

Guiomar Perez de Nanclares

List of Publications by Year in descending order

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128
papers

3,300
citations

126907

33
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168389

53
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138
all docs

138
docs citations

138
times ranked

3527
citing authors

#	ARTICLE	IF	CITATIONS
1	Preimplantation genetic testing for a chr14q32 microdeletion in a family with Kagami-Ogata syndrome and Temple syndrome. <i>Journal of Medical Genetics</i> , 2022, 59, 253-261.	3.2	5
2	Congenital cutaneous ossification. <i>Journal of Paediatrics and Child Health</i> , 2022, 58, 1262-1264.	0.8	1
3	Blood β 2-Synuclein and Neurofilament Light Chain During the Course of Prion Disease. <i>Neurology</i> , 2022, , 10.1212/WNL.0000000000200002.	1.1	11
4	Description of the first Spanish case of Gerstmann-Strussler-Scheinker disease with A117V variant: clinical, histopathological and biochemical characterization. <i>Journal of Neurology</i> , 2022, , .	3.6	3
5	Albright's hereditary osteodystrophy: an entity to recognize. <i>Rheumatology</i> , 2022, 61, e356-e357.	1.9	2
6	Novel genetic variants of KHDC3L and other members of the subcortical maternal complex associated with Beckwith-Wiedemann syndrome or Pseudohypoparathyroidism 1B and multi-locus imprinting disturbances. <i>Clinical Epigenetics</i> , 2022, 14, .	4.1	7
7	Inactivating PTH/PTHrP signaling disorders (iPPSDs): evaluation of the new classification in a multicenter large series of 544 molecularly characterized patients. <i>European Journal of Endocrinology</i> , 2021, 184, 311-320.	3.7	13
8	Design and Validation of a Process Based on Cationic Niosomes for Gene Delivery into Novel Urine-Derived Mesenchymal Stem Cells. <i>Pharmaceutics</i> , 2021, 13, 696.	4.5	3
9	Sporadic Creutzfeldt-Jakob disease with extremely long 14-year survival period. <i>European Journal of Neurology</i> , 2021, 28, 2901-2906.	3.3	3
10	Glucose and galactose malabsorption: A new case in Spain. <i>Anales De Pediatr�a (English Edition)</i> , 2020, 92, 104-105.	0.2	0
11	The role of ZFP57 and additional KRAB-zinc finger proteins in the maintenance of human imprinted methylation and multi-locus imprinting disturbances. <i>Nucleic Acids Research</i> , 2020, 48, 11394-11407.	14.5	32
12	Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients. <i>Hormone Research in Paediatrics</i> , 2020, 93, 182-196.	1.8	42
13	Prenatal and foetal autopsy findings in glutaric aciduria type II. <i>Birth Defects Research</i> , 2020, 112, 1738-1749.	1.5	0
14	Novel Variant in PLAG1 in a Familial Case with Silver-Russell Syndrome Suspicion. <i>Genes</i> , 2020, 11, 1461.	2.4	9
15	Hereditary Spastic Paraplegia and Intellectual Disability: Clinicogenetic Lessons From a Family Suggesting a Dual Genetics Diagnosis. <i>Frontiers in Neurology</i> , 2020, 11, 41.	2.4	2
16	Clinical and Molecular Description of 16 Families With Heterozygous <i>IHH</i> Variants. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 2654-2666.	3.6	6
17	Adenocarcinoma de endometrio en una familia: variante de significado incierto en MSH6 en presencia de fenocopia, �mo resolverlo?. <i>Revista Espanola De Patologia</i> , 2020, , .	0.2	0
18	Implication in Paediatrics of the First International Consensus Statement for the Diagnosis and management of pseudohypoparathyroidism and related disorders. <i>Anales De Pediatr�a (English)</i> Tj ETQq0 0 0 rgBT0 Overlock10 Tf 50 5		

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19	Genetic analyses of aplastic anemia and idiopathic pulmonary fibrosis patients with short telomeres, possible implication of DNA-repair genes. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 82.	2.7	21
20	The most recurrent monogenic disorders that overlap with the phenotype of Rett syndrome. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 609-620.	1.6	27
21	Differences in expression rather than methylation at placenta-specific imprinted loci is associated with intrauterine growth restriction. <i>Clinical Epigenetics</i> , 2019, 11, 35.	4.1	29
22	Impaired proteostasis in rare neurological diseases. <i>Seminars in Cell and Developmental Biology</i> , 2019, 93, 164-177.	5.0	14
23	Transient neonatal diabetes mellitus and hypomethylation at additional imprinted loci: novel ZFP57 mutation and review on the literature. <i>Acta Diabetologica</i> , 2019, 56, 301-307.	2.5	22
24	Progressive osseous heteroplasia caused by a mosaic <i>GNAS</i> mutation. <i>Clinical Endocrinology</i> , 2018, 88, 993-995.	2.4	6
25	Genetic and Epigenetic Defects at the <i>GNAS</i> Locus Lead to Distinct Patterns of Skeletal Growth but Similar Early-Onset Obesity. <i>Journal of Bone and Mineral Research</i> , 2018, 33, 1480-1488.	2.8	41
26	Brachydactyly type C due to a nonsense mutation in the <i>GDF5</i> gene. <i>Anales De PediatrĀa (English)</i> Tj ETQq0 0 0 rgBT/Overlock 10 Tf 50	0.2	0
27	What to consider when pseudohypoparathyroidism is ruled out: iPPSD and differential diagnosis. <i>BMC Medical Genetics</i> , 2018, 19, 32.	2.1	1
28	The Use of Methylation-Sensitive Multiplex Ligation-Dependent Probe Amplification for Quantification of Imprinted Methylation. <i>Methods in Molecular Biology</i> , 2018, 1766, 109-121.	0.9	2
29	Wind of change in pseudohypoparathyroidism and related disorders: New classification and first international management consensus. <i>EndocrinologĀa Diabetes Y NutriciĀn (English Ed)</i> , 2018, 65, 425-427.	0.2	0
30	Cri-du-chat syndrome mimics Silver-Russell syndrome depending on the size of the deletion: a case report. <i>BMC Medical Genomics</i> , 2018, 11, 124.	1.5	0
31	Wind of change in pseudohypoparathyroidism and related disorders: New classification and first international management consensus. <i>Endocrinologia, Diabetes Y NutriciĀn</i> , 2018, 65, 425-427.	0.3	0
32	Parathyroid hormone resistance syndromes â€“ Inactivating PTH/PTHrP signaling disorders (iPPSDs). <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2018, 32, 941-954.	4.7	15
33	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. <i>Nature Reviews Endocrinology</i> , 2018, 14, 476-500.	9.6	224
34	Head and neck manifestations of an undiagnosed McCune-Albright syndrome: clinicopathological description and literature review. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2018, 473, 645-648.	2.8	7
35	The p.R56* mutation in <i>PTHLH</i> causes variable brachydactyly type E. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 816-819.	1.2	12
36	Familial Progressive Hyperpigmentation, Cutaneous Mastocytosis, and Gastrointestinal Stromal Tumor as Clinical Manifestations of Mutations in the <i>KIT</i> Receptor Gene. <i>Pediatric Dermatology</i> , 2017, 34, 84-89.	0.9	7

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37	Mutations causing acrodysostosis-2 facilitate activation of phosphodiesterase 4D3. <i>Human Molecular Genetics</i> , 2017, 26, 3883-3894.	2.9	17
38	Intratumoral activating GNAS (R201C) mutation in two unrelated patients with virilizing ovarian Leydig cell tumors. <i>Endocrinología, Diabetes Y Nutrición</i> , 2017, 64, 335-337.	0.3	2
39	The Importance of Networking in Pseudohypoparathyroidism: EuroPHP Network and Patient Support Associations. <i>Pediatric Endocrinology Reviews</i> , 2017, 15, 92-97.	1.2	2
40	Marfan Syndrome Caused by Somatic Mosaicism in an FBN1 Splicing Mutation. <i>Revista Española De Cardiología (English Ed)</i> , 2016, 69, 520-521.	0.6	4
41	Genome-wide DNA methylation analysis of pseudohypoparathyroidism patients with GNAS imprinting defects. <i>Clinical Epigenetics</i> , 2016, 8, 10.	4.1	53
42	Clinical and molecular analyses of Beckwith-Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2740-2749.	1.2	30
43	The Prevalence of GNAS Deficiency-Related Diseases in a Large Cohort of Patients Characterized by the EuroPHP Network. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 3657-3668.	3.6	66
44	From pseudohypoparathyroidism to inactivating PTH/PTHrP signalling disorder (iPPSD), a novel classification proposed by the EuroPHP network. <i>European Journal of Endocrinology</i> , 2016, 175, P1-P17.	3.7	117
45	Síndrome de Marfan causado por mosaicismo somático de una mutación en splicing en FBN1. <i>Revista Española De Cardiología</i> , 2016, 69, 520-521.	1.2	6
46	Report of two novel mutations in <i>PTHLH</i> associated with brachydactyly type E and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 734-742.	1.2	28
47	Pseudopseudohypoparathyroidism vs progressive osseous heteroplasia in absence of family history. <i>Medicina Clínica (English Edition)</i> , 2015, 145, e25-e27.	0.2	0
48	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. <i>Clinical Epigenetics</i> , 2015, 7, 123.	4.1	174
49	Novel Microdeletions Affecting the GNAS Locus in Pseudohypoparathyroidism: Characterization of the Underlying Mechanisms. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E681-E687.	3.6	23
50	Multilocus methylation defects in imprinting disorders. <i>Biomolecular Concepts</i> , 2015, 6, 47-57.	2.2	34
51	European guidance for the molecular diagnosis of pseudohypoparathyroidism not caused by point genetic variants at GNAS: an EQA study. <i>European Journal of Human Genetics</i> , 2015, 23, 438-444.	2.8	27
52	Pseudohypoparathyroidism vs. tricho-rhino-phalangeal syndrome: patient reclassification. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014, 27, 1089-94.	0.9	3
53	<i>GATA4</i> Mutations Are a Cause of Neonatal and Childhood-Onset Diabetes. <i>Diabetes</i> , 2014, 63, 2888-2894.	0.6	108
54	Clinical utility gene card for: Transient Neonatal Diabetes Mellitus, 6q24-related. <i>European Journal of Human Genetics</i> , 2014, 22, 1153-1153.	2.8	20

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55	Clinical, electrophysiological and magnetic resonance findings in a family with hereditary neuropathy with liability to pressure palsies caused by a novel PMP22 mutation. <i>Neuromuscular Disorders</i> , 2014, 24, 56-62.	0.6	19
56	Maternal Hypomethylation of KvDMR in a Monozygotic Male Twin Pair Discordant for Beckwith-Wiedemann Syndrome. <i>Molecular Syndromology</i> , 2014, 5, 41-46.	0.8	7
57	Brachydactyly E: isolated or as a feature of a syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 141.	2.7	37
58	Genome-Wide Allelic Methylation Analysis Reveals Disease-Specific Susceptibility to Multiple Methylation Defects in Imprinting Syndromes. <i>Human Mutation</i> , 2013, 34, n/a-n/a.	2.5	96
59	Simultaneous Hyper- and Hypomethylation at Imprinted Loci in a Subset of Patients with <i>GNAS</i> Epimutations Underlies a Complex and Different Mechanism of Multilocus Methylation Defect in Pseudohypoparathyroidism Type 1b. <i>Human Mutation</i> , 2013, 34, 1172-1180.	2.5	43
60	Clinical utility gene card for: Pseudohypoparathyroidism. <i>European Journal of Human Genetics</i> , 2013, 21, 5-5.	2.8	20
61	Endocrine Profile and Phenotype-(Epi)Genotype Correlation in Spanish Patients with Pseudohypoparathyroidism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, E996-E1006.	3.6	40
62	Disomy as the Genetic Underlying Mechanisms of Loss of Heterozygosity in SDHD-Paragangliomas. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, E1012-E1016.	3.6	4
63	GNAS (GNAS complex locus). <i>Atlas of Genetics and Cytogenetics in Oncology and Haematology</i> , 2013, , .	0.1	0
64	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. <i>Clinical Dysmorphology</i> , 2012, 21, 172-176.	0.3	3
65	Detection of Hypomethylation Syndrome among Patients with Epigenetic Alterations at the <i>GNAS</i> Locus. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, E1060-E1067.	3.6	37
66	<i>PRKAR1A</i> and <i>PDE4D</i> Mutations Cause Acrodysostosis but Two Distinct Syndromes with or without GPCR-Signaling Hormone Resistance. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, E2328-E2338.	3.6	100
67	Permanent Neonatal Diabetes Caused by Creation of an Ectopic Splice Site within the INS Gene. <i>PLoS ONE</i> , 2012, 7, e29205.	2.5	27
68	Novel mutations in <i>MEN1</i> , <i>CDKN1B</i> and <i>AIP</i> genes in patients with multiple endocrine neoplasia type 1 syndrome in Spain. <i>Clinical Endocrinology</i> , 2012, 76, 719-724.	2.4	63
69	Familial hypocalciuric hypercalcemia: new mutation in the CASR gene converting valine 697 to methionine. <i>European Journal of Pediatrics</i> , 2012, 171, 147-150.	2.7	5
70	Gs α activity is reduced in erythrocyte membranes of patients with pseudohypoparathyroidism due to epigenetic alterations at the <i>GNAS</i> locus. <i>Journal of Bone and Mineral Research</i> , 2011, 26, 1864-1870.	2.8	52
71	Exclusion of the <i>GNAS</i> locus in PHP-1b patients with broad <i>GNAS</i> methylation changes: Evidence for an autosomal recessive form of PHP-1b?. <i>Journal of Bone and Mineral Research</i> , 2011, 26, 1854-1863.	2.8	34
72	Constitutional mosaic genome-wide uniparental disomy due to diploidisation: an unusual cancer-predisposing mechanism. <i>Journal of Medical Genetics</i> , 2011, 48, 212-216.	3.2	32

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73	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. <i>Endocrine Reviews</i> , 2010, 31, 135-135.	20.1	0
74	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. <i>Molecular Endocrinology</i> , 2010, 24, 276-277.	3.7	0
75	Clinical characterization of a girl with trisomy 20q13.2qter and monosomy 13q33.1qter: Delineating phenotype-genotype correlations. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2901-2905.	1.2	0
76	The first clinical case of a mutation at residue K185 of Kir6.2 (KCNJ11): a major ATP-binding residue. <i>Diabetic Medicine</i> , 2010, 27, 225-229.	2.3	15
77	Heterozygous glucokinase mutations and birth weight in Spanish children. <i>Diabetic Medicine</i> , 2010, 27, 608-610.	2.3	11
78	Recessive mutations in the <i>INS</i> gene result in neonatal diabetes through reduced insulin biosynthesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 3105-3110.	7.1	185
79	New mechanisms involved in paternal 20q disomy associated with pseudohypoparathyroidism. <i>European Journal of Endocrinology</i> , 2010, 163, 953-962.	3.7	69
80	Coexistence of two different pseudohypoparathyroidism subtypes (Ia and Ib) in the same kindred with independent Gs α coding mutations and GNAS imprinting defects. <i>Journal of Medical Genetics</i> , 2010, 47, 276-280.	3.2	38
81	Familial Hyperinsulinism-Hyperammonemia Syndrome in a Family with Seizures: Case Report. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2010, 23, 827-30.	0.9	11
82	Neonatal Diabetes Caused by Mutations in Sulfonylurea Receptor 1: Interplay between Expression and Mg-Nucleotide Gating Defects of ATP-Sensitive Potassium Channels. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, E473-E478.	3.6	27
83	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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, 765-771.	3.6	38
84	Neonatal Diabetes Caused by Mutations in Sulfonylurea Receptor 1: Interplay between Expression and Mg-Nucleotide Gating Defects of ATP-Sensitive Potassium Channels. <i>Molecular Endocrinology</i> , 2010, 24, 2070-2070.	3.7	0
85	Neonatal Diabetes Caused by Mutations in Sulfonylurea Receptor 1: Interplay between Expression and Mg-Nucleotide Gating Defects of ATP-Sensitive Potassium Channels. <i>Endocrine Reviews</i> , 2010, 31, 779-779.	20.1	0
86	Two cases of deletion 2q37 associated with segregation of an unbalanced translocation 2;21: choanal atresia leading to misdiagnosis of CHARGE syndrome. <i>European Journal of Endocrinology</i> , 2009, 160, 711-717.	3.7	9
87	ACTH-dependent precocious pseudopuberty in an infant with DAX1 gene mutation. <i>European Journal of Pediatrics</i> , 2009, 168, 65-69.	2.7	12
88	Glibenclamide treatment in relapsed transient neonatal diabetes as a result of a <i>KCNJ11</i> activating mutation (N48D). <i>Diabetic Medicine</i> , 2009, 26, 567-569.	2.3	3
89	Mutations in <i>MAFA</i> and <i>LAPP</i> are not a common cause of monogenic diabetes. <i>Diabetic Medicine</i> , 2009, 26, 746-748.	2.3	3
90	HETEROZYGOUS GLUCOKINASE MUTATIONS AND BIRTH WEIGHT IN SPANISH CHILDREN.. <i>Diabetic Medicine</i> , 2009, , .	2.3	0

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91	Haploinsufficiency at <i>GCK</i> gene is not a frequent event in MODY2 patients. <i>Clinical Endocrinology</i> , 2008, 68, 873-878.	2.4	25
92	New mutation type in pseudohypoparathyroidism type Ia. <i>Clinical Endocrinology</i> , 2008, 69, 705-712.	2.4	13
93	Genética del pseudohipoparatiroidismo: bases para el consejo genético. <i>Endocrinología Y Nutricion: Organo De La Sociedad Espanola De Endocrinología Y Nutricion</i> , 2008, 55, 476-483.	0.8	2
94	Neonatal Diabetes With End-Stage Nephropathy: Pancreas transplantation decision. <i>Diabetes Care</i> , 2008, 31, 2116-2117.	8.6	6
95	Panhypopituitarism: Genetic Versus Acquired Etiological Factors. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2007, 20, 27-36.	0.9	41
96	New <i>ABCC8</i> Mutations in Relapsing Neonatal Diabetes and Clinical Features. <i>Diabetes</i> , 2007, 56, 1737-1741.	0.6	83
97	Two-year follow-up of anti-transglutaminase autoantibodies among celiac children on gluten-free diet: Comparison of IgG and IgA. <i>Autoimmunity</i> , 2007, 40, 117-121.	2.6	20
98	Epigenetic Defects of <i>GNAS</i> in Patients with Pseudohypoparathyroidism and Mild Features of Albright's Hereditary Osteodystrophy. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2007, 92, 2370-2373.	3.6	157
99	Association Study of 69 Genes in the Ret Pathway Identifies Low-penetrance Loci in Sporadic Medullary Thyroid Carcinoma. <i>Cancer Research</i> , 2007, 67, 9561-9567.	0.9	36
100	Association of <i>KIR2DL5B</i> gene with celiac disease supports the susceptibility locus on 19q13.4. <i>Genes and Immunity</i> , 2007, 8, 171-176.	4.1	20
101	The majority of cases of neonatal diabetes in Spain can be explained by known genetic abnormalities. <i>Diabetic Medicine</i> , 2007, 24, 707-713.	2.3	15
102	Mutations in <i>GCK</i> and <i>HNF-1β</i> explain the majority of cases with clinical diagnosis of MODY in Spain. <i>Clinical Endocrinology</i> , 2007, 67, 070615230707001-???	2.4	70
103	Killer Cell Immunoglobulin-Like Receptor (KIR) Genes in the Basque Population: Association Study of KIR Gene Contents With Type 1 Diabetes Mellitus. <i>Human Immunology</i> , 2006, 67, 118-124.	2.4	42
104	Contribution of <i>MIC-A</i> Polymorphism to Type 1 Diabetes Mellitus in Basques. <i>Annals of the New York Academy of Sciences</i> , 2006, 958, 321-324.	3.8	23
105	Conserved extended haplotypes discriminate HLA-DR3-homozygous Basque patients with type 1 diabetes mellitus and celiac disease. <i>Genes and Immunity</i> , 2006, 7, 550-554.	4.1	48
106	No Association of <i>TLR2</i> and <i>TLR4</i> Polymorphisms with Type I Diabetes Mellitus in the Basque Population. <i>Annals of the New York Academy of Sciences</i> , 2006, 1079, 268-272.	3.8	15
107	Functional analysis of six <i>Kir6.2</i> (<i>KCNJ11</i>) mutations causing neonatal diabetes. <i>Pflugers Archiv European Journal of Physiology</i> , 2006, 453, 323-332.	2.8	53
108	Functional Study of a Novel Single Deletion in the <i>TITF1/NKX2.1</i> Homeobox Gene That Produces Congenital Hypothyroidism and Benign Chorea But Not Pulmonary Distress. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006, 91, 1832-1841.	3.6	75

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109	Heterogeneity of vitamin D receptor gene association with celiac disease and type 1 diabetes mellitus. <i>Autoimmunity</i> , 2005, 38, 439-444.	2.6	48
110	Neoplasia endocrina múltiple: estudio genético. <i>Endocrinología Y Nutricion: Organo De La Sociedad Espanola De Endocrinología Y Nutricion</i> , 2005, 52, 199-201.	0.8	0
111	No Association of INS-VNTR Genotype and IAA Autoantibodies. <i>Annals of the New York Academy of Sciences</i> , 2004, 1037, 127-130.	3.8	7
112	Short Communication No Evidence of Association of CTLA4 Polymorphisms with Addison's Disease. <i>Autoimmunity</i> , 2004, 37, 453-456.	2.6	10
113	HLA-DRB1 and MICAI in Autoimmunity. <i>Annals of the New York Academy of Sciences</i> , 2003, 1005, 314-318.	3.8	37
114	5' Insulin Gene VNTR Polymorphism Is Specific for Type 1 Diabetes. <i>Annals of the New York Academy of Sciences</i> , 2003, 1005, 319-323.	3.8	16
115	A submicroscopic deletion of 11p13 associated with the WAGR syndrome. <i>Clinical Genetics</i> , 2003, 63, 319-322.	2.0	2
116	No Association of CTLA4 Gene With Celiac Disease in the Basque Population. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2003, 37, 142-145.	1.8	27
117	Molecular Analysis of Frasier Syndrome: Mutation in the WT1 Gene in a Girl with Gonadal Dysgenesis and Nephronophthisis. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2002, 15, 1047-50.	0.9	10
118	Familial hypercalcemia and hypercalciuria: no mutations in the Ca ²⁺ -sensing receptor gene. <i>Pediatric Nephrology</i> , 2001, 16, 748-751.	1.7	4
119	Analysis of Chromosome 6q in Basque Families with Type 1 Diabetes. <i>Autoimmunity</i> , 2001, 33, 33-36.	2.6	3
120	Molecular Analysis of Hereditary Hyperferritinemia-Cataract Syndrome in a Large Basque Family. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2001, 14, 295-300.	0.9	19
121	Excess Iron Storage in Patients with Type 2 Diabetes Unrelated to Primary Hemochromatosis. <i>New England Journal of Medicine</i> , 2000, 343, 891-891.	27.0	4
122	Multiple endocrine neoplasia type 1 (MEN1): clinical heterogeneity in a large family with a nonsense mutation in the MEN1 gene (Trp471Stop). <i>Clinical Endocrinology</i> , 1999, 50, 309-313.	2.4	12
123	No evidence of association of chromosome 2 q with Type I diabetes in the Basque population. <i>Diabetologia</i> , 1999, 42, 119-120.	6.3	6
124	A case of Prader-Willi syndrome associated with mosaicism: Cytogenetic and FISH study.. <i>Genes and Genetic Systems</i> , 1996, 71, 31-36.	0.7	1
125	From pseudohypoparathyroidism to inactivating PTH/PTHrP signaling disorder (iPPSD), a novel classification proposed by the European EuroPHP-network. <i>Endocrine Abstracts</i> , 0, , .	0.0	0
126	The prevalence of GNAS deficiency-related diseases in a large cohort of patients characterized by the EuroPHP network. <i>Endocrine Abstracts</i> , 0, , .	0.0	0

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127	Craniofacial fibrous dysplasia and long-term untreated GH excess in McCune-Albright syndrome. Endocrine Abstracts, 0, , .	0.0	0
128	Gernutik lortutako zelula ama mesenkimalak (hUSC) pseudohipoparatiroidismoaren (PHP) terapia geniko ez-biralerako. , 0, , .		0