Miguel Angel Moreno-Pelayo

List of Publications by Year
in descending order

41 papers	3,020 citations	331670 21 h-index	302126 39 g-index
44 all docs	44 docs citations	44 times ranked	3462 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.	27.0	557
2	Mutations in the seed region of human miR-96 are responsible for nonsyndromic progressive hearing loss. Nature Genetics, 2009, 41, 609-613.	21.4	483
3	An ENU-induced mutation of miR-96 associated with progressive hearing loss in mice. Nature Genetics, 2009, 41, 614-618.	21.4	281
4	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.	6.2	269
5	Auditory neuropathy in patients carrying mutations in the otoferlin gene (OTOF). Human Mutation, 2003, 22, 451-456.	2.5	181
6	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.	2.5	155
7	KCNQ4 K+ channels tune mechanoreceptors for normal touch sensation in mouse and man. Nature Neuroscience, 2012, 15, 138-145.	14.8	95
8	DFNA8/12 caused by TECTA mutations is the most identified subtype of nonsyndromic autosomal dominant hearing loss. Human Mutation, 2011, 32, 825-834.	2.5	73
9	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.	1.2	68
10	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
11	Differential Biological Role of CD3 Chains Revealed by Human Immunodeficiencies. Journal of Immunology, 2007, 178, 2556-2564.	0.8	64
12	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.	2.9	64
13	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.	6.2	57
14	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.	2.4	56
15	Allelic Mutations of KITLG, Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. American Journal of Human Genetics, 2015, 97, 647-660.	6.2	55
16	A leaky mutation in CD3D differentially affects Î±Î² and Î³Î´ T cells and leads to a TÎ±Î²â€“TÎ³Î´+B+NK+ human SCID. Journal of Clinical Investigation, 2011, 121, 3872-3876.	8.2	46
17	A novel KCNQ4 pore-region mutation (p.G296S) causes deafness by impairing cell-surface channel expression. Human Genetics, 2008, 123, 41-53.	3.8	45
18	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.	1.2	36

#	Article	IF	Citations
19	CSVS, a crowdsourcing database of the Spanish population genetic variability. Nucleic Acids Research, 2021, 49, D1130-D1137.	14.5	34
20	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
21	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.	1.2	25
22	Gly111Ser mutation in CD8A gene causing CD8 immunodeficiency is found in Spanish Gypsies. Molecular Immunology, 2008, 45, 479-484.	2.2	25
23	Genetic etiology of non-syndromic hearing loss in Europe. Human Genetics, 2022, 141, 683-696.	3.8	23
24	A novel locus for autosomal dominant nonsyndromic hearing loss (DFNA44) maps to chromosome 3q28-29. Human Genetics, 2003, 112, 24-28.	3.8	21
25	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.	1.2	20
26	Simple Protocol for Generating and Genotyping Genomeâ€Edited Mice With CRISPRâ€Cas9 Reagents. Current Protocols in Mouse Biology, 2020, 10, e69.	1.2	18
27	Primary T-cell immunodeficiency with functional revertant somatic mosaicism in CD247. Journal of Allergy and Clinical Immunology, 2017, 139, 347-349.e8.	2.9	17
28	A Novel Splice-Site Mutation in the GJB2 Gene Causing Mild Postlingual Hearing Impairment. PLoS ONE, 2013, 8, e73566.	2.5	17
29	Characterization of a Spontaneous, Recessive, Missense Mutation Arising in the Tecta Gene. JARO - Journal of the Association for Research in Otolaryngology, 2008, 9, 202-214.	1.8	16
30	Mutations in PRPS1 causing syndromic or nonsyndromic hearing impairment: intrafamilial phenotypic variation complicates genetic counseling. Pediatric Research, 2015, 78, 97-102.	2.3	15
31	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. Scientific Reports, 2020, 10, 6213.	3.3	15
32	Therapeutic Potential of EWSR1â€“FLI1 Inactivation by CRISPR/Cas9 in Ewing Sarcoma. Cancers, 2021, 13, 3783.	3.7	15
33	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.	4.4	14
34	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.	0.6	13
35	Genetic and phenotypic heterogeneity in two novel cases of Waardenburg syndrome type IV. American Journal of Medical Genetics, Part A, 2009, 149A, 2296-2302.	1.2	12
36	Novel mutations in the KCNJ10 gene associated to a distinctive ataxia, sensorineural hearing loss and spasticity clinical phenotype. Neurogenetics, 2020, 21, 135-143.	1.4	9

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37	Three New Mutations and Mild, Asymmetrical Phenotype in the Highly Distinctive LAMM Syndrome: A Report of Eight Further Cases. Genes, 2019, 10, 529.	2.4	8
38	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.	2.4	8
39	A Novel Truncating Mutation in HOMER2 Causes Nonsyndromic Progressive DFNA68 Hearing Loss in a Spanish Family. Genes, 2021, 12, 411.	2.4	5
40	CRISPR/Cas9-Mediated Allele-Specific Disruption of a Dominant COL6A1 Pathogenic Variant Improves Collagen VI Network in Patient Fibroblasts. International Journal of Molecular Sciences, 2022, 23, 4410.	4.1	5
41	Actin Mutations and Deafness. , 2012, , 169-180.		0