V Martinez-Glez

List of Publications by Year
in descending order

76	2,048	218592	254106
papers	citations	26 h-index	43 g-index

81	81	81	3203
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of a Frameshift Mutation in Osterix in a Patient with Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 87, 110-114.	2.6	246
2	Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. Human Mutation, 2012, 33, 343-350.	1.1	178
3	Somatic activating mutations in PIK3CA cause generalized lymphatic anomaly. Journal of Experimental Medicine, 2019, 216, 407-418.	4.2	96
4	New microdeletion and microduplication syndromes: a comprehensive review. Genetics and Molecular Biology, 2014, 37, 210-219.	0.6	84
5	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome-Osteogenesis imperfecta phenotypic spectrum. Human Mutation, 2012, 33, 1444-1449.	1.1	77
6	Simpson-Golabi-Behmel syndrome types I and II. Orphanet Journal of Rare Diseases, 2014, 9, 138.	1.2	75
7	Genetic and epigenetic alteration of theNF2gene in sporadic meningiomas. Genes Chromosomes and Cancer, 2005, 42, 314-319.	1.5	67
8	Macrocephalyâ€“capillary malformation: Analysis of 13 patients and review of the diagnostic criteria. American Journal of Medical Genetics, Part A, 2010, 152A, 3101-3106.	0.7	57
9	A deletion and a duplication in distal 22q11.2 deletion syndrome region. Clinical implications and review. BMC Medical Genetics, 2009, 10, 48.	2.1	54
10	CLAPO syndrome: identification of somatic activating PIK3CA mutations and delineation of the natural history and phenotype. Genetics in Medicine, 2018, 20, 882-889.	1.1	52
11	CDKN1C (p57^{Kip2}) analysis in Beckwithâ€“Wiedemann syndrome (BWS) patients: Genotypeâ€“phenotype correlations, novel mutations, and polymorphisms. American Journal of Medical Genetics, Part A, 2010, 152A, 1390-1397.	0.7	50
12	Beckwithâ€“Wiedemann syndrome and uniparental disomy 11p: fine mapping of the recombination breakpoints and evaluation of several techniques. European Journal of Human Genetics, 2011, 19, 416-421.	1.4	44
13	CDKN1C Mutations in HELLP/Preeclamptic Mothers of Beckwithâ€“Wiedemann Syndrome (BWS) Patients. Placenta, 2009, 30, 551-554.	0.7	43
14	Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2013, 161, 1354-1369.	0.7	42
15	Characterization of a 8q21.11 Microdeletion Syndrome Associated with Intellectual Disability and a Recognizable Phenotype. American Journal of Human Genetics, 2011, 89, 295-301.	2.6	37
16	Two mutations in IFITM5 causing distinct forms of osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2014, 164, 1136-1142.	0.7	37
17	Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. Molecular Genetics & Genomic Medicine, 2017, 5, 28-39.	0.6	37
18	mTOR mutations in Smithâ€Kingsmore syndrome: Four additional patients and a review. Clinical Genetics, 2018, 93, 762-775.	1.0	36

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19	Clinical presentation of a variant of Axenfeldâ€“Rieger syndrome associated with subtelomeric 6p deletion. European Journal of Medical Genetics, 2007, 50, 120-127.	0.7	35
20	Allelic status of 1p and 19q in oligodendrogliomas and glioblastomas: multiplex ligation-dependent probe amplification versus loss of heterozygosity. Cancer Genetics and Cytogenetics, 2009, 190, 93-96.	1.0	34
21	Variants in members of the cadherinâ€“catenin complex, CDH1 and CTNND1, cause blepharocheilodontic syndrome. European Journal of Human Genetics, 2018, 26, 210-219.	1.4	34
22	A six-attribute classification of geneticmosaicism. Genetics in Medicine, 2020, 22, 1743-1757.	1.1	34
23	A New Overgrowth Syndrome is due to Mutations inRNF125. Human Mutation, 2014, 35, 1436-1441.	1.1	33
24	Constitutional mosaic genome-wide uniparental disomy due to diploidisation: an unusual cancer-predisposing mechanism. Journal of Medical Genetics, 2011, 48, 212-216.	1.5	32
25	Customized high resolution CGHâ€array for clinical diagnosis reveals additional genomic imbalances in previous wellâ€defined pathological samples. American Journal of Medical Genetics, Part A, 2013, 161, 1950-1960.	0.7	32
26	Hypoinsulinaemic, hypoketotic hypoglycaemia due to mosaic genetic activation of PI3-kinase. European Journal of Endocrinology, 2017, 177, 175-186.	1.9	32
27	Clinical and molecular analyses of Beckwithâ€“Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. American Journal of Medical Genetics, Part A, 2016, 170, 2740-2749.	0.7	30
28	Report of a newly indentified patient with mutations in BMP1 and underlying pathogenetic aspects. American Journal of Medical Genetics, Part A, 2014, 164, 1143-1150.	0.7	27
29	Biomarkers in Vestibular Schwannomaâ€“Associated Hearing Loss. Frontiers in Neurology, 2019, 10, 978.	1.1	26
30	DNA methylation pattern in 16 tumor-related genes in schwannomas. Cancer Genetics and Cytogenetics, 2007, 172, 84-86.	1.0	25
31	Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. Nature Communications, 2019, 10, 797.	5.8	24
32	Analysis of invdupdel(8p) rearrangement: Clinical, cytogenetic and molecular characterization. American Journal of Medical Genetics, Part A, 2015, 167, 1018-1025.	0.7	22
33	A standard of care for individuals with <scp>PIK3CA</scp>â€related disorders: An international expert consensus statement. Clinical Genetics, 2022, 101, 32-47.	1.0	21
34	Real-Time Quantitative PCR Analysis of Gene Dosages Reveals Gene Amplification in Low-Grade Oligodendrogliomas. American Journal of Clinical Pathology, 2005, 123, 900-906.	0.4	19
35	Sotos syndrome is associated with leukemia/lymphoma. American Journal of Medical Genetics, Part A, 2007, 143A, 1244-1245.	0.7	17
36	EGFR sequence variations and real-time quantitative polymerase chain reaction analysis of gene dosage in brain metastases of solid tumors. Cancer Genetics and Cytogenetics, 2007, 173, 63-67.	1.0	16

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37	Genomic deletions at 1p and 14q are associated with an abnormal cDNA microarray gene expression pattern in meningiomas but not in schwannomas. Cancer Genetics and Cytogenetics, 2010, 196, 1-6.	1.0	15
38	Mutational analysis of the DAL-1/4.1B tumour-suppressor gene locus in meningiomas. International Journal of Molecular Medicine, 2005, 16, 771-4.	1.8	15
39	Microarray Gene Expression Profiling in Meningiomas and Schwannomas. Current Medicinal Chemistry, 2008, 15, 826-833.	1.2	14
40	Multiplex ligation-dependent probe amplification (MLPA) screening in meningioma. Cancer Genetics and Cytogenetics, 2007, 173, 170-172.	1.0	13
41	cDNA microarray expression profile in vestibular schwannoma: correlation with clinical and radiological features. Cancer Genetics and Cytogenetics, 2009, 194, 125-127.	1.0	13
42	Schuursâ€“Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. Genes, 2021, 12, 738.	1.0	13
43	Impact of NGS in the medical sciences: genetic syndromes with an increased risk of developing cancer as an example of the use of new technologies. Genetics and Molecular Biology, 2014, 37, 241-249.	0.6	12
44	Meningiomas and schwannomas: molecular subgroup classification found by expression arrays. International Journal of Oncology, 2009, 34, 493-504.	1.4	12
45	Pitfalls of trio-based exome sequencing: imprinted genes and parental mosaicismâ€”MAGEL2 as an example. Genetics in Medicine, 2017, 19, 1283-1285.	1.1	10
46	Constitutional mosaicism in RASA1â€related capillary malformationâ€arteriovenous malformation. Clinical Genetics, 2019, 95, 516-519.	1.0	10
47	DAPK1 promoter hypermethylaiton in brain metastases and peripheral blood. Neoplasma, 2007, 54, 123-6.	0.7	10
48	RASSF1A methylation and cyclin D1 expression in vestibular schwannomas. Acta Neuropathologica, 2007, 114, 431-433.	3.9	8
49	Array CGH Analysis of Paired Blood and Tumor Samples from Patients with Sporadic Wilms Tumor. PLoS ONE, 2015, 10, e0136812.	1.1	8
50	Phenotypic Variation in Patients with Homozygous c.1678G>T Mutation in EVC Gene: Report of Two Mexican Families with Ellis-van Creveld Syndrome. American Journal of Case Reports, 2017, 18, 1325-1329.	0.3	8
51	A case of naevus vascularis mixtus with hypotrophy and hypotrichosis due to mosaic GNA11 mutation. Journal of the European Academy of Dermatology and Venereology, 2020, 34, e420-e422.	1.3	8
52	Segmental undergrowth is associated with pathogenic variants in vascular malformation genes: A retrospective caseâ€series study. Clinical Genetics, 2022, 101, 296-306.	1.0	7
53	Expression analysis of tumor-related genes involved in critical regulatory pathways in schwannomas. Clinical and Translational Oncology, 2013, 15, 409-411.	1.2	6
54	Cutaneous and ophthalmic signs as a clue to early diagnosis of severe neurofibromatosis type 2: report of a novel mutation that predicts this poor prognosis. Clinical and Experimental Dermatology, 2014, 39, 557-559.	0.6	6

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55	Real-Time Quantitative PCR Analysis of Gene Dosages Reveals Gene Amplification in Low-Grade Oligodendrogliomas. American Journal of Clinical Pathology, 2005, 123, 900-906.	0.4	6
56	Familial hypercholesterolemia: A single-nucleotide variant (SNV) in mosaic at the low density lipoprotein receptor (LDLR). Atherosclerosis, 2020, 311, 37-43.	0.4	5
57	Larotrectinib as an Effective Therapy in Congenital Infantile Fibrosarcoma: Report of Two Cases. European Journal of Pediatric Surgery Reports, 2022, 10, e76-e79.	0.1	5
58	Gene dosage and mutational analyses of EGFR in oligodendrogliomas. International Journal of Oncology, 0, , .	1.4	4
59	Mutational analysis of the CITED4 gene in glioblastomas. Cancer Genetics and Cytogenetics, 2008, 185, 114-116.	1.0	4
60	Gene expression analysis of aberrant signaling pathways in meningiomas. Oncology Letters, 2013, 6, 275-279.	0.8	4
61	Clinical overlap between CLAPO syndrome and macrocephalyâ€capillary malformation syndrome. JDDG - Journal of the German Society of Dermatology, 2020, 18, 479-482.	0.4	4
62	Mietensâ€“Weber syndrome: two new patients and a review. Clinical Dysmorphology, 2006, 15, 175-177.	0.1	3
63	IdentificaciÃ³n de alteraciones genÃ©ticas en oligodendrogliomas mediante amplificaciÃ³n dependiente de ligasa de mÃºltiples sondas (MLPA) (multiple ligationdependent probe amplification). Neurocirugia, 2009, 20, 117-123.	0.2	3
64	<scp>KRAS</scp> mutation identified in a patient with melorheostosis and extended lymphangiomatosis treated with sirolimus and trametinib. Clinical Genetics, 2021, 100, 484-485.	1.0	3
65	Mixed vascular naevus syndrome: report of three children with somatic GNA11 mutation and new systemic associations. Clinical and Experimental Dermatology, 2021, , .	0.6	3
66	Detection of gene amplification and copy gains in brain metastases of solid tumors using quantitative real-time polymerase chain reaction. Cancer Genetics and Cytogenetics, 2008, 182, 61-62.	1.0	2
67	Phenotypic and molecular characterization of five patients with <scp>PIK3CA</scp>â€related overgrowth spectrum (<scp>PROS</scp>). American Journal of Medical Genetics, Part A, 2022, 188, 1792-1800.	0.7	2
68	OSX/SP7 Mutations and Osteogenesis Imperfecta. , 2014, , 173-179.		1
69	BMP1 Mutations in Autosomal Recessive Osteogenesis Imperfecta. , 2014, , 181-186.		1
70	In-frame Variants in FLNA Proximal Rod 1 Domain Associate With a Predominant Cardiac Valvular Phenotype. Revista Espanola De Cardiologia (English Ed), 2018, 71, 545-552.	0.4	1
71	Capillary malformation with segmental distribution and central atrophy: A series of 7 cases. Journal of the American Academy of Dermatology, 2020, 83, 213-214.	0.6	1
72	BiologÃa molecular de los glioblastomas. Neurocirugia, 2007, 18, .	0.2	0

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73	Brain Metastases: Gene Amplification Using Quantitative Real-Time Polymerase Chain Reaction Analysis. , 2011, , 65-69.		0
74	Schwannomas: Role of Molecular Genetics and Epigenetic Mechanisms. , 2012, , 217-223.		0
75	An asymptomatic stain on the forehead. JDDG - Journal of the German Society of Dermatology, 2020, 18, 1511-1514.	0.4	0
76	Thoracic venous malformation: a particular form of a visceral variant. BMJ Case Reports, 2022, 15, e250307.	0.2	0