Juan Carlos Zenteno

List of Publications by Year
in descending order

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92	1,778	304743 22	330143 37
papers	citations	h-index	g-index

93	93	93	4979
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. Human Genetics, 2014, 133, 331-345.	3.8	204
2	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. Nature Genetics, 2015, 47, 387-392.	21.4	97
3	Review and update on the molecular basis of Leber congenital amaurosis. World Journal of Clinical Cases, 2015, 3, 112.	0.8	94
4	A Novel Mutation Confirms MFRP as the Gene Causing the Syndrome of Nanophthalmosâ€“Renititis Pigmentosaâ€“Foveoschisisâ€“Optic Disk Drusen. American Journal of Ophthalmology, 2008, 146, 323-328.e1.	3.3	86
5	A new autosomal recessive syndrome consisting of posterior microphthalmos, retinitis pigmentosa, foveoschisis, and optic disc drusen is caused by a MFRP gene mutation. Molecular Vision, 2006, 12, 1483-9.	1.1	80
6	Two SRY -negative XX male brothers without genital ambiguity. Human Genetics, 1997, 100, 606-610.	3.8	56
7	Mutations in a novel serine protease PRSS56 in families with nanophthalmos. Molecular Vision, 2011, 17, 1850-61.	1.1	43
8	Contiguous gene syndrome due to deletion of the first three exons of the Kallmann gene and complete deletion of the steroid sulphatase gene. Clinical Endocrinology, 1998, 48, 713-718.	2.4	41
9	Anophthalmia-esophageal atresia syndrome caused by anSOX2 gene deletion in monozygotic twin brothers with markedly discordant phenotypes. American Journal of Medical Genetics, Part A, 2006, 140A, 1899-1903.	1.2	40
10	Expansion of the phenotypic spectrum and description of molecular findings in a cohort of patients with oculocutaneous mosaic RASopathies. Molecular Genetics & Genomic Medicine, 2019, 7, e625.	1.2	39
11	Homozygosity mapping identifies the Crumbs homologue 1 (Crb1) gene as responsible for a recessive syndrome of retinitis pigmentosa and nanophthalmos. American Journal of Medical Genetics, Part A, 2011, 155, 1001-1006.	1.2	38
12	Compound heterozygosity for a novel and a recurrent MFRP gene mutation in a family with the nanophthalmos-retinitis pigmentosa complex. Molecular Vision, 2009, 15, 1794-8.	1.1	38
13	Gene Therapy for Retinitis Pigmentosa Caused by MFRP Mutations: Human Phenotype and Preliminary Proof of Concept. Human Gene Therapy, 2012, 23, 367-376.	2.7	35
14	Next generation sequencing uncovers a missense mutation in COL4A1 as the cause of familial retinal arteriolar tortuosity. Graefe's Archive for Clinical and Experimental Ophthalmology, 2014, 252, 1789-1794.	1.9	33
15	Homozygous FOXE3 mutations cause non-syndromic, bilateral, total sclerocornea, aphakia, microphthalmia and optic disc coloboma. Molecular Vision, 2010, 16, 1162-8.	1.1	33
16	CHM gene molecular analysis and Xâ€chromosome inactivation pattern determination in two families with choroideremia. American Journal of Medical Genetics, Part A, 2009, 149A, 2134-2140.	1.2	32
17	Partially deleted SRY gene confined to testicular tissue in a 46,XX true hermaphrodite without SRY in leukocytic DNA. American Journal of Medical Genetics Part A, 2000, 93, 417-420.	2.4	31
18	Prevalence of high-risk alleles in theLOXL1gene and its association with pseudoexfoliation syndrome and exfoliation glaucoma in a Latin American population. Ophthalmic Genetics, 2012, 33, 12-17.	1.2	31

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19	Clinical and genetic features of TGFBI-linked corneal dystrophies in Mexican population: Description of novel mutations and novel genotypeâ€“phenotype correlations. Experimental Eye Research, 2009, 89, 172-177.	2.6	30
20	Extensive genic and allelic heterogeneity underlying inherited retinal dystrophies in Mexican patients molecularly analyzed by nextâ€generation sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, .	1.2	30
21	Mutation update: TGFBI pathogenic and likely pathogenic variants in corneal dystrophies. Human Mutation, 2019, 40, 675-693.	2.5	28
22	Novel CYP4V2 Gene Mutation in a Mexican Patient with Bietti's Crystalline Corneoretinal Dystrophy. Current Eye Research, 2008, 33, 313-318.	1.5	23
23	An atypical contiguous gene syndrome: molecular studies in a family with X-linked Kallmann's syndrome and X-linked ichthyosis. Clinical Endocrinology, 1999, 50, 157-162.	2.4	22
24	Next generation sequencingâ€based molecular diagnosis in familial congenital cataract expands the mutational spectrum in known congenital cataract genes. American Journal of Medical Genetics, Part A, 2018, 176, 2637-2645.	1.2	22
25	Contribution of CYP1B1 Mutations and Founder Effect to Primary Congenital Glaucoma in Mexico. Journal of Glaucoma, 2008, 17, 189-192.	1.6	20
26	Expanding the phenotype of gingival fibromatosisâ€“mental retardationâ€“hypertrichosis (Zimmermannâ€“Laband) syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1716-1720.	1.2	19
27	Expanding the mutational spectrum in TGFBI-linked corneal dystrophies: Identification of a novel and unusual mutation (Val113Ile) in a family with granular dystrophy. Molecular Vision, 2006, 12, 331-5.	1.1	19
28	Phenotypic Heterogeneity Associated with Identical Mutations in Residue 870 of the Androgen Receptor. Hormone Research in Paediatrics, 2002, 57, 90-93.	1.8	18
29	Association study of multiple gene polymorphisms with the risk of adult-onset primary open-angle glaucoma in a Mexican population. Experimental Eye Research, 2013, 107, 59-64.	2.6	18
30	Novel and recurrent NDP gene mutations in familial cases of Norrie disease and Xâ€linked exudative vitreoretinopathy. Clinical and Experimental Ophthalmology, 2010, 38, 367-374.	2.6	17
31	Identification of unsuspected Wolfram syndrome cases through clinical assessment and WFS1 gene screening in type 1 diabetes mellitus patients. Gene, 2015, 566, 63-67.	2.2	16
32	Novel FREM1 mutations in a patient with MOTA syndrome: Clinical findings, mutation update and review of FREM1 -related disorders literature. European Journal of Medical Genetics, 2017, 60, 190-194.	1.3	16
33	Epidemiological and Molecular Characterization of a Mexican Population Isolate with High Prevalence of Limb-Girdle Muscular Dystrophy Type 2A Due to a Novel Calpain-3 Mutation. PLoS ONE, 2017, 12, e0170280.	2.5	16
34	Identification of novel pathogenic variants and novel gene-phenotype correlations in Mexican subjects with microphthalmia and/or anophthalmia by next-generation sequencing. Journal of Human Genetics, 2018, 63, 1169-1180.	2.3	16
35	Molecular Screening ofRhodopsinandPeripherin/RDSGenes in Mexican Families with Autosomal Dominant Retinitis Pigmentosa. Current Eye Research, 2009, 34, 1050-1056.	1.5	15
36	CFH haplotypes and ARMS2, C2, C3, and CFB alleles show association with susceptibility to age-related macular degeneration in Mexicans. Molecular Vision, 2014, 20, 105-16.	1.1	15

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37	Mutational Screening ofFOXE3, GDF3, ATOH7, andALDH1A3in Congenital Ocular Malformations. Possible Contribution of the FOXE3 p.VAL201MET Variant to the Risk of Severe Eye Malformations. Ophthalmic Genetics, 2014, 35, 190-192.	1.2	14
38	Clinical expression and SRY gene analysis in XY subjects lacking gonadal tissue. American Journal of Medical Genetics Part A, 2001, 99, 244-247.	2.4	13
39	Exome sequencing identifies RDH12 compound heterozygous mutations in a family with severe retinitis pigmentosa. Gene, 2013, 528, 178-182.	2.2	13
40	Rhodopsin p.N78I dominant mutation causing sectorial retinitis pigmentosa in a pedigree with intrafamilial clinical heterogeneity. Gene, 2013, 519, 173-176.	2.2	13
41	TGFBI, CHST6, and GSN gene analysis in Mexican patients with stromal corneal dystrophies. Graefe's Archive for Clinical and Experimental Ophthalmology, 2014, 252, 1267-1272.	1.9	13
42	Submicroscopic Deletions at 13q32.1 Cause Congenital Microcoria. American Journal of Human Genetics, 2015, 96, 631-639.	6.2	13
43	Membrane frizzled-related protein gene-related ophthalmological syndrome: 30-month follow-up of a sporadic case and review of genotype-phenotype correlation in the literature. Molecular Vision, 2012, 18, 2623-32.	1.1	12
44	The TGFBI A546D mutation causes an atypical type of lattice corneal dystrophy. Molecular Vision, 2007, 13, 1695-700.	1.1	12
45	Nasopalpebral Lipomaâ€Coloboma syndrome: Clinical, radiological, and histopathological description of a novel sporadic case. American Journal of Medical Genetics, Part A, 2013, 161, 1470-1474.	1.2	11
46	Tietz/Waardenburg type 2A syndrome associated with posterior microphthalmos in two unrelated patients with novel MITF gene mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3294-3297.	1.2	11
47	D14 repeat polymorphism of the asporin gene is associated with primary osteoarthritis of the knee in a Mexican Mestizo population. International Journal of Rheumatic Diseases, 2017, 20, 1935-1941.	1.9	11
48	Genome-wide mRNA analysis reveals a TUBD1 isoform profile as a potential biomarker for diabetic retinopathy development. Experimental Eye Research, 2017, 155, 99-106.	2.6	10
49	Long-Term Effects of Gene Therapy in a Novel Mouse Model of HumanMFRP-Associated Retinopathy. Human Gene Therapy, 2019, 30, 632-650.	2.7	10
50	Familial Wolfram syndrome due to compound heterozygosity for two novel WFS1 mutations. Molecular Vision, 2008, 14, 1353-7.	1.1	10
51	Molecular and clinical analysis in a series of patients with Pyknodysostosis reveals some uncommon phenotypic findings. International Journal of Clinical and Experimental Medicine, 2014, 7, 3915-23.	1.3	9
52	OCT findings in young asymptomatic subjects carrying familialBEST1gene mutations. Ophthalmic Genetics, 2011, 32, 24-30.	1.2	8
53	SOX2 anophthalmia syndrome and dental anomalies. American Journal of Medical Genetics, Part A, 2015, 167, 2830-2833.	1.2	8
54	Clinical and molecular evidence of possible digenic inheritance for MFN2/GDAP1 genes in Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2020, 30, 986-990.	0.6	8

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55	The T allele of lysyl oxidase-like 1 rs41435250 is a novel risk factor for pseudoexfoliation syndrome and pseudoexfoliation glaucoma independently and through intragenic epistatic interaction. Molecular Vision, 2013, 19, 1937-44.	1.1	8
56	An intellectually disabled patient with the 5q14.3q15 microdeletion syndrome associated with an apparently de novo t(2;5)(q13;q14). American Journal of Medical Genetics, Part A, 2012, 158A, 942-946.	1.2	7
57	Expansion of the Clinical Ocular Spectrum of Wolfram Syndrome in a Family Carrying a NovelWFS1Gene Deletion. Ophthalmic Genetics, 2013, 34, 243-248.	1.2	7
58	Polymorphism analysis and new JAG1 gene mutations of Alagille syndrome in Mexican population. Meta Gene, 2014, 2, 32-40.	0.6	7
59	The clinical implications of molecular monitoring and analyses of inherited retinal diseases. Expert Review of Molecular Diagnostics, 2017, 17, 1009-1021.	3.1	7
60	Extension of the mutational and clinical spectrum of SOX2 related disorders: Description of six new cases and a novel association with suprasellar teratoma. American Journal of Medical Genetics, Part A, 2018, 176, 2710-2719.	1.2	7
61	Previously undescribed phenotypic findings and novel ACTG1 gene pathogenic variants in Baraitser-Winter cerebrofrontofacial syndrome. European Journal of Medical Genetics, 2020, 63, 103877.	1.3	7
62	Extracranial midline defects in a patient with craniofrontonasal syndrome with a novel EFNB1 mutation. American Journal of Medical Genetics, Part A, 2020, 182, 1223-1229.	1.2	7
63	An epidemiological investigation of a Forkhead box protein E3 founder mutation underlying the high frequency of sclerocornea, aphakia, and microphthalmia in a Mexican village. Molecular Vision, 2013, 19, 1866-70.	1.1	7
64	PAX6 gene intragenic deletions in Mexican patients with congenital aniridia. Molecular Vision, 2006, 12, 318-23.	1.1	7
65	Clinical and Genetic Characteristics of Mexican Patients with Juvenile Presentation of Niemann-Pick Type C Disease. Case Reports in Neurological Medicine, 2014, 2014, 1-7.	0.4	6
66	Clinical characterization and identification of five novel FOXL2 pathogenic variants in a cohort of 12 Mexican subjects with the syndrome of blepharophimosis-ptosis-epicanthus inversus. Gene, 2019, 706, 62-68.	2.2	6
67	Characterization of novel GCDH pathogenic variants causing glutaric aciduria type 1 in the southeast of Mexico. Molecular Genetics and Metabolism Reports, 2019, 21, 100533.	1.1	6
68	Clinical and molecular features of familial and sporadic cases of von Hippelâ€Lindau disease from Mexico. Clinical and Experimental Ophthalmology, 2010, 38, 277-283.	2.6	5
69	Homozygosity mapping identifies a GALK1 mutation as the cause of autosomal recessive congenital cataracts in 4 adult siblings. Gene, 2014, 534, 218-221.	2.2	5
70	Identification and expression analysis of a novel intragenic EFNB1 mutation causing craniofrontonasal syndrome. Meta Gene, 2014, 2, 25-31.	0.6	5
71	<scp>PAX6</scp> allelic heterogeneity in <scp>Mexican</scp> congenital aniridia patients: expanding the mutational spectrum with seven novel pathogenic variants. Clinical and Experimental Ophthalmology, 2017, 45, 875-883.	2.6	5
72	Characterization and mRNA expression analysis of a novel ARG1 splicing mutation causing hyperargininemia. Clinical Biochemistry, 2015, 48, 1273-1276.	1.9	4

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73	Exome sequencing identifies a de novo frameshift mutation in the imprinted gene ZDBF2 in a sporadic patient with Nasopalpebral Lipomaâ€coloboma syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1934-1937.	1.2	4
74	TP63 mutation in a patient with acroâ€dermoâ€ungualâ€lacrimalâ€tooth syndrome: Additional evidence of molecular overlap of the ADULT and EEC syndromes. American Journal of Medical Genetics, Part A, 2016, 170, 1635-1638.	1.2	4
75	Vortex Pattern of Corneal Deposits in Granular Corneal Dystrophy Associated With the p.(Arg555Trp) Mutation in TGFBI. Cornea, 2017, 36, 210-216.	1.7	4
76	Exome sequencing identifies a SREBF1 recurrent ARG557CYS mutation as the cause of hereditary mucoepithelial dysplasia in a family with high clinical variability. American Journal of Medical Genetics, Part A, 2020, 182, 2773-2777.	1.2	4
77	Clinical, histopathological, and in silico pathogenicity analyses in a pedigree with familial amyloidosis of the Finnish type (Meretoja syndrome) caused by a novel gelsolin mutation. Molecular Vision, 2020, 26, 345-354.	1.1	4
78	Acroâ€spondyloâ€pubic dysostosis associated with cataracts, microcephaly, and normal intelligence. American Journal of Medical Genetics, Part A, 2015, 167, 282-286.	1.2	3
79	The PTPN22 R620W polymorphism in anti-neutrophil cytoplasmic antibodyâ€“associated vasculitis in Mexican Mestizos: Table 1. Rheumatology, 2016, 55, 2271-2273.	1.9	3
80	Significant Association Between Variant in SGCD and Age-Related Macular Degeneration. Genes, 2018, 9, 467.	2.4	2
81	Assessment of CFH and HTRA1 polymorphisms in age-related macular degeneration using classic and machine-learning approaches. Ophthalmic Genetics, 2020, 41, 539-547.	1.2	2
82	Autofluorescence in female carriers with choroideremia: A familial case with a novel mutation in the CHM gene. Ophthalmic Genetics, 2020, 41, 625-628.	1.2	2
83	Partially deleted SRY gene confined to testicular tissue in a 46,XX true hermaphrodite without SRY in leukocytic DNA. American Journal of Medical Genetics Part A, 2000, 93, 417-420.	2.4	2
84	The Relevance of Cataract as a Risk Factor for Age-Related Macular Degeneration: A Machine Learning Approach. Applied Sciences (Switzerland), 2019, 9, 5550.	2.5	1
85	Next-Generation Sequencing Identifies a Homozygous Nonsense p.Tyr370* Mutation of the TMC6 Gene in a Mexican Pedigree with Epidermodysplasia Verruciformis. Revista De Investigacion Clinica, 2021, 73, .	0.4	1
86	Machine Learning Method to Establish the Connection Between Age Related Macular Degeneration and Some Genetic Variations. Lecture Notes in Computer Science, 2016, , 28-39.	1.3	1
87	Giant Ocular Lipodermoid Cyst in Encephalocraniocutaneous Lipomatosis: Surgical Treatment and Genetic Analysis. American Journal of Case Reports, 2019, 20, 1566-1571.	0.8	1
88	Novel CHRDL1 mutation causing X-linked megalocornea in a family with mild anterior segment manifestations in carrier females. Ophthalmic Genetics, 2022, 43, 224-229.	1.2	1
89	Cerebral hemihypoplasia and nevus flammeus in a child with oromandibular limb hypogenesis syndrome type III. Journal of Pediatric Genetics, 2015, 02, 043-047.	0.7	0
90	Autosomal Dominant MÃ¼ller Cell Sheen Dystrophy. Retina, 2022, Publish Ahead of Print, .	1.7	0

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91	A new missense variant in RAB3GAP2 in a family with muscular dystrophyâ€“short stature and defective autophagy: An expansion of the micro/Martsolf spectrum or a new phenotype?. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0
92	Detailed phenotypic description of stromal corneal dystrophy in a large pedigree carrying the uncommon TGFBI p.Ala546Asp pathogenic variant. Ophthalmic Genetics, 2022, , 1-5.	1.2	0