Berenice Bilharinho Mendonca

List of Publications by Year
in descending order

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485 papers	19,444 citations	11639 70 h-index	20943 115 g-index
537 all docs	537 docs citations	537 times ranked	12450 citing authors

#	Article	IF	Citations
1	Male pseudohermaphroditism caused by mutations of testicular 17Î²â€“hydroxysteroid dehydrogenase 3. Nature Genetics, 1994, 7, 34-39.	9.4	547
2	AGPR54-Activating Mutation in a Patient with Central Precocious Puberty. New England Journal of Medicine, 2008, 358, 709-715.	13.9	507
3	Comprehensive Pan-Genomic Characterization of Adrenocortical Carcinoma. Cancer Cell, 2016, 29, 723-736.	7.7	482
4	Mutant P450 oxidoreductase causes disordered steroidogenesis with and without Antley-Bixler syndrome. Nature Genetics, 2004, 36, 228-230.	9.4	462
5	Central Precocious Puberty Caused by Mutations in the Imprinted Gene MKRN3. New England Journal of Medicine, 2013, 368, 2467-2475.	13.9	450
6	Molecular genetics of steroid 5 alpha-reductase 2 deficiency.. Journal of Clinical Investigation, 1992, 90, 799-809.	3.9	362
7	Testicular and Ovarian Resistance to Luteinizing Hormone Caused by Inactivating Mutations of the Luteinizing Hormoneâ€“Receptor Gene. New England Journal of Medicine, 1996, 334, 507-512.	13.9	350
8	The genetic and functional basis of isolated 17,20â€“lyase deficiency. Nature Genetics, 1997, 17, 201-205.	9.4	306
9	Mutations of the KISS1 Gene in Disorders of Puberty. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 2276-2280.	1.8	301
10	TAC3/TACR3 Mutations Reveal Preferential Activation of Gonadotropin-Releasing Hormone Release by Neurokinin B in Neonatal Life Followed by Reversal in Adulthood. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 2857-2867.	1.8	250
11	Ectopic Adrenocorticotropic Hormone Syndrome. Endocrine Reviews, 1994, 15, 752-787.	8.9	217
12	An Inherited Mutation Outside the Highly Conserved DNA-Binding Domain of the p53 Tumor Suppressor Protein in Children and Adults with Sporadic Adrenocortical Tumors. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 4970-4973.	1.8	183
13	Cushingâ€™s Syndrome Secondary to Adrenocorticotropin-Independent Macronodular Adrenocortical Hyperplasia due to Activating Mutations ofGNAS1Gene. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 2147-2151.	1.8	174
14	Molecular genetics and pathophysiology of 17 beta-hydroxysteroid dehydrogenase 3 deficiency. Journal of Clinical Endocrinology and Metabolism, 1996, 81, 130-136.	1.8	168
15	Natural Mutagenesis Study of the Human Steroid 5.alpha.-Reductase 2 Isoenzyme. Biochemistry, 1994, 33, 1265-1270.	1.2	166
16	The essential role of zinc in growth. Nutrition Research, 1995, 15, 335-358.	1.3	163
17	The PROP1 2-Base Pair Deletion Is a Common Cause of Combined Pituitary Hormone Deficiency1. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 3346-3349.	1.8	163
18	21-Hydroxylaseâ€“deficient nonclassic adrenal hyperplasia is a progressive disorder: A multicenter study. American Journal of Obstetrics and Gynecology, 2000, 183, 1468-1474.	0.7	163

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19	Clinical, Hormonal, Behavioral, and Genetic Characteristics of Androgen Insensitivity Syndrome in a Brazilian Cohort: Five Novel Mutations in the Androgen Receptor Gene. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3241-3250.	1.8	158
20	Expression of Insulin-Like Growth Factor-II and Its Receptor in Pediatric and Adult Adrenocortical Tumors. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3524-3531.	1.8	149
21	Reproductive Outcome of Women with 21-Hydroxylase-Deficient Nonclassic Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 3451-3456.	1.8	146
22	Paternally Inherited DLK1 Deletion Associated With Familial Central Precocious Puberty. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1557-1567.	1.8	145
23	Molecular mechanisms of pituitary organogenesis: In search of novel regulatory genes. Molecular and Cellular Endocrinology, 2010, 323, 4-19.	1.6	140
24	The desmopressin stimulation test in the differential diagnosis of Cushing's syndrome. Clinical Endocrinology, 1993, 38, 463-472.	1.2	137
25	Growth Hormone (GH) Pharmacogenetics: Influence of GH Receptor Exon 3 Retention or Deletion on First-Year Growth Response and Final Height in Patients with Severe GH Deficiency. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 1076-1080.	1.8	136
26	The PROP1 2-Base Pair Deletion Is a Common Cause of Combined Pituitary Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 3346-3349.	1.8	136
27	17Â-Hydroxysteroid Dehydrogenase-3 Deficiency: Diagnosis, Phenotypic Variability, Population Genetics, and Worldwide Distribution of Ancient and de Novo Mutations. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 4713-4721.	1.8	136
28	A Microdeletion in the Ligand Binding Domain of Human Steroidogenic Factor 1 Causes XY Sex Reversal without Adrenal Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 1767-1772.	1.8	131
29	Longitudinal Hormonal and Pituitary Imaging Changes in Two Females with Combined Pituitary Hormone Deficiency due to Deletion of A301,G302 in the PROP1 Gene1. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 942-945.	1.8	130
30	Central Precocious Puberty That Appears to Be Sporadic Caused by Paternally Inherited Mutations in the Imprinted Gene Makorin Ring Finger 3. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1097-E1103.	1.8	126
31	Male Pseudohermaphroditism Due to Steroid 5Î±-Reductase 2 Deficiency Diagnosis, Psychological Evaluation, and Management. Medicine (United States), 1996, 75, 64-76.	0.4	123
32	Steroid 5Î±-reductase 2 deficiency. Journal of Steroid Biochemistry and Molecular Biology, 2016, 163, 206-211.	1.2	123
33	Pituitary Magnetic Resonance Imaging and Function in Patients with Growth Hormone Deficiency with and without Mutations inGHRH-R,GH-1, orPROP-1Genes. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 5076-5084.	1.8	122
34	Ectopic ACTH syndrome: our experience with 25 cases. European Journal of Endocrinology, 2006, 155, 725-733.	1.9	121
35	Longitudinal Hormonal and Pituitary Imaging Changes in Two Females with Combined Pituitary Hormone Deficiency due to Deletion of A301,G302 in the PROP1 Gene. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 942-945.	1.8	121
36	ARMC5Mutations Are a Frequent Cause of Primary Macronodular Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1501-E1509.	1.8	120

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37	A Homozygous Microdeletion in Helix 7 of the Luteinizing Hormone Receptor Associated with Familial Testicular and Ovarian Resistance Is Due to Both Decreased Cell Surface Expression and Impaired Effector Activation by the Cell Surface Receptor. Molecular Endocrinology, 1998, 12, 442-450.	3.7	119
38	Female Pseudohermaphroditism Caused by a Novel Homozygous Missense Mutation of the GR Gene. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 1805-1809.	1.8	110
39	A homozygous mutation in HESX1 is associated with evolving hypopituitarism due to impaired repressor-corepressor interaction. Journal of Clinical Investigation, 2003, 112, 1192-1201.	3.9	110
40	Founder effect for the highly prevalent R337H mutation of tumor suppressor p53 in Brazilian patients with adrenocortical tumors. Arquivos Brasileiros De Endocrinologia E Metabologia, 2004, 48, 647-650.	1.3	109
41	Loss-of-Function Mutations in the Genes Encoding Prokineticin-2 or Prokineticin Receptor-2 Cause Autosomal Recessive Kallmann Syndrome. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 4113-4118.	1.8	106
42	Clinical, genetic, and structural basis of congenital adrenal hyperplasia due to 11Î²-hydroxylase deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E1933-E1940.	3.3	106
43	P450c17 Deficiency in Brazilian Patients: Biochemical Diagnosis through Progesterone Levels Confirmed by CYP17 Genotyping. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 5739-5746.	1.8	104
44	46,XY disorders of sex development (DSD). Clinical Endocrinology, 2009, 70, 173-187.	1.2	103
45	Diagnostic Value of Fluorometric Assays in the Evaluation of Precocious Puberty. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 3539-3544.	1.8	102
46	Activating Mutation of the Stimulatory G Protein (gsp) as a Putative Cause of Ovarian and Testicular Human Stromal Leydig Cell Tumors1. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 2074-2078.	1.8	100
47	Male Pseudohermaphroditism due to 17Î²-Hydroxysteroid Dehydrogenase 3 Deficiency: Diagnosis, Psychological Evaluation, and Management. Medicine (United States), 2000, 79, 299-309.	0.4	100
48	Androgen insensitivity syndrome: a review. Archives of Endocrinology and Metabolism, 2018, 62, 227-235.	0.3	100
49	Novel Fibroblast Growth Factor Receptor 1 Mutations in Patients with Congenital Hypogonadotropic Hypogonadism with and without Anosmia. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 4006-4012.	1.8	97
50	Pheochromocytoma: Study of 50 Cases. Journal of Urology, 1997, 157, 1208-1212.	0.2	96
51	Clinical, Hormonal and Pathological Findings in a Comparative Study of Adrenocortical Neoplasms in Childhood and Adulthood. Journal of Urology, 1995, 154, 2004-2009.	0.2	93
52	Height and bone mineral density in androgen insensitivity syndrome with mutations in the androgen receptor gene. Osteoporosis International, 2007, 18, 369-374.	1.3	92
53	Molecular Genotyping in Brazilian Patients with the Classical and Nonclassical Forms of 21-Hydroxylase Deficiency1. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 4416-4419.	1.8	91
54	Novel Heterozygous Nonsense GLI2 Mutations in Patients with Hypopituitarism and Ectopic Posterior Pituitary Lobe without Holoprosencephaly. Journal of Clinical Endocrinology and Metabolism, 2010, 95, E384-E391.	1.8	91

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55	An update of genetic basis of PCOS pathogenesis. Archives of Endocrinology and Metabolism, 2018, 62, 352-361.	0.3	88
56	A novel mutation of the luteinizing hormone receptor gene causing male gonadotropin-independent precocious puberty. Journal of Clinical Endocrinology and Metabolism, 1995, 80, 2490-2494.	1.8	86
57	Management of ambiguous genitalia in pseudohermaphrodites: New perspectives on vaginal dilation. Fertility and Sterility, 1997, 67, 229-232.	0.5	84
58	A Single Luteinizing Hormone Determination 2 Hours after Depot Leuprolide Is Useful for Therapy Monitoring of Gonadotropin-Dependent Precocious Puberty in Girls. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 4338-4342.	1.8	84
59	PTPN11 (Protein Tyrosine Phosphatase, Nonreceptor Type 11) Mutations and Response to Growth Hormone Therapy in Children with Noonan Syndrome. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 5156-5160.	1.8	83
60	Refining Hormonal Diagnosis of Type II 3Î²-Hydroxysteroid Dehydrogenase Deficiency in Patients with Premature Pubarche and Hirsutism Based on HSD3B2 Genotyping. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 1287-1293.	1.8	81
61	Combined expression of BUB1B, DLGAP5, and PINK1 as predictors of poor outcome in adrenocortical tumors: validation in a Brazilian cohort of adult and pediatric patients. European Journal of Endocrinology, 2012, 166, 61-67.	1.9	81
62	Disorders of sex development: effect of molecular diagnostics. Nature Reviews Endocrinology, 2015, 11, 478-488.	4.3	81
63	Extraadrenal 21-Hydroxylation by CYP2C19 and CYP3A4: Effect on 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 89-95.	1.8	78
64	Influence of different genotypes on 17-hydroxyprogesterone levels in patients with nonclassical congenital adrenal hyperplasia due to 21-hydroxylase deficiency. Clinical Endocrinology, 2000, 52, 601-607.	1.2	76
65	Wide spectrum of NR5A1â€related phenotypes in 46,XY and 46,XX individuals. Birth Defects Research Part C: Embryo Today Reviews, 2016, 108, 309-320.	3.6	76
66	Mutation analysis of the 2 kb 5' to SRY in XY females and XY intersex subjects.. Journal of Medical Genetics, 1996, 33, 465-468.	1.5	75
67	Menstrual disturbances in patients with systemic lupus erythematosus without alkylating therapy: clinical, hormonal and therapeutic associations. Lupus, 2002, 11, 175-180.	0.8	75
68	SHOX mutations in idiopathic short stature and Leri-Weill dyschondrosteosis: frequency and phenotypic variability. Clinical Endocrinology, 2006, 66, 061031010617004-???.	1.2	75
69	Update on the etiology, diagnosis and therapeutic management of sexual precocity. Arquivos Brasileiros De Endocrinologia E Metabologia, 2008, 52, 18-31.	1.3	75
70	DLK1 Is a Novel Link Between Reproduction and Metabolism. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2112-2120.	1.8	75
71	No evidence for oncogenic mutations in the adrenocorticotropin receptor gene in human adrenocortical neoplasms. Journal of Clinical Endocrinology and Metabolism, 1995, 80, 875-877.	1.8	74
72	Molecular Genotyping in Brazilian Patients with the Classical and Nonclassical Forms of 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 4416-4419.	1.8	72

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73	A Unique Constitutively Activating Mutation in Third Transmembrane Helix of Luteinizing Hormone Receptor Causes Sporadic Male Gonadotropin-Independent Precocious Puberty. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 2435-2440.	1.8	72
74	A study of patients with Nelson's syndrome. Clinical Endocrinology, 1998, 49, 533-539.	1.2	71
75	Noonan Syndrome and Related Disorders: A Review of Clinical Features and Mutations in Genes of the RAS/MAPK Pathway. Hormone Research in Paediatrics, 2009, 71, 185-193.	0.8	71
76	Nonsense Mutations inFGF8Gene Causing Different Degrees of Human Gonadotropin-Releasing Deficiency. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3491-3496.	1.8	70
77	Two Novel Mutations in the Gonadotropin-Releasing Hormone Receptor Gene in Brazilian Patients with Hypogonadotropic Hypogonadism and Normal Olfaction. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 2680-2686.	1.8	70
78	New genetic findings in a large cohort of congenital hypogonadotropic hypogonadism. European Journal of Endocrinology, 2019, 181, 103-119.	1.9	70
79	Ectopic ACTH syndrome. Journal of Steroid Biochemistry and Molecular Biology, 1995, 53, 139-151.	1.2	69
80	Apparent mineralocorticoid excess in a Brazilian kindred. Journal of Hypertension, 1997, 15, 1397-1402.	0.3	68
81	Adrenocorticotropin-Dependent Precocious Puberty of Testicular Origin in a Boy with X-Linked Adrenal Hypoplasia Congenita Due to a Novel Mutation in the DAX1 Gene. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 4068-4071.	1.8	68
82	Management of 46,XY Differences/Disorders of Sex Development (DSD) Throughout Life. Endocrine Reviews, 2019, 40, 1547-1572.	8.9	68
83	No evidence of the inactivating mutation (C566T) in the follicle-stimulating hormone receptor gene in Brazilian women with premature ovarian failure. Fertility and Sterility, 1998, 70, 565-567.	0.5	67
84	Steroidogenic Factor 1 Overexpression and Gene Amplification Are More Frequent in Adrenocortical Tumors from Children than from Adults. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 1458-1462.	1.8	66
85	46,XY disorder of sex development (DSD) due to 17Î²-hydroxysteroid dehydrogenase type 3 deficiency. Journal of Steroid Biochemistry and Molecular Biology, 2017, 165, 79-85.	1.2	66
86	A novel missense mutation (S18N) in the 5â€² non-HMG box region of the SRY gene in a patient with partial gonadal dysgenesis and his normal male relatives. Human Genetics, 1998, 102, 213-215.	1.8	65
87	High Frequency of <i>MKRN3</i> Mutations in Male Central Precocious Puberty Previously Classified as Idiopathic. Neuroendocrinology, 2017, 105, 17-25.	1.2	65
88	Report of a del22q11 in a patient with Mayer-Rokitansky-KÃ¼ster-Hauser (MRKH) anomaly and exclusion ofWNT-4,RAR-gamma, andRXR-alpha as major genes determining MRKH anomaly in a study of 25 affected women. American Journal of Medical Genetics, Part A, 2006, 140A, 1339-1342.	0.7	64
89	An Unusual Phenotype of Frasier Syndrome due to IVS9 +4C>T Mutation in the WT1 Gene: Predominantly Male Ambiguous Genitalia and Absence of Gonadal Dysgenesis. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 2500-2505.	1.8	62
90	Regional rearrangements in chromosome 15q21 cause formation of cryptic promoters for the CYP19 (aromatase) gene. Human Molecular Genetics, 2007, 16, 2529-2541.	1.4	62

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91	Molecular CYP21A2 diagnosis in 480 Brazilian patients with congenital adrenal hyperplasia before newborn screening introduction. European Journal of Endocrinology, 2016, 175, 107-116.	1.9	60
92	Combined Pituitary Hormone Deficiency Caused by a Novel Mutation of a Highly Conserved Residue (F88S) in the Homeodomain of PROP-1¹. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 2779-2785.	1.8	59
93	46,XY DSD due to impaired androgen production. Best Practice and Research in Clinical Endocrinology and Metabolism, 2010, 24, 243-262.	2.2	58
94	Three Novel Mutations in CYP21 Gene in Brazilian Patients with the Classical Form of 21-Hydroxylase Deficiency Due to a Founder Effect. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4314-4317.	1.8	57
95	DSD Due to 5Î±-Reductase 2 Deficiency - from Diagnosis to Long Term Outcome. Seminars in Reproductive Medicine, 2012, 30, 427-431.	0.5	57
96	Growth standards of patients with Noonan and Noonanâ€like syndromes with mutations in the RAS/MAPK pathway. American Journal of Medical Genetics, Part A, 2012, 158A, 2700-2706.	0.7	56
97	Pituitary Apoplexy During Therapy with Cabergoline in an Adolescent Male with Prolactin-Secreting Macroadenoma. Pituitary, 2004, 7, 83-87.	1.6	55
98	The role of desmopressin in bilateral and simultaneous inferior petrosal sinus sampling for differential diagnosis of ACTH-dependent Cushing's syndrome. Clinical Endocrinology, 2006, 66, 061120012318003-???.	1.2	55
99	GAPO syndrome (McKusick 23074)â€"A connective tissue disorder: Report on two affected sibs and on the pathologic findings in the older. American Journal of Medical Genetics Part A, 1990, 37, 213-223.	2.4	54
100	Factors Determining Normal Adult Height in Girls with Gonadotropin-Dependent Precocious Puberty Treated with Depot Gonadotropin-Releasing Hormone Analogs. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 2662-2669.	1.8	54
101	Male pseudohermaphroditism due to nonsalt-losing 3Î²-hydroxysteroid dehydrogenase deficiency: Gender role change and absence of gynecomastia at puberty. The Journal of Steroid Biochemistry, 1987, 28, 669-675.	1.3	53
102	A Novel Nonsense Mutation in the First Zinc Finger of the Vitamin D Receptor Causing Hereditary 1,25-Dihydroxyvitamin D₃-Resistant Rickets. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 3892-3894.	1.8	53
103	Mutations in the SRY, DAX1, SF1 and WNT4 genes in Brazilian sex-reversed patients. Brazilian Journal of Medical and Biological Research, 2004, 37, 145-150.	0.7	53
104	Familial Hyperestrogenism in Both Sexes: Clinical, Hormonal, and Molecular Studies of Two Siblings. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3027-3034.	1.8	52
105	Combined Pituitary Hormone Deficiency Caused by a Novel Mutation of a Highly Conserved Residue (F88S) in the Homeodomain of PROP-1. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 2779-2785.	1.8	52
106	Clinical and hormonal features of selective follicle-stimulating hormone (FSH) deficiency due to FSH beta-subunit gene mutations in both sexes. Fertility and Sterility, 2005, 83, 466-470.	0.5	51
107	Targeted Assessment of G0S2 Methylation Identifies a Rapidly Recurrent, Routinely Fatal Molecular Subtype of Adrenocortical Carcinoma. Clinical Cancer Research, 2019, 25, 3276-3288.	3.2	51
108	Gonadal agenesis in XX and XY sisters: Evidence for the involvement of an autosomal gene. American Journal of Medical Genetics Part A, 1994, 52, 39-43.	2.4	50

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109	Microconversion betweenCYP21A2andCYP21A1PPromoter Regions Causes the Nonclassical Form of 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 4028-4034.	1.8	50
110	A novel homozygous splice acceptor site mutation of KISS1R in two siblings with normosmic isolated hypogonadotropic hypogonadism. European Journal of Endocrinology, 2010, 163, 29-34.	1.9	50
111	Role of GLI2 in hypopituitarism phenotype. Journal of Molecular Endocrinology, 2015, 54, R141-R150.	1.1	50
112	Malignant testicular germ cell tumors in postpubertal individuals with androgen insensitivity: prevalence, pathology and relevance of single nucleotide polymorphism-based susceptibility profiling. Human Reproduction, 2017, 32, 2561-2573.	0.4	50
113	Gonadotropin-Independent Precocious Puberty Due to Luteinizing Hormone Receptor Mutations in Brazilian Boys: A Novel Constitutively Activating Mutation in the First Transmembrane Helix. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 4799-4805.	1.8	50
114	Gonadotropin-Independent Precocious Puberty Due to Luteinizing Hormone Receptor Mutations in Brazilian Boys: A Novel Constitutively Activating Mutation in the First Transmembrane Helix1. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 4799-4805.	1.8	49
115	Novel inactivating mutations in the GH secretagogue receptor gene in patients with constitutional delay of growth and puberty. European Journal of Endocrinology, 2011, 165, 233-241.	1.9	49
116	The Common P450 Oxidoreductase Variant A503V Is Not a Modifier Gene for 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 2913-2916.	1.8	48
117	Diagnosis of prolactinoma in two male-to-female transsexual subjects following high-dose cross-sex hormone therapy. Andrologia, 2015, 47, 680-684.	1.0	48
118	A clinico-genetic investigation of Leydig cell hypoplasia. American Journal of Medical Genetics Part A, 1987, 26, 337-344.	2.4	47
119	Role of gonadotropin-releasing hormone receptor mutations in patients with a wide spectrum of pubertal delay. Fertility and Sterility, 2014, 102, 838-846.e2.	0.5	47
120	Long-Term Surgical Outcome of Masculinizing Genitoplasty in Large Cohort of Patients With Disorders of Sex Development. Journal of Urology, 2010, 184, 1122-1127.	0.2	46
121	Mutational analysis of TAC3 and TACR3 genes in patients with idiopathic central pubertal disorders. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 646-652.	1.3	46
122	Detection of Y-specific sequences in 122 patients with Turner syndrome: Nested PCR is not a reliable method. American Journal of Medical Genetics Part A, 2002, 107, 299-305.	2.4	45
123	The â^'202 A Allele of Insulin-Like Growth Factor Binding Protein-3 (IGFBP3) Promoter Polymorphism Is Associated with Higher IGFBP-3 Serum Levels and Better Growth Response to Growth Hormone Treatment in Patients with Severe Growth Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 588-595.	1.8	45
124	The Sitting Height/Height Ratio for Age in Healthy and Short Individuals and Its Potential Role in Selecting Short Children for <i>SHOX</i> Analysis. Hormone Research in Paediatrics, 2013, 80, 449-456.	0.8	45
125	Successful Live Birth in a Woman With 17Î±-Hydroxylase Deficiency Through IVF Frozen-Thawed Embryo Transfer. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 345-348.	1.8	45
126	Genetics of Primary Ovarian Insufficiency in the Next-Generation Sequencing Era. Journal of the Endocrine Society, 2020, 4, bvz037.	0.1	45

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127	Variable ACTH-Stimulated 17-Hydroxyprogesterone Values in 21-Hydroxylase Deficiency Carriers Are Not Related to the Different CYP21 Gene Mutations. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 786-790.	1.8	44
128	Role for postoperative cortisol response to desmopressin in predicting the risk for recurrent Cushing's disease. Clinical Endocrinology, 2008, 69, 117-122.	1.2	44
129	Effectiveness of treating ovarian hyperstimulation syndrome with cabergoline in two patients with gonadotropin-producing pituitary adenomas. Fertility and Sterility, 2006, 86, 719.e15-719.e18.	0.5	43
130	Longâ€term treatment of familial maleâ€limited precocious puberty (testotoxicosis) with cyproterone acetate or ketoconazole. Clinical Endocrinology, 2008, 69, 93-98.	1.2	42
131	KCNJ5 Somatic Mutation Is a Predictor of Hypertension Remission After Adrenalectomy for Unilateral Primary Aldosteronism. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4695-4702.	1.8	42
132	Anatomical and functional outcomes of feminizing genitoplasty for ambiguous genitalia in patients with virilizing congenital adrenal hyperplasia. Clinics, 2006, 61, 209-14.	0.6	41
133	Genetics of primary macronodular adrenal hyperplasia. Journal of Endocrinology, 2015, 224, R31-R43.	1.2	41
134	17Î²-Hydroxysteroid Dehydrogenase 3 Deficiency in Women¹. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 802-804.	1.8	39
135	Long-Term Followup of a Large Cohort of Patients with Ovotesticular Disorder of Sex Development. Journal of Urology, 2014, 191, 1532-1536.	0.2	39
136	Nonâ€coding variation in disorders of sex development. Clinical Genetics, 2017, 91, 163-172.	1.0	39
137	Classic congenital adrenal hyperplasia and its impact on reproduction. Fertility and Sterility, 2019, 111, 7-12.	0.5	39
138	A meiotic recombination in a new isolated familial somatotropinoma kindred. European Journal of Endocrinology, 2004, 150, 643-648.	1.9	38
139	The role of ARMC5 in human cell cultures from nodules of primary macronodular adrenocortical hyperplasia (PMAH). Molecular and Cellular Endocrinology, 2018, 460, 36-46.	1.6	38
140	17Â-Hydroxysteroid Dehydrogenase 3 Deficiency in Women. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 802-804.	1.8	38
141	Discriminating between virilizing ovary tumors and ovary hyperthecosis in postmenopausal women: clinical data, hormonal profiles and image studies. European Journal of Endocrinology, 2017, 177, 93-102.	1.9	37
142	XAF1 as a modifier of p53 function and cancer susceptibility. Science Advances, 2020, 6, eaba3231.	4.7	37
143	Poor Reproducibility of IGF-I and IGF Binding Protein-3 Generation Test in Children with Short Stature and Normal Coding Region of the GH Receptor Gene. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 469-472.	1.8	36
144	GH Values after Clonidine Stimulation Measured by Immunofluorometric Assay in Normal Prepubertal Children and GH-Deficient Patients. Hormone Research in Paediatrics, 2003, 59, 229-233.	0.8	36

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146	Homozygous Inactivating Mutation inNANOS3in Two Sisters with Primary Ovarian Insufficiency. BioMed Research International, 2014, 2014, 1-8.	0.9	36
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