Giorgia Girotto

List of Publications by Year
in descending order

86 papers	7,877 citations	147801 31 h-index	64796 79 g-index
93 all docs	93 docs citations	93 times ranked	15434 citing authors

#	Article	IF	Citations
1	Chronic Rhinosinusitis: T2r38 Genotyping and Nasal Cytology in Primary Ciliary Dyskinesia. Laryngoscope, 2023, 133, 248-254.	2.0	6
2	Meta-GWAS Reveals Novel Genetic Variants Associated with Urinary Excretion of Uromodulin. Journal of the American Society of Nephrology: JASN, 2022, 33, 511-529.	6.1	14
3	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
4	Sensory Capacities and Eating Behavior: Intriguing Results from a Large Cohort of Italian Individuals. Foods, 2022, 11, 735.	4.3	2
5	There Is More Than Meets the Eye: Identification of Dual Molecular Diagnosis in Patients Affected by Hearing Loss. Biomedicines, 2022, 10, 12.	3.2	2
6	Genetic Dissection of Temperament Personality Traits in Italian Isolates. Genes, 2022, 13, 4.	2.4	2
7	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.	6.2	27
8	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
9	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. Communications Biology, 2022, 5, .	4.4	17
10	Hearing loss. , 2021, , 305-322.		2
11	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
12	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
13	Natural human knockouts and Mendelian disorders: deep phenotyping in Italian isolates. European Journal of Human Genetics, 2021, 29, 1272-1281.	2.8	6
14	The Role of Knockout Olfactory Receptor Genes in Odor Discrimination. Genes, 2021, 12, 631.	2.4	3
15	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
16	Non-Syndromic Autosomal Dominant Hearing Loss: The First Italian Family Carrying a Mutation in the NCOA3 Gene. Genes, 2021, 12, 1043.	2.4	3
17	Identification of 371 genetic variants for age at first sex and birth linked to externalising behaviour. Nature Human Behaviour, 2021, 5, 1717-1730.	12.0	62
18	Hearing Function: Identification of New Candidate Genes Further Explaining the Complexity of This Sensory Ability. Genes, 2021, 12, 1228.	2.4	1

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19	Benefit of cochlear implantation in a patient with Myhre syndrome. BMJ Case Reports, 2021, 14, e243164.	0.5	1
20	Genetics, odor perception and food liking: The intriguing role of cinnamon. Food Quality and Preference, 2021, 93, 104277.	4.6	2
21	Pendred Syndrome, or Not Pendred Syndrome? That Is the Question. Genes, 2021, 12, 1569.	2.4	5
22	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	27.8	353
23	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
24	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
25	Lights and Shadows in the Genetics of Syndromic and Non-Syndromic Hearing Loss in the Italian Population. Genes, 2020, 11, 1237.	2.4	13
26	Molecular testing for the study of non-syndromic hearing loss. Hearing, Balance and Communication, 2020, 18, 270-277.	0.4	5
27	Hearing loss and brain abnormalities due to pathogenic mutations in ADGRV1 gene: a case report. Hearing, Balance and Communication, 2020, 18, 196-198.	0.4	2
28	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
29	Functional analysis of candidate genes from genome-wide association studies of hearing. Hearing Research, 2020, 387, 107879.	2.0	13
30	Mutations in PLS1 , encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. Human Mutation, 2019, 40, 2286-2295.	2.5	23
31	Next generation sequencing study in a cohort of Italian patients with syndromic hearing loss. Hearing Research, 2019, 381, 107769.	2.0	7
32	Effects of Calcium, Magnesium, and Potassium Concentrations on Ventricular Repolarization in Unselected Individuals. Journal of the American College of Cardiology, 2019, 73, 3118-3131.	2.8	27
33	Genome-wide association meta-analysis identifies five novel loci for age-related hearing impairment. Scientific Reports, 2019, 9, 15192.	3.3	32
34	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	12.8	84
35	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. Nature Genetics, 2019, 51, 1459-1474.	21.4	251
36	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	21.4	549

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37	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
38	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469.	21.4	89
39	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
40	TBL1Y: a new gene involved in syndromic hearing loss. European Journal of Human Genetics, 2019, 27, 466-474.	2.8	17
41	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
42	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
43	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
44	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
45	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
46	Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway. Redox Biology, 2018, 19, 301-317.	9.0	50
47	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
48	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	27.8	544
49	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
50	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. Scientific Reports, 2017, 7, 45040.	3.3	98
51	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
52	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
53	Genome-wide association study identifies 74 loci associated with educational attainment. Nature, 2016, 533, 539-542.	27.8	1,204
54	PSIP1/LEDGF: a new gene likely involved in sensorineural progressive hearing loss. Scientific Reports, 2016, 5, 18568.	3.3	7

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55	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. Nature Genetics, 2016, 48, 1462-1472.	21.4	284
56	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
57	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
58	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.	0.4	0
59	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.	1.5	3
60	The p.Cys169Tyr variant of connexin 26 is not a polymorphism. Human Molecular Genetics, 2015, 24, 2641-2648.	2.9	14
61	Usher syndrome: An effective sequencing approach to establish a genetic and clinical diagnosis. Hearing Research, 2015, 320, 18-23.	2.0	26
62	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
63	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	27.8	173
64	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
65	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
66	Salt-inducible kinase 3, SIK3, is a new gene associated with hearing. Human Molecular Genetics, 2014, 23, 6407-6418.	2.9	30
67	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
68	Consanguinity and Hereditary Hearing Loss in Qatar. Human Heredity, 2014, 77, 175-182.	0.8	15
69	Genetic landscape of populations along the Silk Road: admixture and migration patterns. BMC Genetics, 2014, 15, 131.	2.7	24
70	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
71	Common Variants in UMOD Associate with Urinary Uromodulin Levels. Journal of the American Society of Nephrology: JASN, 2014, 25, 1869-1882.	6.1	85
72	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. Gene, 2014, 534, 236-239.	2.2	50

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73	Expression and Replication Studies to Identify New Candidate Genes Involved in Normal Hearing Function. PLoS ONE, 2014, 9, e85352.	2.5	39
74	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
75	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
76	Meta-Analysis of Genome-Wide Association Studies Identifies Six New Loci for Serum Calcium Concentrations. PLoS Genetics, 2013, 9, e1003796.	3.5	142
77	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
78	Linkage Study and Exome Sequencing Identify a BDP1 Mutation Associated with Hereditary Hearing Loss. PLoS ONE, 2013, 8, e80323.	2.5	50
79	Evidence of Inbreeding Depression on Human Height. PLoS Genetics, 2012, 8, e1002655.	3.5	79
80	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	27.8	320
81	Genome-wide meta-analysis of common variant differences between men and women. Human Molecular Genetics, 2012, 21, 4805-4815.	2.9	33
82	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.	1.7	27
83	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
84	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
85	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.	2.2	32
86	MYH9related disease: A novel missense Ala95Asp mutation of theMYH9gene. Platelets, 2009, 20, 598-602.	2.3	10