Feliciano J Ramos

List of Publications by Year
in descending order

69 papers	3,488 citations	201385 27 h-index	149479 56 g-index
75 all docs	75 docs citations	75 times ranked	4382 citing authors

#	Article	IF	Citations
1	Subclinical myocardial dysfunction is revealed by speckle tracking echocardiography in patients with Cornelia de Lange syndrome. International Journal of Cardiovascular Imaging, 2022, 38, 2291-2302.	0.2	1
2	Schuursâ€“Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. Genes, 2021, 12, 738.	1.0	13
3	Clinical relevance of postzygotic mosaicism in Cornelia de Lange syndrome and purifying selection of NIPBL variants in blood. Scientific Reports, 2021, 11, 15459.	1.6	11
4	Tenorio syndrome: Description of 14 novel cases and review of the clinical and molecular features. Clinical Genetics, 2021, 100, 405-411.	1.0	2
5	Disruption of NIPBL/Scc2 in Cornelia de Lange Syndrome provokes cohesin genome-wide redistribution with an impact in the transcriptome. Nature Communications, 2021, 12, 4551.	5.8	20
6	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. Applied Sciences (Switzerland), 2021, 11, 710.	1.3	2
7	Things are not always what they seem: From Cornelia de Lange to KBG phenotype in a girl with genetic variants in NIPBL and ANKRD11. Molecular Genetics & Genomic Medicine, 2021, 9, e1826.	0.6	2
8	Heterozygous de novo variants in <scp>CSNK1G1</scp> are associated with syndromic developmental delay and autism spectrum disorder. Clinical Genetics, 2020, 98, 571-576.	1.0	10
9	MAU2 and NIPBL Variants Impair the Heterodimerization of the Cohesin Loader Subunits and Cause Cornelia de Lange Syndrome. Cell Reports, 2020, 31, 107647.	2.9	36
10	Quantifying the economic impact of caregiving for Duchenne muscular dystrophy (DMD) in Spain. European Journal of Health Economics, 2020, 21, 1015-1023.	1.4	10
11	Pathogenic variants in <scp>EP300</scp> and <scp>ANKRD11</scp> in patients with phenotypes overlapping Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1690-1696.	0.7	34
12	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	1.8	24
13	Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. International Journal of Molecular Sciences, 2020, 21, 1042.	1.8	40
14	MRX93 syndrome (BRWD3 gene): five new patients with novel mutations. Clinical Genetics, 2019, 95, 726-731.	1.0	13
15	Rare Variants in 48 Genes Account for 42% of Cases of Epilepsy With or Without Neurodevelopmental Delay in 246 Pediatric Patients. Frontiers in Neuroscience, 2019, 13, 1135.	1.4	39
16	Development, behaviour and autism in individuals with SMC1A variants. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2019, 60, 305-313.	3.1	13
17	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666.	7.7	223
18	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Human Genetics, 2017, 136, 307-320.	1.8	61

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19	Two-step ATP-driven opening of cohesin head. Scientific Reports, 2017, 7, 3266.	1.6	19
20	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	0.7	69
21	mRNA Quantification of NIPBL Isoforms A and B in Adult and Fetal Human Tissues, and a Potentially Pathological Variant Affecting Only Isoform A in Two Patients with Cornelia de Lange Syndrome. International Journal of Molecular Sciences, 2017, 18, 481.	1.8	1
22	Identification and Functional Characterization of Two IntronicNIPBLMutations in Two Patients with Cornelia de Lange Syndrome. BioMed Research International, 2016, 2016, 1-8.	0.9	12
23	Expanding the clinical spectrum of the â€˜<scp>HDAC8</scp>â€phenotypeâ€™ â€“ implications for molecular diagnostics, counseling and risk prediction. Clinical Genetics, 2016, 89, 564-573.	1.0	38
24	A Novel Domain-Specific Mutation in a Sclerosteosis Patient Suggests a Role of LRP4 as an Anchor for Sclerostin in Human Bone. Journal of Bone and Mineral Research, 2016, 31, 874-881.	3.1	65
25	A view on clinical genetics and genomics in Spain: of challenges and opportunities. Molecular Genetics & Genomic Medicine, 2016, 4, 376-391.	0.6	8
26	Special cases in Cornelia de Lange syndrome: The Spanish experience. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 198-205.	0.7	19
27	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	1.1	72
28	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	1.4	37
29	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	2.6	61
30	Functional Characterization of NIPBL Physiological Splice Variants and Eight Splicing Mutations in Patients with Cornelia de Lange Syndrome. International Journal of Molecular Sciences, 2014, 15, 10350-10364.	1.8	22
31	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.	0.9	13
32	Somatic mosaicism in a Cornelia de Lange syndrome patient with <scp>NIPBL</scp> mutation identified by different next generation sequencing approaches. Clinical Genetics, 2014, 86, 595-597.	1.0	17
33	Could a patient with <scp>SMC1A</scp> duplication beÂclassified as a human cohesinopathy?. Clinical Genetics, 2014, 85, 446-451.	1.0	12
34	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	1.4	120
35	Severe ipsilateral musculoskeletal involvement in a Cornelia de Lange patient with a novel NIPBL mutation. European Journal of Medical Genetics, 2014, 57, 503-509.	0.7	8
36	Cornelia de Lange syndrome with NIPBL mutation and mosaic Turner syndrome in the same individual. BMC Medical Genetics, 2012, 13, 43.	2.1	12

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37	CDKL5 gene status in female patients with epilepsy and Rett-like features: two new mutations in the catalytic domain. BMC Medical Genetics, 2012, 13, 68.	2.1	8
38	Human variome project country nodes: Documenting genetic information within a country. Human Mutation, 2012, 33, 1513-1519.	1.1	10
39	Characterization of splice variants of the genes encoding human mitochondrial HMG-CoA lyase and HMG-CoA synthase, the main enzymes of the ketogenesis pathway. Molecular Biology Reports, 2012, 39, 4777-4785.	1.0	24
40	New ocular findings in two sisters with Yunisâ€“VarÃ³n syndrome and literature review. European Journal of Medical Genetics, 2011, 54, 76-81.	0.7	9
41	Epigenetic Modification of the FMR1 Gene in Fragile X Syndrome Is Associated with Differential Response to the mGluR5 Antagonist AFQ056. Science Translational Medicine, 2011, 3, 64ra1.	5.8	344
42	The importance of rare diseases: from the gene to society. Archives of Disease in Childhood, 2011, 96, 791-792.	1.0	52
43	Differential HMGâ€CoA lyase expression in human tissues provides clues about 3â€hydroxyâ€3â€methylglutaric aciduria. Journal of Inherited Metabolic Disease, 2010, 33, 405-410.	1.7	20
44	Mutations and variants in the cohesion factor genes NIPBL, SMC1A, and SMC3 in a cohort of 30 unrelated patients with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 924-929.	0.7	72
45	Facial diagnosis of mild and variant CdLS: Insights from a dysmorphologist survey. American Journal of Medical Genetics, Part A, 2010, 152A, 1641-1653.	0.7	75
46	Risk of cognitive impairment in female premutation carriers of fragile X premutation: Analysis by means of robust segmented linear regression models. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 262-270.	1.1	14
47	Antisense therapeutics for neurofibromatosis type 1 caused by deep intronic mutations. Human Mutation, 2009, 30, 454-462.	1.1	46
48	Ten novelHMGCLmutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. Human Mutation, 2009, 30, E520-E529.	1.1	21
49	Severe infantile-onset cardiomyopathy associated with a homozygous deletion in desmin. Neuromuscular Disorders, 2009, 19, 418-422.	0.3	58
50	A doubleâ€blind, parallel, multicenter comparison of <scp>L</scp>â€acetylcarnitine with placebo on the attention deficit hyperactivity disorder in fragile X syndrome boys. American Journal of Medical Genetics, Part A, 2008, 146A, 803-812.	0.7	91
51	Analysis of the molecular parameters that could predict the risk of manifesting premature ovarian failure in female premutation carriers of fragile X syndrome. Menopause, 2008, 15, 945-949.	0.8	53
52	Molecular genetics of HMG-CoA lyase deficiency. Molecular Genetics and Metabolism, 2007, 92, 198-209.	0.5	64
53	Mutations in Cohesin Complex Members SMC3 and SMC1A Cause a Mild Variant of Cornelia de Lange Syndrome with Predominant Mental Retardation. American Journal of Human Genetics, 2007, 80, 485-494.	2.6	445
54	A further case of opsismodysplasia with hydrocephalus. European Journal of Medical Genetics, 2006, 49, 93-100.	0.7	5

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55	Late-onset treatment in Menkes disease: is there a correlation between genotype and response to therapy?. Clinical Genetics, 2006, 69, 363-366.	1.0	5
56	DMP1 mutations in autosomal recessive hypophosphatemia implicate a bone matrix protein in the regulation of phosphate homeostasis. Nature Genetics, 2006, 38, 1248-1250.	9.4	487
57	Enlarged parietal foramina caused by mutations in the homeobox genes ALX4 and MSX2: from genotype to phenotype. European Journal of Human Genetics, 2006, 14, 151-158.	1.4	67
58	Autoimmune Thyroiditis After Bone Marrow Transplantation in a Boy With Wiskott-Aldrich Syndrome. Journal of Pediatric Hematology/Oncology, 2002, 24, 772-776.	0.3	10
59	Detection of the fragile X syndrome protein for the evaluation of FMR1 intermediate alleles. Human Genetics, 2000, 107, 195-196.	1.8	8
60	Epidermal naevus syndrome and hypophosphataemic rickets: description of a patient with central nervous system anomalies and review of the literature. European Journal of Pediatrics, 1999, 158, 103-107.	1.3	40
61	Further evidence that the Hajdu-Cheney syndrome and the ?serpentine fibula-polycystic kidney syndrome? are a single entity. , 1998, 78, 474-481.		32
62	Seasonally of cryptosporidiosis in children. European Journal of Clinical Microbiology and Infectious Diseases, 1996, 15, 77-79.	1.3	28
63	Cystic kidney disease in Hajdu-Cheney syndrome. American Journal of Medical Genetics Part A, 1995, 56, 25-30.	2.4	37
64	Saethre-Chotzen syndrome with familial translocation at chromosome 7p22. American Journal of Medical Genetics Part A, 1993, 47, 637-639.	2.4	51
65	Direct DNA Testing for Fragile X Syndrome. JAMA Pediatrics, 1993, 147, 1231.	3.6	1
66	Frequency of the common fragile site at Xq27.2 under conditions of thymidylate stress: Implications for cytogenetic diagnosis of the fragile-X syndrome. American Journal of Medical Genetics Part A, 1992, 42, 835-838.	2.4	4
67	Characterization of a highly polymorphic dinucleotide repeat 150 KB proximal to the fragile X site. American Journal of Medical Genetics Part A, 1992, 43, 237-243.	2.4	82
68	Tricho-Rhino-Phalangeal syndrome type II (Langer-Giedion) with persistent cloaca and prune belly sequence in a girl with 8q interstitial deletion. American Journal of Medical Genetics Part A, 1992, 44, 790-794.	2.4	23
69	Giant meningioma in a 5-month-old infant. Child's Nervous System, 1988, 4, 112-115.	0.6	11