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	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
2	Genetic etiology of non-syndromic hearing loss in Europe. Human Genetics, 2022, 141, 683-696.	3.8	23
3	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
4	CSVS, a crowdsourcing database of the Spanish population genetic variability. Nucleic Acids Research, 2021, 49, D1130-D1137.	14.5	34
5	A Novel Truncating Mutation in HOMER2 Causes Nonsyndromic Progressive DFNA68 Hearing Loss in a Spanish Family. Genes, 2021, 12, 411.	2.4	5
6	Therapeutic Potential of EWSR1â€“FLI1 Inactivation by CRISPR/Cas9 in Ewing Sarcoma. Cancers, 2021, 13, 3783.	3.7	15
7	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
8	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. Scientific Reports, 2020, 10, 6213.	3.3	15
9	Simple Protocol for Generating and Genotyping Genomeâ€Edited Mice With CRISPRâ€Cas9 Reagents. Current Protocols in Mouse Biology, 2020, 10, e69.	1.2	18
10	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
11	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
12	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
13	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
14	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
15	Mutations in PRPS1 causing syndromic or nonsyndromic hearing impairment: intrafamilial phenotypic variation complicates genetic counseling. Pediatric Research, 2015, 78, 97-102.	2.3	15
16	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
17	A Novel Splice-Site Mutation in the GJB2 Gene Causing Mild Postlingual Hearing Impairment. PLoS ONE, 2013, 8, e73566.	2.5	17
18	KCNQ4 K+ channels tune mechanoreceptors for normal touch sensation in mouse and man. Nature Neuroscience, 2012, 15, 138-145.	14.8	95

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19	Actin Mutations and Deafness. , 2012, , 169-180.		0
20	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
21	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
22	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
23	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
24	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
25	An ENU-induced mutation of miR-96 associated with progressive hearing loss in mice. Nature Genetics, 2009, 41, 614-618.	21.4	281
26	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
27	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
28	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
29	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
30	Gly111Ser mutation in CD8A gene causing CD8 immunodeficiency is found in Spanish Gypsies. Molecular Immunology, 2008, 45, 479-484.	2.2	25
31	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
32	Differential Biological Role of CD3 Chains Revealed by Human Immunodeficiencies. Journal of Immunology, 2007, 178, 2556-2564.	0.8	64
33	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
34	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
35	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
36	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

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37	A novel locus for autosomal dominant nonsyndromic hearing loss (DFNA44) maps to chromosome 3q28-29. Human Genetics, 2003, 112, 24-28.	3.8	21
38	Auditory neuropathy in patients carrying mutations in the otoferlin gene (OTOF). Human Mutation, 2003, 22, 451-456.	2.5	181
39	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
40	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
41	A Deletion Involving the Connexin 30 Gene in Nonsyndromic Hearing Impairment. New England Journal of Medicine, 2002, 346, 243-249.	27.0	557