

# Teresa Jaijo

## List of Publications by Year in descending order

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Version: 2024-02-01

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	An Update on the Genetics of Usher Syndrome. <i>Journal of Ophthalmology</i> , 2011, 2011, 1-8.	1.3	160
2	Vocal cord paresis and diaphragmatic dysfunction are severe and frequent symptoms of GDAP1-associated neuropathy. <i>Brain</i> , 2008, 131, 3051-3061.	7.6	89
3	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. <i>Scientific Reports</i> , 2021, 11, 1526.	3.3	71
4	Altered Antioxidant-Oxidant Status in the Aqueous Humor and Peripheral Blood of Patients with Retinitis Pigmentosa. <i>PLoS ONE</i> , 2013, 8, e74223.	2.5	64
5	A Genetic Basis for Mechanosensory Traits in Humans. <i>PLoS Biology</i> , 2012, 10, e1001318.	5.6	61
6	Targeted next generation sequencing for molecular diagnosis of Usher syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 168.	2.7	61
7	Clinical Aspects of Usher Syndrome and the <i>USH2A</i> Gene in a Cohort of 433 Patients. <i>JAMA Ophthalmology</i> , 2015, 133, 157.	2.5	59
8	Microarray-Based Mutation Analysis of 183 Spanish Families with Usher Syndrome. , 2010, 51, 1311.		57
9	USH2A Gene Editing Using the CRISPR System. <i>Molecular Therapy - Nucleic Acids</i> , 2017, 8, 529-541.	5.1	56
10	The <i>USH2A</i> c.2299delG mutation: dating its common origin in a Southern European population. <i>European Journal of Human Genetics</i> , 2010, 18, 788-793.	2.8	47
11	Mutational screening of the <i>USH2A</i> gene in Spanish USH patients reveals 23 novel pathogenic mutations. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 65.	2.7	47
12	Usher Syndrome: Genetics of a Human Ciliopathy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6723.	4.1	38
13	High-throughput sequencing for the molecular diagnosis of Usher syndrome reveals 42 novel mutations and consolidates <i>CEP250</i> as Usher-like disease causative. <i>Scientific Reports</i> , 2018, 8, 17113.	3.3	30
14	Identification of Large Rearrangements of the <i>PCDH15</i> Gene by Combined MLPA and a CGH: Large Duplications Are Responsible for Usher Syndrome. , 2010, 51, 5480.		28
15	Expanding the Clinical and Molecular Heterogeneity of Nonsyndromic Inherited Retinal Dystrophies. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 532-543.	2.8	27
16	Phosphodiesterase inhibition induces retinal degeneration, oxidative stress and inflammation in cone-enriched cultures of porcine retina. <i>Experimental Eye Research</i> , 2013, 111, 122-133.	2.6	24
17	Study of <i>USH1</i> Splicing Variants through Minigenes and Transcript Analysis from Nasal Epithelial Cells. <i>PLoS ONE</i> , 2013, 8, e57506.	2.5	21
18	Updating the Genetic Landscape of Inherited Retinal Dystrophies. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 645600.	3.7	18

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19	The importance of biochemical and genetic findings in the diagnosis of atypical Norrie disease. <i>Clinical Chemistry and Laboratory Medicine</i> , 2018, 56, 229-235.	2.3	17
20	Improving the Management of Patients with Hearing Loss by the Implementation of an NGS Panel in Clinical Practice. <i>Genes</i> , 2020, 11, 1467.	2.4	16
21	Sequence variants of the DFNB31 gene among Usher syndrome patients of diverse origin. <i>Molecular Vision</i> , 2010, 16, 495-500.	1.1	13
22	Novel deletions involving the USH2A gene in patients with Usher syndrome and retinitis pigmentosa. <i>Molecular Vision</i> , 2014, 20, 1398-410.	1.1	12
23	Screening of the USH1G Gene among Spanish Patients with Usher Syndrome. Lack of Mutations and Evidence of a Minor Role in the Pathogenesis of the Syndrome. <i>Ophthalmic Genetics</i> , 2007, 28, 151-155.	1.2	11
24	Mutation screening of the PCDH15 gene in Spanish patients with Usher syndrome type I. <i>Molecular Vision</i> , 2012, 18, 1719-26.	1.1	11
25	Expanding the Genetic Landscape of Usher-Like Phenotypes. , 2019, 60, 4701.		9
26	Two novel disease-causing mutations in the CLRN1 gene in patients with Usher syndrome type 3. <i>Molecular Vision</i> , 2012, 18, 3070-8.	1.1	9
27	Genotype-phenotype correlation in patients with Usher syndrome and pathogenic variants in <i>MYO7A</i> : implications for future clinical trials. <i>Acta Ophthalmologica</i> , 2021, 99, 922-930.	1.1	8
28	Epilepsy, status epilepticus, and hemiplegic migraine coexisting with a novel SLC4A4 mutation. <i>Neurological Sciences</i> , 2021, 42, 3647-3654.	1.9	8
29	Genetic Screening of the Usher Syndrome in Cuba. <i>Frontiers in Genetics</i> , 2019, 10, 501.	2.3	7
30	Clinical and genetic characteristics of 21 Spanish patients with biallelic pathogenic SPG7 mutations. <i>Journal of the Neurological Sciences</i> , 2021, 429, 118062.	0.6	5
31	Novel mutations in the USH1C gene in Usher syndrome patients. <i>Molecular Vision</i> , 2010, 16, 2948-54.	1.1	5
32	Functional assays of non-canonical splice-site variants in inherited retinal dystrophies genes. <i>Scientific Reports</i> , 2022, 12, 68.	3.3	5
33	Prevalent ALMS1 Pathogenic Variants in Spanish Alstr�m Patients. <i>Genes</i> , 2021, 12, 282.	2.4	4
34	Hereditary Spastic Paraplegia 7 Presenting as Multifocal Dystonia with Prominent Cranio-Cervical Involvement. <i>Movement Disorders Clinical Practice</i> , 2021, 8, 966-968.	1.5	4
35	Presenilin-1 Mutations Are a Cause of Primary Lateral Sclerosis-Like Syndrome. <i>Frontiers in Molecular Neuroscience</i> , 2021, 14, 721047.	2.9	3
36	CONCOMITANT MUTATIONS IN INHERITED RETINAL DYSTROPHIES. <i>Retina</i> , 2021, 41, 1966-1975.	1.7	2

#	ARTICLE	IF	CITATIONS
37	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. <i>Molecular Vision</i> , 2020, 26, 216-225.	1.1	2
38	Axonemal Symmetry Break, a New Ultrastructural Diagnostic Tool for Primary Ciliary Dyskinesia?. <i>Diagnostics</i> , 2022, 12, 129.	2.6	2
39	Modeling a Novel Variant of Glycogenosis IXa Using a Clonal Inducible Reprogramming System to Generate "Diseased" Hepatocytes for Accurate Diagnosis. <i>Journal of Personalized Medicine</i> , 2022, 12, 1111.	2.5	2