Daniel Grinberg

List of Publications by Year
in descending order

168 papers	4,678 citations	109311 35 h-index	138468 58 g-index
188 all docs	188 docs citations	188 times ranked	6636 citing authors

#	Article	IF	Citations
1	Wholeâ€genome sequencing identifies EN1 as a determinant of bone density and fracture. Nature, 2015, 526, 112-117.	27.8	483
2	Differential Genetic Effects of <EMPH TYPE="ITAL">ESR1</EMPH> Gene Polymorphisms on Osteoporosis Outcomes. JAMA - Journal of the American Medical Association, 2004, 292, 2105.	7.4	265
3	The Association between Common Vitamin D Receptor Gene Variations and Osteoporosis: A Participant-Level Meta-Analysis. Annals of Internal Medicine, 2006, 145, 255.	3.9	219
4	Glucocerebrosidase mutations confer a greater risk of dementia during Parkinson's disease course. Movement Disorders, 2012, 27, 393-399.	3.9	144
5	Unusual expression of Gaucher's disease: cardiovascular calcifications in three sibs homozygous for the D409H mutation.. Journal of Medical Genetics, 1995, 32, 740-742.	3.2	110
6	Cholesterol Regulates Syntaxin 6 Trafficking at trans-Golgi Network Endosomal Boundaries. Cell Reports, 2014, 7, 883-897.	6.4	104
7	Two New Single-Nucleotide Polymorphisms in the COL1A1 Upstream Regulatory Region and Their Relationship to Bone Mineral Density. Journal of Bone and Mineral Research, 2002, 17, 384-393.	2.8	91
8	Functional analysis of 13GBAmutant alleles identified in Gaucher disease patients: Pathogenic changes and â€œmodifierâ€•polymorphisms. Human Mutation, 2004, 23, 567-575.	2.5	80
9	Simvastatin and atorvastatin enhance gene expression of collagen type 1 and osteocalcin in primary human osteoblasts and MG-63 cultures. Journal of Cellular Biochemistry, 2007, 101, 1430-1438.	2.6	80
10	Homozygous tandem duplication within the gene encoding the Î²-subunit of rod phosphodiesterase as a cause for autosomal recessive retinitis pigmentosa. Human Mutation, 1995, 5, 228-234.	2.5	68
11	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
12	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. Human Mutation, 2006, 27, 1060-1060.	2.5	59
13	Regulation of CYP19 gene expression in primary human osteoblasts: effects of vitamin D and other treatments. European Journal of Endocrinology, 2003, 148, 519-526.	3.7	58
14	A New Locus for Autosomal Recessive Retinitis Pigmentosa (RP19) Maps to 1p13â€“1p21. Genomics, 1997, 40, 142-146.	2.9	56
15	COL1A1, ESR1, VDR and TGFB1 polymorphisms and haplotypes in relation to BMD in Spanish postmenopausal women. Osteoporosis International, 2007, 18, 235-243.	3.1	56
16	Haplotypes Defined by Promoter and Intron 1 Polymorphisms of the COLIA1 Gene Regulate Bone Mineral Density in Women. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 3575-3583.	3.6	55
17	Identification and characterization ofSMPD1mutations causing Niemann-Pick types A and B in Spanish patients. Human Mutation, 2009, 30, 1117-1122.	2.5	54
18	Molecular analysis and clinical findings in the Spanish Gaucher disease population: Putative haplotype of the N370S ancestral chromosome. Human Mutation, 1998, 11, 295-305.	2.5	52

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19	GGPS1 Mutation and Atypical Femoral Fractures with Bisphosphonates. New England Journal of Medicine, 2017, 376, 1794-1795.	27.0	50
20	Antisense oligonucleotide treatment for a pseudoexon-generating mutation in theNPC1gene causing Niemann-Pick type C disease. Human Mutation, 2009, 30, E993-E1001.	2.5	47
21	Î²-Glucocerebrosidase gene mutations in two cohorts of Greek patients with sporadic Parkinson's disease. Molecular Genetics and Metabolism, 2011, 104, 149-152.	1.1	47
22	Gaucher disease in Spanish patients: Analysis of eight mutations. Human Mutation, 1995, 5, 303-309.	2.5	46
23	Polymorphisms in the interleukin-6 receptor gene are associated with bone mineral density and body mass index in Spanish postmenopausal women. European Journal of Endocrinology, 2007, 157, 677-684.	3.7	42
24	Therapeutic strategies based on modified U1 snRNAs and chaperones for Sanfilippo C splicing mutations. Orphanet Journal of Rare Diseases, 2014, 9, 180.	2.7	42
25	A mutation within the saposin D domain in a Gaucher disease patient with normal glucocerebrosidase activity. Human Genetics, 2005, 117, 275-277.	3.8	41
26	Identification of 14 novel GLB1 mutations, including five deletions, in 19 patients with GM1 gangliosidosis from South America. Clinical Genetics, 2007, 71, 273-279.	2.0	41
27	Molecular analysis of 30 Niemann-Pick type C patients from Spain. Clinical Genetics, 2011, 80, 39-49.	2.0	40
28	Mapping the genetic and clinical characteristics of Gaucher disease in the Iberian Peninsula. Orphanet Journal of Rare Diseases, 2012, 7, 17.	2.7	40
29	Mutations in the EXT1 and EXT2 genes in Spanish patients with multiple osteochondromas. Scientific Reports, 2013, 3, 1346.	3.3	39
30	Present and future of antisense therapy for splicing modulation in inherited metabolic disease. Journal of Inherited Metabolic Disease, 2010, 33, 397-403.	3.6	38
31	Genetic fine localization of the Î²-glucocerebrosidase (GBA) and prosaposin (PSAP) genes: implications for Gaucher disease. Human Genetics, 1997, 100, 75-79.	3.8	37
32	Mucopolysaccharidosis type VI phenotypes-genotypes and antibody response to galsulfase. Orphanet Journal of Rare Diseases, 2013, 8, 51.	2.7	37
33	Genetic determinants of aromatase inhibitor-related arthralgia: the B-ABLE cohort study. Breast Cancer Research and Treatment, 2013, 140, 385-395.	2.5	37
34	Mutation 1091delC is highly prevalent in Spanish Sanfilippo syndrome type A patients. Human Mutation, 1998, 12, 274-279.	2.5	36
35	In vitro functional assay of alleles and haplotypes of two -promoter SNPs. Bone, 2005, 36, 902-908.	2.9	36
36	Promising results of the chaperone effect caused by iminosugars and aminocyclitol derivatives on mutant glucocerebrosidases causing Gaucher disease. Blood Cells, Molecules, and Diseases, 2009, 42, 159-166.	1.4	36

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37	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
38	An evolutionary and structureâ€based docking model for glucocerebrosidaseâ€“saposin C and glucocerebrosidaseâ€“substrate interactionsâ€”Relevance for Gaucher disease. Proteins: Structure, Function and Bioinformatics, 2008, 70, 882-891.	2.6	35
39	Screening of <scp>CACNA1A</scp> and <scp>ATP1A2</scp> genes in hemiplegic migraine: clinical, genetic, and functional studies. Molecular Genetics & Genomic Medicine, 2013, 1, 206-222.	1.2	35
40	A new autosomal recessive retinitis pigmentosa locus maps on chromosome 2q31-q33.. Journal of Medical Genetics, 1998, 35, 141-145.	3.2	34
41	Homocysteine and the MTHFR 677Câ†'T allele in premature coronary artery disease. Case control and family studies. European Journal of Clinical Investigation, 2001, 31, 24-30.	3.4	34
42	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. European Journal of Endocrinology, 2005, 153, 981-988.	3.7	34
43	Evaluation of Aminoglycoside and Non-Aminoglycoside Compounds for Stop-Codon Readthrough Therapy in Four Lysosomal Storage Diseases. PLoS ONE, 2015, 10, e0135873.	2.5	33
44	Promoter 2 -1025 T/C Polymorphism in the RUNX2 Gene Is Associated with Femoral Neck BMD in Spanish Postmenopausal Women. Calcified Tissue International, 2007, 81, 327-332.	3.1	32
45	Maroteauxâ€“Lamy syndrome: Functional characterization of pathogenic mutations and polymorphisms in the arylsulfatase B gene. Molecular Genetics and Metabolism, 2008, 94, 305-312.	1.1	31
46	Activity and High-Order Effective Connectivity Alterations in Sanfilippo C Patient-Specific Neuronal Networks. Stem Cell Reports, 2015, 5, 546-557.	4.8	31
47	Mutation analysis of Gaucher disease patients from Argentina: High prevalence of the RecNciI mutation. , 1998, 80, 343-351.		30
48	Identification of the molecular defects in Spanish and Argentinian mucopolysaccharidosis VI (Maroteauxâ€“Lamy syndrome) patients, including 9 novel mutations. Molecular Genetics and Metabolism, 2007, 92, 122-130.	1.1	30
49	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. Blood Cells, Molecules, and Diseases, 2005, 35, 253-258.	1.4	29
50	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
51	A new SNP in a negative regulatory region of the CYP19A1 gene is associated with lumbar spine BMD in postmenopausal women. Bone, 2006, 38, 738-743.	2.9	27
52	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
53	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
54	Two new mild homozygous mutations in Gaucher disease patients: Clinical signs and biochemical analyses. , 1997, 70, 437-443.		26

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55	Expression and characterization of 14 GLB1 mutant alleles found in GM1-gangliosidosis and Morquio B patients. Journal of Lipid Research, 2007, 48, 2275-2282.	4.2	26
56	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
57	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
58	EXTL2 and EXTL3 inhibition with siRNAs as a promising substrate reduction therapy for Sanfilippo C syndrome. Scientific Reports, 2015, 5, 13654.	3.3	24
59	Molecular characterization of five patients with homocystinuria due to severe methylenetetrahydrofolate reductase deficiency. Clinical Genetics, 2010, 78, 441-448.	2.0	23
60	New murine Niemann-Pick type C models bearing a pseudoexon-generating mutation recapitulate the main neurobehavioural and molecular features of the disease. Scientific Reports, 2017, 7, 41931.	3.3	23
61	Sanfilippo Syndrome: Molecular Basis, Disease Models and Therapeutic Approaches. International Journal of Molecular Sciences, 2020, 21, 7819.	4.1	23
62	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
63	A novel mutation in exon 17 of the ?-subunit of rod phosphodiesterase in two RP sisters of a consanguineous family. Human Genetics, 1996, 97, 35-8.	3.8	21
64	Gaucher Disease: The N370S Mutation in Ashkenazi Jewish and Spanish Patients has a Common Origin and Arose Several Thousand Years Ago. American Journal of Human Genetics, 1999, 64, 1233-1238.	6.2	21
65	High prevalence ofCBS p.T191M mutation in homocystinuric patients from Colombia. Human Mutation, 2006, 27, 296-296.	2.5	21
66	Polymorphisms and haplotypes across the osteoprotegerin gene associated with bone mineral density and osteoporotic fractures. Osteoporosis International, 2010, 21, 287-296.	3.1	21
67	Molecular analysis of Sanfilippo syndrome type C in Spain: seven novel HGSNAT mutations and characterization of the mutant alleles. Clinical Genetics, 2011, 80, 367-374.	2.0	21
68	CYP11A1 expression in bone is associated with aromatase inhibitor-related bone loss. Journal of Molecular Endocrinology, 2015, 55, 69-79.	2.5	21
69	Functional Characterization of a GGPPS Variant Identified in Atypical Femoral Fracture Patients and Delineation of the Role of GGPPS in Bone-Relevant Cell Types. Journal of Bone and Mineral Research, 2018, 33, 2091-2098.	2.8	21
70	Identification of positive and negative regulatory elements involved in the retinoic acid/cAMP induction of Fgf-3 transcription in F9 cells. Nucleic Acids Research, 1993, 21, 5351-5359.	14.5	20
71	Evidence against involvement of recoverin in autosomal recessive retinitis pigmentosa in 42 Spanish families. Human Genetics, 1995, 96, 89-94.	3.8	20
72	A New Geneâ€“Pseudogene Fusion Allele Due to a Recombination in Intron 2 of the Glucocerebrosidase Gene Causes Gaucher Disease. Blood Cells, Molecules, and Diseases, 2000, 26, 409-416.	1.4	20

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73	Pro-osteoporotic miR-320a impairs osteoblast function and induces oxidative stress. PLoS ONE, 2018, 13, e0208131.	2.5	20
74	RNAi-mediated inhibition of the glucosylceramide synthase (GCS) gene: A preliminary study towards a therapeutic strategy for Gaucher disease and other glycosphingolipid storage diseases. Blood Cells, Molecules, and Diseases, 2006, 37, 197-203.	1.4	19
75	A folate-rich diet is as effective as folic acid from supplements in decreasing plasma homocysteine concentrations. International Journal of Medical Sciences, 2005, 2, 58-63.	2.5	19
76	Neuronopathic and non-neuronopathic presentation of Gaucher disease in patients with the third most common mutation (D409H) in Spain. Journal of Inherited Metabolic Disease, 1996, 19, 798-800.	3.6	18
77	Mutation and haplotype analyses in 26 Spanish Sanfilippo syndrome type A patients: Possible single origin for 1091delC mutation. American Journal of Medical Genetics Part A, 2001, 100, 223-228.	2.4	18
78	Homozygosity for the double D409H+H255Q allele in type II Gaucher disease. Journal of Inherited Metabolic Disease, 2006, 29, 591-591.	3.6	18
79	Haplotype analysis suggests a single Balkan origin for the Gaucher disease [D409H;H255Q] double mutant allele. Human Mutation, 2008, 29, E58-E67.	2.5	18
80	A Haplotype-Based Analysis of the LRP5 Gene in Relation to Osteoporosis Phenotypes in Spanish Postmenopausal Women. Journal of Bone and Mineral Research, 2008, 23, 1954-1963.	2.8	18
81	Characterisation of two deletions involving NPC1 and flanking genes in Niemannâ€“Pick Type C disease patients. Molecular Genetics and Metabolism, 2012, 107, 716-720.	1.1	18
82	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
83	Recurrence of the D409H mutation in Spanish Gaucher disease patients: description of a new homozygous patient and haplotype analysis.. Journal of Medical Genetics, 1998, 35, 775-777.	3.2	17
84	Identification and functional analyses of CBS alleles in Spanish and Argentinian homocystinuric patients. Human Mutation, 2011, 32, 835-842.	2.5	17
85	COL1A1 haplotypes and hip fracture. Journal of Bone and Mineral Research, 2012, 27, 950-953.	2.8	17
86	Clinical and mutational characterization of three patients with multiple sulfatase deficiency: Report of a new splicing mutation. Molecular Genetics and Metabolism, 2005, 86, 206-211.	1.1	16
87	A broad spectrum of genomic changes in latinamerican patients with EXT1/EXT2-CDG. Scientific Reports, 2014, 4, 6407.	3.3	16
88	Functional assays testing pathogenicity of 14 cystathionine-beta synthase mutations. Human Mutation, 2006, 27, 211-211.	2.5	15
89	Identification of a novel pseudodeficiency allele in the GLB1 gene in a carrier of GM1 gangliosidosis. Clinical Genetics, 2007, 72, 109-111.	2.0	15
90	Frequency of the arylsulphatase A pseudodeficiency allele in the Spanish population. Clinical Genetics, 1993, 44, 320-323.	2.0	15

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91	Antisense Mediated Splicing Modulation For Inherited Metabolic Diseases: Challenges for Delivery. Nucleic Acid Therapeutics, 2014, 24, 48-56.	3.6	15
92	Expression profiling of microRNAs in human bone tissue from postmenopausal women. Human Cell, 2018, 31, 33-41.	2.7	15
93	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
94	Autosomal recessive retinitis pigmentosa in Spain: evaluation of four genes and two loci involved in the disease. Clinical Genetics, 1996, 50, 380-387.	2.0	14
95	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
96	Genetic Analysis of High Bone Mass Cases from the BARCOS Cohort of Spanish Postmenopausal Women. PLoS ONE, 2014, 9, e94607.	2.5	14
97	Common and rare variants of WNT16, DKK1 and SOST and their relationship with bone mineral density. Scientific Reports, 2018, 8, 10951.	3.3	14
98	Wnt Pathway Extracellular Components and Their Essential Roles in Bone Homeostasis. Genes, 2022, 13, 138.	2.4	14
99	Identification of a novel R552Q mutation in exon 13 of the Î²-subunit of rod phosphodiesterase gene in a Spanish family with autosomal recessive retinitis pigmentosa. Human Mutation, 1996, 8, 393-394.	2.5	13
100	Unsuccessful chimeraplast strategy for the correction of a mutation causing Gaucher disease. Blood Cells, Molecules, and Diseases, 2003, 31, 183-186.	1.4	13
101	Molecular characterization of a new deletion of the GBA1 gene due to an inter Alu recombination event. Molecular Genetics and Metabolism, 2011, 102, 226-228.	1.1	13
102	Glucocerebrosidase Enhancers for Selected Gaucher Disease Genotypes by Modification of Î±â€²â€²Câ€²Substituted Iminoâ€²<scp>D</scp>â€²xylitols (DIXs) by Click Chemistry. ChemMedChem, 2014, 9, 1744-1754.	3.2	13
103	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
104	Mutational spectrum by phenotype: panelâ€based NGS testing of patients with clinical suspicion of RASopathy and children with multiple cafÃ©â€auâ€lait macules. Clinical Genetics, 2020, 97, 264-275.	2.0	13
105	A Roadmap to Gene Discoveries and Novel Therapies in Monogenic Low and High Bone Mass Disorders. Frontiers in Endocrinology, 2021, 12, 709711.	3.5	13
106	Two novel (1098insA and Y313H) and one rare (R359Q) mutations detected in exon 8 of the Î²â€glucocerebrosidase gene in Gaucher's disease patients. Human Mutation, 1996, 7, 272-274.	2.5	12
107	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. Blood Cells, Molecules, and Diseases, 2001, 27, 950-959.	1.4	12
108	The Spectrum of Niemann-Pick Type C Disease in Greece. JIMD Reports, 2017, 36, 41-48.	1.5	12

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109	Functional characterization of the C7ORF76 genomic region, a prominent GWAS signal for osteoporosis in 7q21.3. Bone, 2019, 123, 39-47.	2.9	12
110	Bone development and remodeling in metabolic disorders. Journal of Inherited Metabolic Disease, 2020, 43, 133-144.	3.6	12
111	Perspective of the GEMSTONE Consortium on Current and Future Approaches to Functional Validation for Skeletal Genetic Disease Using Cellular, Molecular and Animal-Modeling Techniques. Frontiers in Endocrinology, 2021, 12, 731217.	3.5	12
112	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
113	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. Scientific Reports, 2018, 8, 694.	3.3	11
114	Putative association of a mutant ROM1 allele with retinitis pigmentosa. Human Genetics, 1997, 99, 827-830.	3.8	10
115	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
116	Neuronal and Astrocytic Differentiation from Sanfilippo C Syndrome iPSCs for Disease Modeling and Drug Development. Journal of Clinical Medicine, 2020, 9, 644.	2.4	10
117	Two successful pregnancies in pyridoxine-nonresponsive homocystinuria. Journal of Inherited Metabolic Disease, 2004, 27, 775-777.	3.6	9
118	Niemannâ€“Pick type C disease: a novel <scp>NPC1</scp> mutation segregating in a Greek island. Clinical Genetics, 2014, 85, 543-547.	2.0	9
119	Stereodivergent synthesis of right- and left-handed iminoxylitol heterodimers and monomers. Study of their impact on Î²-glucocerebrosidase activity. Organic and Biomolecular Chemistry, 2017, 15, 3681-3705.	2.8	9
120	Generation of two compound heterozygous HGSNAT-mutated lines from healthy induced pluripotent stem cells using CRISPR/Cas9 to model Sanfilippo C syndrome. Stem Cell Research, 2019, 41, 101616.	0.7	9
121	Transcriptional regulation of the int-2 gene in embryonal carcinoma cells. Cell Growth & Differentiation: the Molecular Biology Journal of the American Association for Cancer Research, 1991, 2, 137-43.	0.8	9
122	Genetic fine localization of the arrestin (S-antigen) gene 4 cM distal from D2S172. Human Genetics, 1994, 94, 193-4.	3.8	8
123	No evidence of linkage to 6p markers in Spanish families with juvenile myoclonic epilepsy. Neuroscience Letters, 2000, 286, 213-217.	2.1	8
124	Analysis of nonsense-mediated mRNA decay in mutant alleles identified in Spanish Gaucher disease patients. Blood Cells, Molecules, and Diseases, 2006, 36, 46-52.	1.4	8
125	Generation of a Human Neuronal Stable Cell Model for Niemann-Pick C Disease by RNA Interference. JIMD Reports, 2011, 4, 29-37.	1.5	8
126	Selective chaperone effect of aminocyclitol derivatives on G202R and other mutant glucocerebrosidases causing Gaucher disease. International Journal of Biochemistry and Cell Biology, 2014, 54, 245-254.	2.8	8

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127	Genetics and Genomics of SOST: Functional Analysis of Variants and Genomic Regulation in Osteoblasts. International Journal of Molecular Sciences, 2021, 22, 489.	4.1	8
128	Expression and functional characterization of human mutant sulfamidase in insect cells. Molecular Genetics and Metabolism, 2004, 83, 246-251.	1.1	7
129	Gaucher disease: Biochemical and molecular findings in 141 patients diagnosed in Greece. Molecular Genetics and Metabolism Reports, 2020, 24, 100614.	1.1	7
130	SR proteins and the nonsense-mediated decay mechanism are involved in human GLB1 gene alternative splicing. BMC Research Notes, 2008, 1, 137.	1.4	6
131	Discrepancy between bone density and bone material strength index in three siblings with Camurati-Engelmann disease. Osteoporosis International, 2017, 28, 3489-3493.	3.1	6
132	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
133	Generation of two NAGLU-mutated homozygous cell lines from healthy induced pluripotent stem cells using CRISPR/Cas9 to model Sanfilippo B syndrome. Stem Cell Research, 2020, 42, 101668.	0.7	6
134	Functional Analyses of Four CYP1A1 Missense Mutations Present in Patients with Atypical Femoral Fractures. International Journal of Molecular Sciences, 2021, 22, 7395.	4.1	6
135	Involvement of Gaucher Disease Mutations in Parkinson Disease. Current Protein and Peptide Science, 2017, 18, 758-764.	1.4	6
136	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	2.8	6
137	Linkage disequilibrium detected between myotonic dystrophy and the anonymous marker D19S63 in the Spanish population. Human Genetics, 1992, 89, 287-91.	3.8	5
138	Bone Mass of a 113-Year-Old Man. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2007, 62, 794-795.	3.6	5
139	The Spectrum of Krabbe Disease in Greece: Biochemical and Molecular Findings. JIMD Reports, 2015, 25, 57-64.	1.5	5
140	Case report of a child bearing a novel deleterious splicing variant in PIGT. Medicine (United States), 2019, 98, e14524.	1.0	5
141	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
142	Functional Assessment of Coding and Regulatory Variants From the <scp>DKK1</scp> Locus. JBMR Plus, 2020, 4, e10423.	2.7	5
143	CRISPR/Cas9-Mediated Allele-Specific Disruption of a Dominant COL6A1 Pathogenic Variant Improves Collagen VI Network in Patient Fibroblasts. International Journal of Molecular Sciences, 2022, 23, 4410.	4.1	5
144	On the Age of the Most Prevalent Gaucher Diseaseâ€"Causing Mutation, N370S. American Journal of Human Genetics, 2000, 66, 2014-2015.	6.2	4

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145	Recent Patents Relating to siRNAs and Therapeutic Strategies for Genetic Diseases. Recent Patents on DNA & Gene Sequences, 2008, 2, 40-43.	0.7	4
146	Perinatal lethal form of Gaucher disease. Clinical and molecular characterization of a Greek case. Blood Cells, Molecules, and Diseases, 2010, 44, 82-83.	1.4	4
147	De Novo PORCN and ZIC2 Mutations in a Highly Consanguineous Family. International Journal of Molecular Sciences, 2021, 22, 1549.	4.1	4
148	Human oocyte meiotic maturation is associated with a specific profile of alternatively spliced transcript isoforms. Molecular Reproduction and Development, 2021, 88, 605-617.	2.0	4
149	Gene rearrangements in the glucocerebrosidase-metaxin region giving rise to disease-causing mutations and polymorphisms. Analysis of 25 RecNciI alleles in Gaucher disease patients. Human Genetics, 2003, 112, 426-429.	3.8	3
150	A Novel Nonsense Mutation of the EXT1 Gene in an Argentinian Patient with Multiple Hereditary Exostoses. Journal of Bone and Joint Surgery - Series A, 2012, 94, e76.	3.0	3
151	Description of extreme longevity in the <scp>B</scp>alearic <scp>I</scp>slands: Exploring a potential Blue Zone in <scp>M</scp>enorca, <scp>S</scp>pain. Geriatrics and Gerontology International, 2014, 14, 620-627.	1.5	3
152	Molecular analysis and clinical findings in the Spanish Gaucher disease population: Putative haplotype of the N370S ancestral chromosome. Human Mutation, 1998, 11, 295-305.	2.5	3
153	Estudio del patrÃ³n de expresiÃ³n de microRNAs en el hueso osteoporÃ³tico. Revista De Osteoporosis Y Metabolismo Mineral, 2016, 8, 5-14.	0.3	3
154	Gene Network of Susceptibility to Atypical Femoral Fractures Related to Bisphosphonate Treatment. Genes, 2022, 13, 146.	2.4	3
155	A response to Kowarz et al.: Gaucher mutation c.680A>G (p.N227S) is associated with myoclonic epilepsy. Human Mutation, 2005, 26, 274-275.	2.5	2
156	Screening of cacna1a and ATP1A2 genes in hemiplegic migraine: clinical, genetic and functional studies. Journal of Headache and Pain, 2013, 14, .	6.0	2
157	Strategies for the Genetic Analysis of Autosomal Recessive Retinitis Pigmentosa in Spanish Families. , 1997, , 263-275.		2
158	Reliable co-segregation analysis for prenatal diagnosis and heterozygote detection in Gaucher disease. , 1998, 18, 207-212.		1
159	Effect of the Tumor Suppressor miR-320a on Viability and Functionality of Human Osteosarcoma Cell Lines Compared to Primary Osteoblasts. Applied Sciences (Switzerland), 2020, 10, 2852.	2.5	1
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