Bacterial and Genomics Consortiu

List of Publications by Year
in descending order

55 papers	1,734 citations	430874 18 h-index	315739 38 g-index
57 all docs	57 docs citations	57 times ranked	4709 citing authors

#	Article	IF	Citations
1	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	12.8	250
2	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	14.8	213
3	Comprehensive analysis via exome sequencing uncovers genetic etiology in autosomal recessive nonsyndromic deafness in a large multiethnic cohort. Genetics in Medicine, 2016, 18, 364-371.	2.4	124
4	A novel mutation in VCP causes Charcotâ€“Marieâ€“Tooth Type 2 disease. Brain, 2014, 137, 2897-2902.	7.6	116
5	Trans-ethnic kidney function association study reveals putative causal genes and effects on kidney-specific disease aetiologies. Nature Communications, 2019, 10, 29.	12.8	113
6	Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents. Human Genetics, 2016, 135, 953-961.	3.8	102
7	Folate pathway and nonsyndromic cleft lip and palate. Birth Defects Research Part A: Clinical and Molecular Teratology, 2011, 91, 50-60.	1.6	72
8	Whole-exome sequencing and its impact in hereditary hearing loss. Genetical Research, 2015, 97, e4.	0.9	43
9	A next-generation sequencing gene panel (MiamiOtoGenes) for comprehensive analysis of deafness genes. Hearing Research, 2016, 333, 179-184.	2.0	38
10	A dominant variant in the PDE1C gene is associated with nonsyndromic hearing loss. Human Genetics, 2018, 137, 437-446.	3.8	36
11	The genetic basis of deafness in populations of African descent. Journal of Genetics and Genomics, 2017, 44, 285-294.	3.9	29
12	COVID19: A Systematic Approach to Early Identification and Healthcare Worker Protection. Frontiers in Public Health, 2020, 8, 205.	2.7	28
13	Ethnic Heterogeneity of IRF6 AP-2a Binding Site Promoter SNP Association with Nonsyndromic Cleft Lip and Palate. Cleft Palate-Craniofacial Journal, 2010, 47, 574-577.	0.9	27
14	MYO3A Causes Human Dominant Deafness and Interacts with Protocadherin 15-CD2 Isoform. Human Mutation, 2016, 37, 481-487.	2.5	27
15	Common genes for non-syndromic deafness are uncommon in sub-Saharan Africa: A report from Nigeria. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 1870-1873.	1.0	26
16	Screening of deafness-causing DNA variants that are common in patients of European ancestry using a microarray-based approach. PLoS ONE, 2017, 12, e0169219.	2.5	26
17	Familyâ€based study shows heterogeneity of a susceptibility locus on chromosome 8q24 for nonsyndromic cleft lip and palate. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 256-259.	1.6	24
18	Functional Assessment of Clubfoot Associated HOXA9, TPM1, and TPM2 Variants Suggests a Potential Gene Regulation Mechanism. Clinical Orthopaedics and Related Research, 2016, 474, 1726-1735.	1.5	21

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19	Genetic variants in LEKR1 and GALNT10 modulate sex-difference in carotid intima-media thickness: A genome-wide interaction study. Atherosclerosis, 2015, 240, 462-467.	0.8	20
20	FOXF2is required for cochlear development in humans and mice. Human Molecular Genetics, 2019, 28, 1286-1297.	2.9	20
21	MPZL2 is a novel gene associated with autosomal recessive nonsyndromic moderate hearing loss. Human Genetics, 2018, 137, 479-486.	3.8	19
22	Precision medicine in hearing loss. Journal of Genetics and Genomics, 2018, 45, 99-109.	3.9	18
23	PBXâ€WNTâ€P63â€IRF6 pathway in nonsyndromic cleft lip and palate. Birth Defects Research, 2020, 112, 234-244.	1.5	18
24	Targeted Resequencing of Deafness Genes Reveals a FounderMYO15AVariant in Northeastern Brazil. Annals of Human Genetics, 2016, 80, 327-331.	0.8	17
25	A nonsense TMEM43 variant leads to disruption of connexin-linked function and autosomal dominant auditory neuropathy spectrum disorder. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	17
26	Sirtuin/Uncoupling Protein Gene Variants and Carotid Plaque Area and Morphology. International Journal of Stroke, 2015, 10, 1247-1252.	5.9	16
27	DNA Methylation Variation Is Identified in Monozygotic Twins Discordant for Non-syndromic Cleft Lip and Palate. Frontiers in Cell and Developmental Biology, 2021, 9, 656865.	3.7	16
28	Review of Genotype-Phenotype Correlations in Usher Syndrome. Ear and Hearing, 2022, 43, 1-8.	2.1	16
29	Novel domain-specific POU3F4 mutations are associated with X-linked deafness: examples from different populations. BMC Medical Genetics, 2015, 16, 9.	2.1	15
30	Apolipoprotein E Gene Polymorphism and Subclinical Carotid Atherosclerosis: The Northern Manhattan Study. Journal of Stroke and Cerebrovascular Diseases, 2018, 27, 645-652.	1.6	15
31	Knockdown of Crispld2 in zebrafish identifies a novel network for nonsyndromic cleft lip with or without cleft palate candidate genes. European Journal of Human Genetics, 2018, 26, 1441-1450.	2.8	15
32	Extrusion pump ABCC1 was first linked with nonsyndromic hearing loss in humans by stepwise genetic analysis. Genetics in Medicine, 2019, 21, 2744-2754.	2.4	15
33	Dysfunction of GRAP, encoding the GRB2-related adaptor protein, is linked to sensorineural hearing loss. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 1347-1352.	7.1	15
34	Usher Syndrome in the Inner Ear: Etiologies and Advances in Gene Therapy. International Journal of Molecular Sciences, 2021, 22, 3910.	4.1	15
35	Genetic basis of hearing loss in Spanish, Hispanic and Latino populations. Gene, 2018, 647, 297-305.	2.2	14
36	Targeted Next-Generation Sequencing of a Deafness Gene Panel (MiamiOtoGenes) Analysis in Families Unsuitable for Linkage Analysis. BioMed Research International, 2018, 2018, 1-7.	1.9	14

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37	Novel genetic variants modify the effect of smoking on carotid plaque burden in Hispanics. Journal of the Neurological Sciences, 2014, 344, 27-31.	0.6	13
38	Recent advancements in understanding the role of epigenetics in the auditory system. Gene, 2020, 761, 144996.	2.2	12
39	Genome-wide scan in Hispanics highlights candidate loci for brain white matter hyperintensities. Neurology: Genetics, 2017, 3, e185.	1.9	11
40	Novel pathogenic variants underlie SLC26A4 -related hearing loss in a multiethnic cohort. International Journal of Pediatric Otorhinolaryngology, 2017, 101, 167-171.	1.0	11
41	Evidence for craniofacial enhancer variation underlying nonsyndromic cleft lip and palate. Human Genetics, 2020, 139, 1261-1272.	3.8	10
42	Genetic screening as an adjunct to universal newborn hearing screening: literature review and implications for non-congenital pre-lingual hearing loss. International Journal of Audiology, 2019, 58, 834-850.	1.7	9
43	Peripheral vestibular system: Age-related vestibular loss and associated deficits. Journal of Otology, 2021, 16, 258-265.	1.0	9
44	Identification of a Novel Gene on 10q22.1 Causing Autosomal Dominant Retinitis Pigmentosa (adRP). Advances in Experimental Medicine and Biology, 2016, 854, 193-200.	1.6	8
45	Spectrum of MYO7A Mutations in an Indigenous South African Population Further Elucidates the Nonsyndromic Autosomal Recessive Phenotype of DFNB2 to Include Both Homozygous and Compound Heterozygous Mutations. Genes, 2021, 12, 274.	2.4	8
46	BRCA1 and BRCA2 gene variants and nonsyndromic cleft lip/palate. Birth Defects Research, 2018, 110, 1043-1048.	1.5	6
47	Sequencing of candidate genes in Dominican families implicates both rare exonic and common non-exonic variants for carotid intima-media thickness at bifurcation. Human Genetics, 2015, 134, 1127-1138.	3.8	5
48	Screening Consanguineous Families for Hearing Loss Using the MiamiOtoGenes Panel. Genetic Testing and Molecular Biomarkers, 2020, 24, 674-680.	0.7	5
49	Pleiotropic Locus 15q24.1 Reveals a Gender-Specific Association with Neovascular but Not Atrophic Age-Related Macular Degeneration (AMD). Cells, 2020, 9, 2257.	4.1	5
50	Diagnostic and therapeutic applications of genomic medicine in progressive, late-onset, nonsyndromic sensorineural hearing loss. Gene, 2020, 747, 144677.	2.2	4
51	Relationship between sirtuin and mitochondrial uncoupling protein genes and carotid artery stiffness. Translational Research, 2015, 165, 358-359.	5.0	3
52	Association of IFT88 gene variants with nonsyndromic cleft lip with or without cleft palate. Birth Defects Research, 2019, 111, 659-665.	1.5	3
53	Identification of a genetic variant underlying familial cases of recurrent benign paroxysmal positional vertigo. PLoS ONE, 2021, 16, e0251386.	2.5	2
54	Targeted sequencing of linkage region in Dominican families implicates PRIMA1 and the SPATA7-PTPN21-ZC3H14-EML5-TTC8 locus in carotid-intima media thickness and atherosclerotic events. Scientific Reports, 2019, 9, 11621.	3.3	0

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55	Application of the ACMG / NSGC genetic referral guidelines for hereditary renal cell carcinoma at the University of Miami, from 2014 to 2017. American Journal of Medical Genetics, Part A, 2021, 185, 3012-3018.	1.2	0