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	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
2	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
3	TGF-Î²1â€“induced migration of bone mesenchymal stem cells couples bone resorption with formation. Nature Medicine, 2009, 15, 757-765.	30.7	1,001
4	Transforming Growth Factor-Î²1 to the Bone. Endocrine Reviews, 2005, 26, 743-774.	20.1	622
5	Extracellular Regulation of BMP Signaling in Vertebrates: A Cocktail of Modulators. Developmental Biology, 2002, 250, 231-250.	2.0	572
6	Dysfunction of lipid sensor GPR120 leads to obesity in both mouse and human. Nature, 2012, 483, 350-354.	27.8	572
7	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
8	Mutations in TNFRSF11A, affecting the signal peptide of RANK, cause familial expansile osteolysis. Nature Genetics, 2000, 24, 45-48.	21.4	457
9	Loss-of-function mutations in LEMD3 result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis. Nature Genetics, 2004, 36, 1213-1218.	21.4	410
10	Domain-specific mutations in sequestosome 1 (SQSTM1) cause familial and sporadic Paget's disease. Human Molecular Genetics, 2002, 11, 2735-2739.	2.9	307
11	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
12	Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. Nature Genetics, 1999, 23, 94-98.	21.4	260
13	Bone Overgrowth-associated Mutations in the LRP4 Gene Impair Sclerostin Facilitator Function. Journal of Biological Chemistry, 2011, 286, 19489-19500.	3.4	255
14	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
15	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
16	A look behind the scenes: the risk and pathogenesis of primary osteoporosis. Nature Reviews Rheumatology, 2015, 11, 462-474.	8.0	204
17	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
18	Molecular basis of multiple exostoses: mutations in the EXT1 and EXT2 genes. Human Mutation, 2000, 15, 220-227.	2.5	189

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19	Genome-wide association identifies three new susceptibility loci for Paget's disease of bone. Nature Genetics, 2011, 43, 685-689.	21.4	158
20	Wnt Signaling and the Control of Human Stem Cell Fate. Stem Cell Reviews and Reports, 2014, 10, 207-229.	5.6	155
21	Van Buchem Disease (Hyperostosis Corticalis Generalisata) Maps to Chromosome 17q12-q21. American Journal of Human Genetics, 1998, 62, 391-399.	6.2	141
22	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
23	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
24	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
25	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
26	Wnt signaling: A win for bone. Archives of Biochemistry and Biophysics, 2008, 473, 112-116.	3.0	110
27	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
28	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
29	The Genetics of Low-Density Lipoprotein Receptor-Related Protein 5 in Bone: A Story of Extremes. Endocrinology, 2007, 148, 2622-2629.	2.8	100
30	Heterozygous Mutations Causing Partial Prohormone Convertase 1 Deficiency Contribute to Human Obesity. Diabetes, 2012, 61, 383-390.	0.6	94
31	PLEKHM1 Regulates Salmonella-Containing Vacuole Biogenesis and Infection. Cell Host and Microbe, 2015, 17, 58-71.	11.0	89
32	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
33	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
34	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
35	Genetics in Endocrinology: Autosomal dominant osteopetrosis revisited: lessons from recent studies. European Journal of Endocrinology, 2013, 169, R39-R57.	3.7	65
36	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

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37	Osteopathia striata with cranial sclerosis owing to WTX gene defect. Journal of Bone and Mineral Research, 2010, 25, 82-90.	2.8	64
38	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
39	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
40	WNT Signaling and Bone: Lessons From Skeletal Dysplasias and Disorders. Frontiers in Endocrinology, 2020, 11, 165.	3.5	61
41	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
42	Genetic control of bone mass. Molecular and Cellular Endocrinology, 2016, 432, 3-13.	3.2	59
43	First missense mutation in the SOST gene causing sclerosteosis by loss of sclerostin function. Human Mutation, 2010, 31, E1526-E1543.	2.5	52
44	Paget's Disease of Bone: Evidence for Complex Pathogenetic Interactions. Seminars in Arthritis and Rheumatism, 2012, 41, 619-641.	3.4	51
45	Assessment of gene-by-sex interaction effect on bone mineral density. Journal of Bone and Mineral Research, 2012, 27, 2051-2064.	2.8	47
46	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
47	Identification of biallelicLRRK1mutations in osteosclerotic metaphyseal dysplasia and evidence for locus heterogeneity. Journal of Medical Genetics, 2016, 53, 568-574.	3.2	43
48	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
49	Association between polymorphisms of the Nesfatin gene, NUCB2, and obesity in men. Molecular Genetics and Metabolism, 2011, 103, 282-286.	1.1	40
50	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
51	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
52	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
53	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
54	The neurology of carbonic anhydrase type II deficiency syndrome. Brain, 2011, 134, 3502-3515.	7.6	37

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55	Clinical, molecular genetics and therapeutic aspects of syndromic obesity. Clinical Genetics, 2019, 95, 23-40.	2.0	36
56	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
57	Identification of Three Novel Genetic Variants in the Melanocortinâ€3 Receptor of Obese Children. Obesity, 2011, 19, 152-159.	3.0	30
58	MECHANISMS IN ENDOCRINOLOGY: Genetics of human bone formation. European Journal of Endocrinology, 2017, 177, R69-R83.	3.7	29
59	Molecular and clinical examination of an Italian DEFECT 11 family. European Journal of Human Genetics, 1999, 7, 579-584.	2.8	27
60	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
61	CNV analysis and mutation screening indicate an important role for the NPY4R gene in human obesity. Obesity, 2016, 24, 970-976.	3.0	27
62	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
63	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
64	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
65	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
66	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
67	Camuratiâ€“Engelmann Disease. Calcified Tissue International, 2019, 104, 554-560.	3.1	25
68	Insights into the multifactorial causation of obesity by integrated genetic and epigenetic analysis. Obesity Reviews, 2020, 21, e13019.	6.5	24
69	Novel SOST gene mutation in a sclerosteosis patient and her parents. Bone, 2013, 52, 707-710.	2.9	21
70	Identification of mutations in the NUCB2/nesfatin gene in children with severe obesity. Molecular Genetics and Metabolism, 2012, 107, 729-734.	1.1	19
71	Identification and Functional Characterization of Novel Mutations in the Melanocortin-4 Receptor. Obesity Facts, 2010, 3, 304-311.	3.4	18
72	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

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73	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
74	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
75	Sclerosing bone dysplasias. Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 707-723.	4.7	15
76	Human Genetics of Sclerosing Bone Disorders. Current Osteoporosis Reports, 2018, 16, 256-268.	3.6	13
77	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
78	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
79	Replication of the SH2B1 rs7498665 Association with Obesity in a Belgian Study Population. Obesity Facts, 2011, 4, 473-477.	3.4	12
80	Prevalence of rare MC3R variants in obese cases and lean controls. Endocrine, 2013, 44, 386-390.	2.3	12
81	Sclerosing Bone Dysplasias: Leads Toward Novel Osteoporosis Treatments. Current Osteoporosis Reports, 2014, 12, 243-251.	3.6	12
82	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
83	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
84	Genetic association study of WNT10B polymorphisms with BMD and adiposity parameters in Danish and Belgian males. Endocrine, 2013, 44, 247-254.	2.3	11
85	Burning down DEFECT11. American Journal of Medical Genetics Part A, 2001, 100, 331-332.	2.4	10
86	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
87	Camuratiâ€"Engelmann Disease (Progressive Diaphyseal Dysplasia): Reports of an Indian Kindred. Calcified Tissue International, 2014, 94, 240-247.	3.1	10
88	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
89	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
90	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

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91	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
92	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
93	Genetic and structural variation in the SH2B1 gene in the Belgian population. Molecular Genetics and Metabolism, 2015, 115, 193-198.	1.1	8
94	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
95	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
96	Prenatal diagnosis of osteopathia striata with cranial sclerosis. Prenatal Diagnosis, 2015, 35, 302-304.	2.3	7
97	Germline mosaicism in osteopathia striata with cranial sclerosis â€“ recurrence in siblings. Clinical Dysmorphology, 2016, 25, 45-49.	0.3	7
98	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
99	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
100	Genetic Screening of WNT4 and WNT5B in Two Populations with Deviating Bone Mineral Densities. Calcified Tissue International, 2017, 100, 244-249.	3.1	6
101	A multi-omics approach expands the mutational spectrum of MAP2K1-related melorheostosis. Bone, 2020, 137, 115406.	2.9	6
102	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
103	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
104	Mutation analysis of WNT10B in obese children, adolescents and adults. Endocrine, 2013, 44, 107-113.	2.3	5
105	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
106	Functional Assessment of Coding and Regulatory Variants From the <scp>DKK1</scp> Locus. JBMR Plus, 2020, 4, e10423.	2.7	5
107	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
108	Mutations in sFRP1 or sFRP4 are not a common cause of craniotubular hyperostosis. Bone, 2013, 52, 292-295.	2.9	4

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109	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
110	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
111	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
112	Recent progress in the molecular genetics of sclerosing bone dysplasias. Fetal and Pediatric Pathology, 2003, 22, 11-22.	0.3	4
113	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
114	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
115	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
116	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
117	Recent progress in the molecular genetics of sclerosing bone dysplasias. Fetal and Pediatric Pathology, 2003, 22, 11-22.	0.3	2
118	Lessons from Sclerosing Bone Dysplasias. Hormone Research in Paediatrics, 2007, 68, 37-39.	1.8	2
119	Sclerosing bone disorders: a lot of knowns but still some unknowns. BoneKEy Reports, 2012, 1, 97.	2.7	2
120	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
121	Nucleotide variation of sFRP5 gene is not associated with obesity in children and adolescents. Molecular Biology Reports, 2016, 43, 1041-1047.	2.3	2
122	De genetische aspecten van obesitas. Bijblijven (Amsterdam, Netherlands), 2016, 32, 25-32.	0.0	2
123	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
124	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
125	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
126	Osteopetrosis: from Animal Models to Human Conditions. Clinical Reviews in Bone and Mineral Metabolism, 2008, 6, 71-81.	0.8	1

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127	Sclerosing Bone Disorders. , 2013, , 361-374.		1
128	Sclerosing Bone Disorders. , 2018, , 507-521.		1
129	A Panel-Based Sequencing Analysis of Patients with Pagetâ€™s Disease of Bone Suggests Enrichment of Rare Genetic Variation in regulators of NF-Î¹B Signaling and Supports the Importance of the 7q33 Locus. Calcified Tissue International, 2021, 109, 656-665.	3.1	1
130	Recent progress in the molecular genetics of sclerosing bone dysplasias. Fetal and Pediatric Pathology, 2003, 22, 11-22.	0.3	1
131	Editorial: Innovative Models in Bone Biology: What can be Learned From Rare Bone Diseases?. Frontiers in Endocrinology, 2022, 13, 892799.	3.5	1
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	The role of genes in the pathogenesis of Paget's disease of bone. IBMS BoneKEy, 2010, 7, 124-133.	0.0	0
134	Sclerosing Bone Dysplasias. , 2012, , 541-556.		0
135	Cover Image, Volume 170A, Number 6, June 2016. , 2016, 170, i-i.		0
136	Screening for genetic and structural variation in the NPY2R gene in obese children and adolescents. Endocrine Abstracts, 0, , .	0.0	0