Daniel Grinberg

List of Publications by Year in Descending Order

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	Genetic Analysis in a Familial Case With High Bone Mineral Density Suggests Additive Effects at Two Loci.. *JBMR Plus*, **2022**, 6, e10602	3.9	
159	Wnt Pathway Extracellular Components and Their Essential Roles in Bone Homeostasis.. *Genes*, **2022**, 13,	4.2	3
158	On the association between Chiari malformation type 1, bone mineral density and bone related genes.. *Bone Reports*, **2022**, 16, 101181	2.6	0
157	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases.. *Journal of Molecular Diagnostics*, **2022**, 24, 529-542	5.1	1
156	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	5.7	1
155	Genetics and Genomics of : Functional Analysis of Variants and Genomic Regulation in Osteoblasts. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
154	and Mutations in a Highly Consanguineous Family. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	2
153	Human oocyte meiotic maturation is associated with a specific profile of alternatively spliced transcript isoforms. *Molecular Reproduction and Development*, **2021**, 88, 605-617	2.6	1
152	A Roadmap to Gene Discoveries and Novel Therapies in Monogenic Low and High Bone Mass Disorders. *Frontiers in Endocrinology*, **2021**, 12, 709711	5.7	3
151	Genome Editing Using Cas9-gRNA Ribonucleoprotein in Human Pluripotent Stem Cells for Disease Modeling. *Methods in Molecular Biology*, **2021**, 1	1.4	
150	Functional Assessment of Coding and Regulatory Variants From the Locus. *JBMR Plus*, **2020**, 4, e10423	3.9	3
149	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. *Genetics in Medicine*, **2020**, 22, 1215-1226	8.1	7
148	Neuronal and Astrocytic Differentiation from Sanfilippo C Syndrome iPSCs for Disease Modeling and Drug Development. *Journal of Clinical Medicine*, **2020**, 9,	5.1	6
147	Gaucher disease: Biochemical and molecular findings in 141 patients diagnosed in Greece. *Molecular Genetics and Metabolism Reports*, **2020**, 24, 100614	1.8	2
146	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	4.2	7
145	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	2.6	0
144	Extending the phenotypic spectrum of Bohring-Opitz syndrome: Mild case confirmed by functional studies. *American Journal of Medical Genetics, Part A*, **2020**, 182, 201-204	2.5	4

143	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	1.6	3
142	Sanfilippo Syndrome: Molecular Basis, Disease Models and Therapeutic Approaches. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	12
141	Bone development and remodeling in metabolic disorders. *Journal of Inherited Metabolic Disease*, **2020**, 43, 133-144	5.4	9
140	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	5.3	5
139	Mutational spectrum by phenotype: panel-based NGS testing of patients with clinical suspicion of RASopathy and children with multiple café-au-lait macules. *Clinical Genetics*, **2020**, 97, 264-275	4	9
138	Functional characterization of the C7ORF76 genomic region, a prominent GWAS signal for osteoporosis in 7q21.3. *Bone*, **2019**, 123, 39-47	4.7	8
137	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	1.6	5
136	Case report of a child bearing a novel deleterious splicing variant in PIGT. *Medicine (United States)*, **2019**, 98, e14524	1.8	4
135	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. *Scientific Reports*, **2018**, 8, 694	4.9	5
134	Expression profiling of microRNAs in human bone tissue from postmenopausal women. *Human Cell*, **2018**, 31, 33-41	4.5	10
133	Common and rare variants of WNT16, DKK1 and SOST and their relationship with bone mineral density. *Scientific Reports*, **2018**, 8, 10951	4.9	9
132	Pro-osteoporotic miR-320a impairs osteoblast function and induces oxidative stress. *PLoS ONE*, **2018**, 13, e0208131	3.7	13
131	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	6.3	12
130	The mutation p.Gly646Trpfs*12 found in a Turkish boy with Bohring-Opitz Syndrome. *Clinical Case Reports (discontinued)*, **2018**, 6, 1452-1456	0.7	5
129	The Spectrum of Niemann-Pick Type C Disease in Greece. *JIMD Reports*, **2017**, 36, 41-48	1.9	8
128	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	4.9	24
127	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	4.9	16
126	Stereodivergent synthesis of right- and left-handed iminoxylitol heterodimers and monomers. Study of their impact on β-glucocerebrosidase activity. *Organic and Biomolecular Chemistry*, **2017**, 15, 3681-3705	3.9	6

125	GGPS1 Mutation and Atypical Femoral Fractures with Bisphosphonates. *New England Journal of Medicine*, **2017**, 376, 1794-1795	59.2	32
124	Discrepancy between bone density and bone material strength index in three siblings with Camurati-Engelmann disease. *Osteoporosis International*, **2017**, 28, 3489-3493	5.3	3
123	Involvement of Gaucher Disease Mutations in Parkinson Disease. *Current Protein and Peptide Science*, **2017**, 18, 758-764	2.8	5
122	The Spectrum of Krabbe Disease in Greece: Biochemical and Molecular Findings. *JIMD Reports*, **2016**, 25, 57-64	1.9	5
121	Estudio del patrï de expresiï de microRNAs en el hueso osteoporï tico. *Revista De Osteoporosis Y Metabolismo Mineral*, **2016**, 8, 5-14	1	3
120	Screening of CD96 and ASXL1 in 11 patients with Opitz C or Bohring-Opitz syndromes. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 24-31	2.5	11
119	CYP11A1 expression in bone is associated with aromatase inhibitor-related bone loss. *Journal of Molecular Endocrinology*, **2015**, 55, 69-79	4.5	19
118	Whole-genome sequencing identifies EN1 as a determinant of bone density and fracture. *Nature*, **2015**, 526, 112-7	50.4	308
117	MiRNA profiling of whole trabecular bone: identification of osteoporosis-related changes in MiRNAs in human hip bones. *BMC Medical Genomics*, **2015**, 8, 75	3.7	50
116	EXTL2 and EXTL3 inhibition with siRNAs as a promising substrate reduction therapy for Sanfilippo C syndrome. *Scientific Reports*, **2015**, 5, 13654	4.9	19
115	Evaluation of Aminoglycoside and Non-Aminoglycoside Compounds for Stop-Codon Readthrough Therapy in Four Lysosomal Storage Diseases. *PLoS ONE*, **2015**, 10, e0135873	3.7	22
114	Activity and High-Order Effective Connectivity Alterations in Sanfilippo C Patient-Specific Neuronal Networks. *Stem Cell Reports*, **2015**, 5, 546-57	8	25
113	Antisense mediated splicing modulation for inherited metabolic diseases: challenges for delivery. *Nucleic Acid Therapeutics*, **2014**, 24, 48-56	4.8	11
112	Niemann-Pick type C disease: a novel NPC1 mutation segregating in a Greek island. *Clinical Genetics*, **2014**, 85, 543-7	4	9
111	Selective chaperone effect of aminocyclitol derivatives on G202R and other mutant glucocerebrosidases causing Gaucher disease. *International Journal of Biochemistry and Cell Biology*, **2014**, 54, 245-54	5.6	8
110	Cholesterol regulates Syntaxin 6 trafficking at trans-Golgi network endosomal boundaries. *Cell Reports*, **2014**, 7, 883-97	10.6	75
109	A broad spectrum of genomic changes in latinamerican patients with EXT1/EXT2-CDG. *Scientific Reports*, **2014**, 4, 6407	4.9	9
108	Genetic analysis of high bone mass cases from the BARCOS cohort of Spanish postmenopausal women. *PLoS ONE*, **2014**, 9, e94607	3.7	12

107	Therapeutic strategies based on modified U1 snRNAs and chaperones for Sanfilippo C splicing mutations. *Orphanet Journal of Rare Diseases*, **2014**, 9, 180	4.2	31
106	Glucocerebrosidase enhancers for selected Gaucher disease genotypes by modification of □1-C-substituted imino-D-xylitols (DIXs) by click chemistry. *ChemMedChem*, **2014**, 9, 1744-54	3.7	10
105	Description of extreme longevity in the Balearic Islands: Exploring a potential Blue Zone in Menorca, Spain. *Geriatrics and Gerontology International*, **2014**, 14, 620-7	2.9	2
104	Mucopolysaccharidosis type VI phenotypes-genotypes and antibody response to galsulfase. *Orphanet Journal of Rare Diseases*, **2013**, 8, 51	4.2	33
103	Screening of cacna1a and ATP1A2 genes in hemiplegic migraine: clinical, genetic and functional studies. *Journal of Headache and Pain*, **2013**, 14,	8.8	2
102	Genetic determinants of aromatase inhibitor-related arthralgia: the B-ABLE cohort study. *Breast Cancer Research and Treatment*, **2013**, 140, 385-95	4.4	34
101	Screening of CACNA1A and ATP1A2 genes in hemiplegic migraine: clinical, genetic, and functional studies. *Molecular Genetics & Genomic Medicine*, **2013**, 1, 206-22	2.3	31
100	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, 2550-60	6.3	11
99	Mutations in the EXT1 and EXT2 genes in Spanish patients with multiple osteochondromas. *Scientific Reports*, **2013**, 3, 1346	4.9	34
98	SNPs en el 3□UTR de gen RANK determinan la fractura osteoporó□tica sitio-dependiente. *Revista De Osteoporosis Y Metabolismo Mineral*, **2013**, 5, 85-92	1	
97	Glucocerebrosidase mutations confer a greater risk of dementia during Parkinson□s disease course. *Movement Disorders*, **2012**, 27, 393-9	7	102
96	Characterisation of two deletions involving NPC1 and flanking genes in Niemann-Pick type C disease patients. *Molecular Genetics and Metabolism*, **2012**, 107, 716-20	3.7	17
95	Mapping the genetic and clinical characteristics of Gaucher disease in the Iberian Peninsula. *Orphanet Journal of Rare Diseases*, **2012**, 7, 17	4.2	33
94	COL1A1 haplotypes and hip fracture. *Journal of Bone and Mineral Research*, **2012**, 27, 950-3	6.3	11
93	Generation of a human neuronal stable cell model for niemann-pick C disease by RNA interference. *JIMD Reports*, **2012**, 4, 29-37	1.9	5
92	A novel nonsense mutation of the EXT1 gene in an Argentinian patient with multiple hereditary exostoses: a case report. *Journal of Bone and Joint Surgery - Series A*, **2012**, 94, e76	5.6	2
91	Molecular characterization of a new deletion of the GBA1 gene due to an inter Alu recombination event. *Molecular Genetics and Metabolism*, **2011**, 102, 226-8	3.7	8
90	□Glucocerebrosidase gene mutations in two cohorts of Greek patients with sporadic Parkinson□s disease. *Molecular Genetics and Metabolism*, **2011**, 104, 149-52	3.7	34

89	Molecular analysis of 30 Niemann-Pick type C patients from Spain. *Clinical Genetics*, **2011**, 80, 39-49	4	35
88	Molecular analysis of Sanfilippo syndrome type C in Spain: seven novel HGSNAT mutations and characterization of the mutant alleles. *Clinical Genetics*, **2011**, 80, 367-74	4	18
87	Identification and functional analyses of CBS alleles in Spanish and Argentinian homocystinuric patients. *Human Mutation*, **2011**, 32, 835-42	4.7	17
86	Functional relevance of the BMD-associated polymorphism rs312009: novel involvement of RUNX2 in LRP5 transcriptional regulation. *Journal of Bone and Mineral Research*, **2011**, 26, 1133-44	6.3	13
85	Molecular characterization of five patients with homocystinuria due to severe methylenetetrahydrofolate reductase deficiency. *Clinical Genetics*, **2010**, 78, 441-8	4	19
84	Perinatal lethal form of Gaucher disease. Clinical and molecular characterization of a Greek case. *Blood Cells, Molecules, and Diseases*, **2010**, 44, 82-3	2.1	3
83	Analysis of three functional polymorphisms in relation to osteoporosis phenotypes: replication in a Spanish cohort. *Calcified Tissue International*, **2010**, 87, 14-24	3.9	23
82	Polymorphisms and haplotypes across the osteoprotegerin gene associated with bone mineral density and osteoporotic fractures. *Osteoporosis International*, **2010**, 21, 287-96	5.3	20
81	Present and future of antisense therapy for splicing modulation in inherited metabolic disease. *Journal of Inherited Metabolic Disease*, **2010**, 33, 397-403	5.4	29
80	Effect of IL-1beta, PGE(2), and TGF-beta1 on the expression of OPG and RANKL in normal and osteoporotic primary human osteoblasts. *Journal of Cellular Biochemistry*, **2010**, 110, 304-10	4.7	26
79	Identification and characterization of SMPD1 mutations causing Niemann-Pick types A and B in Spanish patients. *Human Mutation*, **2009**, 30, 1117-22	4.7	39
78	Antisense oligonucleotide treatment for a pseudoexon-generating mutation in the NPC1 gene causing Niemann-Pick type C disease. *Human Mutation*, **2009**, 30, E993-E1001	4.7	41
77	Promising results of the chaperone effect caused by imino sugars and aminocyclitol derivatives on mutant glucocerebrosidases causing Gaucher disease. *Blood Cells, Molecules, and Diseases*, **2009**, 42, 159-66	2.1	35
76	SR proteins and the nonsense-mediated decay mechanism are involved in human GLB1 gene alternative splicing. *BMC Research Notes*, **2008**, 1, 137	2.3	6
75	Maroteaux-Lamy syndrome: functional characterization of pathogenic mutations and polymorphisms in the arylsulfatase B gene. *Molecular Genetics and Metabolism*, **2008**, 94, 305-12	3.7	24
74	Recent patents relating to siRNAs and therapeutic strategies for genetic diseases. *Recent Patents on DNA & Gene Sequences*, **2008**, 2, 40-3		3
73	An evolutionary and structure-based docking model for glucocerebrosidase-saposin C and glucocerebrosidase-substrate interactions - relevance for Gaucher disease. *Proteins: Structure, Function and Bioinformatics*, **2008**, 70, 882-91	4.2	31
72	Haplotype analysis suggests a single Balkan origin for the Gaucher disease [D409H;H255Q] double mutant allele. *Human Mutation*, **2008**, 29, E58-67	4.7	14

71	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, 1954-63	6.3	17
70	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, 1430-8	4.7	73
69	A CBS haplotype and a polymorphism at the MSR gene are associated with cardiovascular disease in a Spanish case-control study. *Clinical Biochemistry*, **2007**, 40, 864-8	3.5	9
68	Identification of 14 novel GLB1 mutations, including five deletions, in 19 patients with GM1 gangliosidosis from South America. *Clinical Genetics*, **2007**, 71, 273-9	4	35
67	Identification of a novel pseudodeficiency allele in the GLB1 gene in a carrier of GM1 gangliosidosis. *Clinical Genetics*, **2007**, 72, 109-11	4	15
66	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, 327-32	3.9	29
65	COL1A1, ESR1, VDR and TGFB1 polymorphisms and haplotypes in relation to BMD in Spanish postmenopausal women. *Osteoporosis International*, **2007**, 18, 235-43	5.3	45
64	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, 677-84	6.5	39
63	Bone mass of a 113-year-old man. *Journals of Gerontology - Series A Biological Sciences and Medical Sciences*, **2007**, 62, 794-5	6.4	5
62	Expression and characterization of 14 GLB1 mutant alleles found in GM1-gangliosidosis and Morquio B patients. *Journal of Lipid Research*, **2007**, 48, 2275-82	6.3	23
61	Identification of the molecular defects in Spanish and Argentinian mucopolysaccharidosis VI (Maroteaux-Lamy syndrome) patients, including 9 novel mutations. *Molecular Genetics and Metabolism*, **2007**, 92, 122-30	3.7	28
60	Functional assays testing pathogenicity of 14 cystathionine-beta synthase mutations. *Human Mutation*, **2006**, 27, 211	4.7	14
59	High prevalence of CBS p.T191M mutation in homocystinuric patients from Colombia. *Human Mutation*, **2006**, 27, 296	4.7	15
58	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	4.7	53
57	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, 3575-83	5.6	48
56	Analysis of nonsense-mediated mRNA decay in mutant alleles identified in Spanish Gaucher disease patients. *Blood Cells, Molecules, and Diseases*, **2006**, 36, 46-52	2.1	8
55	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	2.1	15
54	A new SNP in a negative regulatory region of the CYP19A1 gene is associated with lumbar spine BMD in postmenopausal women. *Bone*, **2006**, 38, 738-43	4.7	24

53	The association between common vitamin D receptor gene variations and osteoporosis: a participant-level meta-analysis. *Annals of Internal Medicine*, **2006**, 145, 255-64	8	185
52	Homozygosity for the double D409H+H255Q allele in type II Gaucher disease. *Journal of Inherited Metabolic Disease*, **2006**, 29, 591	5.4	15
51	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	4.3	20
50	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, 253-8	2.1	21
49	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	2.4	33
48	In vitro functional assay of alleles and haplotypes of two COL1A1-promoter SNPs. *Bone*, **2005**, 36, 902-8	4.7	32
47	Clinical and mutational characterization of three patients with multiple sulfatase deficiency: report of a new splicing mutation. *Molecular Genetics and Metabolism*, **2005**, 86, 206-11	3.7	16
46	A response to Kowarz et al.: Gaucher mutation c.680A>G (p.N227S) is associated with myoclonic epilepsy. *Human Mutation*, **2005**, 26, 274-275	4.7	2
45	A mutation within the saposin D domain in a Gaucher disease patient with normal glucocerebrosidase activity. *Human Genetics*, **2005**, 117, 275-7	6.3	37
44	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, 981-8	6.5	26
43	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	3.7	15
42	Differential genetic effects of ESR1 gene polymorphisms on osteoporosis outcomes. *JAMA - Journal of the American Medical Association*, **2004**, 292, 2105-14	27.4	238
41	Two successful pregnancies in pyridoxine-nonresponsive homocystinuria. *Journal of Inherited Metabolic Disease*, **2004**, 27, 775-7	5.4	7
40	Functional analysis of 13 GBA mutant alleles identified in Gaucher disease patients: Pathogenic changes and "modifier" polymorphisms. *Human Mutation*, **2004**, 23, 567-75	4.7	64
39	Expression and functional characterization of human mutant sulfamidase in insect cells. *Molecular Genetics and Metabolism*, **2004**, 83, 246-51	3.7	6
38	Gene rearrangements in the glucocerebrosidase-metaxin region giving rise to disease-causing mutations and polymorphisms. Analysis of 25 Rec NciI alleles in Gaucher disease patients. *Human Genetics*, **2003**, 112, 426-9	6.3	1
37	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	4.7	21
36	Unsuccessful chimeraplast strategy for the correction of a mutation causing Gaucher disease. *Blood Cells, Molecules, and Diseases*, **2003**, 31, 183-6	2.1	12

35	Regulation of CYP19 gene expression in primary human osteoblasts: effects of vitamin D and other treatments. *European Journal of Endocrinology*, **2003**, 148, 519-26	6.5	45
34	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, 384-93	6.3	77
33	Homocysteine and the MTHFR 677C-->T allele in premature coronary artery disease. Case control and family studies. *European Journal of Clinical Investigation*, **2001**, 31, 24-30	4.6	24
32	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, 223-8		14
31	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, 950-9	2.1	9
30	A new gene-pseudogene fusion allele due to a recombination in intron 2 of the glucocerebrosidase gene causes Gaucher disease. *Blood Cells, Molecules, and Diseases*, **2000**, 26, 409-16	2.1	19
29	No evidence of linkage to 6p markers in spanish families with juvenile myoclonic epilepsy. *Neuroscience Letters*, **2000**, 286, 213-7	3.3	7
28	On the age of the most prevalent Gaucher disease-causing mutation, N370S. *American Journal of Human Genetics*, **2000**, 66, 2014-5	11	4
27	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, 1233-8	11	16
26	Reliable co-segregation analysis for prenatal diagnosis and heterozygote detection in Gaucher disease **1998**, 18, 207-212		1
25	Mutation analysis of Gaucher disease patients from Argentina: high prevalence of the RecNciI mutation. *American Journal of Medical Genetics Part A*, **1998**, 80, 343-51		27
24	Molecular analysis and clinical findings in the Spanish Gaucher disease population: putative haplotype of the N370S ancestral chromosome. *Human Mutation*, **1998**, 11, 295-305	4.7	44
23	Mutation 1091delC is highly prevalent in Spanish Sanfilippo syndrome type A patients. *Human Mutation*, **1998**, 12, 274-9	4.7	28
22	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, 775-7	5.8	13
21	A new autosomal recessive retinitis pigmentosa locus maps on chromosome 2q31-q33. *Journal of Medical Genetics*, **1998**, 35, 141-5	5.8	25
20	Molecular analysis and clinical findings in the Spanish Gaucher disease population: Putative haplotype of the N370S ancestral chromosome **1998**, 11, 295		1
19	A new locus for autosomal recessive retinitis pigmentosa (RP19) maps to 1p13-1p21. *Genomics*, **1997**, 40, 142-6	4.3	46
18	Putative association of a mutant ROM1 allele with retinitis pigmentosa. *Human Genetics*, **1997**, 99, 827-30	4.3	7

17	Genetic fine localization of the beta-glucocerebrosidase (GBA) and prosaposin (PSAP) genes: implications for Gaucher disease. *Human Genetics*, **1997**, 100, 75-9	6.3	29
16	Two new mild homozygous mutations in Gaucher disease patients: clinical signs and biochemical analyses. *American Journal of Medical Genetics Part A*, **1997**, 70, 437-43		22
15	Strategies for the Genetic Analysis of Autosomal Recessive Retinitis Pigmentosa in Spanish Families **1997**, 263-275		2
14	Autosomal recessive retinitis pigmentosa in Spain: evaluation of four genes and two loci involved in the disease. *Clinical Genetics*, **1996**, 50, 380-7	4	8
13	A novel mutation in exon 17 of the beta-subunit of rod phosphodiesterase in two RP sisters of a consanguineous family. *Human Genetics*, **1996**, 97, 35-8	6.3	18
12	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	5.4	17
11	A novel mutation in the BRCA1 gene in a German early-onset breast cancer family. *Human Mutation*, **1996**, 8, 393	4.7	12
10	Two novel (1098insA and Y313H) and one rare (R359Q) mutations detected in exon 8 of the beta-glucocerebrosidase gene in Gaucher© disease patients. *Human Mutation*, **1996**, 7, 272-4	4.7	11
9	Evidence against involvement of recoverin in autosomal recessive retinitis pigmentosa in 42 Spanish families. *Human Genetics*, **1995**, 96, 89-94	6.3	16
8	Unusual expression of Gaucher© disease: cardiovascular calcifications in three sibs homozygous for the D409H mutation. *Journal of Medical Genetics*, **1995**, 32, 740-2	5.8	89
7	Homozygous tandem duplication within the gene encoding the beta-subunit of rod phosphodiesterase as a cause for autosomal recessive retinitis pigmentosa. *Human Mutation*, **1995**, 5, 228-34	4.7	65
6	Gaucher disease in Spanish patients: analysis of eight mutations. *Human Mutation*, **1995**, 5, 303-9	4.7	45
5	Genetic fine localization of the arrestin (S-antigen) gene 4 cM distal from D2S172. *Human Genetics*, **1994**, 94, 193-4	6.3	7
4	Frequency of the arylsulphatase A pseudodeficiency allele in the Spanish population. *Clinical Genetics*, **1993**, 44, 320-3	4	11
3	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, 5351-9	20.1	18
2	Linkage disequilibrium detected between myotonic dystrophy and the anonymous marker D19S63 in the Spanish population. *Human Genetics*, **1992**, 89, 287-91	6.3	5
1	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		8