Guiomar Perez de Nanclares

List of Publications by Year
in descending order

128 papers	3,300 citations	126907 33 h-index	168389 53 g-index
138 all docs	138 docs citations	138 times ranked	3527 citing authors

#	Article	IF	Citations
1	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	9.6	224
2	Recessive mutations in the INS gene result in neonatal diabetes through reduced insulin biosynthesis. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 3105-3110.	7.1	185
3	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123.	4.1	174
4	Epigenetic Defects ofGNASin Patients with Pseudohypoparathyroidism and Mild Features of Albrightâ€™s Hereditary Osteodystrophy. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 2370-2373.	3.6	157
5	From pseudohypoparathyroidism to inactivating PTH/PTHrP signalling disorder (iPPSD), a novel classification proposed by the EuroPHP network. European Journal of Endocrinology, 2016, 175, P1-P17.	3.7	117
6	GATA4 Mutations Are a Cause of Neonatal and Childhood-Onset Diabetes. Diabetes, 2014, 63, 2888-2894.	0.6	108
7	PRKAR1AandPDE4DMutations Cause Acrodysostosis but Two Distinct Syndromes with or without GPCR-Signaling Hormone Resistance. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E2328-E2338.	3.6	100
8	Genome-Wide Allelic Methylation Analysis Reveals Disease-Specific Susceptibility to Multiple Methylation Defects in Imprinting Syndromes. Human Mutation, 2013, 34, n/a-n/a.	2.5	96
9	New ABCC8 Mutations in Relapsing Neonatal Diabetes and Clinical Features. Diabetes, 2007, 56, 1737-1741.	0.6	83
10	Functional Study of a Novel Single Deletion in theTITF1/NKX2.1Homeobox Gene That Produces Congenital Hypothyroidism and Benign Chorea But Not Pulmonary Distress. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 1832-1841.	3.6	75
11	Mutations in GCK and HNF-1? explain the majority of cases with clinical diagnosis of MODY in Spain. Clinical Endocrinology, 2007, 67, 070615230707001-???.	2.4	70
12	New mechanisms involved in paternal 20q disomy associated with pseudohypoparathyroidism. European Journal of Endocrinology, 2010, 163, 953-962.	3.7	69
13	The Prevalence of GNAS Deficiency-Related Diseases in a Large Cohort of Patients Characterized by the EuroPHP Network. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 3657-3668.	3.6	66
14	Novel mutations in MEN1, CDKN1B and AIP genes in patients with multiple endocrine neoplasia type 1 syndrome in Spain. Clinical Endocrinology, 2012, 76, 719-724.	2.4	63
15	Functional analysis of six Kir6.2 (KCNJ11) mutations causing neonatal diabetes. Pflugers Archiv European Journal of Physiology, 2006, 453, 323-332.	2.8	53
16	Genome-wide DNA methylation analysis of pseudohypoparathyroidism patients with GNAS imprinting defects. Clinical Epigenetics, 2016, 8, 10.	4.1	53
17	GsÎ± activity is reduced in erythrocyte membranes of patients with psedohypoparathyroidism due to epigenetic alterations at the GNAS locus. Journal of Bone and Mineral Research, 2011, 26, 1864-1870.	2.8	52
18	Heterogeneity of vitamin D receptor gene association with celiac disease and type 1 diabetes mellitus. Autoimmunity, 2005, 38, 439-444.	2.6	48

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19	Conserved extended haplotypes discriminate HLA-DR3-homozygous Basque patients with type 1 diabetes mellitus and celiac disease. Genes and Immunity, 2006, 7, 550-554.	4.1	48
20	Simultaneous Hyper- and Hypomethylation at Imprinted Loci in a Subset of Patients withGNASEpimutations Underlies a Complex and Different Mechanism of Multilocus Methylation Defect in Pseudohypoparathyroidism Type 1b. Human Mutation, 2013, 34, 1172-1180.	2.5	43
21	Killer Cell Immunoglobulin-Like Receptor (KIR) Genes in the Basque Population: Association Study of KIR Gene Contents With Type 1 Diabetes Mellitus. Human Immunology, 2006, 67, 118-124.	2.4	42
22	Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients. Hormone Research in Paediatrics, 2020, 93, 182-196.	1.8	42
23	Panhypopituitarism: Genetic Versus Acquired Etiological Factors. Journal of Pediatric Endocrinology and Metabolism, 2007, 20, 27-36.	0.9	41
24	Genetic and Epigenetic Defects at the GNAS Locus Lead to Distinct Patterns of Skeletal Growth but Similar Early-Onset Obesity. Journal of Bone and Mineral Research, 2018, 33, 1480-1488.	2.8	41
25	Endocrine Profile and Phenotype-(Epi)Genotype Correlation in Spanish Patients with Pseudohypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E996-E1006.	3.6	40
26	Coexistence of two different pseudohypoparathyroidism subtypes (Ia and Ib) in the same kindred with independent GsÂ coding mutations and GNAS imprinting defects. Journal of Medical Genetics, 2010, 47, 276-280.	3.2	38
27	IntragenicGNASDeletion Involving Exon A/B in Pseudohypoparathyroidism Type 1A Resulting in an Apparent Loss of Exon A/B Methylation: Potential for Misdiagnosis of Pseudohypoparathyroidism Type 1B. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 765-771.	3.6	38
28	HLA-DRB1 andMICAin Autoimmunity. Annals of the New York Academy of Sciences, 2003, 1005, 314-318.	3.8	37
29	Detection of Hypomethylation Syndrome among Patients with Epigenetic Alterations at theGNASLocus. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1060-E1067.	3.6	37
30	Brachydactyly E: isolated or as a feature of a syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 141.	2.7	37
31	Association Study of 69 Genes in the Ret Pathway Identifies Low-penetrance Loci in Sporadic Medullary Thyroid Carcinoma. Cancer Research, 2007, 67, 9561-9567.	0.9	36
32	Exclusion of the GNAS locus in PHP-Ib patients with broad GNAS methylation changes: Evidence for an autosomal recessive form of PHP-Ib?. Journal of Bone and Mineral Research, 2011, 26, 1854-1863.	2.8	34
33	Multilocus methylation defects in imprinting disorders. Biomolecular Concepts, 2015, 6, 47-57.	2.2	34
34	Constitutional mosaic genome-wide uniparental disomy due to diploidisation: an unusual cancer-predisposing mechanism. Journal of Medical Genetics, 2011, 48, 212-216.	3.2	32
35	The role of ZFP57 and additional KRAB-zinc finger proteins in the maintenance of human imprinted methylation and multi-locus imprinting disturbances. Nucleic Acids Research, 2020, 48, 11394-11407.	14.5	32
36	Clinical and molecular analyses of Beckwithâ€“Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. American Journal of Medical Genetics, Part A, 2016, 170, 2740-2749.	1.2	30

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37	Differences in expression rather than methylation at placenta-specific imprinted loci is associated with intrauterine growth restriction. Clinical Epigenetics, 2019, 11, 35.	4.1	29
38	Report of two novel mutations in PTHLH associated with brachydactyly type E and literature review. American Journal of Medical Genetics, Part A, 2016, 170, 734-742.	1.2	28
39	No Association of CTLA4 Gene With Celiac Disease in the Basque Population. Journal of Pediatric Gastroenterology and Nutrition, 2003, 37, 142-145.	1.8	27
40	Neonatal Diabetes Caused by Mutations in Sulfonylurea Receptor 1: Interplay between Expression and Mg-Nucleotide Gating Defects of ATP-Sensitive Potassium Channels. Journal of Clinical Endocrinology and Metabolism, 2010, 95, E473-E478.	3.6	27
41	Permanent Neonatal Diabetes Caused by Creation of an Ectopic Splice Site within the INS Gene. PLoS ONE, 2012, 7, e29205.	2.5	27
42	European guidance for the molecular diagnosis of pseudohypoparathyroidism not caused by point genetic variants at GNAS: an EQA study. European Journal of Human Genetics, 2015, 23, 438-444.	2.8	27
43	The most recurrent monogenic disorders that overlap with the phenotype of Rett syndrome. European Journal of Paediatric Neurology, 2019, 23, 609-620.	1.6	27
44	Haploinsufficiency at GCK gene is not a frequent event in MODY2 patients. Clinical Endocrinology, 2008, 68, 873-878.	2.4	25
45	Contribution of MIC-A Polymorphism to Type 1 Diabetes Mellitus in Basques. Annals of the New York Academy of Sciences, 2006, 958, 321-324.	3.8	23
46	Novel Microdeletions Affecting the GNAS Locus in Pseudohypoparathyroidism: Characterization of the Underlying Mechanisms. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E681-E687.	3.6	23
47	Transient neonatal diabetes mellitus and hypomethylation at additional imprinted loci: novel ZFP57 mutation and review on the literature. Acta Diabetologica, 2019, 56, 301-307.	2.5	22
48	Genetic analyses of aplastic anemia and idiopathic pulmonary fibrosis patients with short telomeres, possible implication of DNA-repair genes. Orphanet Journal of Rare Diseases, 2019, 14, 82.	2.7	21
49	Two-year follow-up of anti-transglutaminase autoantibodies among celiac children on gluten-free diet: Comparison of IgG and IgA. Autoimmunity, 2007, 40, 117-121.	2.6	20
50	Association of KIR2DL5B gene with celiac disease supports the susceptibility locus on 19q13.4. Genes and Immunity, 2007, 8, 171-176.	4.1	20
51	Clinical utility gene card for: Pseudohypoparathyroidism. European Journal of Human Genetics, 2013, 21, 5-5.	2.8	20
52	Clinical utility gene card for: Transient Neonatal Diabetes Mellitus, 6q24-related. European Journal of Human Genetics, 2014, 22, 1153-1153.	2.8	20
53	Molecular Analysis of Hereditary Hyperferritinemia-Cataract Syndrome in a Large Basque Family. Journal of Pediatric Endocrinology and Metabolism, 2001, 14, 295-300.	0.9	19
54	Clinical, electrophysiological and magnetic resonance findings in a family with hereditary neuropathy with liability to pressure palsies caused by a novel PMP22 mutation. Neuromuscular Disorders, 2014, 24, 56-62.	0.6	19

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55	Mutations causing acrodysostosis-2 facilitate activation of phosphodiesterase 4D3. Human Molecular Genetics, 2017, 26, 3883-3894.	2.9	17
56	5â€²-Insulin Gene VNTR Polymorphism Is Specific for Type 1 Diabetes. Annals of the New York Academy of Sciences, 2003, 1005, 319-323.	3.8	16
57	No Association of TLR2 and TLR4 Polymorphisms with Type I Diabetes Mellitus in the Basque Population. Annals of the New York Academy of Sciences, 2006, 1079, 268-272.	3.8	15
58	The majority of cases of neonatal diabetes in Spain can be explained by known genetic abnormalities. Diabetic Medicine, 2007, 24, 707-713.	2.3	15
59	The first clinical case of a mutation at residue K185 of Kir6.2 (KCNJ11): a major ATPâ€binding residue. Diabetic Medicine, 2010, 27, 225-229.	2.3	15
60	Parathyroid hormone resistance syndromes â€“ Inactivating PTH/PTHrP signaling disorders (iPPSDs). Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 941-954.	4.7	15
61	Impaired proteostasis in rare neurological diseases. Seminars in Cell and Developmental Biology, 2019, 93, 164-177.	5.0	14
62	New mutation type in pseudohypoparathyroidism type Ia. Clinical Endocrinology, 2008, 69, 705-712.	2.4	13
63	Inactivating PTH/PTHrP signaling disorders (iPPSDs): evaluation of the new classification in a multicenter large series of 544 molecularly characterized patients. European Journal of Endocrinology, 2021, 184, 311-320.	3.7	13
64	Multiple endocrine neoplasia type 1 (MEN1): clinical heterogeneity in a large family with a nonsense mutation in the MEN1 gene (Trp471Stop). Clinical Endocrinology, 1999, 50, 309-313.	2.4	12
65	ACTH-dependent precocious pseudopuberty in an infant with DAX1 gene mutation. European Journal of Pediatrics, 2009, 168, 65-69.	2.7	12
66	The p.R56* mutation in PTHLH causes variable brachydactyly type E. American Journal of Medical Genetics, Part A, 2017, 173, 816-819.	1.2	12
67	Heterozygous glucokinase mutations and birth weight in Spanish children. Diabetic Medicine, 2010, 27, 608-610.	2.3	11
68	Familial Hyperinsulinism-Hyperammonemia Syndrome in a Family with Seizures: Case Report. Journal of Pediatric Endocrinology and Metabolism, 2010, 23, 827-30.	0.9	11
69	Blood Î²-Synuclein and Neurofilament Light Chain During the Course of Prion Disease. Neurology, 2022, , 10.1212/WNL.0000000000200002.	1.1	11
70	Molecular Analysis of Frasier Syndrome: Mutation in the WT1 Gene in a Girl with Gonadal Dysgenesis and Nephronophthisis. Journal of Pediatric Endocrinology and Metabolism, 2002, 15, 1047-50.	0.9	10
71	Short CommunicationNo Evidence of Association ofCTLA4Polymorphisms with Addison's Disease. Autoimmunity, 2004, 37, 453-456.	2.6	10
72	Two cases of deletion 2q37 associated with segregation of an unbalanced translocation 2;21: choanal atresia leading to misdiagnosis of CHARGE syndrome. European Journal of Endocrinology, 2009, 160, 711-717.	3.7	9

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73	Novel Variant in PLAG1 in a Familial Case with Silverâ€“Russell Syndrome Suspicion. Genes, 2020, 11, 1461.	2.4	9
74	No Association ofINS-VNTR Genotype and IAA Autoantibodies. Annals of the New York Academy of Sciences, 2004, 1037, 127-130.	3.8	7
75	Maternal Hypomethylation of KvDMR in a Monozygotic Male Twin Pair Discordant for Beckwith-Wiedemann Syndrome. Molecular Syndromology, 2014, 5, 41-46.	0.8	7
76	Familial Progressive Hyperpigmentation, Cutaneous Mastocytosis, and Gastrointestinal Stromal Tumor as Clinical Manifestations of Mutations in the câ€<scp>KIT</scp> Receptor Gene. Pediatric Dermatology, 2017, 34, 84-89.	0.9	7
77	Head and neck manifestations of an undiagnosed McCune-Albright syndrome: clinicopathological description and literature review. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2018, 473, 645-648.	2.8	7
78	Novel genetic variants of KHDC3L and other members of the subcortical maternal complex associated with Beckwithâ€“Wiedemann syndrome or Pseudohypoparathyroidism 1B and multi-locus imprinting disturbances. Clinical Epigenetics, 2022, 14, .	4.1	7
79	No evidence of association of chromosome 2 q with Type I diabetes in the Basque population. Diabetologia, 1999, 42, 119-120.	6.3	6
80	Neonatal Diabetes With End-Stage Nephropathy: Pancreas transplantation decision. Diabetes Care, 2008, 31, 2116-2117.	8.6	6
81	SÃndrome de Marfan causado por mosaicismo somÃ¡tico de una mutaciÃ³n en splicing en FBN1. Revista Espanola De Cardiologia, 2016, 69, 520-521.	1.2	6
82	Progressive osseous heteroplasia caused by a mosaic <scp>GNAS</scp> mutation. Clinical Endocrinology, 2018, 88, 993-995.	2.4	6
83	Clinical and Molecular Description of 16 Families With Heterozygous IHH Variants. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2654-2666.	3.6	6
84	Familial hypocalciuric hypercalcemia: new mutation in the CASR gene converting valine 697 to methionine. European Journal of Pediatrics, 2012, 171, 147-150.	2.7	5
85	Preimplantation genetic testing for a chr14q32 microdeletion in a family with Kagami-Ogata syndrome and Temple syndrome. Journal of Medical Genetics, 2022, 59, 253-261.	3.2	5
86	Excess Iron Storage in Patients with Type 2 Diabetes Unrelated to Primary Hemochromatosis. New England Journal of Medicine, 2000, 343, 891-891.	27.0	4
87	Familial hypercalcemia and hypercalciuria: no mutations in the Ca 2+ -sensing receptor gene. Pediatric Nephrology, 2001, 16, 748-751.	1.7	4
88	Disomy as the Genetic Underlying Mechanisms of Loss of Heterozigosity in SDHD-Paragangliomas. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1012-E1016.	3.6	4
89	Marfan Syndrome Caused by Somatic Mosaicism in an FBN1 Splicing Mutation. Revista Espanola De Cardiologia (English Ed), 2016, 69, 520-521.	0.6	4
90	Analysis of Chromosome 6q in Basque Families with Type 1 Diabetes. Autoimmunity, 2001, 33, 33-36.	2.6	3

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91	Glibenclamide treatment in relapsed transient neonatal diabetes as a result of a KCNJ11 activating mutation (N48D). Diabetic Medicine, 2009, 26, 567-569.	2.3	3
92	Mutations in MAFA and IAPP are not a common cause of monogenic diabetes. Diabetic Medicine, 2009, 26, 746-748.	2.3	3
93	Array-based characterization of an interstitial de-novo deletion of chromosome 4q in a patient with a neuronal migration defect and hypocalcemia plus a literature review. Clinical Dysmorphology, 2012, 21, 172-176.	0.3	3
94	Pseudohypoparathyrodism vs. tricho-rhino-phalangeal syndrome: patient reclassification. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 1089-94.	0.9	3
95	Design and Validation of a Process Based on Cationic Niosomes for Gene Delivery into Novel Urine-Derived Mesenchymal Stem Cells. Pharmaceutics, 2021, 13, 696.	4.5	3
96	Sporadic Creutzfeldtâ€“Jakob disease with extremely long 14â€year survival period. European Journal of Neurology, 2021, 28, 2901-2906.	3.3	3
97	Description of the first Spanish case of Gerstmannâ€“StrÃ¤sslerâ€“Scheinker disease with A117V variant: clinical, histopathological and biochemical characterization. Journal of Neurology, 2022, , .	3.6	3
98	A submicroscopic deletion of 11p13 associated with the WAGR syndrome. Clinical Genetics, 2003, 63, 319-322.	2.0	2
99	GenÃ©tica del seudohipoparatiroidismo: bases para el consejo genÃ©tico. Endocrinologia Y Nutricion: Organo De La Sociedad Espanola De Endocrinologia Y Nutricion, 2008, 55, 476-483.	0.8	2
100	Intratumoral activating GNAS (R201C) mutation in two unrelated patients with virilizing ovarian Leydig cell tumors. Endocrinologia, Diabetes Y NutriciÃ“n, 2017, 64, 335-337.	0.3	2
101	The Use of Methylation-Sensitive Multiplex Ligation-Dependent Probe Amplification for Quantification of Imprinted Methylation. Methods in Molecular Biology, 2018, 1766, 109-121.	0.9	2
102	Hereditary Spastic Paraplegia and Intellectual Disability: Clinicogenetic Lessons From a Family Suggesting a Dual Genetics Diagnosis. Frontiers in Neurology, 2020, 11, 41.	2.4	2
103	The Importance of Networking in Pseudohypoparathyroidism: EuroPHP Network and Patient Support Associations. Pediatric Endocrinology Reviews, 2017, 15, 92-97.	1.2	2
104	Albrightâ€™s hereditary osteodystrophy: an entity to recognize. Rheumatology, 2022, 61, e356-e357.	1.9	2
105	A case of Prader-Willi syndrome associated with mosaicism: Cytogenetic and FISH study.. Genes and Genetic Systems, 1996, 71, 31-36.	0.7	1
106	What to consider when pseudohypoparathyroidism is ruled out: iPPSD and differential diagnosis. BMC Medical Genetics, 2018, 19, 32.	2.1	1
107	Implication in Paediatrics of the First International Consensus Statement for the Diagnosis and management of pseudohypoparathyroidism and related disorders. Anales De PediatrÃa (English) Tj ETQq1 1 0.784314 rgBT /Overlock	0.4	1
108	Congenital cutaneous ossification. Journal of Paediatrics and Child Health, 2022, 58, 1262-1264.	0.8	1

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109	Neoplasia endocrina mÃºltiple: estudio genÃ©tico. Endocrinologia Y Nutricion: Organo De La Sociedad Espanola De Endocrinologia Y Nutricion, 2005, 52, 199-201.	0.8	0
110	Intragenic GNAS Deletion Involving Exon A/B in Pseudohypoparathyroidism Type 1A Resulting in an Apparent Loss of Exon A/B Methylation: Potential for Misdiagnosis of Pseudohypoparathyroidism Type 1B. Endocrine Reviews, 2010, 31, 135-135.	20.1	0
111	Intragenic GNAS Deletion Involving Exon A/B in Pseudohypoparathyroidism Type 1A Resulting in an Apparent Loss of Exon A/B Methylation: Potential for Misdiagnosis of Pseudohypoparathyroidism Type 1B. Molecular Endocrinology, 2010, 24, 276-277.	3.7	0
112	Clinical characterization of a girl with trisomy 20q13.2qter and monosomy 13q33.1qter: Delineating phenotypeâ€“genotype correlations. American Journal of Medical Genetics, Part A, 2010, 152A, 2901-2905.	1.2	0
113	Pseudopseudohypoparathyroidism vs progressive osseous heteroplasia in absence of family history. Medicina ClÃnica (English Edition), 2015, 145, e25-e27.	0.2	0
114	Brachydactyly type C due to a nonsense mutation in the GDF5 gene. Anales De PediatrÃa (English) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.2	0
115	Wind of change in pseudohypoparathyroidism and related disorders: New classification and first international management consensus. EndocrinologÃa Diabetes Y NutriciÃ³n (English Ed), 2018, 65, 425-427.	0.2	0
116	Cri-du-chat syndrome mimics Silver-Russell syndrome depending on the size of the deletion: a case report. BMC Medical Genomics, 2018, 11, 124.	1.5	0
117	Wind of change in pseudohypoparathyroidism and related disorders: New classification and first international management consensus. Endocrinologia, Diabetes Y NutriciÃ“n, 2018, 65, 425-427.	0.3	0
118	Glucose and galactose malabsorption: A new case in Spain. Anales De PediatrÃa (English Edition), 2020, 92, 104-105.	0.2	0
119	Prenatal and foetal autopsy findings in glutaric aciduria type II. Birth Defects Research, 2020, 112, 1738-1749.	1.5	0
120	Neonatal Diabetes Caused by Mutations in Sulfonylurea Receptor 1: Interplay between Expression and Mg-Nucleotide Gating Defects of ATP-Sensitive Potassium Channels. Molecular Endocrinology, 2010, 24, 2070-2070.	3.7	0
121	Neonatal Diabetes Caused by Mutations in Sulfonylurea Receptor 1: Interplay between Expression and Mg-Nucleotide Gating Defects of ATP-Sensitive Potassium Channels. Endocrine Reviews, 2010, 31, 779-779.	20.1	0
122	GNAS (GNAS complex locus). Atlas of Genetics and Cytogenetics in Oncology and Haematology, 2013, , .	0.1	0
123	From pseudohypoparathyroidism to inactivating PTH/PTHrP signaling disorder (iPPSD), a novel classification proposed by the European EuroPHP-network. Endocrine Abstracts, 0, , .	0.0	0
124	The prevalence of GNAS deficiency-related diseases in a large cohort of patients characterized by the EuroPHP network. Endocrine Abstracts, 0, , .	0.0	0
125	Craniofacial fibrous dysplasia and long-term untreated GH excess in McCune-Albright syndrome. Endocrine Abstracts, 0, , .	0.0	0
126	Gernutik lortutako zelula ama mesenkimalak (hUSC) pseudohipoparatiroidismoaren (PHP) terapia geniko ez-biralerako. , 0, , .		0

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127	Adenocarcinoma de endometrio en una familia: variante de significado incierto en MSH6 en presencia de fenocopia, Â¿cÃ³mo resolverlo?. Revista Espanola De Patologia, 2020, , .	0.2	0
128	HETEROZYGOUS GLUCOKINASE MUTATIONS AND BIRTH WEIGHT IN SPANISH CHILDREN.. Diabetic Medicine, 2009, , .	2.3	0