Ignacio del Castillo

List of Publications by Year
in descending order

83 papers	5,739 citations	87723 38 h-index	76769 74 g-index
85 all docs	85 docs citations	85 times ranked	5078 citing authors

#	Article	IF	Citations
1	A Deletion Involving the Connexin 30 Gene in Nonsyndromic Hearing Impairment. New England Journal of Medicine, 2002, 346, 243-249.	13.9	557
2	Mutations in the seed region of human miR-96 are responsible for nonsyndromic progressive hearing loss. Nature Genetics, 2009, 41, 609-613.	9.4	483
3	GJB2 Mutations and Degree of Hearing Loss: A Multicenter Study. American Journal of Human Genetics, 2005, 77, 945-957.	2.6	455
4	A novel deletion involving the connexin-30 gene, del(GJB6-d13s1854), found in trans with mutations in the GJB2 gene (connexin-26) in subjects with DFNB1 non-syndromic hearing impairment. Journal of Medical Genetics, 2005, 42, 588-594.	1.5	282
5	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. American Journal of Human Genetics, 2003, 73, 1452-1458.	2.6	269
6	Mutation in TRMU Related to Transfer RNA Modification Modulates the Phenotypic Expression of the Deafness-Associated Mitochondrial 12S Ribosomal RNA Mutations. American Journal of Human Genetics, 2006, 79, 291-302.	2.6	212
7	X chromosome-linked Kallmann syndrome: stop mutations validate the candidate gene.. Proceedings of the National Academy of Sciences of the United States of America, 1992, 89, 8190-8194.	3.3	182
8	Auditory neuropathy in patients carrying mutations in the otoferlin gene (OTOF). Human Mutation, 2003, 22, 451-456.	1.1	181
9	A genotype-phenotype correlation for GJB2 (connexin 26) deafness. Journal of Medical Genetics, 2004, 41, 147-154.	1.5	178
10	Mutations in a new gene encoding a protein of the hair bundle cause non-syndromic deafness at the DFNB16 locus. Nature Genetics, 2001, 29, 345-349.	9.4	159
11	A multicenter study on the prevalence and spectrum of mutations in the otoferlin gene (OTOF) in subjects with nonsyndromic hearing impairment and auditory neuropathy. Human Mutation, 2008, 29, 823-831.	1.1	155
12	Structure of the Xâ€“linked Kallmann syndrome gene and its homologous pseudogene on the Y chromosome. Nature Genetics, 1992, 2, 305-310.	9.4	115
13	The A1555G Mutation in the 12S rRNA Gene of Human mtDNA: Recurrent Origins and Founder Events in Families Affected by Sensorineural Deafness. American Journal of Human Genetics, 1999, 65, 1349-1358.	2.6	111
14	Q829X, a novel mutation in the gene encoding otoferlin (OTOF), is frequently found in Spanish patients with prelingual non-syndromic hearing loss. Journal of Medical Genetics, 2002, 39, 502-506.	1.5	107
15	Heteroplasmy for the 1555A>G mutation in the mitochondrial 12S rRNA gene in six Spanish families with non-syndromic hearing loss. Journal of Medical Genetics, 2003, 40, 632-636.	1.5	107
16	The Escherichia coli Kâ€12 sheA gene encodes a 34â€kDa secreted haemolysin. Molecular Microbiology, 1997, 25, 107-115.	1.2	98
17	A novel DFNB1 deletion allele supports the existence of a distant cisâ€regulatory region that controls GJB2 and GJB6 expression. Clinical Genetics, 2010, 78, 267-274.	1.0	75
18	DFNA8/12 caused by TECTA mutations is the most identified subtype of nonsyndromic autosomal dominant hearing loss. Human Mutation, 2011, 32, 825-834.	1.1	73

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19	An unusual mechanism for resistance to the antibiotic coumermycin A1.. Proceedings of the National Academy of Sciences of the United States of America, 1991, 88, 8860-8864.	3.3	72
20	GJB2: The spectrum of deafness-causing allele variants and their phenotype. Human Mutation, 2004, 24, 305-311.	1.1	72
21	Nonsense Mutations in SMPX, Encoding a Protein Responsive to Physical Force, Result in X-Chromosomal Hearing Loss. American Journal of Human Genetics, 2011, 88, 621-627.	2.6	70
22	Mutations of the Gene Encoding Otogelin Are a Cause of Autosomal-Recessive Nonsyndromic Moderate Hearing Impairment. American Journal of Human Genetics, 2012, 91, 883-889.	2.6	69
23	High prevalence of theW24X mutation in the gene encoding connexin-26 (GJB2) in Spanish Romani (gypsies) with autosomal recessive non-syndromic hearing loss. American Journal of Medical Genetics, Part A, 2005, 137A, 255-258.	0.7	68
24	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	1.1	67
25	DFNB1 Non-syndromic Hearing Impairment: Diversity of Mutations and Associated Phenotypes. Frontiers in Molecular Neuroscience, 2017, 10, 428.	1.4	66
26	In vivo and in vitro effects of two novel gamma-actin (ACTG1) mutations that cause DFNA20/26 hearing impairment. Human Molecular Genetics, 2009, 18, 3075-3089.	1.4	64
27	Construction and Characterization of Mutations at Codon 751 of the Escherichia coli gyrB Gene That Confer Resistance to the Antimicrobial Peptide Microcin B17 and Alter the Activity of DNA Gyrase. Journal of Bacteriology, 2001, 183, 2137-2140.	1.0	61
28	Abnormal Cochlear Potentials from Deaf Patients with Mutations in the Otoferlin Gene. JARO - Journal of the Association for Research in Otolaryngology, 2009, 10, 545-556.	0.9	61
29	A Mutation in CCDC50, a Gene Encoding an Effector of Epidermal Growth Factorâ€“Mediated Cell Signaling, Causes Progressive Hearing Loss. American Journal of Human Genetics, 2007, 80, 1076-1089.	2.6	57
30	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genetics in Medicine, 2019, 21, 2442-2452.	1.1	56
31	Audibility, speech perception and processing of temporal cues in ribbon synaptic disorders due to OTOF mutations. Hearing Research, 2015, 330, 200-212.	0.9	55
32	Tryptophanâ€rich basic protein (<scp>WRB</scp>) mediates insertion of the tailâ€anchored protein otoferlin and is required for hair cell exocytosis and hearing. EMBO Journal, 2016, 35, 2536-2552.	3.5	55
33	Progressive hearing loss and vestibular dysfunction caused by a homozygous nonsense mutation in CLIC5. European Journal of Human Genetics, 2015, 23, 189-194.	1.4	49
34	The DFNB1 subtype of autosomal recessive non-syndromic hearing impairment. Frontiers in Bioscience - Landmark, 2011, 16, 3252.	3.0	47
35	Phenotypic variability of patients homozygous for the GJB2 mutation 35delG cannot be explained by the influence of one major modifier gene. European Journal of Human Genetics, 2009, 17, 517-524.	1.4	46
36	A novel KCNQ4 pore-region mutation (p.G296S) causes deafness by impairing cell-surface channel expression. Human Genetics, 2008, 123, 41-53.	1.8	45

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37	Characterization of the Chicken and Quail Homologues of the Human Gene Responsible for the X-Linked Kallmann Syndrome. Genomics, 1993, 17, 516-518.	1.3	42
38	A cysteine substitution in the zona pellucida domain of alpha-tectorin results in autosomal dominant, postlingual, progressive, mid frequency hearing loss in a Spanish family. Journal of Medical Genetics, 2001, 38, 13e-13.	1.5	42
39	Novel mutation in the gene encoding the GATA3 transcription factor in a Spanish familial case of hypoparathyroidism, deafness, and renal dysplasia (HDR) syndrome with female genital tract malformations. American Journal of Medical Genetics, Part A, 2007, 143A, 757-762.	0.7	36
40	A novel locus for non-syndromic sensorineural deafness (DFN6) maps to chromosome Xp22. Human Molecular Genetics, 1996, 5, 1383-1387.	1.4	34
41	MPZL2, Encoding the Epithelial Junctional Protein Myelin Protein Zero-like 2, Is Essential for Hearing in Man and Mouse. American Journal of Human Genetics, 2018, 103, 74-88.	2.6	34
42	CSVS, a crowdsourcing database of the Spanish population genetic variability. Nucleic Acids Research, 2021, 49, D1130-D1137.	6.5	34
43	Maternally inherited non-syndromic hearing impairment in a Spanish family with the 7510T>C mutation in the mitochondrial tRNASer(UCN) gene. Journal of Medical Genetics, 2002, 39, 82e-82.	1.5	31
44	De novo mutation in the gene encoding connexin-26 (GJB2) in a sporadic case of keratitis-ichthyosis-deafness (KID) syndrome. , 2003, 117A, 89-91.		31
45	Sensorineural Hearing Loss and Mondini Dysplasia Caused by a Deletion at Locus DFN3. JAMA Otolaryngology, 2000, 126, 1065.	1.5	28
46	EMQN Best Practice guidelines for diagnostic testing of mutations causing non-syndromic hearing impairment at the DFNB1 locus. European Journal of Human Genetics, 2013, 21, 1325-1329.	1.4	28
47	A novel locus for autosomal dominant nonsyndromic hearing loss, DFNA50, maps to chromosome 7q32 between the DFNB17 and DFNB13 deafness loci. Journal of Medical Genetics, 2004, 41, 14e-14.	1.5	27
48	Deafness Locus DFNB16 Is Located on Chromosome 15q13-q21 within a 5-cM Interval Flanked by Markers D15S994 and D15S132. American Journal of Human Genetics, 1999, 64, 1238-1241.	2.6	25
49	A novel mutation in the gene encoding TIMM8a, a component of the mitochondrial protein translocase complexes, in a Spanish familial case of deafness-dystonia (Mohrâ€“Tranebjaerg) syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 392-397.	0.7	25
50	Genetics of isolated auditory neuropathies. Frontiers in Bioscience - Landmark, 2012, 17, 1251.	3.0	25
51	Genetic etiology of non-syndromic hearing loss in Europe. Human Genetics, 2022, 141, 683-696.	1.8	23
52	A novel missense mutation in the ESRRB gene causes DFNB35 hearing loss in a Tunisian family. European Journal of Medical Genetics, 2011, 54, e535-e541.	0.7	22
53	A novel locus for autosomal dominant nonsyndromic hearing loss (DFNA44) maps to chromosome 3q28-29. Human Genetics, 2003, 112, 24-28.	1.8	21
54	Similar Phenotypes Caused by Mutations in OTOG and OTOGL. Ear and Hearing, 2014, 35, e84-e91.	1.0	21

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55	A de novo missense mutation in the gene encoding the SOX10 transcription factor in a Spanish sporadic case of Waardenburg syndrome type IV. American Journal of Medical Genetics, Part A, 2008, 146A, 1032-1037.	0.7	20
56	Isolation and characterization of the gene responsible for the X chromosome-linked Kallmann syndrome. Biomedicine and Pharmacotherapy, 1994, 48, 241-246.	2.5	19
57	Secretion of the Escherichia coli K-12 SheA hemolysin is independent of its cytolytic activity. FEMS Microbiology Letters, 2001, 204, 281-285.	0.7	17
58	Uniparental disomy of chromosome 13q causing homozygosity for the 35delG mutation in the gene encoding connexin26 (GJB2) results in prelingual hearing impairment in two unrelated Spanish patients. Journal of Medical Genetics, 2003, 40, 636-639.	1.5	17
59	Stickler and branchioâ€otoâ€renal syndromes in a patient with mutations in EYA1 and COL2A1 genes. Clinical Genetics, 2008, 73, 262-267.	1.0	17
60	A Novel Splice-Site Mutation in the GJB2 Gene Causing Mild Postlingual Hearing Impairment. PLoS ONE, 2013, 8, e73566.	1.1	17
61	Genetics of Deafness. Genetics Research International, 2012, 2012, 1-1.	2.0	15
62	Mutations in PRPS1 causing syndromic or nonsyndromic hearing impairment: intrafamilial phenotypic variation complicates genetic counseling. Pediatric Research, 2015, 78, 97-102.	1.1	15
63	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. Scientific Reports, 2020, 10, 6213.	1.6	15
64	Haplogroup analysis supports a pathogenic role for the 7510T>C mutation of mitochondrial tRNA^{Ser(UCN)} in sensorineural hearing loss. Clinical Genetics, 2008, 73, 50-54.	1.0	14
65	Perrault syndrome with neurological features in a compound heterozygote for two TWNK mutations: overlap of TWNK-related recessive disorders. Journal of Translational Medicine, 2019, 17, 290.	1.8	14
66	Characterization of the genes encoding the SheA haemolysin in Escherichia coli O157:H7 and Shigella flexneri 2a. Research in Microbiology, 2000, 151, 229-230.	1.0	13
67	A Spanish sporadic case of deafnessâ€“dystonia (Mohr-Tranebjaerg) syndrome with a novel mutation in the gene encoding TIMM8a, a component of the mitochondrial protein translocase complexes. Neuromuscular Disorders, 2008, 18, 979-981.	0.3	13
68	Auditory neuropathies and electrocochleography. Hearing, Balance and Communication, 2013, 11, 130-137.	0.1	13
69	DFNA49, a novel locus for autosomal dominant non-syndromic hearing loss, maps proximal to DFNA7/DFNM1 region on chromosome 1q21-q23. Journal of Medical Genetics, 2003, 40, 832-836.	1.5	12
70	Genetic and phenotypic heterogeneity in two novel cases of Waardenburg syndrome type IV. American Journal of Medical Genetics, Part A, 2009, 149A, 2296-2302.	0.7	12
71	Hypothesizing an Ancient Greek Origin of theGJB235delG Mutation: Can Science Meet History?. Genetic Testing and Molecular Biomarkers, 2010, 14, 183-187.	0.3	12
72	Characterization of the promoter of the human KAL gene, responsible for the X-chromosome-linked Kallmann syndrome. Gene, 1995, 164, 235-242.	1.0	10

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73	DFNB66 and DFNB67 loci are non allelic and rarely contribute to autosomal recessive nonsyndromic hearing loss. European Journal of Medical Genetics, 2011, 54, e565-e569.	0.7	10
74	Presynaptic and postsynaptic mechanisms underlying auditory neuropathy in patients with mutations in theOTOForOPA1gene. Audiological Medicine, 2011, 9, 59-66.	0.4	10
75	The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call. Genes, 2021, 12, 1590.	1.0	8
76	Novel Pathogenic Variants in PJVK, the Gene Encoding Pejvakin, in Subjects with Autosomal Recessive Non-Syndromic Hearing Impairment and Auditory Neuropathy Spectrum Disorder. Genes, 2022, 13, 149.	1.0	8
77	Cochlear Synaptopathy due to Mutations in OTOF Gene May Result in Stable Mild Hearing Loss and Severe Impairment of Speech Perception. Ear and Hearing, 2021, 42, 1627-1639.	1.0	7
78	A Novel Truncating Mutation in HOMER2 Causes Nonsyndromic Progressive DFNA68 Hearing Loss in a Spanish Family. Genes, 2021, 12, 411.	1.0	5
79	Novel splice-site mutation c.1615-2A>G (IVS14-2A>G) in the SLC26A4 gene causing Pendred syndrome in a consanguineous Portuguese family. , 2011, 155, 924-927.		4
80	Identification of a SNP in a Regulatory Region of GJB2 Associated With Idiopathic Nonsyndromic Autosomal Recessive Hearing Loss in a Multicenter Study. Otology and Neurotology, 2013, 34, 650-656.	0.7	3
81	Dinucleotide repeat polymorphisms at the D5S1356, D5S1357 and D7S1480 loci. Human Molecular Genetics, 1994, 3, 1441-1441.	1.4	2
82	Electrocochleography in Auditory Neuropathy Related to Mutations in the OTOF or OPA1 Gene. Audiology Research, 2021, 11, 639-652.	0.8	2
83	Genetics of Hearing Impairment. Genes, 2022, 13, 852.	1.0	0