Francisco Martinez

List of Publications by Year
in descending order

114 papers	3,249 citations	172443 29 h-index	182417 51 g-index
123 all docs	123 docs citations	123 times ranked	5862 citing authors

#	Article	IF	Citations
1	Extending the clinical phenotype of <scp>SPTAN1</scp>: From <scp>DEE5</scp> to migraine, epilepsy, and subependymal heterotopias without intellectual disability. American Journal of Medical Genetics, Part A, 2022, 188, 147-159.	1.2	7
2	Mitochondrial developmental encephalopathy with bilateral optic neuropathy related to homozygous variants in<scp>IMMT</scp>gene. Clinical Genetics, 2022, 101, 233-241.	2.0	8
3	Extending the Phenotype Related to SCN1A Gene: Arthrogryposis, Movement Disorders, and Malformations of Cortical Development. Journal of Child Neurology, 2022, 37, 340-350.	1.4	1
4	Derivation of healthy hepatocyte-like cells from a female patient with ornithine transcarbamylase deficiency through X-inactivation selection. Scientific Reports, 2022, 12, 2308.	3.3	1
5	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
6	Germline variant in Ctcf links mental retardation to Wilms tumor predisposition. European Journal of Human Genetics, 2022, , .	2.8	1
7	CfDNA Measurement as a Diagnostic Tool for the Detection of Brain Somatic Mutations in Refractory Epilepsy. International Journal of Molecular Sciences, 2022, 23, 4879.	4.1	2
8	Hidden etiology of cerebral palsy: genetic and clinical heterogeneity and efficient diagnosis by next-generation sequencing. Pediatric Research, 2021, 90, 284-288.	2.3	20
9	Autosomal recessive woolly hair and hypotrichosis in two Caucasian dizygotic twins. Description of a novel biallelic mutation in the LPAR6 gene. International Journal of Dermatology, 2021, 60, e68-e70.	1.0	1
10	Prevalence of pathogenic copy number variants among children conceived by donor oocyte. Scientific Reports, 2021, 11, 6752.	3.3	1
11	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
12	Candidate Genes for Eyelid Myoclonia with Absences, Review of the Literature. International Journal of Molecular Sciences, 2021, 22, 5609.	4.1	13
13	Genetics of Paroxysmal Dyskinesia: Novel Variants Corroborate the Role of KCNA1 in Paroxysmal Dyskinesia and Highlight the Diverse Phenotypic Spectrum of KCNA1- and SLC2A1-Related Disorders. Frontiers in Neurology, 2021, 12, 701351.	2.4	3
14	Case Report: Novel Homozygous Likely Pathogenic SCN1A Variant With Autosomal Recessive Inheritance and Review of the Literature. Frontiers in Neurology, 2021, 12, 784892.	2.4	2
15	Molecular characterization of Spanish patients with MECP2 duplication syndrome. Clinical Genetics, 2020, 97, 610-620.	2.0	16
16	Apparent Radiological Improvement in an Infant With Labrune Syndrome Treated With Bevacizumab. Pediatric Neurology, 2020, 112, 53-55.	2.1	7
17	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
18	Mixed Phenotype of Langerâ€“Giedion's and Cornelia de Lange's Syndromes in an 8q23.3-q24.1 Microdeletion without TRPS1 Deletion. Journal of Pediatric Genetics, 2020, 09, 053-057.	0.7	0

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19	Role of mitochondrial DNA variants in the development of fragile X-associated tremor/ataxia syndrome. Mitochondrion, 2020, 52, 157-162.	3.4	4
20	Mutations in PMM2 gene in four unrelated Spanish families with polycystic kidney disease and hyperinsulinemic hypoglycemia. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1283-1288.	0.9	7
21	Molecular and Clinical Characterization of a Novel Nonsense Variant in Exon 1 of the UPF3B Gene Found in a Large Spanish Basque Family (MRX82). Frontiers in Genetics, 2019, 10, 1074.	2.3	14
22	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
23	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
24	Clinical implication of FMR1 intermediate alleles in a Spanish population. Clinical Genetics, 2018, 94, 153-158.	2.0	5
25	A Novel Mutation of MAGEL2 in a Patient with Schaaf-Yang Syndrome and Hypopituitarism. International Journal of Endocrinology and Metabolism, 2018, In Press, e67329.	1.0	9
26	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
27	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
28	X chromosome dosage and presence of SRY shape sex-specific differences in DNA methylation at an autosomal region in human cells. Biology of Sex Differences, 2018, 9, 10.	4.1	20
29	HUWE1 variants cause dominant X-linked intellectual disability: a clinical study of 21 patients. European Journal of Human Genetics, 2018, 26, 64-74.	2.8	72
30	High diagnostic yield of syndromic intellectual disability by targeted next-generation sequencing. Journal of Medical Genetics, 2017, 54, 87-92.	3.2	93
31	Artificial reproductive techniques and epigenetic alterations: Additional comments to the article by Arcosâ€Machancoses et al. (). American Journal of Medical Genetics, Part A, 2017, 173, 1983-1984.	1.2	1
32	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. Scientific Reports, 2017, 7, 12288.	3.3	23
33	Chimeric Genes in Deletions and Duplications Associated with Intellectual Disability. International Journal of Genomics, 2017, 2017, 1-11.	1.6	10
34	De novo mutations in genes of mediator complex causing syndromic intellectual disability: mediatorpathy or transcriptomopathy?. Pediatric Research, 2016, 80, 809-815.	2.3	27
35	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	2.5	92
36	Multiâ€system involvement in a severe variant of fibrodysplasia ossificans progressiva (ACVR1) Tj ETQq0 0 0 rgBT /Overlock 10 T 2265-2271.	1.2	33

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37	Silverâ€Russell syndrome caused by epigenetic alteration in a child conceived by intrauterine insemination from donor sperm. American Journal of Medical Genetics, Part A, 2015, 167, 2861-2864.	1.2	2
38	In Pursuit of New Imprinting Syndromes by Epimutation Screening in Idiopathic Neurodevelopmental Disorder Patients. BioMed Research International, 2015, 2015, 1-8.	1.9	3
39	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
40	Haploinsufficiency of the MYT1L gene causes intellectual disability frequently associated with behavioral disorder. Genetics in Medicine, 2015, 17, 683-684.	2.4	10
41	Infectious and Immunologic Phenotype of MECP2 Duplication Syndrome. Journal of Clinical Immunology, 2015, 35, 168-181.	3.8	35
42	Pure duplication of 19p13.3 in three members of a family with intellectual disability and literature review. Definition of a new microduplication syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1614-1620.	1.2	12
43	A novel missense mutation in the NSDHL gene identified in a Lithuanian family by targeted nextâ€generation sequencing causes CK syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1342-1348.	1.2	6
44	Novel mutations of NFIX gene causing Marshall-Smith syndrome or Sotos-like syndrome: one gene, two phenotypes. Pediatric Research, 2015, 78, 533-539.	2.3	35
45	Prenatal Diagnosis of a Female Fetus with Ring Chromosome 9, 46,XX,r(9)(p24q34), and a de novo Interstitial 9p Deletion. Cytogenetic and Genome Research, 2014, 144, 275-279.	1.1	8
46	Molecular Testing for Fragile X: Analysis of 5062 Tests from 1105 Fragile X Familiesâ€"Performed in 12 Clinical Laboratories in Spain. BioMed Research International, 2014, 2014, 1-8.	1.9	13
47	Duplication at Xq13.3â€"q21.1 with syndromic intellectual disability, a probable role for the ATRX gene. American Journal of Medical Genetics, Part A, 2014, 164, 918-923.	1.2	10
48	Screening individuals with intellectual disability, autism and Tourette's syndrome for KCNK9 mutations and aberrant DNA methylation within the 8q24 imprinted cluster.. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 472-478.	1.7	13
49	Phenotype profiling of patients with intellectual disability and copy number variations. European Journal of Paediatric Neurology, 2014, 18, 558-566.	1.6	18
50	Mutation screening of AURKB and SYCP3 in patients with reproductive problems. Molecular Human Reproduction, 2013, 19, 102-108.	2.8	14
51	Microphthalmia with Linear Skin Defects Syndrome. Pediatric Dermatology, 2013, 30, e230-1.	0.9	11
52	Detection of Hypomethylation Syndrome among Patients with Epigenetic Alterations at theGNASLocus. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1060-E1067.	3.6	37
53	Hypomethylation of the KCNQ1OT1 imprinting center of chromosome 11 associated to Sotos-like features. Journal of Human Genetics, 2012, 57, 153-156.	2.3	4
54	Copy-Number Gains of HUWE1 Due to Replication- and Recombination-Based Rearrangements. American Journal of Human Genetics, 2012, 91, 252-264.	6.2	71

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55	Clinical, molecular and biochemical characterization of nine Spanish families with Conradi-HÃ¼nermann-Happle syndrome: new insights into X-linked dominant chondrodysplasia punctata with a comprehensive review of the literature. British Journal of Dermatology, 2012, 166, 830-838.	1.5	40
56	Large deletion in the Factor <scp>VIII</scp> gene (<scp>F</scp>8) involving segmental duplications in int22h shows no haematological phenotype in female carriers, but may be embryonic lethal in males. British Journal of Haematology, 2012, 158, 138-140.	2.5	7
57	De novo Interstitial Triplication of <i>MECP2</i> in a Girl with Neurodevelopmental Disorder and Random X Chromosome Inactivation. Cytogenetic and Genome Research, 2011, 135, 93-101.	1.1	22
58	Partial Duplication of 18q Including a Distal Critical Region for Edwards Syndrome in a Patient with Normal Phenotype and Oligoasthenospermia: Case Report. Cytogenetic and Genome Research, 2011, 133, 78-83.	1.1	7
59	Intronic mutations affecting splicing of MBTPS2 cause ichthyosis follicularis, alopecia and photophobia (IFAP) syndrome. Experimental Dermatology, 2011, 20, 447-449.	2.9	19
60	Intermediate FMR1 alleles and cognitive and/or behavioural phenotypes. European Journal of Human Genetics, 2011, 19, 921-923.	2.8	17
61	Minimal disease detection in peripheral blood and bone marrow from patients with non-metastatic neuroblastoma. Journal of Cancer Research and Clinical Oncology, 2011, 137, 1263-1272.	2.5	19
62	Hypermethylation of apoptotic genes as independent prognostic factor in neuroblastoma disease. Molecular Carcinogenesis, 2011, 50, 153-162.	2.7	39
63	Epigenetic alterations in disseminated neuroblastoma tumour cells: influence of TMS1 gene hypermethylation in relapse risk in NB patients. Journal of Cancer Research and Clinical Oncology, 2010, 136, 1415-1421.	2.5	20
64	Common pathological mutations inPQBP1induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. Human Mutation, 2010, 31, 90-98.	2.5	18
65	Enrichment of ultraconserved elements among genomic imbalances causing mental delay and congenital anomalies. BMC Medical Genomics, 2010, 3, 54.	1.5	18
66	IntragenicGNASDeletion Involving Exon A/B in Pseudohypoparathyroidism Type 1A Resulting in an Apparent Loss of Exon A/B Methylation: Potential for Misdiagnosis of Pseudohypoparathyroidism Type 1B. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 765-771.	3.6	38
67	Prenatal study of common submicroscopic â€œgenomic disordersâ€•using MLPA with subtelomeric/microdeletion syndrome probe mixes, among gestations with ultrasound abnormalities in the first trimester. European Journal of Medical Genetics, 2010, 53, 76-79.	1.3	5
68	Submicroscopic Duplication of the Wolf-Hirschhorn Critical Region with a 4p Terminal Deletion. Cytogenetic and Genome Research, 2009, 125, 103-108.	1.1	14
69	Corpus Callosum Abnormalities and the Controversy about the Candidate Genes Located in 1q44. Cytogenetic and Genome Research, 2009, 127, 5-8.	1.1	29
70	Novel UBE3A mutations causing Angelman syndrome: Different parental origin for single nucleotide changes and multiple nucleotide deletions or insertions. American Journal of Medical Genetics, Part A, 2009, 149A, 343-348.	1.2	18
71	MAGE-A1 expression is associated with good prognosis in neuroblastoma tumors. Journal of Cancer Research and Clinical Oncology, 2009, 135, 523-531.	2.5	15
72	Subtelomeric analysis of pediatric astrocytoma: subchromosomal instability is a distinctive feature of pleomorphic xanthoastrocytoma. Journal of Neuro-Oncology, 2009, 93, 175-182.	2.9	11

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73	Molecular Analysis of 250 Patients with Autosomal Recessive Congenital Ichthyosis: Evidence for Mutation Hotspots in ALOXE3 and Allelic Heterogeneity in ALOX12B. Journal of Investigative Dermatology, 2009, 129, 1421-1428.	0.7	96
74	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	21.4	528
75	Tyrosinemia type 1 and Angelman syndrome due to paternal uniparental isodisomy 15. Journal of Inherited Metabolic Disease, 2009, 32, 349-353.	3.6	4
76	Identification of deletion carriers in hemophilia B: quantitative real-time polymerase chain reaction or multiple ligation probe amplification. Translational Research, 2009, 153, 114-117.	5.0	10
77	Duplication of the Williams-Beuren critical region: case report and further delineation of the phenotypic spectrum. BMJ Case Reports, 2009, 2009, bcr0820080665-bcr0820080665.	0.5	0
78	Autosomalâ€dominant hypohidrotic ectodermal dysplasia caused by a novel mutation. Journal of the European Academy of Dermatology and Venereology, 2008, 22, 1508-1510.	2.4	6
79	Founder haplotype associated with the factor VIII Asp1241Glu polymorphism in a cohort of mild hemophilia A patients. Journal of Thrombosis and Haemostasis, 2008, 6, 1428-1430.	3.8	6
80	Detection of known and novel genomic rearrangements by array based comparative genomic hybridisation: deletion of ZNF533 and duplication of CHARGE syndrome genes. Journal of Medical Genetics, 2008, 45, 432-437.	3.2	47
81	X-linked infantile spinal muscular atrophy: Clinical definition and molecular mapping. Genetics in Medicine, 2007, 9, 52-60.	2.4	27
82	MLPA as first screening method for the detection of microduplications and microdeletions in patients with X-linked mental retardation. Genetics in Medicine, 2007, 9, 117-122.	2.4	34
83	Duplication of the Williams-Beuren critical region: case report and further delineation of the phenotypic spectrum. Journal of Medical Genetics, 2007, 45, 187-189.	3.2	24
84	Duplication of 14q11.2 associates with short stature and mild mental retardation: A putative relation with quantitative trait loci. American Journal of Medical Genetics, Part A, 2007, 143A, 382-384.	1.2	12
85	A two base pair deletion in the PQBP1 gene is associated with microphthalmia, microcephaly, and mental retardation. European Journal of Human Genetics, 2007, 15, 29-34.	2.8	34
86	Clinical findings and molecular characterization of six subtelomeric imbalances. Clinical Genetics, 2007, 71, 474-479.	2.0	3
87	X-chromosome tiling path array detection of copy number variants in patients with chromosome X-linked mental retardation. BMC Genomics, 2007, 8, 443.	2.8	57
88	Robust, Easy, and Dose-Sensitive Methylation Test for the Diagnosis of Praderâ€“Willi and Angelman Syndromes. Genetic Testing and Molecular Biomarkers, 2006, 10, 174-177.	1.7	10
89	Evaluation of MLPA for the detection of cryptic subtelomeric rearrangements. Translational Research, 2006, 147, 295-300.	2.3	33
90	A subtelomeric translocation apparently implied in multiple abortions. Journal of Assisted Reproduction and Genetics, 2006, 23, 97-101.	2.5	5

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91	Recombinant X chromosome in a prenatal diagnosis. Cytogenetic and Genome Research, 2006, 112, 337-340.	1.1	3
92	The Doublecortin Gene, A New Molecular Marker to Detect Minimal Residual Disease in Neuroblastoma. Diagnostic Molecular Pathology, 2005, 14, 53-57.	2.1	41
93	There Is No Evidence That the SDHB Gene Is Involved in Neuroblastoma Development. Oncology Research, 2005, 15, 393-398.	1.5	15
94	Screening for microdeletions of the X-chromosome in non-specific mental retardation. , 2004, 124A, 99-101.		0
95	Localization of MRX82: A new nonsyndromic X-linked mental retardation locus to Xq24-q25 in a Basque family. , 2004, 131A, 174-178.		5
96	A submicroscopic deletion of 11p13 associated with the WAGR syndrome. Clinical Genetics, 2003, 63, 319-322.	2.0	2
97	Intronic L1 insertion and F268S, novel mutations in RPS6KA3 (RSK2) causing Coffin-Lowry syndrome. Clinical Genetics, 2003, 64, 491-496.	2.0	31
98	X-linked spermine synthase gene (SMS) defect: the first polyamine deficiency syndrome. European Journal of Human Genetics, 2003, 11, 937-944.	2.8	134
99	Significant linkage and non-linkage of type 1 von Willebrand disease to the von Willebrand factor gene. British Journal of Haematology, 2001, 115, 692-700.	2.5	30
100	Localization of non-specific X-linked mental retardation gene (MRX73) to Xp22.2. American Journal of Medical Genetics Part A, 2001, 102, 200-204.	2.4	8
101	Q1311X: a novel nonsense mutation of putative ancient origin in the von Willebrand factor gene. SHORT REPORT. British Journal of Haematology, 2000, 111, 552-555.	2.5	15
102	Screening for FMR1 mutations among the mentally retarded: prevalence of the fragile X syndrome in Spain. Clinical Genetics, 1999, 56, 98-99.	2.0	10
103	Identification of a new candidate mutation, G1629R, in a family with type 2A von Willebrand disease. , 1999, 60, 309-310.		4
104	Mutation of the XNP/ATR-X Gene in a Family with Severe Mental Retardation, Spastic Paraplegia and Skewed Pattern of X Inactivation: Demonstration that the Mutation is Involved in the Inactivation Bias. American Journal of Human Genetics, 1999, 65, 558-562.	6.2	62
105	X-Linked Anhidrotic (Hypohidrotic) Ectodermal Dysplasia Caused by a Novel Mutation in EDA1 Gene: 406T>G (Leu55Arg). Journal of Investigative Dermatology, 1999, 113, 285-286.	0.7	18
106	Search for mutations in a segment of the exon 28 of the human von Willebrand factor gene: New mutations, R1315C and R1341W, associated with type 2M and 2B variants. American Journal of Hematology, 1998, 59, 57-63.	4.1	29
107	Phenotype Correlation and Intergenerational Dynamics of the Friedreich Ataxia GAA Trinucleotide Repeat. American Journal of Human Genetics, 1997, 61, 101-110.	6.2	161
108	Mutational analysis of the MPZ, PMP22 and Cx32 genes in patients of Spanish ancestry with Charcot-Marie-Tooth disease and hereditary neuropathy with liability to pressure palsies. Human Genetics, 1997, 99, 746-754.	3.8	137

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109	INSTABILITY OF A VARIABLE NUMBER TANDEM REPEAT IN INTRON 40 OF THE HUMAN VON WILLEBRAND FACTOR GENE. British Journal of Haematology, 1995, 91, 255-256.	2.5	6
110	Localization of a gene for X-linked nonspecific mental retardation (MRX24) in Xp22.2-p22.3. American Journal of Medical Genetics Part A, 1995, 55, 387-390.	2.4	17
111	An autosomal dominant retinitis pigmentosa family with close linkage to D7S480 on 7q. Human Genetics, 1995, 96, 216-218.	3.8	14
112	Characteristics of the transmission of the FMR1 gene from carrier females in a prospective sample of conceptuses. American Journal of Medical Genetics Part A, 1994, 51, 503-506.	2.4	12
113	A fragile X family with high penetrance in females: risk heterogeneity?. Clinical Genetics, 1992, 42, 22-26.	2.0	5
114	Variant in CACNA1G as a Possible Genetic Modifier of Neonatal Epilepsy in an Infant with a De Novo SCN2A Mutation. Journal of Pediatric Genetics, 0, , .	0.7	2