Giorgia Girotto

List of Publications by Year in Descending Order

78 papers	4,534 citations	27 h-index	67 g-index
93 ext. papers	6,417 ext. citations	11 avg, IF	3.69 L-index

#	Paper	IF	Citations
78	Meta-GWAS Reveals Novel Genetic Variants Associated with Urinary Excretion of Uromodulin.. *Journal of the American Society of Nephrology: JASN*, **2022**, 33, 511-529	12.7	2
77	Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals.. *Nature Genetics*, **2022**,	36.3	7
76	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. *Communications Biology*, **2022**, 5,	6.7	1
75	Gene-educational attainment interactions in a multi-ancestry genome-wide meta-analysis identify novel blood pressure loci. *Molecular Psychiatry*, **2021**, 26, 2111-2125	15.1	3
74	The power of genetic diversity in genome-wide association studies of lipids. *Nature*, **2021**,	50.4	24
73	Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. *Science Advances*, **2021**, 7,	14.3	11
72	Natural human knockouts and Mendelian disorders: deep phenotyping in Italian isolates. *European Journal of Human Genetics*, **2021**, 29, 1272-1281	5.3	1
71	Variants in USP48 encoding ubiquitin hydrolase are associated with autosomal dominant non-syndromic hereditary hearing loss. *Human Molecular Genetics*, **2021**, 30, 1785-1796	5.6	1
70	Hearing loss **2021**, 305-322		0
69	Non-Syndromic Sensorineural Prelingual and Postlingual Hearing Loss due to COL11A1 Gene Mutation. *Journal of International Advanced Otology*, **2021**, 17, 81-83	1.1	
68	Non-Syndromic Autosomal Dominant Hearing Loss: The First Italian Family Carrying a Mutation in the Gene. *Genes*, **2021**, 12,	4.2	2
67	Identification of 371 genetic variants for age at first sex and birth linked to externalising behaviour. *Nature Human Behaviour*, **2021**,	12.8	5
66	Benefit of cochlear implantation in a patient with Myhre syndrome. *BMJ Case Reports*, **2021**, 14,	0.9	
65	Genetics, odor perception and food liking: The intriguing role of cinnamon. *Food Quality and Preference*, **2021**, 93, 104277	5.8	1
64	Functional analysis of candidate genes from genome-wide association studies of hearing. *Hearing Research*, **2020**, 387, 107879	3.9	4
63	A bird'ƃ-eye view of Italian genomic variation through whole-genome sequencing. *European Journal of Human Genetics*, **2020**, 28, 435-444	5.3	16
62	Lights and Shadows in the Genetics of Syndromic and Non-Syndromic Hearing Loss in the Italian Population. *Genes*, **2020**, 11,	4.2	7

61	Molecular testing for the study of non-syndromic hearing loss. *Hearing, Balance and Communication*, **2020**, 18, 270-277	0.7	3
60	Hearing loss and brain abnormalities due to pathogenic mutations in ADGRV1 gene: a case report. *Hearing, Balance and Communication*, **2020**, 18, 196-198	0.7	0
59	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics*, **2019**, 51, 1459-1474	36.3	122
58	A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics*, **2019**, 51, 957-972	36.3	217
57	Next Generation Sequencing and Animal Models Reveal as a New Gene Involved in Human Age-Related Hearing Loss. *Frontiers in Genetics*, **2019**, 10, 142	4.5	6
56	Mutations in PLS1, encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. *Human Mutation*, **2019**, 40, 2286-2295	4.7	14
55	Next generation sequencing study in a cohort of Italian patients with syndromic hearing loss. *Hearing Research*, **2019**, 381, 107769	3.9	2
54	Effects of Calcium, Magnesium, and Potassium Concentrations on Ventricular Repolarization in Unselected Individuals. *Journal of the American College of Cardiology*, **2019**, 73, 3118-3131	15.1	12
53	Genome-wide association meta-analysis identifies five novel loci for age-related hearing impairment. *Scientific Reports*, **2019**, 9, 15192	4.9	14
52	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
51	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. *Nature Genetics*, **2019**, 51, 452-469	36.3	44
50	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	5.3	14
49	TBL1Y: a new gene involved in syndromic hearing loss. *European Journal of Human Genetics*, **2019**, 27, 466-474	5.3	8
48	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	36.3	59
47	Mutations in L-type amino acid transporter-2 support as a novel gene involved in age-related hearing loss. *ELife*, **2018**, 7,	8.9	27
46	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	5.3	14
45	Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway. *Redox Biology*, **2018**, 19, 301-317	11.3	28
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	4.5	13

43	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. *Nature Genetics*, **2018**, 50, 1412-1425	36.3	386
42	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. *Nature Genetics*, **2017**, 49, 403-415	36.3	313
41	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
40	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	3.3	13
39	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Scientific Reports*, **2017**, 7, 45040	4.9	70
38	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. *Hypertension*, **2017**,	8.5	85
37	A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman. *Journal of Human Genetics*, **2017**, 62, 259-264	4.3	18
36	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. *Nature Genetics*, **2016**, 48, 1462-1472	36.3	198
35	Genetic variants linked to education predict longevity. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, 13366-13371	11.5	90
34	Pharmacogenetics driving personalized medicine: analysis of genetic polymorphisms related to breast cancer medications in Italian isolated populations. *Journal of Translational Medicine*, **2016**, 14, 22	8.5	3
33	Genome-wide association study identifies 74 loci associated with educational attainment. *Nature*, **2016**, 533, 539-42	50.4	850
32	Increased rate of deleterious variants in long runs of homozygosity of an inbred population from Qatar. *Human Heredity*, **2015**, 79, 14-9	1.1	22
31	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	50.4	119
30	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
29	Genome-wide association analysis on normal hearing function identifies PCDH20 and SLC28A3 as candidates for hearing function and loss. *Human Molecular Genetics*, **2015**, 24, 5655-64	5.6	24
28	PSIP1/LEDGF: a new gene likely involved in sensorineural progressive hearing loss. *Scientific Reports*, **2015**, 5, 18568	4.9	4
27	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	2.4	
26	Association of SNPs in LCP1 and CTIF with hearing in 11 year 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	3.7	2

25	The p.Cys169Tyr variant of connexin 26 is not a polymorphism. *Human Molecular Genetics*, **2015**, 24, 2641-8	5.8	9
24	Usher syndrome: an effective sequencing approach to establish a genetic and clinical diagnosis. *Hearing Research*, **2015**, 320, 18-23	3.9	22
23	Common variants in UMOD associate with urinary uromodulin levels: a meta-analysis. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 1869-82	12.7	71
22	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. *Gene*, **2014**, 534, 236-9	3.8	25
21	Salt-inducible kinase 3, SIK3, is a new gene associated with hearing. *Human Molecular Genetics*, **2014**, 23, 6407-18	5.6	23
20	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	0.7	3
19	Consanguinity and hereditary hearing loss in Qatar. *Human Heredity*, **2014**, 77, 175-82	1.1	13
18	Genetic landscape of populations along the Silk Road: admixture and migration patterns. *BMC Genetics*, **2014**, 15, 131	2.6	15
17	Hereditary hearing loss: a 96 gene targeted sequencing protocol reveals novel alleles in a series of Italian and Qatari patients. *Gene*, **2014**, 542, 209-16	3.8	45
16	Expression and replication studies to identify new candidate genes involved in normal hearing function. *PLoS ONE*, **2014**, 9, e85352	3.7	30
15	Estrogen-related receptor gamma and hearing function: evidence of a role in humans and mice. *Neurobiology of Aging*, **2013**, 34, 2077.e1-9	5.6	42
14	Maps of open chromatin highlight cell type-restricted patterns of regulatory sequence variation at hematological trait loci. *Genome Research*, **2013**, 23, 1130-41	9.7	31
13	Meta-analysis of genome-wide association studies identifies six new Loci for serum calcium concentrations. *PLoS Genetics*, **2013**, 9, e1003796	6	100
12	Lifestyle and normal hearing function in Italy and Central Asia: The potential role of coffee. *Hearing, Balance and Communication*, **2013**, 11, 218-223	0.7	5
11	Linkage study and exome sequencing identify a BDP1 mutation associated with hereditary hearing loss. *PLoS ONE*, **2013**, 8, e80323	3.7	43
10	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257
9	Genome-wide meta-analysis of common variant differences between men and women. *Human Molecular Genetics*, **2012**, 21, 4805-15	5.6	24
8	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, 181-5	2.6	25

7	Evidence of inbreeding depression on human height. *PLoS Genetics*, **2012**, 8, e1002655	6	62
6	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		12
5	Hearing function and thresholds: a genome-wide association study in European isolated populations identifies new loci and pathways. *Journal of Medical Genetics*, **2011**, 48, 369-74	5.8	59
4	MYH9 related disease: four novel mutations of the tail domain of myosin-9 correlating with a mild clinical phenotype. *European Journal of Haematology*, **2010**, 84, 291-7	3.8	27
3	MYH9 related disease: a novel missense Ala95Asp mutation of the MYH9 gene. *Platelets*, **2009**, 20, 598-602	3.2	10
2	Genetic analysis of over one million people identifies 535 novel loci for blood pressure		4
1	Protein-Coding Variants Implicate Novel Genes Related to Lipid Homeostasis Contributing to Body Fat Distribution		1