Berenice Bilharinho Mendonca

List of Publications by Citations

446 papers	16,076 citations	69 h-index	105 g-index
527 ext. papers	18,529 ext. citations	4.5 avg, IF	6.45 L-index

#	Paper	IF	Citations
446	Male pseudohermaphroditism caused by mutations of testicular 17 beta-hydroxysteroid dehydrogenase 3. *Nature Genetics*, **1994**, 7, 34-9	36.3	482
445	A GPR54-activating mutation in a patient with central precocious puberty. *New England Journal of Medicine*, **2008**, 358, 709-15	59.2	415
444	Mutant P450 oxidoreductase causes disordered steroidogenesis with and without Antley-Bixler syndrome. *Nature Genetics*, **2004**, 36, 228-30	36.3	396
443	Central precocious puberty caused by mutations in the imprinted gene MKRN3. *New England Journal of Medicine*, **2013**, 368, 2467-75	59.2	351
442	Comprehensive Pan-Genomic Characterization of Adrenocortical Carcinoma. *Cancer Cell*, **2016**, 29, 723-736	24.3	324
441	Brief report: testicular and ovarian resistance to luteinizing hormone caused by inactivating mutations of the luteinizing hormone-receptor gene. *New England Journal of Medicine*, **1996**, 334, 507-12	59.2	312
440	Molecular genetics of steroid 5 alpha-reductase 2 deficiency. *Journal of Clinical Investigation*, **1992**, 90, 799-809	15.9	284
439	The genetic and functional basis of isolated 17,20-lyase deficiency. *Nature Genetics*, **1997**, 17, 201-5	36.3	277
438	Mutations of the KISS1 gene in disorders of puberty. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 2276-80	5.6	236
437	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, 2857-67	5.6	212
436	Ectopic adrenocorticotropic hormone syndrome. *Endocrine Reviews*, **1994**, 15, 752-87	27.2	175
435	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, 4970-3	5.6	151
434	The PROP1 2-base pair deletion is a common cause of combined pituitary hormone deficiency. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 3346-9	5.6	146
433	Cushing's syndrome secondary to adrenocorticotropin-independent macronodular adrenocortical hyperplasia due to activating mutations of GNAS1 gene. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 2147-51	5.6	144
432	Molecular genetics and pathophysiology of 17 beta-hydroxysteroid dehydrogenase 3 deficiency. *Journal of Clinical Endocrinology and Metabolism*, **1996**, 81, 130-136	5.6	142
431	Natural mutagenesis study of the human steroid 5 alpha-reductase 2 isozyme. *Biochemistry*, **1994**, 33, 1265-70	3.2	141
430	The essential role of zinc in growth. *Nutrition Research*, **1995**, 15, 335-358	4	140

429	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, 3241-50	5.6	132
428	21-Hydroxylase-deficient nonclassic adrenal hyperplasia is a progressive disorder: a multicenter study. *American Journal of Obstetrics and Gynecology*, **2000**, 183, 1468-74	6.4	128
427	Expression of insulin-like growth factor-II and its receptor in pediatric and adult adrenocortical tumors. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 3524-31	5.6	123
426	Molecular mechanisms of pituitary organogenesis: In search of novel regulatory genes. *Molecular and Cellular Endocrinology*, **2010**, 323, 4-19	4.4	122
425	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	5.6	121
424	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, 1076-80	5.6	118
423	17´-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	5.6	118
422	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, 1767-72	5.6	117
421	The desmopressin stimulation test in the differential diagnosis of Cushing's syndrome. *Clinical Endocrinology*, **1993**, 38, 463-72	3.4	116
420	Reproductive outcome of women with 21-hydroxylase-deficient nonclassic adrenal hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2006**, 91, 3451-6	5.6	113
419	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, 942-5	5.6	113
418	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	5.6	108
417	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, 442-50		105
416	Pituitary magnetic resonance imaging and function in patients with growth hormone deficiency with and without mutations in GHRH-R, GH-1, or PROP-1 genes. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 5076-84	5.6	104
415	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, E1097-103	5.6	102
414	Ectopic ACTH syndrome: our experience with 25 cases. *European Journal of Endocrinology*, **2006**, 155, 725-33	6.5	98
413	Female pseudohermaphroditism caused by a novel homozygous missense mutation of the GR gene. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 1805-9	5.6	97
412	Male pseudohermaphroditism due to steroid 5alpha-reductase 2 deficiency. Diagnosis, psychological evaluation, and management. *Medicine (United States)*, **1996**, 75, 64-76	1.8	94

411	Paternally Inherited DLK1 Deletion Associated With Familial Central Precocious Puberty. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 1557-1567	5.6	88
410	A homozygous mutation in HESX1 is associated with evolving hypopituitarism due to impaired repressor-corepressor interaction. *Journal of Clinical Investigation*, **2003**, 112, 1192-201	15.9	88
409	ARMC5 mutations are a frequent cause of primary macronodular adrenal Hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1501-9	5.6	87
408	Activating mutation of the stimulatory G protein (gsp) as a putative cause of ovarian and testicular human stromal Leydig cell tumors. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 2074-8	5.6	86
407	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, 4113-8	5.6	85
406	P450c17 deficiency in Brazilian patients: biochemical diagnosis through progesterone levels confirmed by CYP17 genotyping. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 5739-46	5.6	85
405	Diagnostic Value of Fluorometric Assays in the Evaluation of Precocious Puberty. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 3539-3544	5.6	84
404	Novel heterozygous nonsense GLI2 mutations in patients with hypopituitarism and ectopic posterior pituitary lobe without holoprosencephaly. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, E384-91	5.6	83
403	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	5.6	82
402	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, 647-50		81
401	Molecular genotyping in Brazilian patients with the classical and nonclassical forms of 21-hydroxylase deficiency. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 4416-9	5.6	80
400	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, 4006-12	5.6	79
399	SAT-155 High Prevalence Alterations on DNA Mismatch Repair Genes Related to Lynch Syndrome in Pediatric Patients with Adrenocortical Tumor Carried of the Germline Mutation on TP53. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
398	SUN-049 Male Pattern Baldness and Waist-Hip Ratio as Markers of Arterial Stiffness in Transgender Men Undergoing Long-Term Testosterone Therapy. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
397	SUN-725 Clinical and Genetic Features of Families with Maternally Inherited Central Precocious Puberty. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
396	SUN-081 High Throughput Genetic Analysis Revealed Novel Genomic Loci and Candidate Genes Involved in Central Precocious Puberty Associated with Complex Phenotypes. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
395	OR06-6 Whole-Exome Sequencing of Patients with Pituitary Stalk Interruption Syndrome (PSIS) Reveals Probably Pathogenic Variants in Novel Candidate Genes.. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78
394	OR04-6 Predictors of Clinical Outcome after Adrenalectomy for Unilateral Primary Aldosteronism. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78

393	SUN-359 Preserved Bone Mineral Density In Adults With Classical Forms Of Congenital Adrenal Hyperplasia Submitted To Long-term Low Glucocorticoid Doses. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78
392	SAT-064 Validation of Furosemide Upright Test in Primary Aldosteronism Diagnosis Using Direct Renin Assay. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78
391	MON-251 Clinical Features of a Large Cohort of Patients with Familial Central Precocious Puberty Caused by Loss-of-Function Mutations in MKRN3. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78
390	SUN-709 MiR-200c Expression Profiles in Plasma of 46,XY DSD Patients of Unknown Etiology. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
389	SUN-085 Clinical and Hormonal Features of 37 Families with Central Precocious Puberty Due to MKRN3 Loss-Of -Function Mutations. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
388	OR15-04 Central Precocious Puberty without Central Nervous System Lesions: Is It Really Idiopathic?. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
387	SAT-560 Usefulness of Contralateral Suppression in Adrenal Venous Sampling to Define Lateralization in Primary Aldosteronism. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
386	SUN-178 Clinical and Anatomopathological Characteristics of Two Atypical Aldosterone-Producing Adenomas. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
385	SUN-061 Anthropometric and Reproductive Outcomes of Patients with Gonadotropin-Independent Precocious Puberty Due to McCune-Albright Syndrome After Treatment with Distinct Therapeutic Agents. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
384	Pheochromocytoma: study of 50 cases. *Journal of Urology*, **1997**, 157, 1208-12	2.5	77
383	Height and bone mineral density in androgen insensitivity syndrome with mutations in the androgen receptor gene. *Osteoporosis International*, **2007**, 18, 369-74	5.3	76
382	Male pseudohermaphroditism due to 17 beta-hydroxysteroid dehydrogenase 3 deficiency. Diagnosis, psychological evaluation, and management. *Medicine (United States)*, **2000**, 79, 299-309	1.8	73
381	Clinical, genetic, and structural basis of congenital adrenal hyperplasia due to 11β-hydroxylase deficiency. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E1933-E1940	11.5	70
380	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, 4338-42	5.6	69
379	No evidence for oncogenic mutations in the adrenocorticotropin receptor gene in human adrenocortical neoplasms. *Journal of Clinical Endocrinology and Metabolism*, **1995**, 80, 875-877	5.6	69
378	Clinical, Hormonal and Pathological Findings in a Comparative Study of Adrenocortical Neoplasms in Childhood and Adulthood. *Journal of Urology*, **1995**, 154, 2004-2009	2.5	67
377	Extraadrenal 21-hydroxylation by CYP2C19 and CYP3A4: effect on 21-hydroxylase deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 89-95	5.6	66
376	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, 5156-60	5.6	66

375	Refining hormonal diagnosis of type II 3beta-hydroxysteroid dehydrogenase deficiency in patients with premature pubarche and hirsutism based on HSD3B2 genotyping. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 1287-93	5.6	65
374	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	5.6	65
373	Androgen insensitivity syndrome: a review. *Archives of Endocrinology and Metabolism*, **2018**, 62, 227-235	2.2	64
372	Management of ambiguous genitalia in pseudohermaphrodites: new perspectives on vaginal dilation. *Fertility and Sterility*, **1997**, 67, 229-32	4.8	64
371	46,XY disorders of sex development (DSD). *Clinical Endocrinology*, **2009**, 70, 173-87	3.4	63
370	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, 61-7	6.5	62
369	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, 565-7	4.8	62
368	Nonsense mutations in FGF8 gene causing different degrees of human gonadotropin-releasing deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 3491-6	5.6	61
367	A study of patients with Nelson's syndrome. *Clinical Endocrinology*, **1998**, 49, 533-9	3.4	60
366	Menstrual disturbances in patients with systemic lupus erythematosus without alkylating therapy: clinical, hormonal and therapeutic associations. *Lupus*, **2002**, 11, 175-80	2.6	60
365	Ectopic ACTH syndrome. *Journal of Steroid Biochemistry and Molecular Biology*, **1995**, 53, 139-51	5.1	60
364	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	5.6	59
363	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	5.6	59
362	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, 4068-71	5.6	57
361	Disorders of sex development: effect of molecular diagnostics. *Nature Reviews Endocrinology*, **2015**, 11, 478-88	15.2	56
360	A novel missense mutation (S18N) in the 5' non-HMG box region of the SRY gene in a patient with partial gonadal dysgenesis and his normal male relatives. *Human Genetics*, **1998**, 102, 213-5	6.3	56
359	Mutation analysis of the 2 kb 5' to SRY in XY females and XY intersex subjects. *Journal of Medical Genetics*, **1996**, 33, 465-8	5.8	56
358	Report of a del22q11 in a patient with Mayer-Rokitansky-K^ Ster-Hauser (MRKH) anomaly and exclusion of WNT-4, RAR-gamma, and RXR-alpha as major genes determining MRKH anomaly in a study of 25 affected women. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1339-42	2.5	55

357	Apparent mineralocorticoid excess in a Brazilian kindred: hypertension in the heterozygote state. *Journal of Hypertension*, **1997**, 15, 1397-402	1.9	54
356	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, 2500-5	5.6	54
355	Influence of different genotypes on 17-hydroxyprogesterone levels in patients with nonclassical congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Clinical Endocrinology*, **2000**, 52, 601-7	3.4	54
354	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, 185-93	3.3	53
353	Regional rearrangements in chromosome 15q21 cause formation of cryptic promoters for the CYP19 (aromatase) gene. *Human Molecular Genetics*, **2007**, 16, 2529-41	5.6	53
352	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, 2779-85	5.6	53
351	Update on the etiology, diagnosis and therapeutic management of sexual precocity. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2008**, 52, 18-31		52
350	High Frequency of MKRN3 Mutations in Male Central Precocious Puberty Previously Classified as Idiopathic. *Neuroendocrinology*, **2017**, 105, 17-25	5.6	50
349	An update of genetic basis of PCOS pathogenesis. *Archives of Endocrinology and Metabolism*, **2018**, 62, 352-361	2.2	50
348	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, 1458-62	5.6	50
347	GAPO syndrome (McKusick 23074)--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, 213-23		49
346	Steroid 5□reductase 2 deficiency. *Journal of Steroid Biochemistry and Molecular Biology*, **2016**, 163, 206-11	5.1	49
345	Familial hyperestrogenism in both sexes: clinical, hormonal, and molecular studies of two siblings. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 3027-34	5.6	48
344	Pituitary apoplexy during therapy with cabergoline in an adolescent male with prolactin-secreting macroadenoma. *Pituitary*, **2004**, 7, 83-7	4.3	48
343	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	6.5	47
342	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, 3892-4	5.6	47
341	SHOX mutations in idiopathic short stature and Leri-Weill dyschondrosteosis: frequency and phenotypic variability. *Clinical Endocrinology*, **2007**, 66, 130-5	3.4	47
340	Mutations in the SRY, DAX1, SF1 and WNT4 genes in Brazilian sex-reversed patients. *Brazilian Journal of Medical and Biological Research*, **2004**, 37, 145-50	2.8	46

339	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, 4314-7	5.6	46
338	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		46
337	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, 466-70	4.8	45
336	DLK1 Is a Novel Link Between Reproduction and Metabolism. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2112-2120	5.6	44
335	The common P450 oxidoreductase variant A503V is not a modifier gene for 21-hydroxylase deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 2913-6	5.6	44
334	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, 4799-805	5.6	44
333	A clinico-genetic investigation of Leydig cell hypoplasia. *American Journal of Medical Genetics Part A* , **1987**, 26, 337-44		44
332	Role of GLI2 in hypopituitarism phenotype. *Journal of Molecular Endocrinology*, **2015**, 54, R141-50	4.5	43
331	The -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, 588-95	5.6	43
330	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		43
329	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	5.6	43
328	46,XY disorder of sex development (DSD) due to 17β-hydroxysteroid dehydrogenase type 3 deficiency. *Journal of Steroid Biochemistry and Molecular Biology*, **2017**, 165, 79-85	5.1	42
327	Diagnosis of prolactinoma in two male-to-female transsexual subjects following high-dose cross-sex hormone therapy. *Andrologia*, **2015**, 47, 680-4	2.4	42
326	Novel inactivating mutations in the GH secretagogue receptor gene in patients with constitutional delay of growth and puberty. *European Journal of Endocrinology*, **2011**, 165, 233-41	6.5	42
325	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	5.6	42
324	The sitting height/height ratio for age in healthy and short individuals and its potential role in selecting short children for SHOX analysis. *Hormone Research in Paediatrics*, **2013**, 80, 449-56	3.3	41
323	DSD due to 5α-reductase 2 deficiency - from diagnosis to long term outcome. *Seminars in Reproductive Medicine*, **2012**, 30, 427-31	1.4	41
322	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, 2662-9	5.6	41

321	Microconversion between CYP21A2 and CYP21A1P promoter regions causes the nonclassical form of 21-hydroxylase deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 4028-34	5.6	41
320	New genetic findings in a large cohort of congenital hypogonadotropic hypogonadism. *European Journal of Endocrinology*, **2019**, 181, 103-119	6.5	41
319	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	5.7	40
318	Growth standards of patients with Noonan and Noonan-like syndromes with mutations in the RAS/MAPK pathway. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2700-6	2.5	40
317	Role of gonadotropin-releasing hormone receptor mutations in patients with a wide spectrum of pubertal delay. *Fertility and Sterility*, **2014**, 102, 838-846.e2	4.8	39
316	46,XY DSD due to impaired androgen production. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2010**, 24, 243-62	6.5	39
315	Male pseudohermaphroditism due to nonsalt-losing 3 beta-hydroxysteroid dehydrogenase deficiency: gender role change and absence of gynecomastia at puberty. *The Journal of Steroid Biochemistry*, **1987**, 28, 669-75		39
314	Long-term surgical outcome of masculinizing genitoplasty in large cohort of patients with disorders of sex development. *Journal of Urology*, **2010**, 184, 1122-7	2.5	37
313	Molecular CYP21A2 diagnosis in 480 Brazilian patients with congenital adrenal hyperplasia before newborn screening introduction. *European Journal of Endocrinology*, **2016**, 175, 107-16	6.5	37
312	Mutational analysis of TAC3 and TACR3 genes in patients with idiopathic central pubertal disorders. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2012**, 56, 646-52		36
311	Long-term treatment of familial male-limited precocious puberty (testotoxicosis) with cyproterone acetate or ketoconazole. *Clinical Endocrinology*, **2008**, 69, 93-8	3.4	35
310	17´-Hydroxysteroid Dehydrogenase 3 Deficiency in Women. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 802-804	5.6	35
309	Combined pituitary hormone deficiency (CPHD) due to a complete PROP1 deletion. *Clinical Endocrinology*, **2006**, 65, 294-300	3.4	33
308	Spironolactone-reversible rickets associated with 11 beta-hydroxysteroid dehydrogenase deficiency syndrome. *Journal of Pediatrics*, **1986**, 109, 989-93	3.6	33
307	Role for postoperative cortisol response to desmopressin in predicting the risk for recurrent Cushing's disease. *Clinical Endocrinology*, **2008**, 69, 117-22	3.4	32
306	Absence of follicle-stimulating hormone receptor activating mutations in women with iatrogenic ovarian hyperstimulation syndrome. *Fertility and Sterility*, **2005**, 83, 1695-9	4.8	32
305	Anatomical and functional outcomes of feminizing genitoplasty for ambiguous genitalia in patients with virilizing congenital adrenal hyperplasia. *Clinics*, **2006**, 61, 209-14	2.3	32
304	Long-term followup of a large cohort of patients with ovotesticular disorder of sex development. *Journal of Urology*, **2014**, 191, 1532-6	2.5	31

303	Homozygous inactivating mutation in NANOS3 in two sisters with primary ovarian insufficiency. *BioMed Research International*, **2014**, 2014, 787465	3	31
302	The role of desmopressin in bilateral and simultaneous inferior petrosal sinus sampling for differential diagnosis of ACTH-dependent Cushing's syndrome. *Clinical Endocrinology*, **2007**, 66, 136-42	3.4	31
301	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, 469-72	5.6	31
300	Genetics of primary macronodular adrenal hyperplasia. *Journal of Endocrinology*, **2015**, 224, R31-43	4.7	30
299	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, 786-90	5.6	30
298	17Beta-hydroxysteroid dehydrogenase 3 deficiency in women. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 802-4	5.6	30
297	Targeted Assessment of Methylation Identifies a Rapidly Recurrent, Routinely Fatal Molecular Subtype of Adrenocortical Carcinoma. *Clinical Cancer Research*, **2019**, 25, 3276-3288	12.9	29
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