Teresa Jaijo

List of Publications by Year in descending order

39 papers	1,113 citations	516710 16 h-index	434195 31 g-index
39 all docs	39 docs citations	39 times ranked	1587 citing authors

#	Article	IF	Citations
1	Axonemal Symmetry Break, a New Ultrastructural Diagnostic Tool for Primary Ciliary Dyskinesia?. Diagnostics, 2022, 12, 129.	2.6	2
2	Functional assays of non-canonical splice-site variants in inherited retinal dystrophies genes. Scientific Reports, 2022, 12, 68.	3.3	5
3	Modeling a Novel Variant of Glycogenosis IXa Using a Clonal Inducible Reprogramming System to Generate â€œDiseasedâ€•Hepatocytes for Accurate Diagnosis. Journal of Personalized Medicine, 2022, 12, 1111.	2.5	2
4	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. Scientific Reports, 2021, 11, 1526.	3.3	71
5	Prevalent ALMS1 Pathogenic Variants in Spanish AlstrÃ¶m Patients. Genes, 2021, 12, 282.	2.4	4
6	Genotypeâ€“phenotype correlation in patients with Usher syndrome and pathogenic variants in MYO7A: implications for future clinical trials. Acta Ophthalmologica, 2021, 99, 922-930.	1.1	8
7	Hereditary Spastic Paraplegia 7 Presenting as Multifocal Dystonia with Prominent <scp>Crânioâ€Cervical</scp> Involvement. Movement Disorders Clinical Practice, 2021, 8, 966-968.	1.5	4
8	Usher Syndrome: Genetics of a Human Ciliopathy. International Journal of Molecular Sciences, 2021, 22, 6723.	4.1	38
9	Updating the Genetic Landscape of Inherited Retinal Dystrophies. Frontiers in Cell and Developmental Biology, 2021, 9, 645600.	3.7	18
10	Presenilin-1 Mutations Are a Cause of Primary Lateral Sclerosis-Like Syndrome. Frontiers in Molecular Neuroscience, 2021, 14, 721047.	2.9	3
11	CONCOMITANT MUTATIONS IN INHERITED RETINAL DYSTROPHIES. Retina, 2021, 41, 1966-1975.	1.7	2
12	Clinical and genetic characteristics of 21 Spanish patients with biallelic pathogenic SPG7 mutations. Journal of the Neurological Sciences, 2021, 429, 118062.	0.6	5
13	Epilepsy, status epilepticus, and hemiplegic migraine coexisting with a novel SLC4A4 mutation. Neurological Sciences, 2021, 42, 3647-3654.	1.9	8
14	Improving the Management of Patients with Hearing Loss by the Implementation of an NGS Panel in Clinical Practice. Genes, 2020, 11, 1467.	2.4	16
15	Expanding the Clinical and Molecular Heterogeneity of Nonsyndromic Inherited RetinalÂDystrophies. Journal of Molecular Diagnostics, 2020, 22, 532-543.	2.8	27
16	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. Molecular Vision, 2020, 26, 216-225.	1.1	2
17	Genetic Screening of the Usher Syndrome in Cuba. Frontiers in Genetics, 2019, 10, 501.	2.3	7
18	Expanding the Genetic Landscape of Usher-Like Phenotypes. , 2019, 60, 4701.		9

#	Article	IF	Citations
19	The importance of biochemical and genetic findings in the diagnosis of atypical Norrie disease. Clinical Chemistry and Laboratory Medicine, 2018, 56, 229-235.	2.3	17
20	High-throughput sequencing for the molecular diagnosis of Usher syndrome reveals 42 novel mutations and consolidates CEP250 as Usher-like disease causative. Scientific Reports, 2018, 8, 17113.	3.3	30
21	USH2A Gene Editing Using the CRISPR System. Molecular Therapy - Nucleic Acids, 2017, 8, 529-541.	5.1	56
22	Clinical Aspects of Usher Syndrome and theUSH2AGene in a Cohort of 433 Patients. JAMA Ophthalmology, 2015, 133, 157.	2.5	59
23	Targeted next generation sequencing for molecular diagnosis of Usher syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 168.	2.7	61
24	Novel deletions involving the USH2A gene in patients with Usher syndrome and retinitis pigmentosa. Molecular Vision, 2014, 20, 1398-410.	1.1	12
25	Phosphodiesterase inhibition induces retinal degeneration, oxidative stress and inflammation in cone-enriched cultures of porcine retina. Experimental Eye Research, 2013, 111, 122-133.	2.6	24
26	Study of USH1 Splicing Variants through Minigenes and Transcript Analysis from Nasal Epithelial Cells. PLoS ONE, 2013, 8, e57506.	2.5	21
27	Altered Antioxidant-Oxidant Status in the Aqueous Humor and Peripheral Blood of Patients with Retinitis Pigmentosa. PLoS ONE, 2013, 8, e74223.	2.5	64
28	A Genetic Basis for Mechanosensory Traits in Humans. PLoS Biology, 2012, 10, e1001318.	5.6	61
29	Mutation screening of the PCDH15 gene in Spanish patients with Usher syndrome type I. Molecular Vision, 2012, 18, 1719-26.	1.1	11
30	Two novel disease-causing mutations in the CLRN1 gene in patients with Usher syndrome type 3. Molecular Vision, 2012, 18, 3070-8.	1.1	9
31	An Update on the Genetics of Usher Syndrome. Journal of Ophthalmology, 2011, 2011, 1-8.	1.3	160
32	Mutational screening of the USH2A gene in Spanish USH patients reveals 23 novel pathogenic mutations. Orphanet Journal of Rare Diseases, 2011, 6, 65.	2.7	47
33	The USH2A c.2299delG mutation: dating its common origin in a Southern European population. European Journal of Human Genetics, 2010, 18, 788-793.	2.8	47
34	Microarray-Based Mutation Analysis of 183 Spanish Families with Usher Syndrome. , 2010, 51, 1311.		57
35	Identification of Large Rearrangements of thePCDH15Gene by Combined MLPA and a CGH: Large Duplications Are Responsible for Usher Syndrome. , 2010, 51, 5480.		28
36	Sequence variants of the DFNB31 gene among Usher syndrome patients of diverse origin. Molecular Vision, 2010, 16, 495-500.	1.1	13

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37	Novel mutations in the USH1C gene in Usher syndrome patients. Molecular Vision, 2010, 16, 2948-54.	1.1	5
38	Vocal cord paresis and diaphragmatic dysfunction are severe and frequent symptoms of GDAP1-associated neuropathy. Brain, 2008, 131, 3051-3061.	7.6	89
39	Screening of theUSH1GGene among Spanish Patients with Usher Syndrome. Lack of Mutations and Evidence of a Minor Role in the Pathogenesis of the Syndrome. Ophthalmic Genetics, 2007, 28, 151-155.	1.2	11