

# Feliciano J Ramos

## List of Publications by Year in descending order

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69  
papers

3,488  
citations

230014

27  
h-index

169272

56  
g-index

75  
all docs

75  
docs citations

75  
times ranked

4716  
citing authors

#	ARTICLE	IF	CITATIONS
1	DMP1 mutations in autosomal recessive hypophosphatemia implicate a bone matrix protein in the regulation of phosphate homeostasis. <i>Nature Genetics</i> , 2006, 38, 1248-1250.	9.4	487
2	Mutations in Cohesin Complex Members SMC3 and SMC1A Cause a Mild Variant of Cornelia de Lange Syndrome with Predominant Mental Retardation. <i>American Journal of Human Genetics</i> , 2007, 80, 485-494.	2.6	445
3	Epigenetic Modification of the <i>FMR1</i> Gene in Fragile X Syndrome Is Associated with Differential Response to the mGluR5 Antagonist AFQ056. <i>Science Translational Medicine</i> , 2011, 3, 64ra1.	5.8	344
4	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. <i>Nature Reviews Genetics</i> , 2018, 19, 649-666.	7.7	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. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	1.4	120
6	A double-blind, parallel, multicenter comparison of L-carnitine with placebo on the attention deficit hyperactivity disorder in fragile X syndrome boys. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 803-812.	0.7	91
7	Characterization of a highly polymorphic dinucleotide repeat 150 KB proximal to the fragile X site. <i>American Journal of Medical Genetics Part A</i> , 1992, 43, 237-243.	2.4	82
8	Facial diagnosis of mild and variant CdLS: Insights from a dysmorphologist survey. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1641-1653.	0.7	75
9	Mutations and variants in the cohesion factor genes <i>NIPBL</i> , <i>SMC1A</i> , and <i>SMC3</i> in a cohort of 30 unrelated patients with Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 924-929.	0.7	72
10	<i>De Novo</i> Heterozygous Mutations in <i>SMC3</i> Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. <i>Human Mutation</i> , 2015, 36, 454-462.	1.1	72
11	Phenotypes and genotypes in individuals with <i>SMC1A</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2108-2125.	0.7	69
12	Enlarged parietal foramina caused by mutations in the homeobox genes <i>ALX4</i> and <i>MSX2</i> : from genotype to phenotype. <i>European Journal of Human Genetics</i> , 2006, 14, 151-158.	1.4	67
13	A Novel Domain-Specific Mutation in a Sclerosteosis Patient Suggests a Role of <i>LRP4</i> as an Anchor for Sclerostin in Human Bone. <i>Journal of Bone and Mineral Research</i> , 2016, 31, 874-881.	3.1	65
14	Molecular genetics of HMG-CoA lyase deficiency. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 198-209.	0.5	64
15	Recurrent Mutations in the Basic Domain of <i>TWIST2</i> Cause Ablepharon Macrostomia and Barber-Say Syndromes. <i>American Journal of Human Genetics</i> , 2015, 97, 99-110.	2.6	61
16	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. <i>Human Genetics</i> , 2017, 136, 307-320.	1.8	61
17	Severe infantile-onset cardiomyopathy associated with a homozygous deletion in desmin. <i>Neuromuscular Disorders</i> , 2009, 19, 418-422.	0.3	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. <i>Menopause</i> , 2008, 15, 945-949.	0.8	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.	1.0	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.	1.1	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.	1.3	40
23	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
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.	1.4	39
25	Expanding the clinical spectrum of the "HDAC8 phenotype" implications for molecular diagnostics, counseling and risk prediction. Clinical Genetics, 2016, 89, 564-573.	1.0	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.	1.4	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.	2.9	36
29	Pathogenic variants in EP300 and ANKRD11 in patients with phenotypes overlapping Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1690-1696.	0.7	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.	1.3	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.	1.0	24
33	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	1.8	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.	1.8	22
36	Ten novelHMGCLmutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. Human Mutation, 2009, 30, E520-E529.	1.1	21

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37	Differential HMGâ€CoA lyase expression in human tissues provides clues about 3â€hydroxyâ€3â€methylglutaric aciduria. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 405-410.	1.7	20
38	Disruption of NIPBL/Sccl2 in Cornelia de Lange Syndrome provokes cohesin genome-wide redistribution with an impact in the transcriptome. <i>Nature Communications</i> , 2021, 12, 4551.	5.8	20
39	Special cases in Cornelia de Lange syndrome: The Spanish experience. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 198-205.	0.7	19
40	Two-step ATP-driven opening of cohesin head. <i>Scientific Reports</i> , 2017, 7, 3266.	1.6	19
41	Somatic mosaicism in a Cornelia de Lange syndrome patient with <i>NIPBL</i> mutation identified by different next generation sequencing approaches. <i>Clinical Genetics</i> , 2014, 86, 595-597.	1.0	17
42	Risk of cognitive impairment in female premutation carriers of fragile X premutation: Analysis by means of robust segmented linear regression models. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2009, 150B, 262-270.	1.1	14
43	Molecular Testing for Fragile X: Analysis of 5062 Tests from 1105 Fragile X Familiesâ€Performed in 12 Clinical Laboratories in Spain. <i>BioMed Research International</i> , 2014, 2014, 1-8.	0.9	13
44	MRX93 syndrome ( <i>BRWD3</i> gene): five new patients with novel mutations. <i>Clinical Genetics</i> , 2019, 95, 726-731.	1.0	13
45	Development, behaviour and autism in individuals with <i>SMC1A</i> variants. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2019, 60, 305-313.	3.1	13
46	Schuursâ€Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. <i>Genes</i> , 2021, 12, 738.	1.0	13
47	Cornelia de Lange syndrome with NIPBL mutation and mosaic Turner syndrome in the same individual. <i>BMC Medical Genetics</i> , 2012, 13, 43.	2.1	12
48	Could a patient with <i>SMC1A</i> duplication be classified as a human cohesinopathy?. <i>Clinical Genetics</i> , 2014, 85, 446-451.	1.0	12
49	Identification and Functional Characterization of Two Intronic NIPBL Mutations in Two Patients with Cornelia de Lange Syndrome. <i>BioMed Research International</i> , 2016, 2016, 1-8.	0.9	12
50	Clinical relevance of postzygotic mosaicism in Cornelia de Lange syndrome and purifying selection of NIPBL variants in blood. <i>Scientific Reports</i> , 2021, 11, 15459.	1.6	11
51	Giant meningioma in a 5-month-old infant. <i>Child's Nervous System</i> , 1988, 4, 112-115.	0.6	11
52	Autoimmune Thyroiditis After Bone Marrow Transplantation in a Boy With Wiskott-Aldrich Syndrome. <i>Journal of Pediatric Hematology/Oncology</i> , 2002, 24, 772-776.	0.3	10
53	Human variome project country nodes: Documenting genetic information within a country. <i>Human Mutation</i> , 2012, 33, 1513-1519.	1.1	10
54	Heterozygous de novo variants in <i>CSNK1G1</i> are associated with syndromic developmental delay and autism spectrum disorder. <i>Clinical Genetics</i> , 2020, 98, 571-576.	1.0	10

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55	Quantifying the economic impact of caregiving for Duchenne muscular dystrophy (DMD) in Spain. <i>European Journal of Health Economics</i> , 2020, 21, 1015-1023.	1.4	10
56	New ocular findings in two sisters with Yunisâ€™VarÃ³n syndrome and literature review. <i>European Journal of Medical Genetics</i> , 2011, 54, 76-81.	0.7	9
57	Detection of the fragile X syndrome protein for the evaluation of FMR1 intermediate alleles. <i>Human Genetics</i> , 2000, 107, 195-196.	1.8	8
58	CDKL5 gene status in female patients with epilepsy and Rett-like features: two new mutations in the catalytic domain. <i>BMC Medical Genetics</i> , 2012, 13, 68.	2.1	8
59	Severe ipsilateral musculoskeletal involvement in a Cornelia de Lange patient with a novel NIPBL mutation. <i>European Journal of Medical Genetics</i> , 2014, 57, 503-509.	0.7	8
60	A view on clinical genetics and genomics in Spain: of challenges and opportunities. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2016, 4, 376-391.	0.6	8
61	A further case of opsismodysplasia with hydrocephalus. <i>European Journal of Medical Genetics</i> , 2006, 49, 93-100.	0.7	5
62	Late-onset treatment in Menkes disease: is there a correlation between genotype and response to therapy?. <i>Clinical Genetics</i> , 2006, 69, 363-366.	1.0	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. <i>American Journal of Medical Genetics Part A</i> , 1992, 42, 835-838.	2.4	4
64	Tenorio syndrome: Description of 14 novel cases and review of the clinical and molecular features. <i>Clinical Genetics</i> , 2021, 100, 405-411.	1.0	2
65	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. <i>Applied Sciences (Switzerland)</i> , 2021, 11, 710.	1.3	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. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1826.	0.6	2
67	Direct DNA Testing for Fragile X Syndrome. <i>JAMA Pediatrics</i> , 1993, 147, 1231.	3.6	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. <i>International Journal of Molecular Sciences</i> , 2017, 18, 481.	1.8	1
69	Subclinical myocardial dysfunction is revealed by speckle tracking echocardiography in patients with Cornelia de Lange syndrome. <i>International Journal of Cardiovascular Imaging</i> , 2022, 38, 2291-2302.	0.2	1