Berenice Bilharinho Mendonca

List of Publications by Year in Descending Order

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	Novel OTX2 loss of function variant associated with congenital hypopituitarism without eye abnormalities.. *Journal of Pediatric Endocrinology and Metabolism*, **2022**,	1.6	
445	Genotype-Phenotype Correlations in Central Precocious Puberty Caused by MKRN3 Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, 1041-1050	5.6	10
444	Genetics of ovarian insufficiency and defects of folliculogenesis. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2021**, 101594	6.5	5
443	Role of the Mevalonate Pathway in Adrenocortical Tumorigenesis. *Hormone and Metabolic Research* , **2021**, 53, 124-131	3.1	0
442	Genetic and clinical aspects of paediatric pheochromocytomas and paragangliomas. *Clinical Endocrinology*, **2021**, 95, 117-124	3.4	4
441	International practice of corticosteroid replacement therapy in congenital adrenal hyperplasia: data from the I-CAH registry. *European Journal of Endocrinology*, **2021**, 184, 553-563	6.5	4
440	The Cost-Effectiveness of Congenital Adrenal Hyperplasia Newborn Screening in Brazil: A Comparison Between Screened and Unscreened Cohorts. *Frontiers in Pediatrics*, **2021**, 9, 659492	3.4	0
439	Low Protein Expression of ATRX and ZNRF3 as a Novel Prognostic Marker of Adult Adrenocortical Carcinoma. *Journal of the Endocrine Society*, **2021**, 5, A87-A88	0.4	
438	Adult Height in 299 Patients with Turner Syndrome with or without Growth Hormone Therapy: Results and Literature Review. *Hormone Research in Paediatrics*, **2021**, 94, 63-70	3.3	0
437	Primary adrenal insufficiency due to bilateral adrenal infarction in COVID-19: a case report. *Journal of Clinical Endocrinology and Metabolism*, **2021**,	5.6	3
436	Genetics, clinical features and outcomes of non-syndromic pituitary gigantism: experience of a single center from Sao Paulo, Brazil. *Pituitary*, **2021**, 24, 252-261	4.3	
435	Management of functioning pediatric adrenal tumors. *Journal of Pediatric Surgery*, **2021**, 56, 768-771	2.6	2
434	Real-World Estimates of Adrenal Insufficiency-Related Adverse Events in Children With Congenital Adrenal Hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, e192-e203	5.6	8
433	Insights from the genetic characterization of central precocious puberty associated with multiple anomalies. *Human Reproduction*, **2021**, 36, 506-518	5.7	6
432	Vasculometabolic effects in patients with congenital growth hormone deficiency with and without GH replacement therapy during adulthood. *Pituitary*, **2021**, 24, 216-228	4.3	3
431	Low Protein Expression of both and as Novel Negative Prognostic Markers of Adult Adrenocortical Carcinoma. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	6
430	SDHB large deletions are associated with absence of MIBG uptake in metastatic lesions of malignant paragangliomas. *Endocrine*, **2021**, 72, 586-590	4	1

429	Phosphodiesterase 2A and 3B variants are associated with primary aldosteronism. *Endocrine-Related Cancer*, **2021**, 28, 1-13	5.7	7
428	Performance of mutation pathogenicity prediction tools on missense variants associated with 46,XY differences of sex development. *Clinics*, **2021**, 76, e2052	2.3	2
427	The phenotypic spectrum associated with OTX2 mutations in humans. *European Journal of Endocrinology*, **2021**, 185, 121-135	6.5	3
426	Anthropometric, metabolic, and reproductive outcomes of patients with central precocious puberty treated with leuprorelin acetate 3-month depot (11.25´mg). *Journal of Pediatric Endocrinology and Metabolism*, **2021**, 34, 1371-1377	1.6	1
425	Adrenal crisis and mortality rate in adrenal insufficiency and congenital adrenal hyperplasia. *Archives of Endocrinology and Metabolism*, **2021**, 65, 488-494	2.2	1
424	High-throughput splicing assays identify missense and silent splice-disruptive POU1F1 variants underlying pituitary hormone deficiency. *American Journal of Human Genetics*, **2021**, 108, 1526-1539	11	5
423	WT1 Pathogenic Variants are Associated with a Broad Spectrum of Differences in Sex Development Phenotypes and Heterogeneous Progression of Renal Disease. *Sexual Development*, **2021**, 1-9	1.6	0
422	Variants in 46,XY DSD-Related Genes in Syndromic and Non-Syndromic Small for Gestational Age Children with Hypospadias. *Sexual Development*, **2021**, 1-7	1.6	0
421	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
420	Steroid Screening Tools Differentiating Nonclassical Congenital Adrenal Hyperplasia and Polycystic Ovary Syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	3
419	Long-term cardio-metabolic outcomes in patients with classical congenital adrenal hyperplasia: is the risk real?. *Current Opinion in Endocrinology, Diabetes and Obesity*, **2020**, 27, 155-161	4	8
418	XAF1 as a modifier of p53 function and cancer susceptibility. *Science Advances*, **2020**, 6, eaba3231	14.3	14
417	Allelic Variants of in Patients With Adrenal Incidentalomas and in Patients With Cushing's Syndrome Associated With Bilateral Adrenal Nodules. *Frontiers in Endocrinology*, **2020**, 11, 36	5.7	2
416	Adverse Outcomes and Economic Burden of Congenital Adrenal Hyperplasia Late Diagnosis in the Newborn Screening Absence. *Journal of the Endocrine Society*, **2020**, 4, bvz013	0.4	3
415	Sterol O-Acyl Transferase 1 as a Prognostic Marker of Adrenocortical Carcinoma. *Cancers*, **2020**, 12,	6.6	16
414	Genetics of Primary Ovarian Insufficiency in the Next-Generation Sequencing Era. *Journal of the Endocrine Society*, **2020**, 4, bvz037	0.4	25
413	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
412	SUN-725 Clinical and Genetic Features of Families with Maternally Inherited Central Precocious Puberty. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78

411	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
410	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. *PLoS ONE*, **2020**, 15, e0240795	3.7	7
409	Anorexia as the first clinical manifestation of von Hippel-Lindau syndrome. *Molecular and Clinical Oncology*, **2020**, 13, 65	1.6	
408	Anorexia as the first clinical manifestation of von Hippel-Lindau syndrome. *Molecular and Clinical Oncology*, **2020**, 13, 1-1	1.6	
407	Integrative and Analytical Review of the 5-Alpha-Reductase Type 2 Deficiency Worldwide. *The Application of Clinical Genetics*, **2020**, 13, 83-96	3.1	9
406	SELAdb: A database of exonic variants in a Brazilian population referred to a quaternary medical center in S^ B Paulo. *Clinics*, **2020**, 75, e1913	2.3	3
405	Adrenal Insufficiency and Glucocorticoid Use During the COVID-19 Pandemic. *Clinics*, **2020**, 75, e2022	2.3	10
404	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
403	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
402	OR15-04 Central Precocious Puberty without Central Nervous System Lesions: Is It Really Idiopathic?. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
401	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
400	SUN-178 Clinical and Anatomopathological Characteristics of Two Atypical Aldosterone-Producing Adenomas. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
399	Impact of schooling in the HIV/AIDS prevalence among Brazilian transgender women. *Archives of Endocrinology and Metabolism*, **2020**, 64, 369-373	2.2	1
398	Outcomes of Patients with Central Precocious Puberty Due to Loss-of-Function Mutations in the MKRN3 Gene after Treatment with Gonadotropin-Releasing Hormone Analog. *Neuroendocrinology*, **2020**, 110, 705-713	5.6	7
397	Clinical and Genetic Characterization of a Constitutional Delay of Growth and Puberty Cohort. *Neuroendocrinology*, **2020**, 110, 959-966	5.6	6
396	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
395	Comprehensive Genetic Analysis of 128 Candidate Genes in a Cohort With Idiopathic, Severe, or Familial Osteoporosis. *Journal of the Endocrine Society*, **2020**, 4, bvaa148	0.4	6
394	A New Insight into the Surgical Treatment of Primary Macronodular Adrenal Hyperplasia. *Journal of the Endocrine Society*, **2020**, 4, bvaa083	0.4	3

393	A Bayesian Approach to Diagnose Growth Hormone Deficiency in Children: Insulin-Like Growth Factor Type 1 Is Valuable for Screening and IGF-Binding Protein Type 3 for Confirmation. *Hormone Research in Paediatrics*, **2020**, 93, 197-205	3.3	3
392	Plasma Renin Measurements are Unrelated to Mineralocorticoid Replacement Dose in Patients With Primary Adrenal Insufficiency. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	15
391	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency **2020**, 15, e0240795		
390	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency **2020**, 15, e0240795		
389	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency **2020**, 15, e0240795		
388	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency **2020**, 15, e0240795		
387	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	5.6	24
386	New Insights Into Pheochromocytoma Surveillance of Young Patients With Missense Mutations. *Journal of the Endocrine Society*, **2019**, 3, 1682-1692	0.4	7
385	IGF-1 assessed by pubertal status has the best positive predictive power for GH deficiency diagnosis in peripubertal children. *Journal of Pediatric Endocrinology and Metabolism*, **2019**, 32, 173-179	1.6	16
384	Premature Pubarche due to Exogenous Testosterone Gel or Intense Diaper Rash Prevention Cream Use: A Case Series. *Hormone Research in Paediatrics*, **2019**, 91, 411-415	3.3	5
383	Persistent Poor Metabolic Profile in Postmenopausal Women With Ovarian Hyperandrogenism After Testosterone Level Normalization. *Journal of the Endocrine Society*, **2019**, 3, 1087-1096	0.4	7
382	Evaluation of SHOX defects in the era of next-generation sequencing. *Clinical Genetics*, **2019**, 96, 261-265	4	5
381	Exome Sequencing Reveals the POLR3H Gene as a Novel Cause of Primary Ovarian Insufficiency. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2827-2841	5.6	17
380	Androgen Biosynthetic Defects: 17-Hydroxysteroid Dehydrogenadse Type 3 and 5-Reductase Type 2 Deficiencies **2019**, 486-491		
379	DLK1 Is a Novel Link Between Reproduction and Metabolism. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2112-2120	5.6	44
378	Psychosexual Aspects, Effects of Prenatal Androgen Exposure, and Gender Change in 46,XY Disorders of Sex Development. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 1160-1170	5.6	13
377	Clinical spectrum of Li-Fraumeni syndrome/Li-Fraumeni-like syndrome in Brazilian individuals with the TP53 p.R337H mutation. *Journal of Steroid Biochemistry and Molecular Biology*, **2019**, 190, 250-255	5.1	14
376	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

375	Two rare loss-of-function variants in the STAG3 gene leading to primary ovarian insufficiency. *European Journal of Medical Genetics*, **2019**, 62, 186-189	2.6	23
374	Impact of Long-Term Dexamethasone Therapy on the Metabolic Profile of Patients With 21-Hydroxylase Deficiency. *Journal of the Endocrine Society*, **2019**, 3, 1574-1582	0.4	3
373	Combined pituitary hormone deficiency caused by PROP1 mutations: update 20 years post-discovery. *Archives of Endocrinology and Metabolism*, **2019**, 63, 167-174	2.2	12
372	Genetic Evidence of the Association of DEAH-Box Helicase 37 Defects With 46,XY Gonadal Dysgenesis Spectrum. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 5923-5934	5.6	14
371	Management of 46,XY Differences/Disorders of Sex Development (DSD) Throughout Life. *Endocrine Reviews*, **2019**, 40, 1547-1572	27.2	23
370	Genetic diagnosis of congenital hypopituitarism by a target gene panel: novel pathogenic variants in GLI2, OTX2 and GHRHR. *Endocrine Connections*, **2019**, 8, 590-595	3.5	6
369	ESR1 polymorphism (rs2234693) influences femoral bone mass in patients with Turner syndrome. *Endocrine Connections*, **2019**, 8, 1513-1519	3.5	3
368	New genetic findings in a large cohort of congenital hypogonadotropic hypogonadism. *European Journal of Endocrinology*, **2019**, 181, 103-119	6.5	41
367	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
366	OR04-6 Predictors of Clinical Outcome after Adrenalectomy for Unilateral Primary Aldosteronism. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78
365	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
364	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
363	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
362	Mobile DNA in Endocrinology: LINE-1 Retrotransposon Causing Partial Androgen Insensitivity Syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 6385-6390	5.6	5
361	Classic congenital adrenal hyperplasia and its impact on reproduction. *Fertility and Sterility*, **2019**, 111, 7-12	4.8	21
360	Mutations in MAP3K1 that cause 46,XY disorders of sex development disrupt distinct structural domains in the protein. *Human Molecular Genetics*, **2019**, 28, 1620-1628	5.6	12
359	A 46,XX testicular disorder of sex development caused by a Wilms' tumour Factor-1 (WT1) pathogenic variant. *Clinical Genetics*, **2019**, 95, 172-176	4	14
358	Long-term outcomes and molecular analysis of a large cohort of patients with 46,XY disorder of sex development due to partial gonadal dysgenesis. *Clinical Endocrinology*, **2018**, 89, 164-177	3.4	4

357	Partial androgen insensitivity syndrome due to somatic mosaicism of the androgen receptor. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 223-228	1.6	8
356	Pathogenic copy number variants in patients with congenital hypopituitarism associated with complex phenotypes. *Clinical Endocrinology*, **2018**, 88, 425-431	3.4	7
355	Spontaneous fertility in a male patient with testotoxicosis despite suppression of FSH levels. *Human Reproduction*, **2018**, 33, 914-918	5.7	2
354	Long-Term Outcomes of Patients with Central Precocious Puberty due to Hypothalamic Hamartoma after GnRHa Treatment: Anthropometric, Metabolic, and Reproductive Aspects. *Neuroendocrinology*, **2018**, 106, 203-210	5.6	10
353	The role of ARMC5 in human cell cultures from nodules of primary macronodular adrenocortical hyperplasia (PMAH). *Molecular and Cellular Endocrinology*, **2018**, 460, 36-46	4.4	22
352	Biallelic and monoallelic ESR2 variants associated with 46,XY disorders of sex development. *Genetics in Medicine*, **2018**, 20, 717-727	8.1	16
351	Identification of the first homozygous 1-bp deletion in GDF9 gene leading to primary ovarian insufficiency by using targeted massively parallel sequencing. *Clinical Genetics*, **2018**, 93, 408-411	4	20
350	A severe phenotype of Kennedy disease associated with a very large CAG repeat expansion. *Muscle and Nerve*, **2018**, 57, E95-E97	3.4	6
349	Androgen insensitivity syndrome: a review. *Archives of Endocrinology and Metabolism*, **2018**, 62, 227-235	2.2	64
348	Applicability of a novel mathematical model for the prediction of adult height and age at menarche in girls with idiopathic central precocious puberty. *Clinics*, **2018**, 73, e480	2.3	3
347	Low estrogen doses normalize testosterone and estradiol levels to the female range in transgender women. *Clinics*, **2018**, 73, e86	2.3	11
346	Assembling the jigsaw puzzle: CBX2 isoform 2 and its targets in disorders/differences of sex development. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 785-795	2.3	14
345	An update of genetic basis of PCOS pathogenesis. *Archives of Endocrinology and Metabolism*, **2018**, 62, 352-361	2.2	50
344	Methylome profiling of healthy and central precocious puberty girls. *Clinical Epigenetics*, **2018**, 10, 146	7.7	17
343	Primary malignant tumors of the adrenal glands. *Clinics*, **2018**, 73, e756s	2.3	15
342	Androgen receptor mRNA analysis from whole blood: a low-cost strategy for detection of androgen receptor gene splicing defects. *Clinical Genetics*, **2018**, 94, 489-490	4	1
341	Central Precocious Puberty Caused by a Heterozygous Deletion in the MKRN3 Promoter Region. *Neuroendocrinology*, **2018**, 107, 127-132	5.6	12
340	High Frequency of MKRN3 Mutations in Male Central Precocious Puberty Previously Classified as Idiopathic. *Neuroendocrinology*, **2017**, 105, 17-25	5.6	50

339	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
338	Persistent Mˆ Ullerian duct syndrome due to a mutation in the anti-Mˆ Ullerian hormone receptor gene (AMHR2). *Anales De Pediatr (English Edition)*, **2017**, 86, 94-95	0.4	
337	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
336	Discordant Genotypic Sex and Phenotype Variations in Two Spanish Siblings with 17β-Hydroxylase/17,20-Lyase Deficiency Carrying the Most Prevalent Mutated CYP17A1 Alleles of Brazilian Patients. *Sexual Development*, **2017**, 11, 70-77	1.6	5
335	Triple A Syndrome: Preliminary Response to the Antioxidant N-Acetylcysteine Treatment in a Child. *Hormone Research in Paediatrics*, **2017**, 88, 167-171	3.3	3
334	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	6.5	23
333	Paternally Inherited DLK1 Deletion Associated With Familial Central Precocious Puberty. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 1557-1567	5.6	88
332	Heterozygous Nonsense Mutation in the Androgen Receptor Gene Associated with Partial Androgen Insensitivity Syndrome in an Individual with 47,XXY Karyotype. *Sexual Development*, **2017**, 11, 78-81	1.6	4
331	Assessment of stress levels in girls with central precocious puberty before and during long-acting gonadotropin-releasing hormone agonist treatment: a pilot study. *Journal of Pediatric Endocrinology and Metabolism*, **2017**, 30, 657-662	1.6	8
330	A Novel Homozygous Missense FSHR Variant Associated with Hypergonadotropic Hypogonadism in Two Siblings from a Brazilian Family. *Sexual Development*, **2017**, 11, 137-142	1.6	21
329	Physiology of Male Gonadotropic Axis and Disorders of Sex Development **2017**, 75-96		
328	Neonatal 17-hydroxyprogesterone levels adjusted according to age at sample collection and birthweight improve the efficacy of congenital adrenal hyperplasia newborn screening. *Clinical Endocrinology*, **2017**, 86, 480-487	3.4	23
327	A novel homozygous 1-bp deletion in the NOBOX gene in two Brazilian sisters with primary ovarian failure. *Endocrine*, **2017**, 58, 442-447	4	12
326	Global Application of the Assessment of Communication Skills of Paediatric Endocrinology Fellows in the Management of Differences in Sex Development Using the ESPE E-Learning.Org Portal. *Hormone Research in Paediatrics*, **2017**, 88, 127-139	3.3	6
325	An activating mutation in the CRHR1 gene is rarely associated with pituitary-dependent hyperadrenocorticism in poodles. *Clinics*, **2017**, 72, 575-581	2.3	0
324	Growth hormone deficiency with advanced bone age: phenotypic interaction between GHRH receptor and CYP21A2 mutations diagnosed by sanger and whole exome sequencing. *Archives of Endocrinology and Metabolism*, **2017**, 61, 633-636	2.2	3
323	Adjuvant radiotherapy for the primary treatment of adrenocortical carcinoma: Are we offering the best?. *International Braz J Urol: Official Journal of the Brazilian Society of Urology*, **2017**, 43, 841-848	2	12
322	A recurrent synonymous mutation in the human androgen receptor gene causing complete androgen insensitivity syndrome. *Journal of Steroid Biochemistry and Molecular Biology*, **2017**, 174, 14-16	5.1	13

321	Molecular analysis of brazilian patients with combined pituitary hormone deficiency and orthotopic posterior pituitary lobe reveals eight different PROP1 alterations with three novel mutations. *Clinical Endocrinology*, **2017**, 87, 725-732	3.4	9
320	Non-coding variation in disorders of sex development. *Clinical Genetics*, **2017**, 91, 163-172	4	27
319	Differential Expression of Stem Cell Markers in Human Adamantinomatous Craniopharyngioma and Pituitary Adenoma. *Neuroendocrinology*, **2017**, 104, 183-193	5.6	16
318	Reprint of "Steroid 5‐reductase 2 deficiency". *Journal of Steroid Biochemistry and Molecular Biology*, **2017**, 165, 95-100	5.1	7
317	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
316	Successful Pregnancies After Adequate Hormonal Replacement in Patients With Combined Pituitary Hormone Deficiencies. *Journal of the Endocrine Society*, **2017**, 1, 1322-1330	0.4	10
315	Effects of Type 1 Insulin-Like Growth Factor Receptor Silencing in a Human Adrenocortical Cell Line. *Hormone and Metabolic Research*, **2016**, 48, 484-8	3.1	3
314	The Use of Three-dimensional Printers for Partial Adrenalectomy: Estimating the Resection Limits. *Urology*, **2016**, 90, 217-20	1.6	23
313	Successful Live Birth in a Woman With 17‐Hydroxylase Deficiency Through IVF Frozen-Thawed Embryo Transfer. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 345-8	5.6	28
312	High 18F-FDG uptake in PMAH correlated with normal expression of Glut1, HK1, HK2, and HK3. *Acta Radiologica*, **2016**, 57, 370-7	2	6
311	Clinical, hormonal, ovarian, and genetic aspects of 46,XX patients with congenital adrenal hyperplasia due to CYP17A1 defects. *Fertility and Sterility*, **2016**, 105, 1612-9	4.8	19
310	Malignant paraganglioma in children treated with embolization prior to surgical excision. *World Journal of Surgical Oncology*, **2016**, 14, 26	3.4	6
309	The Presence of Clitoromegaly in the Nonclassical Form of 21-Hydroxylase Deficiency Could Be Partially Modulated by the CAG Polymorphic Tract of the Androgen Receptor Gene. *PLoS ONE*, **2016**, 11, e0148548	3.7	8
308	Mutation analysis of NANOS3 in Brazilian women with primary ovarian failure. *Clinics*, **2016**, 71, 695-698	2.3	5
307	Long-term follow-up of a female with congenital adrenal hyperplasia due to P450-oxidoreductase deficiency. *Archives of Endocrinology and Metabolism*, **2016**, 60, 500-504	2.2	11
306	Long-term Results after CT-Guided Percutaneous Ethanol Ablation for the Treatment of Hyperfunctioning Adrenal Disorders. *Clinics*, **2016**, 71, 600-605	2.3	4
305	Surgical Treatment after Failed Primary Correction of Urogenital Sinus in Female Patients with Virilizing Congenital Adrenal Hyperplasia: Are Good Results Possible?. *Frontiers in Pediatrics*, **2016**, 4, 118	3.4	5
304	Steroid 5‐reductase 2 deficiency. *Journal of Steroid Biochemistry and Molecular Biology*, **2016**, 163, 206-11	5.1	49

303	HESX1 mutations in patients with congenital hypopituitarism: variable phenotypes with the same genotype. *Clinical Endocrinology*, **2016**, 85, 408-14	3.4	20
302	Negative correlation between tumour size and cortisol/ACTH ratios in patients with Cushing's disease harbouring microadenomas or macroadenomas. *Journal of Endocrinological Investigation*, **2016**, 39, 1401-1409	5.2	9
301	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
300	Comprehensive Pan-Genomic Characterization of Adrenocortical Carcinoma. *Cancer Cell*, **2016**, 29, 723-736	24.3	324
299	A homozygous point mutation in the GH1 promoter (c.-223C>T) leads to reduced GH1 expression in siblings with isolated GH deficiency (IGHD). *European Journal of Endocrinology*, **2016**, 175, K7-K15	6.5	4
298	A Single Nucleotide Variant in the Promoter Region of 17HSD Type 5 Gene Influences External Genitalia Virilization in Females with 21-Hydroxylase Deficiency. *Hormone Research in Paediatrics*, **2016**, 85, 333-8	3.3	4
297	Wide spectrum of NR5A1-related phenotypes in 46,XY and 46,XX individuals. *Birth Defects Research Part C: Embryo Today Reviews*, **2016**, 108, 309-320		28
296	Expression of LIN28 and its regulatory microRNAs in adult adrenocortical cancer. *Clinical Endocrinology*, **2015**, 82, 481-8	3.4	20
295	Frequent development of combined pituitary hormone deficiency in patients initially diagnosed as isolated growth hormone deficiency: a long term follow-up of patients from a single center. *Pituitary*, **2015**, 18, 561-7	4.3	23
294	Pitfalls in hormonal diagnosis of 17-beta hydroxysteroid dehydrogenase III deficiency. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 623-8	1.6	21
293	Disorders of sex development: effect of molecular diagnostics. *Nature Reviews Endocrinology*, **2015**, 11, 478-88	15.2	56
292	Genetics of primary macronodular adrenal hyperplasia. *Journal of Endocrinology*, **2015**, 224, R31-43	4.7	30
291	DAX1 Overexpression in Pediatric Adrenocortical Tumors: A Synergic Role with SF1 in Tumorigenesis. *Hormone and Metabolic Research*, **2015**, 47, 656-61	3.1	8
290	Role of GLI2 in hypopituitarism phenotype. *Journal of Molecular Endocrinology*, **2015**, 54, R141-50	4.5	43
289	FGFR1 and PROKR2 rare variants found in patients with combined pituitary hormone deficiencies. *Endocrine Connections*, **2015**, 4, 100-7	3.5	28
288	Quality of life in a large cohort of adult Brazilian patients with 46,XX and 46,XY disorders of sex development from a single tertiary centre. *Clinical Endocrinology*, **2015**, 82, 274-9	3.4	20
287	Quality of life of patients with 46,XX and 46,XY disorders of sex development. *Clinical Endocrinology*, **2015**, 82, 159-64	3.4	24
286	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

285	Low DICER1 expression is associated with poor clinical outcome in adrenocortical carcinoma. *Oncotarget*, **2015**, 6, 22724-33	3.3	15
284	Association Study of GWAS-Derived Loci with Height in Brazilian Children: Importance of MAP3K3, MMP24 and IGF1R Polymorphisms for Height Variation. *Hormone Research in Paediatrics*, **2015**, 84, 248-53	3.3	4
283	Validation of an immunoassay for anti-Mˆ Ilerian hormone measurements and reference intervals in healthy Brazilian subjects. *Annals of Clinical Biochemistry*, **2015**, 52, 67-75	2.2	3
282	The clinical, structural, and biological features of neovaginas: a comparison of the Frank and the McIndoe techniques. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2015**, 186, 12-6	2.4	5
281	46,XY DSD due to 17Hydroxysteroid Dehydrogenase 3 Deficiency **2014**, 191-197		
280	Autosomal recessive form of isolated growth hormone deficiency is more frequent than the autosomal dominant form in a Brazilian cohort. *Growth Hormone and IGF Research*, **2014**, 24, 180-6	2	5
279	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
278	Increased expression of ACTH (MC2R) and androgen (AR) receptors in giant bilateral myelolipomas from patients with congenital adrenal hyperplasia. *BMC Endocrine Disorders*, **2014**, 14, 42	3.3	14
277	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
276	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
275	ARMC5 mutations are a frequent cause of primary macronodular adrenal Hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1501-9	5.6	87
274	Clinical management of transsexual subjects. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2014**, 58, 188-96		14
273	Association of glucocorticoid receptor polymorphisms with clinical and metabolic profiles in polycystic ovary syndrome. *Clinics*, **2014**, 69, 179-84	2.3	6
272	Amplification of the insulin-like growth factor 1 receptor gene is a rare event in adrenocortical adenocarcinomas: searching for potential mechanisms of overexpression. *BioMed Research International*, **2014**, 2014, 936031	3	7
271	Influence of the A3669G Glucocorticoid Receptor Gene Polymorphism on the Metabolic Profile of Pediatric Patients with Congenital Adrenal Hyperplasia. *International Journal of Endocrinology*, **2014** , 2014, 594710	2.7	7
270	Homozygous inactivating mutation in NANOS3 in two sisters with primary ovarian insufficiency. *BioMed Research International*, **2014**, 2014, 787465	3	31
269	Genetic predictors of long-term response to growth hormone (GH) therapy in children with GH deficiency and Turner syndrome: the influence of a SOCS2 polymorphism. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1808-13	5.6	14
268	Gender assignment in patients with disorder of sex development. *Current Opinion in Endocrinology, Diabetes and Obesity*, **2014**, 21, 511-4	4	15

267	Molecular and gene network analysis of thyroid transcription factor 1 (TTF1) and enhanced at puberty (EAP1) genes in patients with GnRH-dependent pubertal disorders. *Hormone Research in Paediatrics*, **2013**, 80, 257-66	3.3	17
266	Relatively high frequency of non-synonymous GLI2 variants in patients with congenital hypopituitarism without holoprosencephaly. *Clinical Endocrinology*, **2013**, 78, 551-7	3.4	28
265	The benign spectrum of hypothalamic hamartomas: infrequent epilepsy and normal cognition in patients presenting with central precocious puberty. *Seizure: the Journal of the British Epilepsy Association*, **2013**, 22, 28-32	3.2	15
264	Analysis of the insulin-like growth factor 1 receptor gene in children born small for gestational age: in vitro characterization of a novel mutation (p.Arg511Trp). *Clinical Endocrinology*, **2013**, 78, 558-63	3.4	12
263	Combined use of multiplex ligation-dependent probe amplification and automatic sequencing for identification of KAL1 defects in patients with Kallmann syndrome. *Fertility and Sterility*, **2013**, 100, 854-9	4.8	7
262	Central precocious puberty caused by mutations in the imprinted gene MKRN3. *New England Journal of Medicine*, **2013**, 368, 2467-75	59.2	351
261	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
260	Obesity and familial predisposition are significant determining factors of an adverse metabolic profile in young patients with congenital adrenal hyperplasia. *Hormone Research in Paediatrics*, **2013**, 80, 111-8	3.3	16
259	CYP21A2 genotypes do not predict the severity of hyperandrogenic manifestations in the nonclassical form of congenital adrenal hyperplasia. *Hormone and Metabolic Research*, **2013**, 45, 301-7	3.1	15
258	The effect of fetal androgen metabolism-related gene variants on external genitalia virilization in congenital adrenal hyperplasia. *Clinical Genetics*, **2013**, 84, 482-8	4	13
257	Y chromosome aberration in a patient with cloacal-bladder exstrophy-epispadias complex: an unusual finding. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2013**, 57, 148-52		
256	Mineralocorticoid replacement during infancy for salt wasting congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Clinics*, **2013**, 68, 147-52	2.3	3
255	Modulatory effect of BclI GR gene polymorphisms on the obesity phenotype in Brazilian patients with Cushing's disease. *Clinics*, **2013**, 68, 579-85	2.3	7
254	PROP1 overexpression in corticotrophinomas: evidence for the role of PROP1 in the maintenance of cells committed to corticotrophic differentiation. *Clinics*, **2013**, 68, 887-91	2.3	8
253	Insulin-like growth factor 1 gene (CA)n repeats and a variable number of tandem repeats of the insulin gene in Brazilian children born small for gestational age. *Clinics*, **2013**, 68, 785-91	2.3	1
252	Absence of inactivating mutations and deletions in the DMRT1 and FGF9 genes in a large cohort of 46,XY patients with gonadal dysgenesis. *European Journal of Medical Genetics*, **2012**, 55, 690-4	2.6	8
251	Laparoscopic adrenalectomy in children. *Journal of Pediatric Urology*, **2012**, 8, 379-85	1.5	22
250	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

249	Impact of glucocorticoid receptor gene polymorphisms on the metabolic profile of adult patients with the classical form of 21-hydroxylase deficiency. *PLoS ONE*, **2012**, 7, e44893	3.7	24
248	Analysis of anti-Mˆ Ilerian hormone (AMH) and its receptor (AMHR2) genes in patients with persistent Mˆ Ilerian duct syndrome. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2012**, 56, 473-8		13
247	Novel mutation in the gonadotropin-releasing hormone receptor (GNRHR) gene in a patient with normosmic isolated hypogonadotropic hypogonadism. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2012**, 56, 540-4		5
246	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
245	Growth hormone pharmacogenetics: the interactive effect of a microsatellite in the IGF1 promoter region with the GHR-exon 3 and -202 A/C IGFBP3 variants on treatment outcomes of children with severe GH deficiency. *Pharmacogenomics Journal*, **2012**, 12, 439-45	3.5	18
244	The role of fibroblast growth factor receptor 4 overexpression and gene amplification as prognostic markers in pediatric and adult adrenocortical tumors. *Endocrine-Related Cancer*, **2012**, 19, L11-3	5.7	16
243	Post-receptor IGF1 insensitivity restricted to the MAPK pathway in a Silver-Russell syndrome patient with hypomethylation at the imprinting control region on chromosome 11. *European Journal of Endocrinology*, **2012**, 166, 543-50	6.5	7
242	DSD due to 5□eductase 2 deficiency - from diagnosis to long term outcome. *Seminars in Reproductive Medicine*, **2012**, 30, 427-31	1.4	41
241	The interactive effect of GHR-exon 3 and -202 A/C IGFBP3 polymorphisms on rhGH responsiveness and treatment outcomes in patients with Turner syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E671-7	5.6	13
240	Potential effects of alendronate on fibroblast growth factor 23 levels and effective control of hypercalciuria in an adult with Jansen's metaphyseal chondrodysplasia. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, 1098-103	5.6	18
239	Advice on the management of ambiguous genitalia to a young endocrinologist from experienced clinicians. *Seminars in Reproductive Medicine*, **2012**, 30, 339-50	1.4	18
238	Preface: disorders of sex development. *Seminars in Reproductive Medicine*, **2012**, 30, 337-8	1.4	
237	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
236	GH-releasing hormone receptor gene: a novel splice-disrupting mutation and study of founder effects. *Hormone Research in Paediatrics*, **2012**, 78, 165-72	3.3	16
235	Absence of functional LIN28B mutations in a large cohort of patients with idiopathic central precocious puberty. *Hormone Research in Paediatrics*, **2012**, 78, 144-50	3.3	28
234	Genotype analysis of the human endostatin variant p.D104N in benign and malignant adrenocortical tumors. *Clinics*, **2012**, 67, 95-8	2.3	2
233	Catch-Up Growth in Patients with Congenital or Acquired Growth Hormone Deficiency After GH Replacement: Clinical Features and Hypothalamic□Pituitary Imaging **2012**, 963-985		
232	Germline mutation analysis of Tpit in Poodle dogs with ACTH-dependent hyperadrenocorticism. *Arquivo Brasileiro De Medicina Veterinaria E Zootecnia*, **2012**, 64, 853-859	0.3	

231	Weight-adjusted neonatal 17OH-progesterone cutoff levels improve the efficiency of newborn screening for congenital adrenal hyperplasia. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2011**, 55, 632-7		20
230	Pharmacogenetics of glucocorticoid replacement could optimize the treatment of congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Clinics*, **2011**, 66, 1361-6	2.3	7
229	Possible role of a radiation-induced p53 mutation in a Nelson's syndrome patient with a fatal outcome. *Pituitary*, **2011**, 14, 400-4	4.3	14
228	A novel WT1 heterozygous nonsense mutation (p.K248X) causing a mild and slightly progressive nephropathy in a 46,XY patient with Denys-Drash syndrome. *Pediatric Nephrology*, **2011**, 26, 1311-5	3.2	8
227	Absence of GH-releasing hormone (GHRH) mutations in selected patients with isolated GH deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E1457-60	5.6	14
226	46,XY disorders of sex development (46,XY DSD) due to androgen receptor defects: androgen insensitivity syndrome. *Advances in Experimental Medicine and Biology*, **2011**, 707, 59-61	3.6	8
225	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
224	Mutational analysis of the necdin gene in patients with congenital isolated hypogonadotropic hypogonadism. *European Journal of Endocrinology*, **2011**, 165, 145-50	6.5	9
223	The role of SRY mutations in the etiology of gonadal dysgenesis in patients with 45,X/46,XY disorder of sex development and variants. *Hormone Research in Paediatrics*, **2011**, 75, 26-31	3.3	6
222	(18)F-FDG-PET/CT imaging of ACTH-independent macronodular adrenocortical hyperplasia (AIMAH) demonstrating increased (18)F-FDG uptake. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, 3300-1	5.6	23
221	46,XY DSD due to 17□HSD3 deficiency and 5□reductase type 2 deficiency. *Advances in Experimental Medicine and Biology*, **2011**, 707, 9-14	3.6	7
220	Clinical and molecular aspects of a pediatric metachronous adrenocortical tumor. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2011**, 55, 72-7		9
219	Frequency of genetic polymorphisms of PXR gene in the Brazilian population. *Clinics*, **2011**, 66, 1041-4	2.3	7
218	PROP1 and CTNNB1 expression in adamantinomatous craniopharyngiomas with or without □catenin mutations. *Clinics*, **2011**, 66, 1849-54	2.3	7
217	Glucose-dependent insulinotropic peptide receptor overexpression in adrenocortical hyperplasia in MEN1 syndrome without loss of heterozygosity at the 11q13 locus. *Clinics*, **2011**, 66, 529-33	2.3	5
216	MAMLD1 (mastermind-like domain containing 1) homozygous gain-of-function missense mutation causing 46,XX disorder of sex development in a virilized female. *Advances in Experimental Medicine and Biology*, **2011**, 707, 129-31	3.6	5
215	Screening of autosomal gene deletions in patients with hypogonadotropic hypogonadism using multiplex ligation-dependent probe amplification: detection of a hemizygosis for the fibroblast growth factor receptor 1. *Clinical Endocrinology*, **2010**, 72, 371-6	3.4	17
214	Isolated familial somatotropinoma: 11q13-loh and gene/protein expression analysis suggests a possible involvement of aip also in non-pituitary tumorigenesis. *Clinics*, **2010**, 65, 407-15	2.3	27

213	A novel GNAS mutation in an infant boy with pseudohypoparathyroidism type Ia and normal serum calcium and phosphate levels. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2010**, 54, 728-31		4
212	Influence of the fibroblast growth factor receptor 4 expression and the G388R functional polymorphism on Cushing's disease outcome. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, E271-9	5.6	12
211	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
210	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
209	Male hypogonadism: childhood diagnosis and future therapies. *Pediatric Health*, **2010**, 4, 539-555		2
208	Could the leukocyte x chromosome inactivation pattern be extrapolated to hair bulbs?. *Hormone Research in Paediatrics*, **2010**, 73, 238-43	3.3	3
207	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
206	Congenital hyperinsulinism in Brazilian neonates: a study of histology, KATP channel genes, and proliferation of □-cells. *Pediatric and Developmental Pathology*, **2010**, 13, 375-84	2.2	7
205	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
204	Effectiveness of the combined recombinant human growth hormone and gonadotropin-releasing hormone analog therapy in pubertal patients with short stature due to SHOX deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 328-32	5.6	29
203	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
202	Mutations of the KISS1 gene in disorders of puberty. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 2276-80	5.6	236
201	Molecular mechanisms of pituitary organogenesis: In search of novel regulatory genes. *Molecular and Cellular Endocrinology*, **2010**, 323, 4-19	4.4	122
200	Usefulness of MLPA in the detection of SHOX deletions. *European Journal of Medical Genetics*, **2010**, 53, 234-8	2.6	20
199	46,XY DSD due to impaired androgen production. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2010**, 24, 243-62	6.5	39
198	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
197	ACTH-Independent Cushing□ Syndrome: Adrenocortical Tumors **2010**, 189-208		
196	A Potential Role of Mevalonate Pathway in Adrenocortical Tumors. *FASEB Journal*, **2010**, 24, 421.1	0.9	

195	Analysis of craniofacial and extremity growth in patients with growth hormone deficiency during growth hormone therapy. *Hormone Research*, **2009**, 71, 173-7		9
194	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
193	Expression profiles of the glucose-dependent insulinotropic peptide receptor and LHCGR in sporadic adrenocortical tumors. *Journal of Endocrinology*, **2009**, 200, 167-75	4.7	3
192	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
191	Novel mutations in CYP11B1 gene leading to 11 beta-hydroxylase deficiency in Brazilian patients. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 3481-5	5.6	21
190	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
189	46,XY disorders of sex development (DSD). *Clinical Endocrinology*, **2009**, 70, 173-187	3.4	18
188	Molecular analysis of CYP21A2 can optimize the follow-up of positive results in newborn screening for congenital adrenal hyperplasia. *Clinical Genetics*, **2009**, 76, 503-10	4	22
187	Comparison between weight-based and IGF-I-based growth hormone (GH) dosing in the treatment of children with GH deficiency and influence of exon 3 deleted GH receptor variant. *Growth Hormone and IGF Research*, **2009**, 19, 179-86	2	14
186	46,XY disorders of sex development (DSD). *Clinical Endocrinology*, **2009**, 70, 173-87	3.4	63
185	Analysis of glucose-dependent insulinotropic peptide receptor (GIPR) and luteinizing hormone receptor (LHCGR) expression in human adrenocortical hyperplasia. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2009**, 53, 326-31		2
184	Long-term treatment of familial male-limited precocious puberty (testotoxicosis) with cyproterone acetate or ketoconazole. *Clinical Endocrinology*, **2008**, 69, 93-8	3.4	35
183	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
182	Analysis of the PTPN11 gene in idiopathic short stature children and Noonan syndrome patients. *Clinical Endocrinology*, **2008**, 69, 426-31	3.4	7
181	Y-chromosomal STR haplotypes in a sample from Sˆ õ Paulo (Brazil). *Forensic Science International: Genetics Supplement Series*, **2008**, 1, 248-249	0.5	2
180	A GPR54-activating mutation in a patient with central precocious puberty. *New England Journal of Medicine*, **2008**, 358, 709-15	59.2	415
179	High prevalence of pituitary magnetic resonance abnormalities and gene mutations in a cohort of Brazilian children with growth hormone deficiency and response to treatment. *Journal of Pediatric Endocrinology and Metabolism*, **2008**, 21, 673-80	1.6	8
178	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

177	Effects of long-term storage of filter paper blood samples on neonatal thyroid stimulating hormone, thyroxin and 17-alpha-hydroxyprogesterone measurements. *Journal of Medical Screening*, **2008**, 15, 109-11	1.4	16
176	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
175	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
174	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
173	Update on the etiology, diagnosis and therapeutic management of sexual precocity. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2008**, 52, 18-31		52
172	Combined 17 alpha-hydroxylase/17,20-lyase deficiency due to a homozygous 25 BP duplication (NT 4157-4181) at exon 5 in the CYP17 resulting in a premature stop codon predicted by molecular modeling. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2008**, 52, 1317-20		4
171	Cryptic intragenic deletion of the SHOX gene in a family with Lˆ Ĥi-Weill dyschondrosteosis detected by Multiplex Ligation-Dependent Probe Amplification (MLPA). *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2008**, 52, 1382-7		7
170	Tall stature and poor breast development after estrogen replacement in a hypergonadotrophic hypogonadic patient with a 45,X/46,X,der(X) karyotype with SHOX gene overdosage. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2008**, 52, 1282-7		6
169	Autosomal dominant familial neurohypophyseal diabetes insipidus caused by a novel mutation in arginine-vasopressin gene in a Brazilian family. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2008**, 52, 1272-6		6
168	The degree of external genitalia virilization in girls with 21-hydroxylase deficiency appears to be influenced by the CAG repeats in the androgen receptor gene. *Clinical Endocrinology*, **2008**, 68, 226-32	3.4	14
167	Population and mutation analysis of Y-STR loci in a sample from the city of Sˆ õ Paulo (Brazil). *Genetics and Molecular Biology*, **2008**, 31, 651-656	2	3
166	Exon 3-deleted genotype of growth hormone receptor (GHRd3) positively influences IGF-1 increase at generation test in children with idiopathic short stature. *Clinical Endocrinology*, **2007**, 67, 500-4	3.4	8
165	Y-STRs in forensic medicine: DNA analysis in semen samples of azoospermic individuals. *Journal of Forensic Sciences*, **2007**, 52, 664-70	1.8	13
164	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
163	Polymorphisms identified in the upstream core polyadenylation signal of IGF1 gene exon 6 do not cause pre- and postnatal growth impairment. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 4889-92	5.6	20
162	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
161	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
160	An inhibin B and estrogen-secreting adrenocortical carcinoma leading to selective FSH suppression. *Hormone Research in Paediatrics*, **2007**, 67, 7-11	3.3	9

159	Molecular analysis of the neuropeptide Y1 receptor gene in human idiopathic gonadotropin-dependent precocious puberty and isolated hypogonadotropic hypogonadism. *Fertility and Sterility*, **2007**, 87, 627-34	4.8	15
158	Hormonal, pituitary magnetic resonance, LHX4 and HESX1 evaluation in patients with hypopituitarism and ectopic posterior pituitary lobe. *Clinical Endocrinology*, **2007**, 66, 95-102	3.4	18
157	SHOX mutations in idiopathic short stature and Leri-Weill dyschondrosteosis: frequency and phenotypic variability. *Clinical Endocrinology*, **2007**, 66, 130-5	3.4	47
156	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
155	Report of a del22q11 in a patient with Mayer-Rokitansky-Kˆ 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
154	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
153	Allelic variants of the gamma-aminobutyric acid-A receptor alpha1-subunit gene (GABRA1) are not associated with idiopathic gonadotropin-dependent precocious puberty in girls with and without electroencephalographic abnormalities. *Journal of Clinical Endocrinology and Metabolism*, **2006**, 91, 2432-6	5.6	14
152	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
151	Ectopic ACTH syndrome: our experience with 25 cases. *European Journal of Endocrinology*, **2006**, 155, 725-33	6.5	98
150	Reproductive outcome of women with 21-hydroxylase-deficient nonclassic adrenal hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2006**, 91, 3451-6	5.6	113
149	Some aspects of the behavior of the hypothalamus-pituitary-adrenal axis in patients with uncomplicated Plasmodium falciparum malaria: Cortisol and dehydroepiandrosterone levels. *Acta Tropica*, **2006**, 98, 270-6	3.2	21
148	Effectiveness of treating ovarian hyperstimulation syndrome with cabergoline in two patients with gonadotropin-producing pituitary adenomas. *Fertility and Sterility*, **2006**, 86, 719.e15-8	4.8	29
147	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
146	Combined pituitary hormone deficiency (CPHD) due to a complete PROP1 deletion. *Clinical Endocrinology*, **2006**, 65, 294-300	3.4	33
145	A single somatic activating Asp578His mutation of the luteinizing hormone receptor causes Leydig cell tumour in boys with gonadotropin-independent precocious puberty. *Clinical Endocrinology*, **2006**, 65, 408-10	3.4	18
144	Preclinical diagnosis of testotoxicosis in a boy with an activating mutation of the luteinizing hormone receptor. *Journal of Pediatric Endocrinology and Metabolism*, **2006**, 19, 541-4	1.6	7
143	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
142	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

141	High degree of discordance between three-dimensional and two-dimensional lumbar spine bone mineral density in Turner's syndrome. *Journal of Clinical Densitometry*, **2005**, 8, 461-6	3.5	15
140	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
139	Craniofacial features with growth hormone treatment. *Journal of Pediatrics*, **2005**, 146, 295; author reply 295-6	3.6	1
138	The laparoscopic management of intersex patients: the preferred approach. *BJU International*, **2005**, 95, 863-7	5.6	29
137	Substitutions in the CYP21A2 promoter explain the simple-virilizing form of 21-hydroxylase deficiency in patients harbouring a P30L mutation. *Clinical Endocrinology*, **2005**, 62, 132-6	3.4	29
136	Letter re: FSH receptor polymorphisms and iatrogenic ovarian hyperstimulation. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 4978; author reply 4978-9	5.6	1
135	Letter re: FSH Receptor Polymorphisms and Iatrogenic Ovarian Hyperstimulation. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 4978-4978	5.6	6
134	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
133	Deletion mapping of chromosome 17 in benign and malignant adrenocortical tumors associated with the Arg337His mutation of the p53 tumor suppressor protein. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 2976-81	5.6	27
132	Frequency of the allelic variant (Trp8Arg/Ile15Thr) of the luteinizing hormone gene in a Brazilian cohort of healthy subjects and in patients with hypogonadotropic hypogonadism. *Clinics*, **2005**, 60, 461-4	2.3	4
131	Use of nonradioactive labeling to detect large gene rearrangements in 21-hydroxylase deficiency. *Revista Do Hospital Das Clinicas*, **2004**, 59, 369-74		
130	Clinical and molecular analysis of human reproductive disorders in Brazilian patients. *Brazilian Journal of Medical and Biological Research*, **2004**, 37, 137-44	2.8	4
129	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
128	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
127	A meiotic recombination in a new isolated familial somatotropinoma kindred. *European Journal of Endocrinology*, **2004**, 150, 643-8	6.5	26
126	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
125	Inhibin alpha-subunit (INHA) gene and locus changes in paediatric adrenocortical tumours from TP53 R337H mutation heterozygote carriers. *Journal of Medical Genetics*, **2004**, 41, 354-9	5.8	19
124	Surgery for adrenal tumours with thrombus in the supra-diaphragmatic infra-atrial inferior vena cava, with no cardiopulmonary bypass. *BJU International*, **2004**, 94, 70-3	5.6	8

123	The first homozygous mutation (S226I) in the highly-conserved WSXWS-like motif of the GH receptor causing Laron syndrome: supression of GH secretion by GnRH analogue therapy not restored by dihydrotestosterone administration. *Clinical Endocrinology*, **2004**, 60, 36-40	3.4	14
122	Mutant P450 oxidoreductase causes disordered steroidogenesis with and without Antley-Bixler syndrome. *Nature Genetics*, **2004**, 36, 228-30	36.3	396
121	Pituitary apoplexy during therapy with cabergoline in an adolescent male with prolactin-secreting macroadenoma. *Pituitary*, **2004**, 7, 83-7	4.3	48
120	Assessment of the role of transcript for GATA-4 as a marker of unfavorable outcome in human adrenocortical neoplasms. *BMC Endocrine Disorders*, **2004**, 4, 3	3.3	12
119	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
118	Adrenal, supra-renal. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2004**, 48, 579-579		
117	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
116	Male Pseudohermaphroditism Due to 5□Reductase 2 Deficiency: Outcome of a Brazilian Cohort **2003**, 13, 201-204		22
115	Authors□Response: Pituitary Magnetic Resonance Imaging in Idiopathic and Genetic Growth Hormone Deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 1911-1912	5.6	4
114	Maternal isodisomy causing homozygosity for a dominant activating mutation of the luteinizing hormone receptor gene in a boy with familial male-limited precocious puberty. *Clinical Endocrinology*, **2003**, 59, 533-4	3.4	2
113	Acromegalic features in growth hormone (GH)-deficient patients after long-term GH therapy. *Clinical Endocrinology*, **2003**, 59, 788-92	3.4	12
112	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
111	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
110	GH values after clonidine stimulation measured by immunofluorometric assay in normal prepubertal children and GH-deficient patients. *Hormone Research in Paediatrics*, **2003**, 59, 229-33	3.3	26
109	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
108	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
107	Gen^ tica molecular do eixo hipot^ □amo-hip^ fise-gonadal. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2003**, 47, 440-452		
106	NON-RADIOMETRIC IMMUNOASSAYS [FLUOROIMMUNOASSAY (FIA) AND FLUOROMETRIC ENZYME IMMUNOASSAY (FEIA)] WITH RADIOIMMUNOASSAY (RIA) FOR EVALUATION OF ADRENAL FUNCTION IN NORMAL AND HYPERCORTISOLEMIC DOGS. *Ciencia Rural*, **2002**, 32, 259-262	1.3	3

105	Aspectos Moleculares da Determina^ ▯▯o e Diferencia^ ▯▯o Sexual. *Arquivos Brasileiros De Endocrinologia E Metabologia*, **2002**, 46, 433-443		4
104	Adrenocortical tumors: results of treatment and study of Weiss's score as a prognostic factor. *Revista Do Hospital Das Clinicas*, **2002**, 57, 251-6		24
103	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
102	Reply to correspondence from Hall▯Detection of Y-specific sequences in patients with Turner syndrome▯*American Journal of Medical Genetics Part A*, **2002**, 113, 115-115		2
101	Zinc supplementation does not inhibit basal and metoclopramide-stimulated prolactinemia secretion in healthy men. *Journal of Trace Elements in Medicine and Biology*, **2002**, 16, 69-73	4.1	
100	Glucocorticoid receptor gene polymorphisms in ACTH-secreting pituitary tumours. *Clinical Endocrinology*, **2002**, 57, 657-62	3.4	23
99	Severe and mild neonatal hypothyroidism mediate opposite effects on Leydig cells of rats. *Thyroid*, **2002**, 12, 13-8	6.2	19
98	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
97	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
96	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
95	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
94	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
93	Menstrual disturbances in patients with systemic lupus erythematosus without alkylating therapy: clinical, hormonal and therapeutic associations. *Lupus*, **2002**, 11, 175-80	2.6	60
92	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
91	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
90	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
89	Nongenetic male pseudohermaphroditism and reduced prenatal growth. *New England Journal of Medicine*, **2001**, 345, 1135	59.2	9
88	Absence of mutations involving the LIM homeobox domain gene LHX9 in 46,XY gonadal agenesis and dysgenesis. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 2465-9	5.6	29

87	Premature thelarche in girls after growth hormone therapy. *Journal of Pediatrics*, **2001**, 138, 448-9	3.6	6
86	Laparoscopic management of intersexual states. *Urologic Clinics of North America*, **2001**, 28, 31-42	2.9	13
85	Normal bone density in male pseudohermaphroditism due to 5alpha- reductase 2 deficiency. *Revista Do Hospital Das Clinicas*, **2001**, 56, 139-42		9
84	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
83	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
82	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
81	The effect of distinct activating mutations of the luteinizing hormone receptor gene on the pituitary-gonadal axis in both sexes. *Clinical Endocrinology*, **2000**, 53, 609-13	3.4	26
80	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
79	21-Hydroxylase deficiency in Brazil. *Brazilian Journal of Medical and Biological Research*, **2000**, 33, 1211-6	2.8	3
78	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
77	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
76	Phenotypic variability and origins of mutations in the gene encoding 3beta-hydroxysteroid dehydrogenase type II. *Journal of Molecular Endocrinology*, **2000**, 24, 75-82	4.5	16
75	Genotyping of the type II 3beta-hydroxysteroid dehydrogenase gene (HSD3B2) in women with hirsutism and elevated ACTH-stimulated delta(5)-steroids. *Fertility and Sterility*, **2000**, 74, 553-7	4.8	11
74	No evidence of somatic activating mutations on gonadotropin receptor genes in sex cord stromal tumors. *Fertility and Sterility*, **2000**, 74, 992-5	4.8	28
73	Mutation analysis of the follicle-stimulating hormone receptor gene in girls with gonadotropin-independent precocious puberty resulting from autonomous cystic ovaries. *Fertility and Sterility*, **2000**, 73, 280-3	4.8	15
72	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
71	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
70	A novel missense mutation, GLY424SER, in Brazilian patients with 21-hydroxylase deficiency. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 2870-2	5.6	28

69	17Beta-hydroxysteroid dehydrogenase 3 deficiency in women. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 802-4	5.6	30
68	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
67	Clinical features of women with resistance to luteinizing hormone. *Clinical Endocrinology*, **1999**, 51, 701-7	3.4	18
66	Effect of zinc administration on thyrotropin releasing hormone-stimulated prolactinemia in healthy men. *BioMetals*, **1999**, 12, 347-52	3.4	8
65	A novel point mutation (R840S) in the androgen receptor in a Brazilian family with partial androgen insensitivity syndrome. *Human Mutation*, **1999**, 14, 353	4.7	9
64	Menstrual disorders and infertility caused by inactivating mutations of the luteinizing hormone receptor gene. *Fertility and Sterility*, **1999**, 71, 597-601	4.8	24
63	Low frequency of CYP2B deletions in Brazilian patients with congenital adrenal hyperplasia due to 21-hydroxylas deficiency. *Human Heredity*, **1999**, 49, 9-14	1.1	25
62	Diagnostic Value of Fluorometric Assays in the Evaluation of Precocious Puberty. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 3539-3544	5.6	84
61	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
60	17´-Hydroxysteroid Dehydrogenase 3 Deficiency in Women. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 802-804	5.6	35
59	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
58	A Novel Missense Mutation, GLY424SER, in Brazilian Patients with 21-Hydroxylase Deficiency. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 2870-2872	5.6	24
57	A novel homozygous nonsense mutations E135* in the type II 3beta-hydroxysteroid dehydrogenase gene in a girl with salt-losing congenital adrenal hyperplasia. Mutations in brief no. 168. Online. *Human Mutation*, **1998**, 12, 139	4.7	7
56	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
55	A study of patients with Nelson's syndrome. *Clinical Endocrinology*, **1998**, 49, 533-9	3.4	60
54	Effects of acute and chronic zinc administration on growth velocity in patients with hypopituitarism. *Nutrition Research*, **1998**, 18, 1865-1877	4	2
53	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
52	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

51	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
50	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
49	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
48	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
47	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
46	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
45	A novel homozygous nonsense mutation E135* in the type II 3□hydroxysteroid dehydrogenase gene in a girl with salt-losing congenital adrenal hyperplasia **1998**, 12, 139		1
44	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
43	Apparent mineralocorticoid excess in a Brazilian kindred: hypertension in the heterozygote state. *Journal of Hypertension*, **1997**, 15, 1397-402	1.9	54
42	Management of ambiguous genitalia in pseudohermaphrodites: new perspectives on vaginal dilation. *Fertility and Sterility*, **1997**, 67, 229-32	4.8	64
41	Ovarian resistance to luteinizing hormone: a novel cause of amenorrhea and infertility. *Fertility and Sterility*, **1997**, 67, 394-7	4.8	17
40	Pheochromocytoma: study of 50 cases. *Journal of Urology*, **1997**, 157, 1208-12	2.5	77
39	The genetic and functional basis of isolated 17,20-lyase deficiency. *Nature Genetics*, **1997**, 17, 201-5	36.3	277
38	Growth Hormone Receptor Messenger Ribonucleic Acid in Normal and Pathologic Human Adrenocortical Tissues--An Analysis by Quantitative Polymerase Chain Reaction Technique. *Journal of Clinical Endocrinology and Metabolism*, **1997**, 82, 2671-2676	5.6	7
37	Male pseudohermaphroditism due to steroid 5alpha-reductase 2 deficiency. Diagnosis, psychological evaluation, and management. *Medicine (United States)*, **1996**, 75, 64-76	1.8	94
36	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
35	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
34	Molecular genetics and pathophysiology of 17 beta-hydroxysteroid dehydrogenase 3 deficiency. *Journal of Clinical Endocrinology and Metabolism*, **1996**, 81, 130-136	5.6	142

33	Cortisol and adrenocorticotropin response to desmopressin in women with Cushing's disease compared with depressive illness. *Journal of Clinical Endocrinology and Metabolism*, **1996**, 81, 2233-2237	5.6	27
32	Selection of adrenal tumor cells in culture demonstrated by interphase cytogenetics. *Cancer Genetics and Cytogenetics*, **1995**, 79, 36-40		13
31	Endocrine interaction between zinc and prolactin. An interpretative review. *Biological Trace Element Research*, **1995**, 49, 139-49	4.5	16
30	Failure of Partial Hypophysectomy as Definitive Treatment in Cushing's Disease Owing to Nodular Corticotrope Hyperplasia: Report of Four Cases. *Endocrine Pathology*, **1995**, 6, 57-66	4.2	1
29	The essential role of zinc in growth. *Nutrition Research*, **1995**, 15, 335-358	4	140
28	Ectopic ACTH syndrome. *Journal of Steroid Biochemistry and Molecular Biology*, **1995**, 53, 139-51	5.1	60
27	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
26	Human androgen insensitivity due to point mutations encoding amino acid substitutions in the androgen receptor steroid-binding domain. *Human Mutation*, **1995**, 6, 152-62	4.7	10
25	Women with steroid 5 alpha-reductase 2 deficiency have normal concentrations of plasma 5 alpha-dihydroprogesterone during the luteal phase. *Journal of Clinical Endocrinology and Metabolism*, **1995**, 80, 3136-3139	5.6	24
24	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
23	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
22	Ectopic adrenocorticotropic hormone syndrome. *Endocrine Reviews*, **1994**, 15, 752-87	27.2	175
21	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
20	Male pseudohermaphroditism caused by mutations of testicular 17 beta-hydroxysteroid dehydrogenase 3. *Nature Genetics*, **1994**, 7, 34-9	36.3	482
19	Natural mutagenesis study of the human steroid 5 alpha-reductase 2 isozyme. *Biochemistry*, **1994**, 33, 1265-70	3.2	141
18	Ectopic adrenocorticotropic hormone syndrome [published erratum appears in Endocr Rev 1995 Aug;16(4):507] **1994**, 15, 752-787		22
17	Calcium-dependent protein kinase-C activity in human adrenocortical neoplasms, hyperplastic adrenals, and normal adrenocortical tissue. *Journal of Clinical Endocrinology and Metabolism*, **1994**, 79, 736-739	5.6	7
16	Long-term treatment of central precocious puberty with a long-acting analogue of luteinizing hormone release hormone (D-Tryp6-GnRH) in monthly injections. Its possible use in normal puberty. *Hormone and Metabolic Research*, **1993**, 25, 105-9	3.1	2

15	Adrenal autografts following bilateral adrenalectomy. *Journal of Urology*, **1993**, 149, 977-9	2.5	17
14	The desmopressin stimulation test in the differential diagnosis of Cushing's syndrome. *Clinical Endocrinology*, **1993**, 38, 463-72	3.4	116
13	Molecular genetics of steroid 5 alpha-reductase 2 deficiency. *Journal of Clinical Investigation*, **1992**, 90, 799-809	15.9	284
12	Zinc acutely and temporarily inhibits adrenal cortisol secretion in humans. A preliminary report. *Biological Trace Element Research*, **1990**, 24, 83-9	4.5	18
11	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
10	Somatomedin levels in the diagnosis and therapy of growth hormone deficiency. *Hormone and Metabolic Research*, **1988**, 20, 175-81	3.1	2
9	Normal expression of the serologically defined H-Y antigen in Leydig cell hypoplasia. *Journal of Urology*, **1988**, 140, 1549-52	2.5	8
8	Low-dose oral clonidine: effective growth hormone releasing agent in children but not in adolescents. *Journal of Pediatrics*, **1987**, 111, 564-7	3.6	14
7	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
6	A clinico-genetic investigation of Leydig cell hypoplasia. *American Journal of Medical Genetics Part A* , **1987**, 26, 337-44		44
5	Spironolactone-reversible rickets associated with 11 beta-hydroxysteroid dehydrogenase deficiency syndrome. *Journal of Pediatrics*, **1986**, 109, 989-93	3.6	33
4	Male pseudohermaphroditism resulting from Leydig cell hypoplasia. *Journal of Pediatrics*, **1985**, 106, 1057	3.6	19
3	Variable ACTH-Stimulated 17-Hydroxyprogesterone Values in 21-Hydroxylase Deficiency Carriers Are Not Related to the Different CYP21 Gene Mutations		7
2	DIFEREN ˆ AS NO DESENVOLVIMENTO SEXUAL: UM ESTUDO DE DIVULGA ˆ D DA CI ˆ NCIA EM UM HOSPITAL. *Ensaio Pesquisa Em Educa Em Ci cias*,22,	2	0
1	The Use of Genetics for Reaching a Diagnosis in XY DSD. *Sexual Development*,1-18	1.6	1