Paolo Gasparini

List of Publications by Year
in descending order

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315	31,070	7568 77	6131 159
papers	citations	h-index	g-index

335	335	335	40264
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genome-wide association studies on Northern Italy isolated populations provide further support concerning genetic susceptibility for major depressive disorder. World Journal of Biological Psychiatry, 2023, 24, 135-148.	2.6	1
2	Combined influence of TAS2R38 genotype and PROP phenotype on the intensity of basic tastes, astringency and pungency in the Italian taste project. Food Quality and Preference, 2022, 95, 104361.	4.6	15
3	Eating disinhibition and food liking are influenced by variants in CAV1 (caveolin 1) gene. Food Quality and Preference, 2022, 96, 104447.	4.6	1
4	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
5	Sensory Capacities and Eating Behavior: Intriguing Results from a Large Cohort of Italian Individuals. Foods, 2022, 11, 735.	4.3	2
6	Genetic variations associated with the soapy flavor perception in Gorgonzola PDO cheese. Food Quality and Preference, 2022, 99, 104569.	4.6	1
7	Genetic Dissection of Temperament Personality Traits in Italian Isolates. Genes, 2022, 13, 4.	2.4	2
8	Large-scale GWAS of food liking reveals genetic determinants and genetic correlations with distinct neurophysiological traits. Nature Communications, 2022, 13, 2743.	12.8	22
9	TTC5 syndrome: Clinical and molecular spectrum of a severe and recognizable condition. American Journal of Medical Genetics, Part A, 2022, 188, 2652-2665.	1.2	4
10	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. Communications Biology, 2022, 5, .	4.4	17
11	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
12	Differences in taste and smell perception between type 2 diabetes mellitus patients and healthy controls. Nutrition, Metabolism and Cardiovascular Diseases, 2021, 31, 193-200.	2.6	19
13	Hearing loss. , 2021, , 305-322.		2
14	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
15	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
16	Runs of homozygosity are associated with staging of periodontitis in isolated populations. Human Molecular Genetics, 2021, 30, 1154-1159.	2.9	3
17	Natural human knockouts and Mendelian disorders: deep phenotyping in Italian isolates. European Journal of Human Genetics, 2021, 29, 1272-1281.	2.8	6
18	Systematic analysis of factors that improve homologous direct repair (HDR) efficiency in CRISPR/Cas9 technique. PLoS ONE, 2021, 16, e0247603.	2.5	19

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19	The Role of Knockout Olfactory Receptor Genes in Odor Discrimination. Genes, 2021, 12, 631.	2.4	3
20	Dietary Macronutrient Composition in Relation to Circulating HDL and Non-HDL Cholesterol: A Federated Individual-Level Analysis of Cross-Sectional Data from Adolescents and Adults in 8 European Studies. Journal of Nutrition, 2021, 151, 2317-2329.	2.9	8
21	Variants in USP48 encoding ubiquitin hydrolase are associated with autosomal dominant non-syndromic hereditary hearing loss. Human Molecular Genetics, 2021, 30, 1785-1796.	2.9	6
22	Relationship between clone metrics and clinical outcome in clonal cytopenia. Blood, 2021, 138, 965-976.	1.4	58
23	Hearing Function: Identification of New Candidate Genes Further Explaining the Complexity of This Sensory Ability. Genes, 2021, 12, 1228.	2.4	1
24	Genetics, odor perception and food liking: The intriguing role of cinnamon. Food Quality and Preference, 2021, 93, 104277.	4.6	2
25	Taste perception and expression in stomach of bitter taste receptor tas2r38 in obese and lean subjects. Appetite, 2021, 166, 105595.	3.7	7
26	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
27	Primary Ciliary Dyskinesia: The Impact of Taste Receptor (TAS2R38) Gene Polymorphisms on Disease Outcome and Severity. International Archives of Allergy and Immunology, 2020, 181, 727-731.	2.1	8
28	Molecular testing for the study of non-syndromic hearing loss. Hearing, Balance and Communication, 2020, 18, 270-277.	0.4	5
29	Gene-educational attainment interactions in a multi-ancestry genome-wide meta-analysis identify novel blood pressure loci. Molecular Psychiatry, 2020, 26, 2111-2125.	7.9	17
30	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. Nature Communications, 2020, 11, 2542.	12.8	59
31	New age-related hearing loss candidate genes in humans: an ongoing challenge. Gene, 2020, 742, 144561.	2.2	9
32	Deleterious variants in genes associated with bone mineral density are linked to susceptibility to periodontitis development. Meta Gene, 2020, 24, 100670.	0.6	2
33	A population-based approach for gene prioritization in understanding complex traits. Human Genetics, 2020, 139, 647-655.	3.8	7
34	Mutations in PLS1 , encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. Human Mutation, 2019, 40, 2286-2295.	2.5	23
35	Next generation sequencing study in a cohort of Italian patients with syndromic hearing loss. Hearing Research, 2019, 381, 107769.	2.0	7
36	A Brief Review of Genetic Approaches to the Study of Food Preferences: Current Knowledge and Future Directions. Nutrients, 2019, 11, 1735.	4.1	20

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37	Genome-wide association meta-analysis identifies five novel loci for age-related hearing impairment. Scientific Reports, 2019, 9, 15192.	3.3	32
38	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	12.8	84
39	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. Nature Genetics, 2019, 51, 1459-1474.	21.4	251
40	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
41	Multiancestry Genome-Wide Association Study of Lipid Levels Incorporating Gene-Alcohol Interactions. American Journal of Epidemiology, 2019, 188, 1033-1054.	3.4	85
42	Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity. Nature Communications, 2019, 10, 376.	12.8	64
43	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	21.4	549
44	TAS2R38 bitter taste genotype is associated with complementary feeding behavior in infants. Genes and Nutrition, 2019, 14, 13.	2.5	11
45	Next Generation Sequencing and Animal Models Reveal SLC9A3R1 as a New Gene Involved in Human Age-Related Hearing Loss. Frontiers in Genetics, 2019, 10, 142.	2.3	11
46	Heterogeneity in Circulating Tumor Cells: The Relevance of the Stem-Cell Subset. Cancers, 2019, 11, 483.	3.7	107
47	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
48	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
49	Two further patients with Warsaw breakage syndrome. Is a mild phenotype possible?. Molecular Genetics & Genomic Medicine, 2019, 7, e639.	1.2	10
50	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469.	21.4	89
51	Factors associated with food liking and their relationship with metabolic traits in Italian cohorts. Food Quality and Preference, 2019, 75, 64-70.	4.6	9
52	Next-generation sequencing identified SPATC1L as a possible candidate gene for both early-onset and age-related hearing loss. European Journal of Human Genetics, 2019, 27, 70-79.	2.8	22
53	Investigation of the link between PROP taste perception and vegetables consumption using FAOSTAT data. International Journal of Food Sciences and Nutrition, 2019, 70, 484-490.	2.8	6
54	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

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55	TBL1Y: a new gene involved in syndromic hearing loss. European Journal of Human Genetics, 2019, 27, 466-474.	2.8	17
56	IBS clinical management in Italy: The AIGO survey. Digestive and Liver Disease, 2019, 51, 782-789.	0.9	19
57	Association of <scp>LTA</scp> gene haploblock with periodontal disease in Italian adults. Journal of Periodontal Research, 2019, 54, 128-133.	2.7	3
58	Joint Data Analysis in Nutritional Epidemiology: Identification of Observational Studies and Minimal Requirements. Journal of Nutrition, 2018, 148, 285-297.	2.9	13
59	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
60	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
61	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
62	19p13 microduplications encompassing NFIX are responsible for intellectual disability, short stature and small head circumference. European Journal of Human Genetics, 2018, 26, 85-93.	2.8	7
63	Genetic Landscape of Slovenians: Past Admixture and Natural Selection Pattern. Frontiers in Genetics, 2018, 9, 551.	2.3	8
64	Genomic Studies in a Large Cohort of Hearing Impaired Italian Patients Revealed Several New Alleles, a Rare Case of Uniparental Disomy (UPD) and the Importance to Search for Copy Number Variations. Frontiers in Genetics, 2018, 9, 681.	2.3	25
65	A genomeâ€wide association study identifies an association between variants in <scp>EFCAB</scp>4B gene and periodontal disease in an Italian isolated population. Journal of Periodontal Research, 2018, 53, 992-998.	2.7	15
66	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
67	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
68	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
69	Exome-chip meta-analysis identifies novel loci associated with cardiac conduction, including ADAMTS6. Genome Biology, 2018, 19, 87.	8.8	47
70	Whole-genome sequencing reveals new insights into age-related hearing loss: cumulative effects, pleiotropy and the role of selection. European Journal of Human Genetics, 2018, 26, 1167-1179.	2.8	22
71	Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway. Redox Biology, 2018, 19, 301-317.	9.0	50
72	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

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73	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
74	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
75	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
76	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	27.8	544
77	NLRC5 polymorphism is associated with susceptibility to chronic periodontitis. Immunobiology, 2017, 222, 704-708.	1.9	17
78	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
79	<scp>LTF</scp> and <scp>DEFB</scp>1 polymorphisms are associated with susceptibility toward chronic periodontitis development. Oral Diseases, 2017, 23, 1001-1008.	3.0	21
80	Targeted sequencing identifies novel variants involved in autosomal recessive hereditary hearing loss in Qatari families. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2017, 800-802, 29-36.	1.0	23
81	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. Scientific Reports, 2017, 7, 45040.	3.3	98
82	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
83	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. American Journal of Human Genetics, 2017, 100, 865-884.	6.2	131
84	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
85	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
86	Genetic structure in the Sherpa and neighboring Nepalese populations. BMC Genomics, 2017, 18, 102.	2.8	21
87	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. Nature Communications, 2017, 8, 15927.	12.8	64
88	A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman. Journal of Human Genetics, 2017, 62, 259-264.	2.3	21
89	Factors Influencing the Phenotypic Characterization of the Oral Marker, PROP. Nutrients, 2017, 9, 1275.	4.1	57
90	A Genome-Wide Association Study in isolated populations reveals new genes associated to common food likings. Reviews in Endocrine and Metabolic Disorders, 2016, 17, 209-219.	5.7	22

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91	Genome-wide association study identifies 74 loci associated with educational attainment. Nature, 2016, 533, 539-542.	27.8	1,204
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	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	21.4	2,421
94	Global diversity in the TAS2R38 bitter taste receptor: revisiting a classic evolutionary PROPosal. Scientific Reports, 2016, 6, 25506.	3.3	69
95	Caries and Innate Immunity: <i>DEFB1</i> Gene Polymorphisms and Caries Susceptibility in Genetic Isolates from North-Eastern Italy. Caries Research, 2016, 50, 589-594.	2.0	19
96	52 Genetic Loci Influencing MyocardialÂMass. Journal of the American College of Cardiology, 2016, 68, 1435-1448.	2.8	113
97	Non-additive genome-wide association scan reveals a new gene associated with habitual coffee consumption. Scientific Reports, 2016, 6, 31590.	3.3	25
98	PSIP1/LEDGF: a new gene likely involved in sensorineural progressive hearing loss. Scientific Reports, 2016, 5, 18568.	3.3	7
99	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. Nature Genetics, 2016, 48, 1462-1472.	21.4	284
100	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
101	Pharmacogenetics driving personalized medicine: analysis of genetic polymorphisms related to breast cancer medications in Italian isolated populations. Journal of Translational Medicine, 2016, 14, 22.	4.4	6
102	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
103	Understanding the role of personality and alexithymia in food preferences and PROP taste perception. Physiology and Behavior, 2016, 157, 72-78.	2.1	37
104	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. Nature Communications, 2016, 7, 10023.	12.8	412
105	Genetic analysis of Italian patients with congenital tufting enteropathy. World Journal of Pediatrics, 2016, 12, 219-224.	1.8	14
106	Clinical and Molecular Cytogenetic Characterisation of Children with Developmental Delay and Dysmorphic Features / KliniÄna in Molekularna Citogenetska Obravnava Otrok Z Razvojnim Zaostankom in DisplastiÄnimi Znaki. Zdravstveno Varstvo, 2015, 54, 69-73.	0.9	0
107	Genetic testing and genomic analysis: a debate on ethical, social and legal issues in the Arab world with a focus on Qatar. Journal of Translational Medicine, 2015, 13, 358.	4.4	22
108	The p.Cys169Tyr variant of connexin 26 is not a polymorphism. Human Molecular Genetics, 2015, 24, 2641-2648.	2.9	14

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109	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
110	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	27.8	173
111	Analysis of functional variants reveals new candidate genes associated with alexithymia. Psychiatry Research, 2015, 227, 363-365.	3.3	12
112	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
113	Genome-wide association analysis on five isolated populations identifies variants of the HLA-DOA gene associated with white wine liking. European Journal of Human Genetics, 2015, 23, 1717-1722.	2.8	12
114	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
115	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
116	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
117	Multicohort analysis of the maternal age effect on recombination. Nature Communications, 2015, 6, 7846.	12.8	29
118	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
119	Population genetic differentiation of height and body mass index across Europe. Nature Genetics, 2015, 47, 1357-1362.	21.4	227
120	Polymorphisms in sweet taste genes (TAS1R2 and GLUT2), sweet liking, and dental caries prevalence in an adult Italian population. Genes and Nutrition, 2015, 10, 485.	2.5	25
121	Connexin 26 variant carriers have a better gastrointestinal health: is this the heterozygote advantage?. European Journal of Human Genetics, 2015, 23, 563-564.	2.8	9
122	Assessment of the Olfactory Function in Italian Patients with Type 3 von Willebrand Disease Caused by a Homozygous 253 Kb Deletion Involving VWF and TMEM16B/ANO2. PLoS ONE, 2015, 10, e0116483.	2.5	7
123	Modulation of Genetic Associations with Serum Urate Levels by Body-Mass-Index in Humans. PLoS ONE, 2015, 10, e0119752.	2.5	64
124	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
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	Salt-inducible kinase 3, SIK3, is a new gene associated with hearing. Human Molecular Genetics, 2014, 23, 6407-6418.	2.9	30

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127	A General Approach for Haplotype Phasing across the Full Spectrum of Relatedness. PLoS Genetics, 2014, 10, e1004234.	3.5	553
128	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
129	Age related hearing loss and level of education: An epidemiological study on a large cohort of isolated populations. Hearing, Balance and Communication, 2014, 12, 94-98.	0.4	5
130	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
131	DNA mismatch repair gene MSH6 implicated in determining age at natural menopause. Human Molecular Genetics, 2014, 23, 2490-2497.	2.9	56
132	Consanguinity and Hereditary Hearing Loss in Qatar. Human Heredity, 2014, 77, 175-182.	0.8	15
133	Genetic landscape of populations along the Silk Road: admixture and migration patterns. BMC Genetics, 2014, 15, 131.	2.7	24
134	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
135	Insight into genetic determinants of resting heart rate. Gene, 2014, 545, 170-174.	2.2	7
136	Common Variants in UMOD Associate with Urinary Uromodulin Levels. Journal of the American Society of Nephrology: JASN, 2014, 25, 1869-1882.	6.1	85
137	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. Nature, 2014, 514, 92-97.	27.8	548
138	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
139	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	21.4	281
140	A novel deletion mutation involving TMEM38B in a patient with autosomal recessive osteogenesis imperfecta. Gene, 2014, 545, 290-292.	2.2	38
141	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. Gene, 2014, 534, 236-239.	2.2	50
142	Expression and Replication Studies to Identify New Candidate Genes Involved in Normal Hearing Function. PLoS ONE, 2014, 9, e85352.	2.5	39
143	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
144	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

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145	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
146	Regulatory evaluation of Glybera in Europe â€" two committees, one mission. Nature Reviews Drug Discovery, 2013, 12, 719-719.	46.4	54
147	A Novel CRYBB2 Missense Mutation Causing Congenital Autosomal Dominant Cataract in an Italian Family. Ophthalmic Genetics, 2013, 34, 115-117.	1.2	10
148	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. Nature Genetics, 2013, 45, 145-154.	21.4	675
149	Molecular cytogenetic characterization of 2p23.2p23.3 deletion in a child with developmental delay, hypotonia and cryptorchism. European Journal of Medical Genetics, 2013, 56, 62-65.	1.3	5
150	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
151	Genetics of eye colours in different rural populations on the Silk Road. European Journal of Human Genetics, 2013, 21, 1320-1323.	2.8	11
152	Congenital hyperinsulinism: Clinical and molecular analysis of a large Italian cohort. Gene, 2013, 521, 160-165.	2.2	21
153	Co-inheritance of two ABCC8 mutations causing an unresponsive congenital hyperinsulinism: Clinical and functional characterization of two novel ABCC8 mutations. Gene, 2013, 516, 122-125.	2.2	11
154	Alagille Syndrome: A New Missense Mutation Detected by Whole-Exome Sequencing in a Case Previously Found to Be Negative by DHPLC and MLPA. Molecular Syndromology, 2013, 4, 207-210.	0.8	10
155	Meta-Analysis of Genome-Wide Association Studies Identifies Six New Loci for Serum Calcium Concentrations. PLoS Genetics, 2013, 9, e1003796.	3.5	142
156	Lifestyle and normal hearing function in Italy and Central Asia: The potential role of coffee. Hearing, Balance and Communication, 2013, 11, 218-223.	0.4	7
157	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
158	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
159	Genome Wide Association Analysis of a Founder Population Identified TAF3 as a Gene for MCHC in Humans. PLoS ONE, 2013, 8, e69206.	2.5	9
160	Linkage Study and Exome Sequencing Identify a BDP1 Mutation Associated with Hereditary Hearing Loss. PLoS ONE, 2013, 8, e80323.	2.5	50
161	Evidence of Inbreeding Depression on Human Height. PLoS Genetics, 2012, 8, e1002655.	3.5	79
162	Genome-Wide Association and Functional Follow-Up Reveals New Loci for Kidney Function. PLoS Genetics, 2012, 8, e1002584.	3.5	166

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163	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
164	Delayed Diagnosis of Glycogen Storage Disease Type III. Journal of Pediatric Gastroenterology and Nutrition, 2012, 54, 122-124.	1.8	1
165	Phylloid Pattern of Hypomelanosis Closely Related to Chromosomal Abnormalities in the 13q Detected by SNP Array Analysis. Dermatology, 2012, 225, 294-297.	2.1	15
166	Identification of a New Mutation (L46P) in the Human <i>NOG</i> Gene in an Italian Patient with Symphalangism Syndrome. Molecular Syndromology, 2012, 3, 21-24.	0.8	6
167	Two Novel <i>COH1</i> Mutations in an Italian Patient with Cohen Syndrome. Molecular Syndromology, 2012, 3, 30-33.	0.8	4
168	Genetics of Food Preferences: A First View from Silk Road Populations. Journal of Food Science, 2012, 77, S413-8.	3.1	45
169	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	27.8	320
170	De novo 911ÂKb interstitial deletion on chromosome 1q43 in a boy with mental retardation and short stature. European Journal of Medical Genetics, 2012, 55, 117-119.	1.3	22
171	Contribution of SNP arrays in diagnosis of deletion 2p11.2â€“p12. Gene, 2012, 492, 315-318.	2.2	5
172	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
173	A meta-analysis of genome-wide association studies of the electrocardiographic early repolarization pattern. Heart Rhythm, 2012, 9, 1627-1634.	0.7	58
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