

Daniel Grinberg

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

160
papers

3,673
citations

32
h-index

51
g-index

188
ext. papers

4,169
ext. citations

5
avg, IF

4.46
L-index

#	Paper	IF	Citations
160	Whole-genome sequencing identifies EN1 as a determinant of bone density and fracture. <i>Nature</i> , 2015 , 526, 112-7	50.4	308
159	Differential genetic effects of ESR1 gene polymorphisms on osteoporosis outcomes. <i>JAMA - Journal of the American Medical Association</i> , 2004 , 292, 2105-14	27.4	238
158	The association between common vitamin D receptor gene variations and osteoporosis: a participant-level meta-analysis. <i>Annals of Internal Medicine</i> , 2006 , 145, 255-64	8	185
157	Glucocerebrosidase mutations confer a greater risk of dementia during Parkinson disease course. <i>Movement Disorders</i> , 2012 , 27, 393-9	7	102
156	Unusual expression of Gaucher disease: cardiovascular calcifications in three sibs homozygous for the D409H mutation. <i>Journal of Medical Genetics</i> , 1995 , 32, 740-2	5.8	89
155	Two new single-nucleotide polymorphisms in the COL1A1 upstream regulatory region and their relationship to bone mineral density. <i>Journal of Bone and Mineral Research</i> , 2002 , 17, 384-93	6.3	77
154	Cholesterol regulates Syntaxin 6 trafficking at trans-Golgi network endosomal boundaries. <i>Cell Reports</i> , 2014 , 7, 883-97	10.6	75
153	Simvastatin and atorvastatin enhance gene expression of collagen type 1 and osteocalcin in primary human osteoblasts and MG-63 cultures. <i>Journal of Cellular Biochemistry</i> , 2007 , 101, 1430-8	4.7	73
152	Homozygous tandem duplication within the gene encoding the beta-subunit of rod phosphodiesterase as a cause for autosomal recessive retinitis pigmentosa. <i>Human Mutation</i> , 1995 , 5, 228-34	4.7	65
151	Functional analysis of 13 GBA mutant alleles identified in Gaucher disease patients: Pathogenic changes and "modifier" polymorphisms. <i>Human Mutation</i> , 2004 , 23, 567-75	4.7	64
150	Twenty-one novel mutations in the GLB1 gene identified in a large group of GM1-gangliosidosis and Morquio B patients: possible common origin for the prevalent p.R59H mutation among gypsies. <i>Human Mutation</i> , 2006 , 27, 1060	4.7	53
149	MiRNA profiling of whole trabecular bone: identification of osteoporosis-related changes in MiRNAs in human hip bones. <i>BMC Medical Genomics</i> , 2015 , 8, 75	3.7	50
148	Haplotypes defined by promoter and intron 1 polymorphisms of the COL1A1 gene regulate bone mineral density in women. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006 , 91, 3575-83	5.6	48
147	A new locus for autosomal recessive retinitis pigmentosa (RP19) maps to 1p13-1p21. <i>Genomics</i> , 1997 , 40, 142-6	4.3	46
146	COL1A1, ESR1, VDR and TGFB1 polymorphisms and haplotypes in relation to BMD in Spanish postmenopausal women. <i>Osteoporosis International</i> , 2007 , 18, 235-43	5.3	45
145	Regulation of CYP19 gene expression in primary human osteoblasts: effects of vitamin D and other treatments. <i>European Journal of Endocrinology</i> , 2003 , 148, 519-26	6.5	45
144	Gaucher disease in Spanish patients: analysis of eight mutations. <i>Human Mutation</i> , 1995 , 5, 303-9	4.7	45

143	Molecular analysis and clinical findings in the Spanish Gaucher disease population: putative haplotype of the N370S ancestral chromosome. <i>Human Mutation</i> , 1998 , 11, 295-305	4.7	44
142	Antisense oligonucleotide treatment for a pseudoexon-generating mutation in the NPC1 gene causing Niemann-Pick type C disease. <i>Human Mutation</i> , 2009 , 30, E993-E1001	4.7	41
141	Identification and characterization of SMPD1 mutations causing Niemann-Pick types A and B in Spanish patients. <i>Human Mutation</i> , 2009 , 30, 1117-22	4.7	39
140	Polymorphisms in the interleukin-6 receptor gene are associated with bone mineral density and body mass index in Spanish postmenopausal women. <i>European Journal of Endocrinology</i> , 2007 , 157, 677-84	4.7	39
139	A mutation within the saposin D domain in a Gaucher disease patient with normal glucocerebrosidase activity. <i>Human Genetics</i> , 2005 , 117, 275-7	6.3	37
138	Molecular analysis of 30 Niemann-Pick type C patients from Spain. <i>Clinical Genetics</i> , 2011 , 80, 39-49	4	35
137	Promising results of the chaperone effect caused by imino sugars and aminocyclitol derivatives on mutant glucocerebrosidases causing Gaucher disease. <i>Blood Cells, Molecules, and Diseases</i> , 2009 , 42, 159-66	2.1	35
136	Identification of 14 novel GLB1 mutations, including five deletions, in 19 patients with GM1 gangliosidosis from South America. <i>Clinical Genetics</i> , 2007 , 71, 273-9	4	35
135	Genetic determinants of aromatase inhibitor-related arthralgia: the B-ABLE cohort study. <i>Breast Cancer Research and Treatment</i> , 2013 , 140, 385-95	4.4	34
134	Mutations in the EXT1 and EXT2 genes in Spanish patients with multiple osteochondromas. <i>Scientific Reports</i> , 2013 , 3, 1346	4.9	34
133	Glucocerebrosidase gene mutations in two cohorts of Greek patients with sporadic Parkinson disease. <i>Molecular Genetics and Metabolism</i> , 2011 , 104, 149-52	3.7	34
132	Mucopolysaccharidosis type VI phenotypes-genotypes and antibody response to galsulfase. <i>Orphanet Journal of Rare Diseases</i> , 2013 , 8, 51	4.2	33
131	Mapping the genetic and clinical characteristics of Gaucher disease in the Iberian Peninsula. <i>Orphanet Journal of Rare Diseases</i> , 2012 , 7, 17	4.2	33
130	Polymorphisms of genes involved in homocysteine metabolism in preeclampsia and in uncomplicated pregnancies. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2005 , 120, 45-52	2.4	33
129	GGPS1 Mutation and Atypical Femoral Fractures with Bisphosphonates. <i>New England Journal of Medicine</i> , 2017 , 376, 1794-1795	59.2	32
128	In vitro functional assay of alleles and haplotypes of two COL1A1-promoter SNPs. <i>Bone</i> , 2005 , 36, 902-8	4.7	32
127	Therapeutic strategies based on modified U1 snRNAs and chaperones for Sanfilippo C splicing mutations. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 180	4.2	31
126	Screening of CACNA1A and ATP1A2 genes in hemiplegic migraine: clinical, genetic, and functional studies. <i>Molecular Genetics & Genomic Medicine</i> , 2013 , 1, 206-22	2.3	31

125	An evolutionary and structure-based docking model for glucocerebrosidase-saposin C and glucocerebrosidase-substrate interactions - relevance for Gaucher disease. <i>Proteins: Structure, Function and Bioinformatics</i> , 2008 , 70, 882-91	4.2	31
124	Present and future of antisense therapy for splicing modulation in inherited metabolic disease. <i>Journal of Inherited Metabolic Disease</i> , 2010 , 33, 397-403	5.4	29
123	Genetic fine localization of the beta-glucocerebrosidase (GBA) and prosaposin (PSAP) genes: implications for Gaucher disease. <i>Human Genetics</i> , 1997 , 100, 75-9	6.3	29
122	Promoter 2 -1025 T/C polymorphism in the RUNX2 gene is associated with femoral neck bmd in Spanish postmenopausal women. <i>Calcified Tissue International</i> , 2007 , 81, 327-32	3.9	29
121	Mutation 1091delC is highly prevalent in Spanish Sanfilippo syndrome type A patients. <i>Human Mutation</i> , 1998 , 12, 274-9	4.7	28
120	Identification of the molecular defects in Spanish and Argentinian mucopolysaccharidosis VI (Maroteaux-Lamy syndrome) patients, including 9 novel mutations. <i>Molecular Genetics and Metabolism</i> , 2007 , 92, 122-30	3.7	28
119	Mutation analysis of Gaucher disease patients from Argentina: high prevalence of the RecNcil mutation. <i>American Journal of Medical Genetics Part A</i> , 1998 , 80, 343-51		27
118	Effect of IL-1beta, PGE(2), and TGF-beta1 on the expression of OPG and RANKL in normal and osteoporotic primary human osteoblasts. <i>Journal of Cellular Biochemistry</i> , 2010 , 110, 304-10	4.7	26
117	Functional analysis of the I.3, I.6, pII and I.4 promoters of CYP19 (aromatase) gene in human osteoblasts and their role in vitamin D and dexamethasone stimulation. <i>European Journal of Endocrinology</i> , 2005 , 153, 981-8	6.5	26
116	Activity and High-Order Effective Connectivity Alterations in Sanfilippo C Patient-Specific Neuronal Networks. <i>Stem Cell Reports</i> , 2015 , 5, 546-57	8	25
115	A new autosomal recessive retinitis pigmentosa locus maps on chromosome 2q31-q33. <i>Journal of Medical Genetics</i> , 1998 , 35, 141-5	5.8	25
114	A De Novo Nonsense Mutation in MAGEL2 in a Patient Initially Diagnosed as Opitz-C: Similarities Between Schaaf-Yang and Opitz-C Syndromes. <i>Scientific Reports</i> , 2017 , 7, 44138	4.9	24
113	Maroteaux-Lamy syndrome: functional characterization of pathogenic mutations and polymorphisms in the arylsulfatase B gene. <i>Molecular Genetics and Metabolism</i> , 2008 , 94, 305-12	3.7	24
112	A new SNP in a negative regulatory region of the CYP19A1 gene is associated with lumbar spine BMD in postmenopausal women. <i>Bone</i> , 2006 , 38, 738-43	4.7	24
111	Homocysteine and the MTHFR 677C-->T allele in premature coronary artery disease. Case control and family studies. <i>European Journal of Clinical Investigation</i> , 2001 , 31, 24-30	4.6	24
110	Analysis of three functional polymorphisms in relation to osteoporosis phenotypes: replication in a Spanish cohort. <i>Calcified Tissue International</i> , 2010 , 87, 14-24	3.9	23
109	Expression and characterization of 14 GLB1 mutant alleles found in GM1-gangliosidosis and Morquio B patients. <i>Journal of Lipid Research</i> , 2007 , 48, 2275-82	6.3	23
108	Evaluation of Aminoglycoside and Non-Aminoglycoside Compounds for Stop-Codon Readthrough Therapy in Four Lysosomal Storage Diseases. <i>PLoS ONE</i> , 2015 , 10, e0135873	3.7	22

107	Two new mild homozygous mutations in Gaucher disease patients: clinical signs and biochemical analyses. <i>American Journal of Medical Genetics Part A</i> , 1997 , 70, 437-43		22
106	Perinatal lethal phenotype with generalized ichthyosis in a type 2 Gaucher disease patient with the [L444P;E326K]/P182L genotype: effect of the E326K change in neonatal and classic forms of the disease. <i>Blood Cells, Molecules, and Diseases</i> , 2005 , 35, 253-8	2.1	21
105	Spectrum of CBS mutations in 16 homocystinuric patients from the Iberian Peninsula: high prevalence of T191M and absence of I278T or G307S. <i>Human Mutation</i> , 2003 , 22, 103	4.7	21
104	Polymorphisms and haplotypes across the osteoprotegerin gene associated with bone mineral density and osteoporotic fractures. <i>Osteoporosis International</i> , 2010 , 21, 287-96	5.3	20
103	The p.T191M mutation of the CBS gene is highly prevalent among homocystinuric patients from Spain, Portugal and South America. <i>Journal of Human Genetics</i> , 2006 , 51, 305-313	4.3	20
102	CYP11A1 expression in bone is associated with aromatase inhibitor-related bone loss. <i>Journal of Molecular Endocrinology</i> , 2015 , 55, 69-79	4.5	19
101	EXTL2 and EXTL3 inhibition with siRNAs as a promising substrate reduction therapy for Sanfilippo C syndrome. <i>Scientific Reports</i> , 2015 , 5, 13654	4.9	19
100	Molecular characterization of five patients with homocystinuria due to severe methylenetetrahydrofolate reductase deficiency. <i>Clinical Genetics</i> , 2010 , 78, 441-8	4	19
99	A new gene-pseudogene fusion allele due to a recombination in intron 2 of the glucocerebrosidase gene causes Gaucher disease. <i>Blood Cells, Molecules, and Diseases</i> , 2000 , 26, 409-16	2.1	19
98	Molecular analysis of Sanfilippo syndrome type C in Spain: seven novel HGSNAT mutations and characterization of the mutant alleles. <i>Clinical Genetics</i> , 2011 , 80, 367-74	4	18
97	A novel mutation in exon 17 of the beta-subunit of rod phosphodiesterase in two RP sisters of a consanguineous family. <i>Human Genetics</i> , 1996 , 97, 35-8	6.3	18
96	Identification of positive and negative regulatory elements involved in the retinoic acid/cAMP induction of Fgf-3 transcription in F9 cells. <i>Nucleic Acids Research</i> , 1993 , 21, 5351-9	20.1	18
95	Characterisation of two deletions involving NPC1 and flanking genes in Niemann-Pick type C disease patients. <i>Molecular Genetics and Metabolism</i> , 2012 , 107, 716-20	3.7	17
94	Identification and functional analyses of CBS alleles in Spanish and Argentinian homocystinuric patients. <i>Human Mutation</i> , 2011 , 32, 835-42	4.7	17
93	A haplotype-based analysis of the LRP5 gene in relation to osteoporosis phenotypes in Spanish postmenopausal women. <i>Journal of Bone and Mineral Research</i> , 2008 , 23, 1954-63	6.3	17
92	Neuronopathic and non-neuronopathic presentation of Gaucher disease in patients with the third most common mutation (D409H) in Spain. <i>Journal of Inherited Metabolic Disease</i> , 1996 , 19, 798-800	5.4	17
91	New murine Niemann-Pick type C models bearing a pseudoexon-generating mutation recapitulate the main neurobehavioural and molecular features of the disease. <i>Scientific Reports</i> , 2017 , 7, 41931	4.9	16
90	Clinical and mutational characterization of three patients with multiple sulfatase deficiency: report of a new splicing mutation. <i>Molecular Genetics and Metabolism</i> , 2005 , 86, 206-11	3.7	16

89	Gaucher disease: the N370S mutation in Ashkenazi Jewish and Spanish patients has a common origin and arose several thousand years ago. <i>American Journal of Human Genetics</i> , 1999 , 64, 1233-8	11	16
88	Evidence against involvement of recoverin in autosomal recessive retinitis pigmentosa in 42 Spanish families. <i>Human Genetics</i> , 1995 , 96, 89-94	6.3	16
87	Identification of a novel pseudodeficiency allele in the GLB1 gene in a carrier of GM1 gangliosidosis. <i>Clinical Genetics</i> , 2007 , 72, 109-11	4	15
86	High prevalence of CBS p.T191M mutation in homocystinuric patients from Colombia. <i>Human Mutation</i> , 2006 , 27, 296	4.7	15
85	RNAi-mediated inhibition of the glucosylceramide synthase (GCS) gene: A preliminary study towards a therapeutic strategy for Gaucher disease and other glycosphingolipid storage diseases. <i>Blood Cells, Molecules, and Diseases</i> , 2006 , 37, 197-203	2.1	15
84	Homozygosity for the double D409H+H255Q allele in type II Gaucher disease. <i>Journal of Inherited Metabolic Disease</i> , 2006 , 29, 591	5.4	15
83	A folate-rich diet is as effective as folic acid from supplements in decreasing plasma homocysteine concentrations. <i>International Journal of Medical Sciences</i> , 2005 , 2, 58-63	3.7	15
82	Haplotype analysis suggests a single Balkan origin for the Gaucher disease [D409H;H255Q] double mutant allele. <i>Human Mutation</i> , 2008 , 29, E58-67	4.7	14
81	Functional assays testing pathogenicity of 14 cystathionine-beta synthase mutations. <i>Human Mutation</i> , 2006 , 27, 211	4.7	14
80	Mutation and haplotype analyses in 26 Spanish Sanfilippo syndrome type A patients: possible single origin for 1091delC mutation. <i>American Journal of Medical Genetics Part A</i> , 2001 , 100, 223-8		14
79	Functional relevance of the BMD-associated polymorphism rs312009: novel involvement of RUNX2 in LRP5 transcriptional regulation. <i>Journal of Bone and Mineral Research</i> , 2011 , 26, 1133-44	6.3	13
78	Recurrence of the D409H mutation in Spanish Gaucher disease patients: description of a new homozygous patient and haplotype analysis. <i>Journal of Medical Genetics</i> , 1998 , 35, 775-7	5.8	13
77	Pro-osteoporotic miR-320a impairs osteoblast function and induces oxidative stress. <i>PLoS ONE</i> , 2018 , 13, e0208131	3.7	13
76	Genetic analysis of high bone mass cases from the BARCOS cohort of Spanish postmenopausal women. <i>PLoS ONE</i> , 2014 , 9, e94607	3.7	12
75	Unsuccessful chimeroplast strategy for the correction of a mutation causing Gaucher disease. <i>Blood Cells, Molecules, and Diseases</i> , 2003 , 31, 183-6	2.1	12
74	A novel mutation in the BRCA1 gene in a German early-onset breast cancer family. <i>Human Mutation</i> , 1996 , 8, 393	4.7	12
73	Sanfilippo Syndrome: Molecular Basis, Disease Models and Therapeutic Approaches. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	12
72	Functional Characterization of a GGPPS Variant Identified in Atypical Femoral Fracture Patients and Delineation of the Role of GGPPS in Bone-Relevant Cell Types. <i>Journal of Bone and Mineral Research</i> , 2018 , 33, 2091-2098	6.3	12

71	Antisense mediated splicing modulation for inherited metabolic diseases: challenges for delivery. <i>Nucleic Acid Therapeutics</i> , 2014 , 24, 48-56	4.8	11
70	COL1A1 haplotypes and hip fracture. <i>Journal of Bone and Mineral Research</i> , 2012 , 27, 950-3	6.3	11
69	Analyses of RANK and RANKL in the post-GWAS context: functional evidence of vitamin D stimulation through a RANKL distal region. <i>Journal of Bone and Mineral Research</i> , 2013 , 28, 2550-60	6.3	11
68	Frequency of the arylsulphatase A pseudodeficiency allele in the Spanish population. <i>Clinical Genetics</i> , 1993 , 44, 320-3	4	11
67	Two novel (1098insA and Y313H) and one rare (R359Q) mutations detected in exon 8 of the beta-glucocerebrosidase gene in Gaucher disease patients. <i>Human Mutation</i> , 1996 , 7, 272-4	4.7	11
66	Screening of CD96 and ASXL1 in 11 patients with Opitz C or Bohring-Opitz syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 24-31	2.5	11
65	Expression profiling of microRNAs in human bone tissue from postmenopausal women. <i>Human Cell</i> , 2018 , 31, 33-41	4.5	10
64	Glucocerebrosidase enhancers for selected Gaucher disease genotypes by modification of E1-C-substituted imino-D-xylitols (DIXs) by click chemistry. <i>ChemMedChem</i> , 2014 , 9, 1744-54	3.7	10
63	Common and rare variants of WNT16, DKK1 and SOST and their relationship with bone mineral density. <i>Scientific Reports</i> , 2018 , 8, 10951	4.9	9
62	Niemann-Pick type C disease: a novel NPC1 mutation segregating in a Greek island. <i>Clinical Genetics</i> , 2014 , 85, 543-7	4	9
61	A broad spectrum of genomic changes in latinamerican patients with EXT1/EXT2-CDG. <i>Scientific Reports</i> , 2014 , 4, 6407	4.9	9
60	A CBS haplotype and a polymorphism at the MSR gene are associated with cardiovascular disease in a Spanish case-control study. <i>Clinical Biochemistry</i> , 2007 , 40, 864-8	3.5	9
59	New insights into the origin of the Gaucher disease-causing mutation N370S: extended haplotype analysis using the 5GC3.2, 5470 G/A, and ITG6.2 polymorphisms. <i>Blood Cells, Molecules, and Diseases</i> , 2001 , 27, 950-9	2.1	9
58	Bone development and remodeling in metabolic disorders. <i>Journal of Inherited Metabolic Disease</i> , 2020 , 43, 133-144	5.4	9
57	Mutational spectrum by phenotype: panel-based NGS testing of patients with clinical suspicion of RASopathy and children with multiple café-au-lait macules. <i>Clinical Genetics</i> , 2020 , 97, 264-275	4	9
56	The Spectrum of Niemann-Pick Type C Disease in Greece. <i>JIMD Reports</i> , 2017 , 36, 41-48	1.9	8
55	Functional characterization of the C7ORF76 genomic region, a prominent GWAS signal for osteoporosis in 7q21.3. <i>Bone</i> , 2019 , 123, 39-47	4.7	8
54	Selective chaperone effect of aminocyclitol derivatives on G202R and other mutant glucocerebrosidases causing Gaucher disease. <i>International Journal of Biochemistry and Cell Biology</i> , 2014 , 54, 245-54	5.6	8

53	Molecular characterization of a new deletion of the GBA1 gene due to an inter Alu recombination event. <i>Molecular Genetics and Metabolism</i> , 2011 , 102, 226-8	3.7	8
52	Autosomal recessive retinitis pigmentosa in Spain: evaluation of four genes and two loci involved in the disease. <i>Clinical Genetics</i> , 1996 , 50, 380-7	4	8
51	Analysis of nonsense-mediated mRNA decay in mutant alleles identified in Spanish Gaucher disease patients. <i>Blood Cells, Molecules, and Diseases</i> , 2006 , 36, 46-52	2.1	8
50	Transcriptional regulation of the int-2 gene in embryonal carcinoma cells. <i>Cell Growth & Differentiation: the Molecular Biology Journal of the American Association for Cancer Research</i> , 1991 , 2, 137-43		8
49	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. <i>Genetics in Medicine</i> , 2020 , 22, 1215-1226	8.1	7
48	Five new cases of syndromic intellectual disability due to KAT6A mutations: widening the molecular and clinical spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 44	4.2	7
47	Putative association of a mutant ROM1 allele with retinitis pigmentosa. <i>Human Genetics</i> , 1997 , 99, 827-30	3	7
46	Two successful pregnancies in pyridoxine-nonresponsive homocystinuria. <i>Journal of Inherited Metabolic Disease</i> , 2004 , 27, 775-7	5.4	7
45	No evidence of linkage to 6p markers in spanish families with juvenile myoclonic epilepsy. <i>Neuroscience Letters</i> , 2000 , 286, 213-7	3.3	7
44	Genetic fine localization of the arrestin (S-antigen) gene 4 cM distal from D2S172. <i>Human Genetics</i> , 1994 , 94, 193-4	6.3	7
43	Stereodivergent synthesis of right- and left-handed iminoxylitol heterodimers and monomers. Study of their impact on Eglucocerebrosidase activity. <i>Organic and Biomolecular Chemistry</i> , 2017 , 15, 3681-3705	3.9	6
42	Neuronal and Astrocytic Differentiation from Sanfilippo C Syndrome iPSCs for Disease Modeling and Drug Development. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	6
41	SR proteins and the nonsense-mediated decay mechanism are involved in human GLB1 gene alternative splicing. <i>BMC Research Notes</i> , 2008 , 1, 137	2.3	6
40	Expression and functional characterization of human mutant sulfamidase in insect cells. <i>Molecular Genetics and Metabolism</i> , 2004 , 83, 246-51	3.7	6
39	The Spectrum of Krabbe Disease in Greece: Biochemical and Molecular Findings. <i>JIMD Reports</i> , 2016 , 25, 57-64	1.9	5
38	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. <i>Scientific Reports</i> , 2018 , 8, 694	4.9	5
37	Generation of a human neuronal stable cell model for niemann-pick C disease by RNA interference. <i>JIMD Reports</i> , 2012 , 4, 29-37	1.9	5
36	Bone mass of a 113-year-old man. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , 2007 , 62, 794-5	6.4	5

35	Linkage disequilibrium detected between myotonic dystrophy and the anonymous marker D19S63 in the Spanish population. <i>Human Genetics</i> , 1992 , 89, 287-91	6.3	5
34	Involvement of Gaucher Disease Mutations in Parkinson Disease. <i>Current Protein and Peptide Science</i> , 2017 , 18, 758-764	2.8	5
33	Generation of two compound heterozygous HGSNAT-mutated lines from healthy induced pluripotent stem cells using CRISPR/Cas9 to model Sanfilippo C syndrome. <i>Stem Cell Research</i> , 2019 , 41, 101616	1.6	5
32	DPH1 syndrome: two novel variants and structural and functional analyses of seven missense variants identified in syndromic patients. <i>European Journal of Human Genetics</i> , 2020 , 28, 64-75	5.3	5
31	The mutation p.Gly646Trpfs*12 found in a Turkish boy with Bohring-Opitz Syndrome. <i>Clinical Case Reports (discontinued)</i> , 2018 , 6, 1452-1456	0.7	5
30	On the age of the most prevalent Gaucher disease-causing mutation, N370S. <i>American Journal of Human Genetics</i> , 2000 , 66, 2014-5	11	4
29	Extending the phenotypic spectrum of Bohring-Opitz syndrome: Mild case confirmed by functional studies. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 201-204	2.5	4
28	Case report of a child bearing a novel deleterious splicing variant in PIGT. <i>Medicine (United States)</i> , 2019 , 98, e14524	1.8	4
27	Functional Assessment of Coding and Regulatory Variants From the Locus. <i>JBMR Plus</i> , 2020 , 4, e10423	3.9	3
26	Discrepancy between bone density and bone material strength index in three siblings with Camurati-Engelmann disease. <i>Osteoporosis International</i> , 2017 , 28, 3489-3493	5.3	3
25	Perinatal lethal form of Gaucher disease. Clinical and molecular characterization of a Greek case. <i>Blood Cells, Molecules, and Diseases</i> , 2010 , 44, 82-3	2.1	3
24	Recent patents relating to siRNAs and therapeutic strategies for genetic diseases. <i>Recent Patents on DNA & Gene Sequences</i> , 2008 , 2, 40-3		3
23	Wnt Pathway Extracellular Components and Their Essential Roles in Bone Homeostasis.. <i>Genes</i> , 2022 , 13,	4.2	3
22	Estudio del patrón de expresión de microRNAs en el hueso osteoporótico. <i>Revista De Osteoporosis Y Metabolismo Mineral</i> , 2016 , 8, 5-14	1	3
21	Generation of two NAGLU-mutated homozygous cell lines from healthy induced pluripotent stem cells using CRISPR/Cas9 to model Sanfilippo B syndrome. <i>Stem Cell Research</i> , 2020 , 42, 101668	1.6	3
20	Genetics and Genomics of : Functional Analysis of Variants and Genomic Regulation in Osteoblasts. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	3
19	A Roadmap to Gene Discoveries and Novel Therapies in Monogenic Low and High Bone Mass Disorders. <i>Frontiers in Endocrinology</i> , 2021 , 12, 709711	5.7	3
18	Gaucher disease: Biochemical and molecular findings in 141 patients diagnosed in Greece. <i>Molecular Genetics and Metabolism Reports</i> , 2020 , 24, 100614	1.8	2

17	Screening of cacna1a and ATP1A2 genes in hemiplegic migraine: clinical, genetic and functional studies. <i>Journal of Headache and Pain</i> , 2013 , 14,	8.8	2
16	Description of extreme longevity in the Balearic Islands: Exploring a potential Blue Zone in Menorca, Spain. <i>Geriatrics and Gerontology International</i> , 2014 , 14, 620-7	2.9	2
15	A novel nonsense mutation of the EXT1 gene in an Argentinian patient with multiple hereditary exostoses: a case report. <i>Journal of Bone and Joint Surgery - Series A</i> , 2012 , 94, e76	5.6	2
14	A response to Kowarz et al.: Gaucher mutation c.680A>G (p.N227S) is associated with myoclonic epilepsy. <i>Human Mutation</i> , 2005 , 26, 274-275	4.7	2
13	and Mutations in a Highly Consanguineous Family. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	2
12	Strategies for the Genetic Analysis of Autosomal Recessive Retinitis Pigmentosa in Spanish Families 1997 , 263-275		2
11	Reliable co-segregation analysis for prenatal diagnosis and heterozygote detection in Gaucher disease 1998 , 18, 207-212		1
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