Feliciano J Ramos

List of Publications by Year
in descending order

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69	3,488	201674	149698
papers	citations	27 h-index	56 g-index

75	75	75	4382
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	DMP1 mutations in autosomal recessive hypophosphatemia implicate a bone matrix protein in the regulation of phosphate homeostasis. Nature Genetics, 2006, 38, 1248-1250.	21.4	487
2	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.	6.2	445
3	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.	12.4	344
4	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666.	16.3	223
5	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.	2.9	120
6	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.	1.2	91
7	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
8	Facial diagnosis of mild and variant CdLS: Insights from a dysmorphologist survey. American Journal of Medical Genetics, Part A, 2010, 152A, 1641-1653.	1.2	75
9	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.	1.2	72
10	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	2.5	72
11	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
12	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.	2.8	67
13	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.	2.8	65
14	Molecular genetics of HMG-CoA lyase deficiency. Molecular Genetics and Metabolism, 2007, 92, 198-209.	1.1	64
15	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	6.2	61
16	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Human Genetics, 2017, 136, 307-320.	3.8	61
17	Severe infantile-onset cardiomyopathy associated with a homozygous deletion in desmin. Neuromuscular Disorders, 2009, 19, 418-422.	0.6	58
18	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.	2.0	53

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19	The importance of rare diseases: from the gene to society. Archives of Disease in Childhood, 2011, 96, 791-792.	1.9	52
20	Saethre-Chotzen syndrome with familial translocation at chromosome 7p22. American Journal of Medical Genetics Part A, 1993, 47, 637-639.	2.4	51
21	Antisense therapeutics for neurofibromatosis type 1 caused by deep intronic mutations. Human Mutation, 2009, 30, 454-462.	2.5	46
22	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.	2.7	40
23	Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. International Journal of Molecular Sciences, 2020, 21, 1042.	4.1	40
24	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.	2.8	39
25	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.	2.0	38
26	Cystic kidney disease in Hajdu-Cheney syndrome. American Journal of Medical Genetics Part A, 1995, 56, 25-30.	2.4	37
27	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	2.8	37
28	MAU2 and NIPBL Variants Impair the Heterodimerization of the Cohesin Loader Subunits and Cause Cornelia de Lange Syndrome. Cell Reports, 2020, 31, 107647.	6.4	36
29	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.	1.2	34
30	Further evidence that the Hajdu-Cheney syndrome and the ?serpentine fibula-polycystic kidney syndrome? are a single entity. , 1998, 78, 474-481.		32
31	Seasonally of cryptosporidiosis in children. European Journal of Clinical Microbiology and Infectious Diseases, 1996, 15, 77-79.	2.9	28
32	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.	2.3	24
33	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
34	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
35	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.	4.1	22
36	Ten novelHMGCLmutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. Human Mutation, 2009, 30, E520-E529.	2.5	21

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37	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.	3.6	20
38	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.	12.8	20
39	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.	1.6	19
40	Two-step ATP-driven opening of cohesin head. Scientific Reports, 2017, 7, 3266.	3.3	19
41	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.	2.0	17
42	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.	1.7	14
43	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
44	MRX93 syndrome (BRWD3 gene): five new patients with novel mutations. Clinical Genetics, 2019, 95, 726-731.	2.0	13
45	Development, behaviour and autism in individuals with SMC1A variants. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2019, 60, 305-313.	5.2	13
46	Schuursâ€"Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. Genes, 2021, 12, 738.	2.4	13
47	Cornelia de Lange syndrome with NIPBL mutation and mosaic Turner syndrome in the same individual. BMC Medical Genetics, 2012, 13, 43.	2.1	12
48	Could a patient with <scp>SMC1A</scp> duplication beÂclassified as a human cohesinopathy?. Clinical Genetics, 2014, 85, 446-451.	2.0	12
49	Identification and Functional Characterization of Two IntronicNIPBLMutations in Two Patients with Cornelia de Lange Syndrome. BioMed Research International, 2016, 2016, 1-8.	1.9	12
50	Clinical relevance of postzygotic mosaicism in Cornelia de Lange syndrome and purifying selection of NIPBL variants in blood. Scientific Reports, 2021, 11, 15459.	3.3	11
51	Giant meningioma in a 5-month-old infant. Child's Nervous System, 1988, 4, 112-115.	1.1	11
52	Autoimmune Thyroiditis After Bone Marrow Transplantation in a Boy With Wiskott-Aldrich Syndrome. Journal of Pediatric Hematology/Oncology, 2002, 24, 772-776.	0.6	10
53	Human variome project country nodes: Documenting genetic information within a country. Human Mutation, 2012, 33, 1513-1519.	2.5	10
54	Heterozygous de novo variants in <scp>CSNK1G1</scp> are associated with syndromic developmental delay and autism spectrum disorder. Clinical Genetics, 2020, 98, 571-576.	2.0	10

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55	Quantifying the economic impact of caregiving for Duchenne muscular dystrophy (DMD) in Spain. European Journal of Health Economics, 2020, 21, 1015-1023.	2.8	10
56	New ocular findings in two sisters with Yunisâ€“VarÃ³n syndrome and literature review. European Journal of Medical Genetics, 2011, 54, 76-81.	1.3	9
57	Detection of the fragile X syndrome protein for the evaluation of FMR1 intermediate alleles. Human Genetics, 2000, 107, 195-196.	3.8	8
58	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
59	Severe ipsilateral musculoskeletal involvement in a Cornelia de Lange patient with a novel NIPBL mutation. European Journal of Medical Genetics, 2014, 57, 503-509.	1.3	8
60	A view on clinical genetics and genomics in Spain: of challenges and opportunities. Molecular Genetics & Genomic Medicine, 2016, 4, 376-391.	1.2	8
61	A further case of opsismodysplasia with hydrocephalus. European Journal of Medical Genetics, 2006, 49, 93-100.	1.3	5
62	Late-onset treatment in Menkes disease: is there a correlation between genotype and response to therapy?. Clinical Genetics, 2006, 69, 363-366.	2.0	5
63	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
64	Tenorio syndrome: Description of 14 novel cases and review of the clinical and molecular features. Clinical Genetics, 2021, 100, 405-411.	2.0	2
65	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. Applied Sciences (Switzerland), 2021, 11, 710.	2.5	2
66	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.	1.2	2
67	Direct DNA Testing for Fragile X Syndrome. JAMA Pediatrics, 1993, 147, 1231.	3.0	1
68	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.	4.1	1
69	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.	0.6	1