Roser Urreizti

List of Publications by Year in descending order

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44	3,215	430874	243625
papers	citations	18 h-index	44 g-index

48	48	48	8786
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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.	1.2	4
2	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	2.8	6
3	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.	3.6	23
4	De Novo PORCN and ZIC2 Mutations in a Highly Consanguineous Family. International Journal of Molecular Sciences, 2021, 22, 1549.	4.1	4
5	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
6	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.	2.8	15
7	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.	1.2	5
8	Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. Acta Neuropathologica, 2020, 140, 971-975.	7.7	24
9	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
10	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	2.8	14
11	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.	2.7	18
12	Understanding the Pathophysiology and Searching for Biomarkers for Rare Genetic Developmental Diseases. Proceedings (mdpi), 2019, 22, 53.	0.2	0
13	C syndrome - what do we know and what could the future hold?. Expert Opinion on Orphan Drugs, 2019, 7, 91-94.	0.8	0
14	Case report of a child bearing a novel deleterious splicing variant in PIGT. Medicine (United States), 2019, 98, e14524.	1.0	5
15	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. Scientific Reports, 2018, 8, 694.	3.3	11
16	The ASXL1 mutation p.Gly646Trpfs*12 found in a Turkish boy with Bohringâ€Opitz Syndrome. Clinical Case Reports (discontinued), 2018, 6, 1452-1456.	0.5	6
17	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.	3.3	29
18	GGPS1 Mutation and Atypical Femoral Fractures with Bisphosphonates. New England Journal of Medicine, 2017, 376, 1794-1795.	27.0	50

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19	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
20	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.	1.2	13
21	MiRNA profiling of whole trabecular bone: identification of osteoporosis-related changes in MiRNAs in human hip bones. BMC Medical Genomics, 2016, 8, 75.	1.5	67
22	Genetic Sharing with Cardiovascular Disease Risk Factors and Diabetes Reveals Novel Bone Mineral Density Loci. PLoS ONE, 2015, 10, e0144531.	2.5	14
23	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	27.8	1,328
24	Genetic Analysis of High Bone Mass Cases from the BARCOS Cohort of Spanish Postmenopausal Women. PLoS ONE, 2014, 9, e94607.	2.5	14
25	A broad spectrum of genomic changes in latinamerican patients with EXT1/EXT2-CDG. Scientific Reports, 2014, 4, 6407.	3.3	16
26	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.	2.8	11
27	Mutations in the EXT1 and EXT2 genes in Spanish patients with multiple osteochondromas. Scientific Reports, 2013, 3, 1346.	3.3	39
28	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.	21.4	1,100
29	Assessment of gene-by-sex interaction effect on bone mineral density. Journal of Bone and Mineral Research, 2012, 27, 2051-2064.	2.8	47
30	COL1A1 haplotypes and hip fracture. Journal of Bone and Mineral Research, 2012, 27, 950-953.	2.8	17
31	Identification and functional analyses of CBS alleles in Spanish and Argentinian homocystinuric patients. Human Mutation, 2011, 32, 835-842.	2.5	17
32	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.	2.8	14
33	Analysis of Three Functional Polymorphisms in Relation to Osteoporosis Phenotypes: Replication in a Spanish Cohort. Calcified Tissue International, 2010, 87, 14-24.	3.1	25
34	Polymorphisms and haplotypes across the osteoprotegerin gene associated with bone mineral density and osteoporotic fractures. Osteoporosis International, 2010, 21, 287-296.	3.1	21
35	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.	2.6	27
36	Molecular characterization of five patients with homocystinuria due to severe methylenetetrahydrofolate reductase deficiency. Clinical Genetics, 2010, 78, 441-448.	2.0	23

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37	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.	1.9	10
38	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.	2.3	27
39	Functional assays testing pathogenicity of 14 cystathionine-beta synthase mutations. Human Mutation, 2006, 27, 211-211.	2.5	15
40	High prevalence ofCBS p.T191M mutation in homocystinuric patients from Colombia. Human Mutation, 2006, 27, 296-296.	2.5	21
41	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.	1.1	35
42	Two successful pregnancies in pyridoxine-nonresponsive homocystinuria. Journal of Inherited Metabolic Disease, 2004, 27, 775-777.	3.6	9
43	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.	2.5	24
44	Hyperhomocysteinemia in children with renal transplants. Pediatric Nephrology, 2002, 17, 718-723.	1.7	13