

# Feliciano J Ramos

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5139588/publications.pdf>

Version: 2024-02-01

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	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
2	Schuursâ€œHoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. <i>Genes</i> , 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. <i>Scientific Reports</i> , 2021, 11, 15459.	1.6	11
4	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
5	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
6	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. <i>Applied Sciences (Switzerland)</i> , 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. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1826.	0.6	2
8	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
9	MAU2 and NIPBL Variants Impair the Heterodimerization of the Cohesin Loader Subunits and Cause Cornelia de Lange Syndrome. <i>Cell Reports</i> , 2020, 31, 107647.	2.9	36
10	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
11	Pathogenic variants in <i>EP300</i> and <i>ANKRD11</i> in patients with phenotypes overlapping Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1690-1696.	0.7	34
12	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. <i>Human Genetics</i> , 2020, 139, 575-592.	1.8	24
13	Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. <i>International Journal of Molecular Sciences</i> , 2020, 21, 1042.	1.8	40
14	MRX93 syndrome ( <i>BRWD3</i> gene): five new patients with novel mutations. <i>Clinical Genetics</i> , 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. <i>Frontiers in Neuroscience</i> , 2019, 13, 1135.	1.4	39
16	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
17	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. <i>Nature Reviews Genetics</i> , 2018, 19, 649-666.	7.7	223
18	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

#	ARTICLE	IF	CITATIONS
19	Two-step ATP-driven opening of cohesin head. <i>Scientific Reports</i> , 2017, 7, 3266.	1.6	19
20	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
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. <i>International Journal of Molecular Sciences</i> , 2017, 18, 481.	1.8	1
22	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
23	Expanding the clinical spectrum of the <i>HDAC8</i> phenotype™ implications for molecular diagnostics, counseling and risk prediction. <i>Clinical Genetics</i> , 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. <i>Journal of Bone and Mineral Research</i> , 2016, 31, 874-881.	3.1	65
25	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
26	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
27	<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
28	Clinical utility gene card for: Cornelia de Lange syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1431-1431.	1.4	37
29	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. <i>American Journal of Human Genetics</i> , 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. <i>International Journal of Molecular Sciences</i> , 2014, 15, 10350-10364.	1.8	22
31	Molecular Testing for Fragile X: Analysis of 5062 Tests from 1105 Fragile X Families Performed in 12 Clinical Laboratories in Spain. <i>BioMed Research International</i> , 2014, 2014, 1-8.	0.9	13
32	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
33	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
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. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	1.4	120
35	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
36	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

#	ARTICLE	IF	CITATIONS
37	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
38	Human variome project country nodes: Documenting genetic information within a country. <i>Human Mutation</i> , 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. <i>Molecular Biology Reports</i> , 2012, 39, 4777-4785.	1.0	24
40	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
41	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
42	The importance of rare diseases: from the gene to society. <i>Archives of Disease in Childhood</i> , 2011, 96, 791-792.	1.0	52
43	Differential HMGâ€“CoA lyase expression in human tissues provides clues about 3â€“hydroxyâ€“methylglutaric aciduria. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 405-410.	1.7	20
44	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
45	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
46	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
47	Antisense therapeutics for neurofibromatosis type 1 caused by deep intronic mutations. <i>Human Mutation</i> , 2009, 30, 454-462.	1.1	46
48	Ten novelHMGCLmutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. <i>Human Mutation</i> , 2009, 30, E520-E529.	1.1	21
49	Severe infantile-onset cardiomyopathy associated with a homozygous deletion in desmin. <i>Neuromuscular Disorders</i> , 2009, 19, 418-422.	0.3	58
50	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
51	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
52	Molecular genetics of HMG-CoA lyase deficiency. <i>Molecular Genetics and Metabolism</i> , 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. <i>American Journal of Human Genetics</i> , 2007, 80, 485-494.	2.6	445
54	A further case of opsismodysplasia with hydrocephalus. <i>European Journal of Medical Genetics</i> , 2006, 49, 93-100.	0.7	5

#	ARTICLE	IF	CITATIONS
55	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
56	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
57	Enlarged parietal foramina caused by mutations in the homeobox genes ALX4 and MSX2: from genotype to phenotype. <i>European Journal of Human Genetics</i> , 2006, 14, 151-158.	1.4	67
58	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
59	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
60	Epidermal naevus syndrome and hypophosphataemic rickets: description of a patient with central nervous system anomalies and review of the literature. <i>European Journal of Pediatrics</i> , 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. <i>European Journal of Clinical Microbiology and Infectious Diseases</i> , 1996, 15, 77-79.	1.3	28
63	Cystic kidney disease in Hajdu-Cheney syndrome. <i>American Journal of Medical Genetics Part A</i> , 1995, 56, 25-30.	2.4	37
64	Saethre-Chotzen syndrome with familial translocation at chromosome 7p22. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 637-639.	2.4	51
65	Direct DNA Testing for Fragile X Syndrome. <i>JAMA Pediatrics</i> , 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. <i>American Journal of Medical Genetics Part A</i> , 1992, 42, 835-838.	2.4	4
67	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
68	Tricho-Rhino-Phalangeal syndrome type II (Langer-Giedion) with persistent cloaca and prune belly sequence in a girl with 8q interstitial deletion. <i>American Journal of Medical Genetics Part A</i> , 1992, 44, 790-794.	2.4	23
69	Giant meningioma in a 5-month-old infant. <i>Child's Nervous System</i> , 1988, 4, 112-115.	0.6	11