Roser Urreizti

List of Publications by Year
in descending order

44 papers	3,215 citations	430754 18 h-index	243529 44 g-index
48 all docs	48 docs citations	48 times ranked	8786 citing authors

#	Article	IF	Citations
1	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
2	Genome-wide meta-analysis identifies 56 bone mineral density loci and reveals 14 loci associated with risk of fracture. Nature Genetics, 2012, 44, 491-501.	9.4	1,100
3	MiRNA profiling of whole trabecular bone: identification of osteoporosis-related changes in MiRNAs in human hip bones. BMC Medical Genomics, 2016, 8, 75.	0.7	67
4	GGPS1 Mutation and Atypical Femoral Fractures with Bisphosphonates. New England Journal of Medicine, 2017, 376, 1794-1795.	13.9	50
5	Assessment of gene-by-sex interaction effect on bone mineral density. Journal of Bone and Mineral Research, 2012, 27, 2051-2064.	3.1	47
6	Mutations in the EXT1 and EXT2 genes in Spanish patients with multiple osteochondromas. Scientific Reports, 2013, 3, 1346.	1.6	39
7	Polymorphisms of genes involved in homocysteine metabolism in preeclampsia and in uncomplicated pregnancies. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2005, 120, 45-52.	0.5	35
8	A De Novo Nonsense Mutation in MAGEL2 in a Patient Initially Diagnosed as Opitz-C: Similarities Between Schaaf-Yang and Opitz-C Syndromes. Scientific Reports, 2017, 7, 44138.	1.6	29
9	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28
10	The p.T191M mutation of the CBS gene is highly prevalent among homocystinuric patients from Spain, Portugal and South America. Journal of Human Genetics, 2006, 51, 305-313.	1.1	27
11	Effect of ILâ€1Î², PGE₂, and TGFâ€Î²1 on the expression of OPG and RANKL in normal and osteoporotic primary human osteoblasts. Journal of Cellular Biochemistry, 2010, 110, 304-310.	1.2	27
12	Analysis of Three Functional Polymorphisms in Relation to Osteoporosis Phenotypes: Replication in a Spanish Cohort. Calcified Tissue International, 2010, 87, 14-24.	1.5	25
13	Spectrum of CBS mutations in 16 homocystinuric patients from the Iberian Peninsula: High prevalence of T191M and absence of I278T or G307S. Human Mutation, 2003, 22, 103-103.	1.1	24
14	Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. Acta Neuropathologica, 2020, 140, 971-975.	3.9	24
15	Molecular characterization of five patients with homocystinuria due to severe methylenetetrahydrofolate reductase deficiency. Clinical Genetics, 2010, 78, 441-448.	1.0	23
16	Delineating the neurological phenotype in children with defects in the <scp>ECHS1</scp> or <scp>HIBCH</scp> gene. Journal of Inherited Metabolic Disease, 2021, 44, 401-414.	1.7	23
17	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
18	High prevalence ofCBS p.T191M mutation in homocystinuric patients from Colombia. Human Mutation, 2006, 27, 296-296.	1.1	21

#	Article	IF	Citations
19	Polymorphisms and haplotypes across the osteoprotegerin gene associated with bone mineral density and osteoporotic fractures. Osteoporosis International, 2010, 21, 287-296.	1.3	21
20	Five new cases of syndromic intellectual disability due to KAT6A mutations: widening the molecular and clinical spectrum. Orphanet Journal of Rare Diseases, 2020, 15, 44.	1.2	18
21	Identification and functional analyses of CBS alleles in Spanish and Argentinian homocystinuric patients. Human Mutation, 2011, 32, 835-842.	1.1	17
22	COL1A1 haplotypes and hip fracture. Journal of Bone and Mineral Research, 2012, 27, 950-953.	3.1	17
23	A broad spectrum of genomic changes in latinamerican patients with EXT1/EXT2-CDG. Scientific Reports, 2014, 4, 6407.	1.6	16
24	Functional assays testing pathogenicity of 14 cystathionine-beta synthase mutations. Human Mutation, 2006, 27, 211-211.	1.1	15
25	DPH1 syndrome: two novel variants and structural and functional analyses of seven missense variants identified in syndromic patients. European Journal of Human Genetics, 2020, 28, 64-75.	1.4	15
26	Functional relevance of the BMD-associated polymorphism rs312009: Novel Involvement of RUNX2 in LRP5 transcriptional regulation. Journal of Bone and Mineral Research, 2011, 26, 1133-1144.	3.1	14
27	Genetic Analysis of High Bone Mass Cases from the BARCOS Cohort of Spanish Postmenopausal Women. PLoS ONE, 2014, 9, e94607.	1.1	14
28	Genetic Sharing with Cardiovascular Disease Risk Factors and Diabetes Reveals Novel Bone Mineral Density Loci. PLoS ONE, 2015, 10, e0144531.	1.1	14
29	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	1.2	14
30	Hyperhomocysteinemia in children with renal transplants. Pediatric Nephrology, 2002, 17, 718-723.	0.9	13
31	Screening of CD96 and ASXL1 in 11 patients with Opitz C or Bohringâ€“Opitz syndromes. American Journal of Medical Genetics, Part A, 2016, 170, 24-31.	0.7	13
32	Analyses of RANK and RANKL in the Post-GWAS Context: Functional Evidence of Vitamin D Stimulation Through a RANKL Distal Region. Journal of Bone and Mineral Research, 2013, 28, 2550-2560.	3.1	11
33	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. Scientific Reports, 2018, 8, 694.	1.6	11
34	A CBS haplotype and a polymorphism at the MSR gene are associated with cardiovascular disease in a Spanish caseâ€“control study. Clinical Biochemistry, 2007, 40, 864-868.	0.8	10
35	Two successful pregnancies in pyridoxine-nonresponsive homocystinuria. Journal of Inherited Metabolic Disease, 2004, 27, 775-777.	1.7	9
36	The ASXL1 mutation p.Gly646Trpfs*12 found in a Turkish boy with Bohringâ€Opitz Syndrome. Clinical Case Reports (discontinued), 2018, 6, 1452-1456.	0.2	6

#	Article	IF	Citations
37	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	1.2	6
38	Case report of a child bearing a novel deleterious splicing variant in PIGT. Medicine (United States), 2019, 98, e14524.	0.4	5
39	Extending the phenotypic spectrum of Bohringâ€Opitz syndrome: Mild case confirmed by functional studies. American Journal of Medical Genetics, Part A, 2020, 182, 201-204.	0.7	5
40	De Novo PORCN and ZIC2 Mutations in a Highly Consanguineous Family. International Journal of Molecular Sciences, 2021, 22, 1549.	1.8	4
41	Heterozygous variants in <scp>ZBTB7A</scp> cause a neurodevelopmental disorder associated with symptomatic overgrowth of pharyngeal lymphoid tissue, macrocephaly, and elevated fetal hemoglobin. American Journal of Medical Genetics, Part A, 2022, 188, 272-282.	0.7	4
42	IdentificaciÃ³n de variantes genÃ©ticas asociadas con la densidad mineral Ã³sea (DMO) en el gen FLJ42280. Revista De Osteoporosis Y Metabolismo Mineral, 2017, 9, 28-34.	0.3	1
43	Understanding the Pathophysiology and Searching for Biomarkers for Rare Genetic Developmental Diseases. Proceedings (mdpi), 2019, 22, 53.	0.2	0
44	C syndrome - what do we know and what could the future hold?. Expert Opinion on Orphan Drugs, 2019, 7, 91-94.	0.5	0