Wim Van Hul

List of Publications by Year
in descending order

136 papers	13,105 citations	53794 45 h-index	22832 112 g-index
161 all docs	161 docs citations	161 times ranked	17695 citing authors

#	Article	IF	Citations
1	Camurati-Engelmann Disease Complicated by Hypopituitarism: Management Challenges and Literature Review of Outcomes With Bisphosphonates. AACE Clinical Case Reports, 2022, 8, 58-64.	1.1	3
2	Broadening the spectrum of loss-of-function variants in NPR-C-related extreme tall stature. Journal of the Endocrine Society, 2022, 6, bvac019.	0.2	2
3	Editorial: Innovative Models in Bone Biology: What can be Learned From Rare Bone Diseases?. Frontiers in Endocrinology, 2022, 13, 892799.	3.5	1
4	Identification of Compound Heterozygous Variants in LRP4 Demonstrates That a Pathogenic Variant outside the Third Î²-Propeller Domain Can Cause Sclerosteosis. Genes, 2022, 13, 80.	2.4	3
5	A Panel-Based Sequencing Analysis of Patients with Pagetâ€™s Disease of Bone Suggests Enrichment of Rare Genetic Variation in regulators of NF-Î°B Signaling and Supports the Importance of the 7q33 Locus. Calcified Tissue International, 2021, 109, 656-665.	3.1	1
6	A targeted multi-omics approach reveals paraoxonase-1 as a determinant of obesity-associated fatty liver disease. Clinical Epigenetics, 2021, 13, 158.	4.1	9
7	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
8	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
9	WNT16 Requires GÎ± Subunits as Intracellular Partners for Both Its Canonical and Non-Canonical WNT Signalling Activity in Osteoblasts. Calcified Tissue International, 2020, 106, 294-302.	3.1	9
10	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	6.2	33
11	DNA Methylation Profiling and Genomic Analysis in 20 Children with Short Stature Who Were Born Small for Gestational Age. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e4730-e4741.	3.6	12
12	Delineation of a new fibrillino-2-pathy with evidence for a role of FBN2 in the pathogenesis of carpal tunnel syndrome. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-107085.	3.2	4
13	Functional Assessment of Coding and Regulatory Variants From the <scp>DKK1</scp> Locus. JBMR Plus, 2020, 4, e10423.	2.7	5
14	Copy number variant analysis and expression profiling of the olfactory receptor-rich 11q11 region in obesity predisposition. Molecular Genetics and Metabolism Reports, 2020, 25, 100656.	1.1	5
15	Insights into the multifactorial causation of obesity by integrated genetic and epigenetic analysis. Obesity Reviews, 2020, 21, e13019.	6.5	24
16	Spondylo-epi-metaphyseal dysplasia due to a homozygous missense mutation in the gene encoding Matrilin-3 (T120M). Bone Reports, 2020, 12, 100245.	0.4	2
17	WNT Signaling and Bone: Lessons From Skeletal Dysplasias and Disorders. Frontiers in Endocrinology, 2020, 11, 165.	3.5	61
18	A multi-omics approach expands the mutational spectrum of MAP2K1-related melorheostosis. Bone, 2020, 137, 115406.	2.9	6

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19	Unmet therapeutic, educational and scientific needs in parathyroid disorders: Consensus Statement from the first European Society of Endocrinology Workshop (PARAT). European Journal of Endocrinology, 2019, 181, P1-P19.	3.7	61
20	Familial Paget's disease of bone: Long-term follow-up of unaffected relatives with and without Sequestosome 1 mutations. Bone, 2019, 128, 115044.	2.9	4
21	Genetic Variation in RIN3 in the Belgian Population Supports Its Involvement in the Pathogenesis of Pagetâ€™s Disease of Bone and Modifies the Age of Onset. Calcified Tissue International, 2019, 104, 613-621.	3.1	5
22	Camuratiâ€“Engelmann Disease. Calcified Tissue International, 2019, 104, 554-560.	3.1	25
23	Clinical, molecular genetics and therapeutic aspects of syndromic obesity. Clinical Genetics, 2019, 95, 23-40.	2.0	36
24	Human Genetics of Sclerosing Bone Disorders. Current Osteoporosis Reports, 2018, 16, 256-268.	3.6	13
25	DNA sequencing and copy number variation analysis of MCHR2 in a cohort of Prader Willi like (PWL) patients. Obesity Research and Clinical Practice, 2018, 12, 158-166.	1.8	2
26	Evaluation of a Role for NPY and NPY2R in the Pathogenesis of Obesity by Mutation and Copy Number Variation Analysis in Obese Children and Adolescents. Annals of Human Genetics, 2018, 82, 1-10.	0.8	10
27	Conditional mouse models support the role of SLC39A14 (ZIP14) in Hyperostosis Cranialis Interna and in bone homeostasis. PLoS Genetics, 2018, 14, e1007321.	3.5	13
28	Sclerosing Bone Disorders. , 2018, , 507-521.		1
29	Sclerosing bone dysplasias. Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 707-723.	4.7	15
30	Bi-allelic Loss-of-Function Mutations in the NPR-C Receptor Result in Enhanced Growth and Connective Tissue Abnormalities. American Journal of Human Genetics, 2018, 103, 288-295.	6.2	25
31	Genetic Screening of WNT4 and WNT5B in Two Populations with Deviating Bone Mineral Densities. Calcified Tissue International, 2017, 100, 244-249.	3.1	6
32	The Lrp4R1170Q Homozygous Knock-In Mouse Recapitulates the Bone Phenotype of Sclerosteosis in Humans. Journal of Bone and Mineral Research, 2017, 32, 1739-1749.	2.8	27
33	MECHANISMS IN ENDOCRINOLOGY: Genetics of human bone formation. European Journal of Endocrinology, 2017, 177, R69-R83.	3.7	29
34	Fibrogenesis Imperfecta Ossium and Response to Human Growth Hormone: A Potential Therapy. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1750-1756.	3.6	7
35	A Novel Domain-Specific Mutation in a Sclerosteosis Patient Suggests a Role of LRP4 as an Anchor for Sclerostin in Human Bone. Journal of Bone and Mineral Research, 2016, 31, 874-881.	2.8	65
36	CNV analysis and mutation screening indicate an important role for the NPY4R gene in human obesity. Obesity, 2016, 24, 970-976.	3.0	27

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37	Germline mosaicism in osteopathia striata with cranial sclerosis â€“ recurrence in siblings. Clinical Dysmorphology, 2016, 25, 45-49.	0.3	7
38	Identification of biallelicLRRK1mutations in osteosclerotic metaphyseal dysplasia and evidence for locus heterogeneity. Journal of Medical Genetics, 2016, 53, 568-574.	3.2	43
39	Nucleotide variation of sFRP5 gene is not associated with obesity in children and adolescents. Molecular Biology Reports, 2016, 43, 1041-1047.	2.3	2
40	Cover Image, Volume 170A, Number 6, June 2016. , 2016, 170, i-i.		0
41	Further delineation of facioaudiosymphalangism syndrome: Description of a family with a novel NOG mutation and without hearing loss. American Journal of Medical Genetics, Part A, 2016, 170, 1479-1484.	1.2	8
42	De genetische aspecten van obesitas. Bijblijven (Amsterdam, Netherlands), 2016, 32, 25-32.	0.0	2
43	Screening for rare variants in the PNPLA3 gene in obese liver biopsy patients. Clinics and Research in Hepatology and Gastroenterology, 2016, 40, 715-721.	1.5	3
44	Copy number variation (CNV) analysis and mutation analysis of the 6q14.1â€“6q16.3 genes SIM1 and MRAP2 in Prader Willi like patients. Molecular Genetics and Metabolism, 2016, 117, 383-388.	1.1	25
45	Investigation of common and rare genetic variation in the BAMBI genomic region in light of human obesity. Endocrine, 2016, 52, 277-286.	2.3	4
46	Genetic control of bone mass. Molecular and Cellular Endocrinology, 2016, 432, 3-13.	3.2	59
47	Association study of PNPLA2 gene with histological parameters of NAFLD in an obese population. Clinics and Research in Hepatology and Gastroenterology, 2016, 40, 333-339.	1.5	7
48	Eight mutations including 5 novel ones in the COL1A1 gene in Czech patients with osteogenesis imperfecta. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2016, 160, 442-447.	0.6	9
49	Buried in the Middle but Guilty: Intronic Mutations in the TCIRG1 Gene Cause Human Autosomal Recessive Osteopetrosis. Journal of Bone and Mineral Research, 2015, 30, 1814-1821.	2.8	39
50	Prenatal diagnosis of osteopathia striata with cranial sclerosis. Prenatal Diagnosis, 2015, 35, 302-304.	2.3	7
51	Mutations in the latent TGF-beta binding protein 3 (LTBP3) gene cause brachyolmia with amelogenesis imperfecta. Human Molecular Genetics, 2015, 24, 3038-3049.	2.9	40
52	PPARÎ± gene expression correlates with severity and histological treatment response in patients with non-alcoholic steatohepatitis. Journal of Hepatology, 2015, 63, 164-173.	3.7	270
53	A look behind the scenes: the risk and pathogenesis of primary osteoporosis. Nature Reviews Rheumatology, 2015, 11, 462-474.	8.0	204
54	Genetic and structural variation in the SH2B1 gene in the Belgian population. Molecular Genetics and Metabolism, 2015, 115, 193-198.	1.1	8

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55	PLEKHM1 Regulates Salmonella-Containing Vacuole Biogenesis and Infection. Cell Host and Microbe, 2015, 17, 58-71.	11.0	89
56	Variation in the Kozak sequence of WNT16 results in an increased translation and is associated with osteoporosis related parameters. Bone, 2014, 59, 57-65.	2.9	39
57	Wnt Signaling and the Control of Human Stem Cell Fate. Stem Cell Reviews and Reports, 2014, 10, 207-229.	5.6	155
58	Camuratiâ€“Engelmann Disease (Progressive Diaphyseal Dysplasia): Reports of an Indian Kindred. Calcified Tissue International, 2014, 94, 240-247.	3.1	10
59	Sclerosing Bone Dysplasias: Leads Toward Novel Osteoporosis Treatments. Current Osteoporosis Reports, 2014, 12, 243-251.	3.6	12
60	No important role for genetic variation in the Chibby gene in monogenic and complex obesity. Molecular Biology Reports, 2013, 40, 4491-4498.	2.3	3
61	Resistin polymorphisms show associations with obesity, but not with bone parameters in men: results from the Odense Androgen Study. Molecular Biology Reports, 2013, 40, 2467-2472.	2.3	10
62	Prevalence of rare MC3R variants in obese cases and lean controls. Endocrine, 2013, 44, 386-390.	2.3	12
63	Genetic association study of WNT10B polymorphisms with BMD and adiposity parameters in Danish and Belgian males. Endocrine, 2013, 44, 247-254.	2.3	11
64	Genetics in Endocrinology: Autosomal dominant osteopetrosis revisited: lessons from recent studies. European Journal of Endocrinology, 2013, 169, R39-R57.	3.7	65
65	A common LRP4 haplotype is associated with bone mineral density and hip geometry in menâ€”Data from the Odense Androgen Study (OAS). Bone, 2013, 53, 414-420.	2.9	17
66	No mutations in the serotonin related TPH1 and HTR1B genes in patients with monogenic sclerosing bone disorders. Bone, 2013, 55, 52-56.	2.9	7
67	Mutation analysis of WNT10B in obese children, adolescents and adults. Endocrine, 2013, 44, 107-113.	2.3	5
68	The role of extracellular modulators of canonical Wnt signaling in bone metabolism and diseases. Seminars in Arthritis and Rheumatism, 2013, 43, 220-240.	3.4	62
69	Mutations in sFRP1 or sFRP4 are not a common cause of craniotubular hyperostosis. Bone, 2013, 52, 292-295.	2.9	4
70	Novel SOST gene mutation in a sclerosteosis patient and her parents. Bone, 2013, 52, 707-710.	2.9	21
71	Negative mutation screening of the NOG, BMPR1B, GDF5, and FGF9 genes indicates further genetic heterogeneity of the facioaudiosymphalangism syndrome. Clinical Dysmorphology, 2013, 22, 1-6.	0.3	2
72	Sclerosing Bone Disorders. , 2013, , 361-374.		1

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73	Loss-of-function mutations in SIM1 contribute to obesity and Prader-Williâ€“like features. Journal of Clinical Investigation, 2013, 123, 3037-3041.	8.2	105
74	Sclerosing bone disorders: a lot of knowns but still some unknowns. BoneKEy Reports, 2012, 1, 97.	2.7	2
75	Heterozygous Mutations Causing Partial Prohormone Convertase 1 Deficiency Contribute to Human Obesity. Diabetes, 2012, 61, 383-390.	0.6	94
76	Identification of mutations in the NUCB2/nesfatin gene in children with severe obesity. Molecular Genetics and Metabolism, 2012, 107, 729-734.	1.1	19
77	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
78	Association study of common variants in the sFRP1 gene region and parameters of bone strength and body composition in two independent healthy Caucasian male cohorts. Molecular Genetics and Metabolism, 2012, 105, 508-515.	1.1	5
79	Single nucleotide polymorphisms in sFRP4 are associated with bone and body composition related parameters in Danish but not in Belgian men. Molecular Genetics and Metabolism, 2012, 106, 366-374.	1.1	9
80	Assessment of gene-by-sex interaction effect on bone mineral density. Journal of Bone and Mineral Research, 2012, 27, 2051-2064.	2.8	47
81	Dysfunction of lipid sensor GPR120 leads to obesity in both mouse and human. Nature, 2012, 483, 350-354.	27.8	572
82	Monogenic and Complex Forms of Obesity: Insights from Genetics Reveal the Leptin-Melanocortin Signaling Pathway as a Common Player. Critical Reviews in Eukaryotic Gene Expression, 2012, 22, 325-343.	0.9	18
83	Sclerosing Bone Dysplasias. , 2012, , 541-556.		0
84	Paget's Disease of Bone: Evidence for Complex Pathogenetic Interactions. Seminars in Arthritis and Rheumatism, 2012, 41, 619-641.	3.4	51
85	Association Study of Polymorphisms in the SOST Gene Region and Parameters of Bone Strength and Body Composition in Both Young and Elderly Men: Data from the Odense Androgen Study. Calcified Tissue International, 2012, 90, 30-39.	3.1	18
86	Replication of the SH2B1 rs7498665 Association with Obesity in a Belgian Study Population. Obesity Facts, 2011, 4, 473-477.	3.4	12
87	Identification of the first deletion in the LRP5 gene in a patient with Autosomal Dominant Osteopetrosis type I. Bone, 2011, 49, 568-571.	2.9	27
88	Association between polymorphisms of the Nesfatin gene, NUCB2, and obesity in men. Molecular Genetics and Metabolism, 2011, 103, 282-286.	1.1	40
89	Indications for a genetic association of a VCP polymorphism with the pathogenesis of sporadic Paget's disease of bone, but not for TNFSF11 (RANKL) and IL-6 polymorphisms. Molecular Genetics and Metabolism, 2011, 103, 287-292.	1.1	26
90	Two novel WTX mutations underscore the unpredictability of male survival in osteopathia striata with cranial sclerosis. Clinical Genetics, 2011, 80, 383-388.	2.0	25

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91	Genome-wide association identifies three new susceptibility loci for Paget's disease of bone. Nature Genetics, 2011, 43, 685-689.	21.4	158
92	Identification of Three Novel Genetic Variants in the Melanocortinâ€3 Receptor of Obese Children. Obesity, 2011, 19, 152-159.	3.0	30
93	Levels of serotonin, sclerostin, bone turnover markers as well as bone density and microarchitecture in patients with high-bone-mass phenotype due to a mutation in Lrp5. Journal of Bone and Mineral Research, 2011, 26, 1721-1728.	2.8	67
94	Identification of genetic modifiers of monogenic (bone) diseases: New tools available, but with limitations. Journal of Bone and Mineral Research, 2011, 26, 918-919.	2.8	5
95	Bone Overgrowth-associated Mutations in the LRP4 Gene Impair Sclerostin Facilitator Function. Journal of Biological Chemistry, 2011, 286, 19489-19500.	3.4	255
96	The neurology of carbonic anhydrase type II deficiency syndrome. Brain, 2011, 134, 3502-3515.	7.6	37
97	Osteopathia striata with cranial sclerosis owing to WTX gene defect. Journal of Bone and Mineral Research, 2010, 25, 82-90.	2.8	64
98	First missense mutation in the SOST gene causing sclerosteosis by loss of sclerostin function. Human Mutation, 2010, 31, E1526-E1543.	2.5	52
99	Genetic variation in the TNFRSF11A gene encoding RANK is associated with susceptibility to Paget's disease of bone. Journal of Bone and Mineral Research, 2010, 25, 2592-2605.	2.8	42
100	The role of genes in the pathogenesis of Paget's disease of bone. IBMS BoneKEy, 2010, 7, 124-133.	0.0	0
101	Identification and Functional Characterization of Novel Mutations in the Melanocortin-4 Receptor. Obesity Facts, 2010, 3, 304-311.	3.4	18
102	The Role of the Leptin-Melanocortin Signalling Pathway in the Control of Food Intake. Critical Reviews in Eukaryotic Gene Expression, 2009, 19, 267-287.	0.9	39
103	TGF-Î²1â€“induced migration of bone mesenchymal stem cells couples bone resorption with formation. Nature Medicine, 2009, 15, 757-765.	30.7	1,001
104	The Binding Between Sclerostin and LRP5 is Altered by DKK1 and by High-Bone Mass LRP5 Mutations. Calcified Tissue International, 2008, 82, 445-453.	3.1	128
105	Osteopetrosis: from Animal Models to Human Conditions. Clinical Reviews in Bone and Mineral Metabolism, 2008, 6, 71-81.	0.8	1
106	Wnt signaling: A win for bone. Archives of Biochemistry and Biophysics, 2008, 473, 112-116.	3.0	110
107	Variants in the FTO gene are associated with common obesity in the Belgian population. Molecular Genetics and Metabolism, 2008, 93, 481-484.	1.1	134
108	Large-Scale Analysis of Association Between <emph type="ital">LRP5</emph> and <emph type="ital">LRP6</emph> Variants and Osteoporosis. JAMA - Journal of the American Medical Association, 2008, 299, 1277.	7.4	246

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109	Lessons from Sclerosing Bone Dysplasias. Hormone Research in Paediatrics, 2007, 68, 37-39.	1.8	2
110	The Genetics of Low-Density Lipoprotein Receptor-Related Protein 5 in Bone: A Story of Extremes. Endocrinology, 2007, 148, 2622-2629.	2.8	100
111	Novel LRP5 Missense Mutation in a Patient With a High Bone Mass Phenotype Results in Decreased DKK1-Mediated Inhibition of Wnt Signaling*. Journal of Bone and Mineral Research, 2007, 22, 708-716.	2.8	82
112	Identification of Sex-Specific Associations Between Polymorphisms of the Osteoprotegerin Gene, TNFRSF11B, and Paget's Disease of Bone. Journal of Bone and Mineral Research, 2007, 22, 1062-1071.	2.8	59
113	Involvement of PLEKHM1 in osteoclastic vesicular transport and osteopetrosis in incisors absent rats and humans. Journal of Clinical Investigation, 2007, 117, 919-930.	8.2	204
114	An Autosomal Dominant High Bone Mass Phenotype in Association With Craniosynostosis in an Extended Family Is Caused by an LRP5 Missense Mutation. Journal of Bone and Mineral Research, 2005, 20, 1254-1260.	2.8	46
115	Transforming Growth Factor-Î²1 to the Bone. Endocrine Reviews, 2005, 26, 743-774.	20.1	622
116	Loss-of-function mutations in LEMD3 result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis. Nature Genetics, 2004, 36, 1213-1218.	21.4	410
117	Six Novel Missense Mutations in the LDL Receptor-Related Protein 5 (LRP5) Gene in Different Conditions with an Increased Bone Density. American Journal of Human Genetics, 2003, 72, 763-771.	6.2	522
118	Recent progress in the molecular genetics of sclerosing bone dysplasias. Fetal and Pediatric Pathology, 2003, 22, 11-22.	0.3	2
119	Transforming Growth Factor-Î²1 Mutations in Camurati-Engelmann Disease Lead to Increased Signaling by Altering either Activation or Secretion of the Mutant Protein. Journal of Biological Chemistry, 2003, 278, 7718-7724.	3.4	102
120	Recent progress in the molecular genetics of sclerosing bone dysplasias. Fetal and Pediatric Pathology, 2003, 22, 11-22.	0.3	4
121	Recent progress in the molecular genetics of sclerosing bone dysplasias. Fetal and Pediatric Pathology, 2003, 22, 11-22.	0.3	1
122	Domain-specific mutations in sequestosome 1 (SQSTM1) cause familial and sporadic Paget's disease. Human Molecular Genetics, 2002, 11, 2735-2739.	2.9	307
123	Extracellular Regulation of BMP Signaling in Vertebrates: A Cocktail of Modulators. Developmental Biology, 2002, 250, 231-250.	2.0	572
124	Localization of the Gene Causing Autosomal Dominant Osteopetrosis Type I to Chromosome 11q12-13. Journal of Bone and Mineral Research, 2002, 17, 1111-1117.	2.8	80
125	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
126	Burning down DEFECT11. American Journal of Medical Genetics Part A, 2001, 100, 331-332.	2.4	10

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127	Molecular basis of multiple exostoses: mutations in the EXT1 and EXT2 genes. Human Mutation, 2000, 15, 220-227.	2.5	189
128	Mutations in TNFRSF11A, affecting the signal peptide of RANK, cause familial expansile osteolysis. Nature Genetics, 2000, 24, 45-48.	21.4	457
129	Mutations in the gene encoding the latency-associated peptide of TGF-Î²1 cause Camurati-Engelmann disease. Nature Genetics, 2000, 26, 273-275.	21.4	205
130	Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. Nature Genetics, 1999, 23, 94-98.	21.4	260
131	Molecular and clinical examination of an Italian DEFECT 11 family. European Journal of Human Genetics, 1999, 7, 579-584.	2.8	27
132	A novel frameshift mutation (1651ins5) in exon 10 of the CFTR gene can be misinterpreted as a ?F508 mutation. Human Mutation, 1999, 14, 271-271.	2.5	0
133	Localization of the Gene for Sclerosteosis to the van Buchem Diseaseâ€“Gene Region on Chromosome 17q12â€“q21. American Journal of Human Genetics, 1999, 64, 1661-1669.	6.2	125
134	Paget's Disease of Bone: Evidence for a Susceptibility Locus on Chromosome 18q and for Genetic Heterogeneity. Journal of Bone and Mineral Research, 1998, 13, 911-917.	2.8	125
135	Van Buchem Disease (Hyperostosis Corticalis Generalisata) Maps to Chromosome 17q12-q21. American Journal of Human Genetics, 1998, 62, 391-399.	6.2	141
136	Screening for genetic and structural variation in the NPY2R gene in obese children and adolescents. Endocrine Abstracts, 0, , .	0.0	0