AndrÃ©a Maciel-Guerra

List of Publications by Year
in descending order

99 papers	1,840 citations	331670 21 h-index	315739 38 g-index
107 all docs	107 docs citations	107 times ranked	2053 citing authors

#	Article	IF	Citations
1	Mutations inNR5A1Associated with Ovarian Insufficiency. New England Journal of Medicine, 2009, 360, 1200-1210.	27.0	339
2	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. American Journal of Human Genetics, 2003, 73, 1452-1458.	6.2	269
3	Mutation update for theNR5A1gene involved in DSD and infertility. Human Mutation, 2020, 41, 58-68.	2.5	52
4	Classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency: a cross-sectional study of factors involved in bone mineral density. Journal of Bone and Mineral Metabolism, 2003, 21, 396-401.	2.7	46
5	XX Maleness and XX True Hermaphroditism inSRY-Negative Monozygotic Twins: Additional Evidence for a Common Origin. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 339-343.	3.6	44
6	Complete gonadal dysgenesis in clinical practice: the 46,XY karyotype accounts for more than one third of cases. Fertility and Sterility, 2011, 96, 1431-1434.	1.0	42
7	Homozygous Inactivating Mutation inNANOS3in Two Sisters with Primary Ovarian Insufficiency. BioMed Research International, 2014, 2014, 1-8.	1.9	36
8	Deafness resulting from mutations in the GJB2 (connexin 26) gene in Brazilian patients. Clinical Genetics, 2002, 61, 354-358.	2.0	35
9	Bone mineralization in Turner syndrome: a transverse study of the determinant factors in 58 patients. Journal of Bone and Mineral Metabolism, 2002, 20, 294-297.	2.7	32
10	Determination of the frequency of the 35delG allele in Brazilian neonates. Clinical Genetics, 2000, 58, 339-340.	2.0	29
11	Clinical and molecular spectrum of patients with 17Î²-hydroxysteroid dehydrogenase type 3 (17-Î²-HSD3) deficiency. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 533-539.	1.3	29
12	Clinical but Not Histological Outcomes in Males With 45,X/46,XY Mosaicism Vary Depending on Reason for Diagnosis. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4366-4381.	3.6	27
13	A Novel Homozygous Missense <i>FSHR</i> Variant Associated with Hypergonadotropic Hypogonadism in Two Siblings from a Brazilian Family. Sexual Development, 2017, 11, 137-142.	2.0	26
14	Turner's Syndrome and Thyroid Disease: A Transverse Study of Pediatric Patients in Brazil. Journal of Pediatric Endocrinology and Metabolism, 2000, 13, 357-62.	0.9	25
15	Clinical and Laboratorial Features That May Differentiate 46,XY DSD due to Partial Androgen Insensitivity and 5Î±-Reductase Type 2 Deficiency. International Journal of Endocrinology, 2012, 2012, 1-7.	1.5	25
16	The novel p.Cys65Tyr mutation in NR5A1gene in three 46,XY siblings with normal testosterone levels and their mother with primary ovarian insufficiency. BMC Medical Genetics, 2014, 15, 7.	2.1	25
17	Morphometry and Histology of Gonads From 13 Children With Dysgenetic Male Pseudohermaphroditism. Archives of Pathology and Laboratory Medicine, 2001, 125, 652-656.	2.5	25
18	True agonadism: Report of a case analyzed with Y-specific DNA probes. American Journal of Medical Genetics Part A, 1991, 41, 444-445.	2.4	23

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19	408 Cases of Genital Ambiguity Followed by Single Multidisciplinary Team during 23 Years: Etiologic Diagnosis and Sex of Rearing. International Journal of Endocrinology, 2016, 2016, 1-9.	1.5	23
20	New approach to phenotypic variability and karyotype-phenotype correlation in Turner syndrome. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 475-9.	0.9	23
21	Analysis of anti-MÃ¼llerian hormone (AMH) and its receptor (AMHR2) genes in patients with persistent MÃ¼llerian duct syndrome. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 473-478.	1.3	21
22	Long-Term Follow-Up of Patients with 46,XY Partial Gonadal Dysgenesis Reared as Males. International Journal of Endocrinology, 2014, 2014, 1-8.	1.5	21
23	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. PLoS ONE, 2020, 15, e0240795.	2.5	21
24	46,XY and 45,X/46,XY testicular dysgenesis: similar gonadal and genital phenotype, different prognosis. Arquivos Brasileiros De Endocrinologia E Metabologia, 2010, 54, 331-334.	1.3	20
25	NPHS2 mutations account for only 15Â% of nephrotic syndrome cases. BMC Medical Genetics, 2015, 16, 88.	2.1	18
26	<i>NR5A1</i> Loss-of-Function Mutations Lead to 46,XY Partial Gonadal Dysgenesis Phenotype: Report of Three Novel Mutations. Sexual Development, 2016, 10, 191-199.	2.0	18
27	Imaging Techniques in the Diagnostic Journey of Disorders of Sex Development. Sexual Development, 2018, 12, 95-99.	2.0	18
28	Mutations in SRY and WT1 genes required for gonadal development are not responsible for XY partial gonadal dysgenesis. Brazilian Journal of Medical and Biological Research, 2005, 38, 17-25.	1.5	17
29	Screening of Y chromosome microdeletions in 46,XY partial gonadal dysgenesis and in patients with a 45,X/46,XY karyotype or its variants. BMC Medical Genetics, 2013, 14, 115.	2.1	17
30	Spontaneous puberty in girls with early diagnosis of Turner syndrome. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 653-657.	1.3	17
31	Clinical and genetic findings of five patients with WT1-related disorders. Arquivos Brasileiros De Endocrinologia E Metabologia, 2008, 52, 1236-1243.	1.3	16
32	O pediatra frente a uma crianÃ§a com ambigÃ¼idade genital. Jornal De Pediatria, 2007, 83, S184-S191.	2.0	15
33	Novel DMRT1 3'UTR+11insT mutation associated to XY partial gonadal dysgenesis. Arquivos Brasileiros De Endocrinologia E Metabologia, 2010, 54, 749-753.	1.3	14
34	Multifunctional role of steroidogenic factor 1 and disorders of sex development. Arquivos Brasileiros De Endocrinologia E Metabologia, 2011, 55, 607-612.	1.3	14
35	Frasier syndrome: four new cases with unusual presentations. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 525-532.	1.3	14
36	Idiopathic male pseudohermaphroditism is associated with prenatal growth retardation. European Journal of Pediatrics, 2005, 164, 287-291.	2.7	13

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37	Diagnosis of 5Î±-Reductase Type 2 Deficiency: Contribution of Anti-MÃ¼llerian Hormone Evaluation. Journal of Pediatric Endocrinology and Metabolism, 2005, 18, 1383-9.	0.9	13
38	Social skills in women with Turner Syndrome. Scandinavian Journal of Psychology, 2011, 52, 440-447.	1.5	13
39	46,XX DSD and Antley-Bixler syndrome due to novel mutations in the cytochrome P450 oxidoreductase gene. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 578-585.	1.3	13
40	Turner syndrome and metabolic derangements: Another example of fetal programming. Early Human Development, 2012, 88, 99-102.	1.8	13
41	Parentsâ€™ experiences of having a baby with ambiguous genitalia. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 833-8.	0.9	13
42	45,X/46,XY ovotesticular disorder of sex development revisited: undifferentiated gonadal tissue may be mistaken as ovarian tissue. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 899-904.	0.9	13
43	Six new cases confirm the clinical molecular profile of complete combined 17Î±-hydroxylase/ 17,20-lyase deficiency in Brazil. Arquivos Brasileiros De Endocrinologia E Metabologia, 2010, 54, 711-716.	1.3	12
44	Two distinct WT1 mutations identified in patients and relatives with isolated nephrotic proteinuria. Biochemical and Biophysical Research Communications, 2013, 441, 371-376.	2.1	12
45	Functional characterization of fiveNR5A1gene mutations found in patients with 46,XY disorders of sex development. Human Mutation, 2018, 39, 114-123.	2.5	12
46	Spontaneous Final Height in Turner's Syndrome in Brazil. Journal of Pediatric Endocrinology and Metabolism, 2007, 20, 1207-14.	0.9	11
47	Severe forms of partial androgen insensitivity syndrome due to p.L830F novel mutation in androgen receptor gene in a Brazilian family. BMC Research Notes, 2011, 4, 173.	1.4	11
48	The use of fluorescence in situ hybridization in the diagnosis of hidden mosaicism: apropos of three cases of sex chromosome anomalies. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 545-551.	1.3	11
49	FSH may be a useful tool to allow early diagnosis of Turner syndrome. BMC Endocrine Disorders, 2018, 18, 8.	2.2	11
50	Turner syndrome: a pediatric diagnosis frequently made by non-pediatricians. Jornal De Pediatria, 2010, 86, 121-125.	2.0	11
51	Growth hormone effect on body composition in Turner syndrome. Endocrine, 2011, 40, 486-491.	2.3	10
52	NPHS1 gene mutations confirm congenital nephrotic syndrome in four Brazilian cases: A novel mutation is described. Nephrology, 2016, 21, 753-757.	1.6	10
53	Prevalence of genital abnormalities in neonates. Jornal De Pediatria, 2012, 88, 489-95.	2.0	10
54	Klinefelter syndrome: an unusual diagnosis in pediatric patients. Jornal De Pediatria, 2012, 88, 323-7.	2.0	9

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55	Functional Impact of Novel Androgen Receptor Mutations on the Clinical Manifestation of Androgen Insensitivity Syndrome. Sexual Development, 2017, 11, 238-247.	2.0	9
56	Three new Brazilian cases of 17Î±-hydroxylase deficiency: clinical, molecular, hormonal, and treatment features. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 937-942.	0.9	9
57	Mutations of androgen receptor gene in Brazilian patients with male pseudohermaphroditism. Brazilian Journal of Medical and Biological Research, 1998, 31, 775-778.	1.5	8
58	Identification of a neocentromere in a rearranged Y chromosome with no detectable DYZ3 centromeric sequence. American Journal of Medical Genetics Part A, 2002, 113, 263-267.	2.4	8
59	Preserved Fertility in a Patient with Gynecomastia Associated with the p.Pro695Ser Mutation in the Androgen Receptor. Sexual Development, 2014, 8, 350-355.	2.0	8
60	Ovotesticular disorder of sex development with unusual karyotype: patient report. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 677-80.	0.9	7
61	A boy with mental retardation, blepharophimosis and hypothyroidism: a diagnostic dilemma between Young-Simpson and Ohdo syndrome. Clinical Dysmorphology, 2000, 9, 199-204.	0.3	6
62	Androgens by immunoassay and mass spectrometry in children with 46,XY disorder of sex development. Endocrine Connections, 2020, 9, 1085-1094.	1.9	6
63	Leberâ€™s hereditary optic neuropathy: Clinical and molecular profile of a Brazilian sample. Ophthalmic Genetics, 2010, 31, 126-128.	1.2	5
64	<i>WT1 </i>Haploinsufficiency Supports Milder Renal Manifestation in Two Patients with Denys-Drash Syndrome. Sexual Development, 2017, 11, 34-39.	2.0	5
65	Genomic Investigation of Balanced Chromosomal Rearrangements in Patients with Abnormal Phenotypes. Molecular Syndromology, 2017, 8, 187-194.	0.8	5
66	A study of splicing mutations in disorders of sex development. Scientific Reports, 2017, 7, 16202.	3.3	5
67	A Search for Disorders of Sex Development among Infertile Men. Sexual Development, 2018, 12, 275-280.	2.0	5
68	Turner's Syndrome and Subclinical Autoimmune Thyroid Disease: A Two-Year Follow-up Study. Journal of Pediatric Endocrinology and Metabolism, 2009, 22, 109-18.	0.9	4
69	Early development of a gonadal tumor in a patient with mixed gonadal dysgenesis. Archives of Endocrinology and Metabolism, 2018, 62, 644-647.	0.6	4
70	Can Non-Coding NR5A1 Gene Variants Explain Phenotypes of Disorders of Sex Development?. Sexual Development, 2022, 16, 252-260.	2.0	4
71	Mutations in NR5A1 Associated With Ovarian Insufficiency. Obstetrical and Gynecological Survey, 2009, 64, 665-666.	0.4	3
72	Molecular Diagnosis of 5Î±-Reductase Type II Deficiency in Brazilian Siblings with 46,XY Disorder of Sex Development. International Journal of Molecular Sciences, 2011, 12, 9471-9480.	4.1	3

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73	Clinical and cytogenetic features of 516 patients with suspected Turner syndrome â€“ a single-center experience. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 167-173.	0.9	3
74	Why pediatricians need to know the disorders of sex development: experience of 709 cases in a specialized service. Jornal De Pediatria, 2020, 96, 607-613.	2.0	3
75	Female counterpart of shawl scrotum in aarskog-scott syndrome. International Braz J Urol: Official Journal of the Brazilian Society of Urology, 2006, 32, 459-461.	1.5	3
76	Intersexo: entre o gene e o gÃªnero. Arquivos Brasileiros De Endocrinologia E Metabologia, 2005, 49, 1-3.	1.3	3
77	The novel p.E89K mutation in the SRY gene inhibits DNA binding and causes the 46,XY disorder of sex development. Brazilian Journal of Medical and Biological Research, 2011, 44, 361-365.	1.5	3
78	Interaction between audiology and genetics in the study of a family: the complexity of molecular diagnosis and genetic counseling. Brazilian Journal of Otorhinolaryngology, 2008, 74, 698-702.	1.0	2
79	NPHS2Mutations: A Closer Look to Latin American Countries. BioMed Research International, 2017, 2017, 1-6.	1.9	2
80	Promises and pitfalls of whole-exome sequencing exemplified by a nephrotic syndrome family. Molecular Genetics and Genomics, 2020, 295, 135-142.	2.1	2
81	Suggested Cutoff Point for Testosterone by Liquid Chromatography with Tandem Mass Spectrometry (LC-MS/MS) after Stimulation with Recombinant Human Chorionic Gonadotropin. Sexual Development, 2022, 16, 266-269.	2.0	2
82	Infertility and marker chromosomes: Application of molecular cytogenetic techniques in a case of inv dup(15). Journal of Applied Genetics, 2006, 47, 89-91.	1.9	1
83	SÃndrome de Turner: um diagnÃ³stico pediÃ¡trico frequentemente realizado por nÃ£o pediatras. Jornal De Pediatria, 2010, 86, 121-125.	2.0	1
84	Sexual difference, identification and object choice in individuals with sex differentiation disorders. Revista Latinoamericana De Psicopatologia Fundamental, 2012, 15, 464-481.	0.0	1
85	Prader-Willi syndrome: a case report with atypical developmental features. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 983-8.	0.9	1
86	Lipomatous nevus and urethral caruncle mistaken for ambiguous genitalia in a female infant. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 233-6.	0.9	1
87	An illustrative case of LÃ©ri-Weill dyschondrosteosis. Genetics and Molecular Biology, 2008, 31, 839-842.	1.3	1
88	Densidade Mineral Ã“ssea: Estudo Transversal em Pacientes com SÃndrome de Turner. Arquivos Brasileiros De Endocrinologia E Metabologia, 2002, 46, 143-149.	1.3	1
89	Leydig and Sertoli cell function in individuals with genital ambiguity, 46,XY karyotype, palpable gonads and normal testosterone secretion: a case-control study. Sao Paulo Medical Journal, 2022, , .	0.9	1
90	Comparison between two inhibin B ELISA assays in 46,XY testicular disorders of sex development (DSD) with normal testosterone secretion. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 191-194.	0.9	0

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91	Response to Letter to the Editor: â€œClinical but Not Histological Outcomes in Males With 45,X/46,XY Mosaicism Vary Depending on Reason for Diagnosisâ€• Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5812-5813.	3.6	0
92	Why pediatricians need to know the disorders of sex development: experience of 709 cases in a specialized service. Jornal De Pediatria (VersÃ£o Em PortuguÃªs), 2020, 96, 607-613.	0.2	0
93	A importÃ¢ncia da interdisciplinaridade no atendimento dos distÃºrbios da diferenciaÃ§Ã£o do sexo em hospital universitÃ¡rio. ServiÃ§o Social E SaÃºde, 2015, 2, 55.	0.1	0
94	Association between Down Syndrome and Disorders of Sex Development: Report of Three Cases and Review of 188 Cases in the Literature. Sexual Development, 2020, 14, 3-11.	2.0	0
95	Trends in Time Regarding Sex Assignment of Patients with Disorders of Sex Development: Experience of an Interdisciplinary Service. Sexual Development, 2022, 16, 236-241.	2.0	0
96	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
97	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
98	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
99	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0