Paolo Gasparini

List of Publications by Year
in descending order

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315 papers	31,070 citations	7568 77 h-index	6130 159 g-index
335 all docs	335 docs citations	335 times ranked	40264 citing authors

#	Article	IF	Citations
1	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	21.4	2,421
2	Genome-wide association study identifies 74 loci associated with educational attainment. Nature, 2016, 533, 539-542.	27.8	1,204
3	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. Nature Genetics, 2018, 50, 1412-1425.	21.4	924
4	The putative forkhead transcription factor FOXL2 is mutated in blepharophimosis/ptosis/epicanthus inversus syndrome. Nature Genetics, 2001, 27, 159-166.	21.4	886
5	The gene TFR2 is mutated in a new type of haemochromatosis mapping to 7q22. Nature Genetics, 2000, 25, 14-15.	21.4	751
6	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. Nature Genetics, 2013, 45, 145-154.	21.4	675
7	Connexin-26 mutations in sporadic and inherited sensorineural deafness. Lancet, The, 1998, 351, 394-398.	13.7	610
8	A General Approach for Haplotype Phasing across the Full Spectrum of Relatedness. PLoS Genetics, 2014, 10, e1004234.	3.5	553
9	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	21.4	549
10	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. Nature, 2014, 514, 92-97.	27.8	548
11	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	27.8	544
12	Genome-wide association analyses of risk tolerance and risky behaviors in over 1 million individuals identify hundreds of loci and shared genetic influences. Nature Genetics, 2019, 51, 245-257.	21.4	536
13	Dating the Origin of the CCR5-Î”32 AIDS-Resistance Allele by the Coalescence of Haplotypes. American Journal of Human Genetics, 1998, 62, 1507-1515.	6.2	507
14	Mutations of SURF-1 in Leigh Disease Associated with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 1609-1621.	6.2	504
15	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. Nature Genetics, 2017, 49, 403-415.	21.4	492
16	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
17	Autosomal-dominant hemochrom-atosis is associated with a mutation in the ferroportin (SLC11A3) gene. Journal of Clinical Investigation, 2001, 108, 619-623.	8.2	429
18	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. Nature Communications, 2016, 7, 10023.	12.8	412

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19	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	21.4	403
20	New gene functions in megakaryopoiesis and platelet formation. Nature, 2011, 480, 201-208.	27.8	401
21	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
22	Cystinuria caused by mutations in rBAT, a gene involved in the transport of cystine. Nature Genetics, 1994, 6, 420-425.	21.4	366
23	High carrier frequency of the 35delG deafness mutation in European populations. European Journal of Human Genetics, 2000, 8, 19-23.	2.8	363
24	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	21.4	357
25	Mutations in GJB6 cause nonsyndromic autosomal dominant deafness at DFNA3 locus. Nature Genetics, 1999, 23, 16-18.	21.4	345
26	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	27.8	320
27	Reelin gene alleles and haplotypes as a factor predisposing to autistic disorder. Molecular Psychiatry, 2001, 6, 150-159.	7.9	314
28	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
29	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	21.4	286
30	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. Nature Genetics, 2016, 48, 1462-1472.	21.4	284
31	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	21.4	281
32	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. Nature Genetics, 1999, 23, 52-57.	21.4	280
33	Genetic Structure of Europeans: A View from the Northâ€“East. PLoS ONE, 2009, 4, e5472.	2.5	279
34	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. American Journal of Human Genetics, 2003, 73, 1452-1458.	6.2	269
35	Hereditary Hemochromatosis in Adults without Pathogenic Mutations in the Hemochromatosis Gene. New England Journal of Medicine, 1999, 341, 725-732.	27.0	257
36	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. Nature Genetics, 2019, 51, 1459-1474.	21.4	251

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37	Comparison between SLC3A1 and SLC7A9 Cystinuria Patients and Carriers. Journal of the American Society of Nephrology: JASN, 2002, 13, 2547-2553.	6.1	234
38	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
39	New mutations inactivating transferrin receptor 2 in hemochromatosis type 3. Blood, 2001, 97, 2555-2560.	1.4	232
40	Juvenile Hemochromatosis Locus Maps to Chromosome 1q. American Journal of Human Genetics, 1999, 64, 1388-1393.	6.2	229
41	Population genetic differentiation of height and body mass index across Europe. Nature Genetics, 2015, 47, 1357-1362.	21.4	227
42	Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals. Nature Genetics, 2022, 54, 437-449.	21.4	215
43	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
44	Molecular genetics of hearing impairment due to mutations in gap junction genes encoding beta connexins. Human Mutation, 2000, 16, 190-202.	2.5	197
45	Mutations in a Novel Gene with Transmembrane Domains Underlie Usher Syndrome Type 3. American Journal of Human Genetics, 2001, 69, 673-684.	6.2	195
46	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. Nature Genetics, 2015, 47, 1272-1281.	21.4	193
47	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
48	Age- And Sex-Related Variations in Platelet Count in Italy: A Proposal of Reference Ranges Based on 40987 Subjects' Data. PLoS ONE, 2013, 8, e54289.	2.5	190
49	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	27.8	173
50	Spectrum of hemojuvelin gene mutations in 1q-linked juvenile hemochromatosis. Blood, 2004, 103, 4317-4321.	1.4	167
51	Genome-Wide Association and Functional Follow-Up Reveals New Loci for Kidney Function. PLoS Genetics, 2012, 8, e1002584.	3.5	166
52	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
53	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
54	Nonmuscle Myosin Heavy-Chain Gene MYH14 Is Expressed in Cochlea and Mutated in Patients Affected by Autosomal Dominant Hearing Impairment (DFNA4). American Journal of Human Genetics, 2004, 74, 770-776.	6.2	150

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55	Natural history of juvenile haemochromatosis. British Journal of Haematology, 2002, 117, 973-979.	2.5	145
56	Meta-Analysis of Genome-Wide Association Studies Identifies Six New Loci for Serum Calcium Concentrations. PLoS Genetics, 2013, 9, e1003796.	3.5	142
57	Stomatocytic haemolysis and macrothrombocytopenia (Mediterranean) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50 667 Td (stoma British Journal of Haematology, 2005, 130, 297-309.	2.5	138
58	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. American Journal of Human Genetics, 2017, 100, 865-884.	6.2	131
59	Exploring influences on food choice in a large population sample: The Italian Taste project. Food Quality and Preference, 2017, 59, 123-140.	4.6	128
60	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
61	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. Hypertension, 2017, 70, .	2.7	123
62	A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure. American Journal of Human Genetics, 2018, 102, 375-400.	6.2	123
63	A functional study of plasma-membrane calcium-pump isoform 2 mutants causing digenic deafness. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 1516-1521.	7.1	116
64	52 Genetic Loci Influencing MyocardialÂMass. Journal of the American College of Cardiology, 2016, 68, 1435-1448.	2.8	113
65	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
66	Multi-ancestry genome-wide geneâ€“smoking interaction study of 387,272 individuals identifies new loci associated with serum lipids. Nature Genetics, 2019, 51, 636-648.	21.4	112
67	Mitochondrial DNA mutations in patients with postlingual, nonsyndromic hearing impairment. European Journal of Human Genetics, 2005, 13, 26-33.	2.8	110
68	Genetic variants linked to education predict longevity. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 13366-13371.	7.1	110
69	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
70	Heterogeneity in Circulating Tumor Cells: The Relevance of the Stem-Cell Subset. Cancers, 2019, 11, 483.	3.7	107
71	Multiple Loci Are Associated with White Blood Cell Phenotypes. PLoS Genetics, 2011, 7, e1002113.	3.5	106
72	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. Scientific Reports, 2017, 7, 45040.	3.3	98

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73	Multiple Mutations ofMYO1A, a Cochlear-Expressed Gene, in Sensorineural Hearing Loss. American Journal of Human Genetics, 2003, 72, 1571-1577.	6.2	97
74	Insights into the Binding of Phenyltiocarbamide (PTC) Agonist to Its Target Human TAS2R38 Bitter Receptor. PLoS ONE, 2010, 5, e12394.	2.5	97
75	Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries. PLoS ONE, 2018, 13, e0198166.	2.5	94
76	Hearing loss: frequency and functional studies of the most common connexin26 alleles. Biochemical and Biophysical Research Communications, 2002, 296, 685-691.	2.1	89
77	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469.	21.4	89
78	Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability. Nature Genetics, 2018, 50, 652-656.	21.4	86
79	Common Variants in UMOD Associate with Urinary Uromodulin Levels. Journal of the American Society of Nephrology: JASN, 2014, 25, 1869-1882.	6.1	85
80	Multiancestry Genome-Wide Association Study of Lipid Levels Incorporating Gene-Alcohol Interactions. American Journal of Epidemiology, 2019, 188, 1033-1054.	3.4	85
81	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	12.8	84
82	Genetic analysis in Italian families with inflammatory bowel disease supports linkage to the IBD1 locus â€“ A GISC study. European Journal of Human Genetics, 1999, 7, 567-573.	2.8	81
83	Screening hepcidin for mutations in juvenile hemochromatosis: identification of a new mutation (C70R). Blood, 2004, 103, 2407-2409.	1.4	80
84	Evidence of Inbreeding Depression on Human Height. PLoS Genetics, 2012, 8, e1002655.	3.5	79
85	Existence of a genetic risk factor on chromosome 5q in Italian Coeliac Disease families. Annals of Human Genetics, 2001, 65, 35-41.	0.8	78
86	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	2.8	71
87	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
88	PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. Nature Communications, 2018, 9, 2904.	12.8	71
89	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. Human Genetics, 1992, 89, 653-658.	3.8	69
90	Global diversity in the TAS2R38 bitter taste receptor: revisiting a classic evolutionary PROPosal. Scientific Reports, 2016, 6, 25506.	3.3	69

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91	Coarse-Grained/Molecular Mechanics of the TAS2R38 Bitter Taste Receptor: Experimentally-Validated Detailed Structural Prediction of Agonist Binding. PLoS ONE, 2013, 8, e64675.	2.5	67
92	Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps. Nature Genetics, 2016, 48, 1303-1312.	21.4	66
93	Osteoporosis in beta-thalassaemia major patients: analysis of the genetic background. British Journal of Haematology, 2000, 111, 461-466.	2.5	66
94	Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients. Journal of Molecular Medicine, 2002, 80, 124-131.	3.9	65
95	Integration of genome-wide association studies with biological knowledge identifies six novel genes related to kidney function. Human Molecular Genetics, 2012, 21, 5329-5343.	2.9	64
96	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
97	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. Nature Communications, 2017, 8, 15927.	12.8	64
98	Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity. Nature Communications, 2019, 10, 376.	12.8	64
99	Modulation of Genetic Associations with Serum Urate Levels by Body-Mass-Index in Humans. PLoS ONE, 2015, 10, e0119752.	2.5	64
100	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
101	Mitochondrial 12S rRNA gene mutations affect RNA secondary structure and lead to variable penetrance in hearing impairment. Biochemical and Biophysical Research Communications, 2006, 341, 950-957.	2.1	61
102	Espin gene (ESPN) mutations associated with autosomal dominant hearing loss cause defects in microvillar elongation or organisation. Journal of Medical Genetics, 2005, 43, 157-161.	3.2	59
103	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. Nature Communications, 2020, 11, 2542.	12.8	59
104	A meta-analysis of genome-wide association studies of the electrocardiographic early repolarization pattern. Heart Rhythm, 2012, 9, 1627-1634.	0.7	58
105	Relationship between clone metrics and clinical outcome in clonal cytopenia. Blood, 2021, 138, 965-976.	1.4	58
106	Factors Influencing the Phenotypic Characterization of the Oral Marker, PROP. Nutrients, 2017, 9, 1275.	4.1	57
107	Analysis of the complete coding region of the CFTR gene in a cohort of CF patients from North-Eastern Italy: identification of 90% of the mutations. Human Genetics, 1995, 95, 397-402.	3.8	56
108	DNA mismatch repair gene MSH6 implicated in determining age at natural menopause. Human Molecular Genetics, 2014, 23, 2490-2497.	2.9	56

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109	Regulatory evaluation of Glybera in Europe â€" two committees, one mission. Nature Reviews Drug Discovery, 2013, 12, 719-719.	46.4	54
110	Autosomal dominant reticuloendothelial iron overload (HFE type 4) due to a new missense mutation in the FERROPORTIN 1 gene (SLC11A3) in a large French-Canadian family. Haematologica, 2003, 88, 824-6.	3.5	54
111	Estrogen-related receptor gamma and hearing function: evidence of a role in humans and mice. Neurobiology of Aging, 2013, 34, 2077.e1-2077.e9.	3.1	53
112	First all-in-one diagnostic tool for DNA intelligence: genome-wide inference of biogeographic ancestry, appearance, relatedness, and sex with the Identitas v1 Forensic Chip. International Journal of Legal Medicine, 2013, 127, 559-572.	2.2	51
113	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. Gene, 2014, 534, 236-239.	2.2	50
114	Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway. Redox Biology, 2018, 19, 301-317.	9.0	50
115	Linkage Study and Exome Sequencing Identify a BDP1 Mutation Associated with Hereditary Hearing Loss. PLoS ONE, 2013, 8, e80323.	2.5	50
116	Fixed drug eruptions with feprazone are linked to HLA-B22. Journal of the American Academy of Dermatology, 1997, 36, 782-784.	1.2	49
117	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
118	Hereditary hearing loss: a 96 gene targeted sequencing protocol reveals novel alleles in a series of Italian and Qatari patients. Gene, 2014, 542, 209-216.	2.2	48
119	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
120	Exome-chip meta-analysis identifies novel loci associated with cardiac conduction, including ADAMTS6. Genome Biology, 2018, 19, 87.	8.8	47
121	Genetics of Food Preferences: A First View from Silk Road Populations. Journal of Food Science, 2012, 77, S413-8.	3.1	45
122	Transforming Growth Factor-Î²1 Gene Polymorphism, Bone Turnover, and Bone Mass in Italian Postmenopausal Women. Journal of Bone and Mineral Research, 2010, 15, 634-639.	2.8	44
123	Genetic evidence for an origin of the Armenians from Bronze Age mixing of multiple populations. European Journal of Human Genetics, 2016, 24, 931-936.	2.8	44
124	A novel mutation in the mitochondrial tRNAVal gene associated with a complex neurological presentation. Annals of Neurology, 1998, 43, 98-101.	5.3	42
125	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
126	Linkage analysis of 6p21 polymorphic markers and the hereditary hemochromatosis: localization of the gene centromeric to HLA-F. Human Molecular Genetics, 1993, 2, 571-576.	2.9	40

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127	GOAL: automated Gene Ontology analysis of expression profiles. Nucleic Acids Research, 2004, 32, W492-W499.	14.5	40
128	Vezatin, an integral membrane protein of adherens junctions, is required for the sound resilience of cochlear hair cells. EMBO Molecular Medicine, 2009, 1, 125-138.	6.9	39
129	SOS2 and ACP1 Loci Identified through Large-Scale Exome Chip Analysis Regulate Kidney Development and Function. Journal of the American Society of Nephrology: JASN, 2017, 28, 981-994.	6.1	39
130	Expression and Replication Studies to Identify New Candidate Genes Involved in Normal Hearing Function. PLoS ONE, 2014, 9, e85352.	2.5	39
131	Polymorphism in intron 4 of HFE does not compromise haemochromatosis mutation results. Nature Genetics, 1999, 23, 271-271.	21.4	38
132	A novel deletion mutation involving TMEM38B in a patient with autosomal recessive osteogenesis imperfecta. Gene, 2014, 545, 290-292.	2.2	38
133	Characterization of 14 novel deletions underlying Rubinsteinâ€“Taybi syndrome: an update of the CREBBP deletion repertoire. Human Genetics, 2015, 134, 613-626.	3.8	38
134	Mutations in L-type amino acid transporter-2 support SLC7A8 as a novel gene involved in age-related hearing loss. ELife, 2018, 7, .	6.0	38
135	Splice-site mutation in thePDS gene may result in intrafamilial variability for deafness in Pendred syndrome. , 1999, 14, 520-526.		37
136	Heritability and Demographic Analyses in the Large Isolated Population of Val Borbera Suggest Advantages in Mapping Complex Traits Genes. PLoS ONE, 2009, 4, e7554.	2.5	37
137	Genome-wide association analysis on normal hearing function identifiesPCDH20andSLC28A3as candidates for hearing function and loss. Human Molecular Genetics, 2015, 24, 5655-5664.	2.9	37
138	Understanding the role of personality and alexithymia in food preferences and PROP taste perception. Physiology and Behavior, 2016, 157, 72-78.	2.1	37
139	Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. Science Advances, 2021, 7, .	10.3	36
140	GABA (Î³-Amino-Butyric Acid) Neurotransmission: Identification and Fine Mapping of the Human GABABReceptor Gene. Biochemical and Biophysical Research Communications, 1998, 250, 240-245.	2.1	35
141	Does epidermal thickening explain GJB2 high carrier frequency and heterozygote advantage?. European Journal of Human Genetics, 2009, 17, 284-286.	2.8	35
142	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
143	A Population-Based Approach to Study the Impact of PROP Perception on Food Liking in Populations along the Silk Road. PLoS ONE, 2014, 9, e91716.	2.5	34
144	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

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145	Genome-Wide Meta-Analysis Unravels Interactions between Magnesium Homeostasis and Metabolic Phenotypes. Journal of the American Society of Nephrology: JASN, 2018, 29, 335-348.	6.1	34
146	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
147	Common Variants in Mendelian Kidney Disease Genes and Their Association with Renal Function. Journal of the American Society of Nephrology: JASN, 2013, 24, 2105-2117.	6.1	33
148	Genome-wide association meta-analysis identifies five novel loci for age-related hearing impairment. Scientific Reports, 2019, 9, 15192.	3.3	32
149	Genetic homogeneity of lysinuric protein intolerance. European Journal of Human Genetics, 1998, 6, 612-615.	2.8	31
150	D184E mutation in aquaporin-4 gene impairs water permeability and links to deafness. Neuroscience, 2011, 197, 80-88.	2.3	31
151	A multi-ancestry genome-wide study incorporating geneâ€“smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. Human Molecular Genetics, 2019, 28, 2615-2633.	2.9	31
152	Assignment of the gene responsible for cystinuria (rBAT) and of markers D2S119 and D2S177 to 2p16 by fluorescence in situ hybridization. Human Genetics, 1995, 95, 633-6.	3.8	30
153	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
154	Salt-inducible kinase 3, SIK3, is a new gene associated with hearing. Human Molecular Genetics, 2014, 23, 6407-6418.	2.9	30
155	An expression atlas of connexin genes in the mouse. Genomics, 2004, 83, 812-820.	2.9	29
156	Multicohort analysis of the maternal age effect on recombination. Nature Communications, 2015, 6, 7846.	12.8	29
157	Genome-wide association meta-analysis of 30,000 samples identifies seven novel loci for quantitative ECG traits. European Journal of Human Genetics, 2019, 27, 952-962.	2.8	29
158	A birdâ€™s-eye view of Italian genomic variation through whole-genome sequencing. European Journal of Human Genetics, 2020, 28, 435-444.	2.8	29
159	Molecular Diagnosis of Usher Syndrome: Application of Two Different Next Generation Sequencing-Based Procedures. PLoS ONE, 2012, 7, e43799.	2.5	29
160	Juvenile hemochromatosis locus maps to chromosome 1q in a French Canadian population. European Journal of Human Genetics, 2003, 11, 585-589.	2.8	28
161	Are MYO1C and MYO1F associated with hearing loss?. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 27-32.	3.8	28
162	Increased Rate of Deleterious Variants in Long Runs of Homozygosity of an Inbred Population from Qatar. Human Heredity, 2015, 79, 14-19.	0.8	28

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163	Rapid sizing of polymorphic microsatellite markers by capillary array electrophoresis. Journal of Chromatography A, 1997, 781, 295-305.	3.7	27
164	Common variants in CLDN14 are associated with differential excretion of magnesium over calcium in urine. Pflugers Archiv European Journal of Physiology, 2017, 469, 91-103.	2.8	27
165	Molecular epidemiology of Usher syndrome in Italy. Molecular Vision, 2011, 17, 1662-8.	1.1	27
166	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
167	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
168	DHPLC analysis of the MECP2 gene in Italian Rett patients. Human Mutation, 2001, 18, 132-140.	2.5	25
169	A second locus mapping to 2q35â€“36 for familial pseudohyperkalaemia. European Journal of Human Genetics, 2004, 12, 1073-1076.	2.8	25
170	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
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