Giorgia Girotto

List of Publications by Year
in descending order

86 papers	7,877 citations	147726 31 h-index	64755 79 g-index
93 all docs	93 docs citations	93 times ranked	15434 citing authors

#	Article	IF	Citations
1	Genome-wide association study identifies 74 loci associated with educational attainment. Nature, 2016, 533, 539-542.	13.7	1,204
2	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. Nature Genetics, 2018, 50, 1412-1425.	9.4	924
3	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	9.4	549
4	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	13.7	544
5	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. Nature Genetics, 2017, 49, 403-415.	9.4	492
6	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	9.4	357
7	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	13.7	353
8	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	13.7	320
9	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. Nature Genetics, 2016, 48, 1462-1472.	9.4	284
10	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. Nature Genetics, 2019, 51, 1459-1474.	9.4	251
11	Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals. Nature Genetics, 2022, 54, 437-449.	9.4	215
12	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	13.7	173
13	Meta-Analysis of Genome-Wide Association Studies Identifies Six New Loci for Serum Calcium Concentrations. PLoS Genetics, 2013, 9, e1003796.	1.5	142
14	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. Hypertension, 2017, 70, .	1.3	123
15	Genetic variants linked to education predict longevity. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 13366-13371.	3.3	110
16	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. Scientific Reports, 2017, 7, 45040.	1.6	98
17	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469.	9.4	89
18	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.	9.4	86

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19	Common Variants in UMOD Associate with Urinary Uromodulin Levels. Journal of the American Society of Nephrology: JASN, 2014, 25, 1869-1882.	3.0	85
20	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	5.8	84
21	Evidence of Inbreeding Depression on Human Height. PLoS Genetics, 2012, 8, e1002655.	1.5	79
22	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.	1.5	71
23	Identification of 371 genetic variants for age at first sex and birth linked to externalising behaviour. Nature Human Behaviour, 2021, 5, 1717-1730.	6.2	62
24	Estrogen-related receptor gamma and hearing function: evidence of a role in humans and mice. Neurobiology of Aging, 2013, 34, 2077.e1-2077.e9.	1.5	53
25	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. Gene, 2014, 534, 236-239.	1.0	50
26	Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway. Redox Biology, 2018, 19, 301-317.	3.9	50
27	Linkage Study and Exome Sequencing Identify a BDP1 Mutation Associated with Hereditary Hearing Loss. PLoS ONE, 2013, 8, e80323.	1.1	50
28	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.	1.0	48
29	Expression and Replication Studies to Identify New Candidate Genes Involved in Normal Hearing Function. PLoS ONE, 2014, 9, e85352.	1.1	39
30	Mutations in L-type amino acid transporter-2 support SLC7A8 as a novel gene involved in age-related hearing loss. ELife, 2018, 7, .	2.8	38
31	Genome-wide association analysis on normal hearing function identifiesPCDH20andSLC28A3as candidates for hearing function and loss. Human Molecular Genetics, 2015, 24, 5655-5664.	1.4	37
32	Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. Science Advances, 2021, 7, .	4.7	36
33	Maps of open chromatin highlight cell typeâ€“restricted patterns of regulatory sequence variation at hematological trait loci. Genome Research, 2013, 23, 1130-1141.	2.4	34
34	Genome-wide meta-analysis of common variant differences between men and women. Human Molecular Genetics, 2012, 21, 4805-4815.	1.4	33
35	MYH9 related disease: four novel mutations of the tail domain of myosinâ€9 correlating with a mild clinical phenotype. European Journal of Haematology, 2010, 84, 291-297.	1.1	32
36	Genome-wide association meta-analysis identifies five novel loci for age-related hearing impairment. Scientific Reports, 2019, 9, 15192.	1.6	32

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37	Salt-inducible kinase 3, SIK3, is a new gene associated with hearing. Human Molecular Genetics, 2014, 23, 6407-6418.	1.4	30
38	A birdâ€™s-eye view of Italian genomic variation through whole-genome sequencing. European Journal of Human Genetics, 2020, 28, 435-444.	1.4	29
39	Increased Rate of Deleterious Variants in Long Runs of Homozygosity of an Inbred Population from Qatar. Human Heredity, 2015, 79, 14-19.	0.4	28
40	GJB2 and GJB6 genes and the A1555G mitochondrial mutation are only minor causes of nonsyndromic hearing loss in the Qatari population. International Journal of Audiology, 2012, 51, 181-185.	0.9	27
41	Effects of Calcium, Magnesium, and Potassium Concentrations on Ventricular Repolarization in Unselected Individuals. Journal of the American College of Cardiology, 2019, 73, 3118-3131.	1.2	27
42	Genome-wide association meta-analysis identifies 48 risk variants and highlights the role of the stria vascularis in hearing loss. American Journal of Human Genetics, 2022, 109, 1077-1091.	2.6	27
43	Usher syndrome: An effective sequencing approach to establish a genetic and clinical diagnosis. Hearing Research, 2015, 320, 18-23.	0.9	26
44	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.	1.1	25
45	Genetic landscape of populations along the Silk Road: admixture and migration patterns. BMC Genetics, 2014, 15, 131.	2.7	24
46	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.	0.4	23
47	Mutations in PLS1 , encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. Human Mutation, 2019, 40, 2286-2295.	1.1	23
48	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.	1.4	22
49	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.	1.4	22
50	A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman. Journal of Human Genetics, 2017, 62, 259-264.	1.1	21
51	TBL1Y: a new gene involved in syndromic hearing loss. European Journal of Human Genetics, 2019, 27, 466-474.	1.4	17
52	Gene-educational attainment interactions in a multi-ancestry genome-wide meta-analysis identify novel blood pressure loci. Molecular Psychiatry, 2020, 26, 2111-2125.	4.1	17
53	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. Communications Biology, 2022, 5, .	2.0	17
54	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

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55	Consanguinity and Hereditary Hearing Loss in Qatar. Human Heredity, 2014, 77, 175-182.	0.4	15
56	The p.Cys169Tyr variant of connexin 26 is not a polymorphism. Human Molecular Genetics, 2015, 24, 2641-2648.	1.4	14
57	Meta-GWAS Reveals Novel Genetic Variants Associated with Urinary Excretion of Uromodulin. Journal of the American Society of Nephrology: JASN, 2022, 33, 511-529.	3.0	14
58	Lights and Shadows in the Genetics of Syndromic and Non-Syndromic Hearing Loss in the Italian Population. Genes, 2020, 11, 1237.	1.0	13
59	Functional analysis of candidate genes from genome-wide association studies of hearing. Hearing Research, 2020, 387, 107879.	0.9	13
60	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.	1.1	11
61	MYH9related disease: A novel missense Ala95Asp mutation of theMYH9gene. Platelets, 2009, 20, 598-602.	1.1	10
62	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.	0.9	8
63	Lifestyle and normal hearing function in Italy and Central Asia: The potential role of coffee. Hearing, Balance and Communication, 2013, 11, 218-223.	0.1	7
64	PSIP1/LEDGF: a new gene likely involved in sensorineural progressive hearing loss. Scientific Reports, 2016, 5, 18568.	1.6	7
65	Next generation sequencing study in a cohort of Italian patients with syndromic hearing loss. Hearing Research, 2019, 381, 107769.	0.9	7
66	Pharmacogenetics driving personalized medicine: analysis of genetic polymorphisms related to breast cancer medications in Italian isolated populations. Journal of Translational Medicine, 2016, 14, 22.	1.8	6
67	Natural human knockouts and Mendelian disorders: deep phenotyping in Italian isolates. European Journal of Human Genetics, 2021, 29, 1272-1281.	1.4	6
68	Variants in USP48 encoding ubiquitin hydrolase are associated with autosomal dominant non-syndromic hereditary hearing loss. Human Molecular Genetics, 2021, 30, 1785-1796.	1.4	6
69	Chronic Rhinosinusitis: T2r38 Genotyping and Nasal Cytology in Primary Ciliary Dyskinesia. Laryngoscope, 2023, 133, 248-254.	1.1	6
70	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.	0.1	5
71	Molecular testing for the study of non-syndromic hearing loss. Hearing, Balance and Communication, 2020, 18, 270-277.	0.1	5
72	Pendred Syndrome, or Not Pendred Syndrome? That Is the Question. Genes, 2021, 12, 1569.	1.0	5

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73	TTC5 syndrome: Clinical and molecular spectrum of a severe and recognizable condition. American Journal of Medical Genetics, Part A, 2022, 188, 2652-2665.	0.7	4
74	Association of SNPs in LCP1 and CTIF with hearing in 11Âyear old children: Findings from the Avon Longitudinal Study of Parents and Children (ALSPAC) birth cohort and the G-EAR consortium. BMC Medical Genomics, 2015, 8, 48.	0.7	3
75	The Role of Knockout Olfactory Receptor Genes in Odor Discrimination. Genes, 2021, 12, 631.	1.0	3
76	Non-Syndromic Autosomal Dominant Hearing Loss: The First Italian Family Carrying a Mutation in the NCOA3 Gene. Genes, 2021, 12, 1043.	1.0	3
77	Hearing loss and brain abnormalities due to pathogenic mutations in ADGRV1 gene: a case report. Hearing, Balance and Communication, 2020, 18, 196-198.	0.1	2
78	Hearing loss. , 2021, , 305-322.		2
79	Genetics, odor perception and food liking: The intriguing role of cinnamon. Food Quality and Preference, 2021, 93, 104277.	2.3	2
80	Sensory Capacities and Eating Behavior: Intriguing Results from a Large Cohort of Italian Individuals. Foods, 2022, 11, 735.	1.9	2
81	There Is More Than Meets the Eye: Identification of Dual Molecular Diagnosis in Patients Affected by Hearing Loss. Biomedicines, 2022, 10, 12.	1.4	2
82	Genetic Dissection of Temperament Personality Traits in Italian Isolates. Genes, 2022, 13, 4.	1.0	2
83	Non-Syndromic Sensorineural Prelingual and Postlingual Hearing Loss due to COL11A1 Gene Mutation. Journal of International Advanced Otology, 2021, 17, 81-83.	1.0	1
84	Hearing Function: Identification of New Candidate Genes Further Explaining the Complexity of This Sensory Ability. Genes, 2021, 12, 1228.	1.0	1
85	Benefit of cochlear implantation in a patient with Myhre syndrome. BMJ Case Reports, 2021, 14, e243164.	0.2	1
86	Large-Scale Genomic Analyses Link Reproductive Aging to Hypothalamic Signaling, Breast Cancer Susceptibility, and BRCA1-Mediated DNA Repair. Obstetrical and Gynecological Survey, 2015, 70, 758-762.	0.2	0