendritic cells therapeutic vaccine reveals an association of <i>CNOT1</i> gene with response to the treatment. <i>Journal of the International AIDS Society</i> , 2014, 17, 18938.	3.0	15
38	Autosomal recessive stickler syndrome due to a loss of function mutation in the <i>COL9A3</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 42-47.	1.2	49
39	DNA mismatch repair gene MSH6 implicated in determining age at natural menopause. <i>Human Molecular Genetics</i> , 2014, 23, 2490-2497.	2.9	56
40	Genetic landscape of populations along the Silk Road: admixture and migration patterns. <i>BMC Genetics</i> , 2014, 15, 131.	2.7	24
41	Efficient mitochondrial biogenesis drives incomplete penetrance in Leber's hereditary optic neuropathy. <i>Brain</i> , 2014, 137, 335-353.	7.6	229
42	Genome-wide association study of sexual maturation in males and females highlights a role for body mass and menarche loci in male puberty. <i>Human Molecular Genetics</i> , 2014, 23, 4452-4464.	2.9	82
43	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. <i>Nature</i> , 2014, 514, 92-97.	27.8	548
44	Next generation sequencing in nonsyndromic intellectual disability: From a negative molecular karyotype to a possible causative mutation detection. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 170-176.	1.2	34
45	Juvenile stroke in combined syndrome of hereditary hemorrhagic telangiectasia and juvenile polyposis. <i>Neurological Sciences</i> , 2014, 35, 1315-1318.	1.9	2
46	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. <i>Nature Genetics</i> , 2014, 46, 826-836.	21.4	281
47	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. <i>Gene</i> , 2014, 534, 236-239.	2.2	50
48	Adult-onset Alexander disease, associated with a mutation in an alternative GFAP transcript, may be phenotypically modulated by a non-neutral HDAC6 variant. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 66.	2.7	21
49	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. <i>European Journal of Human Genetics</i> , 2013, 21, 659-665.	2.8	64
50	Genome-wide association and longitudinal analyses reveal genetic loci linking pubertal height growth, pubertal timing and childhood adiposity. <i>Human Molecular Genetics</i> , 2013, 22, 2735-2747.	2.9	188
51	A Novel CRYBB2 Missense Mutation Causing Congenital Autosomal Dominant Cataract in an Italian Family. <i>Ophthalmic Genetics</i> , 2013, 34, 115-117.	1.2	10
52	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. <i>Nature Genetics</i> , 2013, 45, 145-154.	21.4	675
53	Maps of open chromatin highlight cell type-restricted patterns of regulatory sequence variation at hematological trait loci. <i>Genome Research</i> , 2013, 23, 1130-1141.	5.5	34
54	Meta-Analysis of Genome-Wide Association Studies Identifies Six New Loci for Serum Calcium Concentrations. <i>PLoS Genetics</i> , 2013, 9, e1003796.	3.5	142

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55	Evidence of Inbreeding Depression on Human Height. <i>PLoS Genetics</i> , 2012, 8, e1002655.	3.5	79
56	A GWAS sequence variant for platelet volume marks an alternative DNMT3 promoter in megakaryocytes near a MEIS1 binding site. <i>Blood</i> , 2012, 120, 4859-4868.	1.4	44
57	Seventy-five genetic loci influencing the human red blood cell. <i>Nature</i> , 2012, 492, 369-375.	27.8	320
58	Genome-wide meta-analysis of common variant differences between men and women. <i>Human Molecular Genetics</i> , 2012, 21, 4805-4815.	2.9	33
59	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. <i>Nature Genetics</i> , 2012, 44, 260-268.	21.4	303
60	Searching for genetic modifiers of Leber's hereditary optic neuropathy penetrance. <i>Mitochondrion</i> , 2012, 12, 572.	3.4	0
61	Meta-analysis identifies multiple loci associated with kidney function-related traits in east Asian populations. <i>Nature Genetics</i> , 2012, 44, 904-909.	21.4	254
62	Influence of age, sex and ethnicity on platelet count in five Italian geographic isolates: mild thrombocytopenia may be physiological. <i>British Journal of Haematology</i> , 2012, 157, 384-387.	2.5	33
63	Genomic profiling by whole-genome single nucleotide polymorphism arrays in Wilms tumor and association with relapse. <i>Genes Chromosomes and Cancer</i> , 2012, 51, 644-653.	2.8	28
64	Does the 1.5-Mb microduplication in chromosome band Xp22.31 have a pathogenetic role? New contribution and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 461-464.	1.2	21
65	Somatic mosaic IDH1 and IDH2 mutations are associated with enchondroma and spindle cell hemangioma in Ollier disease and Maffucci syndrome. <i>Nature Genetics</i> , 2011, 43, 1256-1261.	21.4	488
66	New gene functions in megakaryopoiesis and platelet formation. <i>Nature</i> , 2011, 480, 201-208.	27.8	401
67	Mutations in TTC19 cause mitochondrial complex III deficiency and neurological impairment in humans and flies. <i>Nature Genetics</i> , 2011, 43, 259-263.	21.4	148
68	Association of a variant in the CHRNA5-A3-B4 gene cluster region to heavy smoking in the Italian population. <i>European Journal of Human Genetics</i> , 2011, 19, 593-596.	2.8	13
69	Frequency of hearing loss in a series of rural communities of five developing countries located along the Silk Road. <i>Audiological Medicine</i> , 2011, 9, 135-140.	0.4	15
70	Hearing function and thresholds: a genome-wide association study in European isolated populations identifies new loci and pathways. <i>Journal of Medical Genetics</i> , 2011, 48, 369-374.	3.2	71
71	Phospholipase C- $\beta$ 3 Is a Key Modulator of IL-8 Expression in Cystic Fibrosis Bronchial Epithelial Cells. <i>Journal of Immunology</i> , 2011, 186, 4946-4958.	0.8	34
72	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. <i>Nature Genetics</i> , 2011, 43, 1005-1011.	21.4	403

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73	Severe X-Linked Mitochondrial Encephalomyopathy Associated with a Mutation in Apoptosis-Inducing Factor. <i>American Journal of Human Genetics</i> , 2010, 86, 639-649.	6.2	199
74	Metabonomics and population studies: age-related amino acids excretion and inferring networks through the study of urine samples in two Italian isolated populations. <i>Amino Acids</i> , 2010, 38, 65-73.	2.7	18
75	Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies. <i>Nature Genetics</i> , 2010, 42, 1077-1085.	21.4	445
76	Genetic Structure of Europeans: A View from the North-East. <i>PLoS ONE</i> , 2009, 4, e5472.	2.5	279
77	Age-related hearing loss in four Italian genetic isolates: An epidemiological study. <i>International Journal of Audiology</i> , 2009, 48, 465-472.	1.7	17
78	Detection of Epidermal Thickening in GJB2 Carriers with Epidermal US. <i>Radiology</i> , 2009, 251, 280-286.	7.3	23
79	Does epidermal thickening explain GJB2 high carrier frequency and heterozygote advantage?. <i>European Journal of Human Genetics</i> , 2009, 17, 284-286.	2.8	35
80	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. <i>Nature Genetics</i> , 2009, 41, 654-656.	21.4	233
81	Genetic Variation in Taste Sensitivity to 6-Propylthiouracil and Its Relationship to Taste Perception and Food Selection. <i>Annals of the New York Academy of Sciences</i> , 2009, 1170, 126-139.	3.8	112
82	Dominant inheritance of a novel integrin $\beta 3$ mutation associated with a hereditary macrothrombocytopenia and platelet dysfunction in two Italian families. <i>Haematologica</i> , 2009, 94, 663-669.	3.5	64
83	Cohen syndrome resulting from a novel large intragenic <i>COH1</i> deletion segregating in an isolated Greek island population. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2221-2226.	1.2	26
84	Severe Infantile Encephalomyopathy Caused by a Mutation in COX6B1, a Nucleus-Encoded Subunit of Cytochrome C Oxidase. <i>American Journal of Human Genetics</i> , 2008, 82, 1281-1289.	6.2	165
85	FASTKD2 Nonsense Mutation in an Infantile Mitochondrial Encephalomyopathy Associated with Cytochrome C Oxidase Deficiency. <i>American Journal of Human Genetics</i> , 2008, 83, 415-423.	6.2	107
86	Variation in the Bitter-taste Receptor Gene <i>TAS2R38</i> , and Adiposity in a Genetically Isolated Population in Southern Italy. <i>Obesity</i> , 2008, 16, 2289-2295.	3.0	165
87	Variation of hemoglobin levels in normal Italian populations from genetic isolates. <i>Haematologica</i> , 2008, 93, 1372-1375.	3.5	25
88	MRX87 family with Aristaless Xdup24bp mutation and implication for polyAlanine expansions. <i>BMC Medical Genetics</i> , 2007, 8, 25.	2.1	20
89	MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. <i>Nature Genetics</i> , 2006, 38, 570-575.	21.4	380
90	Identification of a Novel Mutation in the Myosin VIIA Motor Domain in a Family with Autosomal Dominant Hearing Loss (DFNA11). <i>Audiology and Neuro-Otology</i> , 2006, 11, 157-164.	1.3	25

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91	A novel GJA1 mutation causes oculodentodigital dysplasia without syndactyly. American Journal of Medical Genetics, Part A, 2005, 133A, 58-60.	1.2	55
92	Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. Journal of Neurology, 2005, 252, 897-900.	3.6	8
93	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
94	A second locus mapping to 2q35â€“36 for familial pseudohyperkalaemia. European Journal of Human Genetics, 2004, 12, 1073-1076.	2.8	25
95	Narrowing of the critical region in autosomal recessive spastic paraplegia linked to the SPG5 locus. Neurogenetics, 2004, 5, 49-54.	1.4	21
96	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
97	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
98	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
99	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
100	Juvenile hemochromatosis locus maps to chromosome 1q in a French Canadian population. European Journal of Human Genetics, 2003, 11, 585-589.	2.8	28
101	Otosclerosis: exclusion of linkage to the OTSC1 and OTSC2 loci in four Italian families: Otoesclerosis: exclusi3n de enlaces entre los loci OTSC1 y OTSC2 en cuatro familias italianas. International Journal of Audiology, 2003, 42, 475-480.	1.7	5
102	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
103	Cystinuria type I: Identification of eight new mutations in SLC3A1. Kidney International, 2001, 59, 1250-1256.	5.2	24
104	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