

Andr a Maciel-Guerra

List of Publications by Year in descending order

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99
papers

1,840
citations

377584

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355658

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107
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107
docs citations

107
times ranked

2138
citing authors

#	ARTICLE	IF	CITATIONS
1	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, , .	0.4	1
2	Trends in Time Regarding Sex Assignment of Patients with Disorders of Sex Development: Experience of an Interdisciplinary Service. Sexual Development, 2022, 16, 236-241.	1.1	0
3	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.	1.1	2
4	Can Non-Coding NR5A1 Gene Variants Explain Phenotypes of Disorders of Sex Development?. Sexual Development, 2022, 16, 252-260.	1.1	4
5	Promises and pitfalls of whole-exome sequencing exemplified by a nephrotic syndrome family. Molecular Genetics and Genomics, 2020, 295, 135-142.	1.0	2
6	Mutation update for the NR5A1 gene involved in DSD and infertility. Human Mutation, 2020, 41, 58-68.	1.1	52
7	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.	0.9	3
8	Why pediatricians need to know the disorders of sex development: experience of 709 cases in a specialized service. Jornal De Pediatria (Versão Em Português), 2020, 96, 607-613.	0.2	0
9	Androgens by immunoassay and mass spectrometry in children with 46,XY disorder of sex development. Endocrine Connections, 2020, 9, 1085-1094.	0.8	6
10	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.	1.1	0
11	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. PLoS ONE, 2020, 15, e0240795.	1.1	21
12	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
13	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
14	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
15	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
16	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.	1.8	27
17	Response to Letter to the Editor: "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, 5812-5813.	1.8	0
18	Imaging Techniques in the Diagnostic Journey of Disorders of Sex Development. Sexual Development, 2018, 12, 95-99.	1.1	18

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19	Clinical and cytogenetic features of 516 patients with suspected Turner syndrome – a single-center experience. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 167-173.	0.4	3
20	Comparison between two inhibin B ELISA assays in 46,XY testicular disorders of sex development (DSD) with normal testosterone secretion. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 191-194.	0.4	0
21	Functional characterization of five <i>NR5A1</i> gene mutations found in patients with 46,XY disorders of sex development. <i>Human Mutation</i> , 2018, 39, 114-123.	1.1	12
22	Early development of a gonadal tumor in a patient with mixed gonadal dysgenesis. <i>Archives of Endocrinology and Metabolism</i> , 2018, 62, 644-647.	0.3	4
23	A Search for Disorders of Sex Development among Infertile Men. <i>Sexual Development</i> , 2018, 12, 275-280.	1.1	5
24	Three new Brazilian cases of 17 β -hydroxylase deficiency: clinical, molecular, hormonal, and treatment features. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 937-942.	0.4	9
25	FSH may be a useful tool to allow early diagnosis of Turner syndrome. <i>BMC Endocrine Disorders</i> , 2018, 18, 8.	0.9	11
26	<i>WT1</i> Haploinsufficiency Supports Milder Renal Manifestation in Two Patients with Denys-Drash Syndrome. <i>Sexual Development</i> , 2017, 11, 34-39.	1.1	5
27	A Novel Homozygous Missense <i>FSHR</i> Variant Associated with Hypergonadotropic Hypogonadism in Two Siblings from a Brazilian Family. <i>Sexual Development</i> , 2017, 11, 137-142.	1.1	26
28	Genomic Investigation of Balanced Chromosomal Rearrangements in Patients with Abnormal Phenotypes. <i>Molecular Syndromology</i> , 2017, 8, 187-194.	0.3	5
29	45,X/46,XY ovotesticular disorder of sex development revisited: undifferentiated gonadal tissue may be mistaken as ovarian tissue. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2017, 30, 899-904.	0.4	13
30	Functional Impact of Novel Androgen Receptor Mutations on the Clinical Manifestation of Androgen Insensitivity Syndrome. <i>Sexual Development</i> , 2017, 11, 238-247.	1.1	9
31	A study of splicing mutations in disorders of sex development. <i>Scientific Reports</i> , 2017, 7, 16202.	1.6	5
32	<i>NPHS2</i> Mutations: A Closer Look to Latin American Countries. <i>BioMed Research International</i> , 2017, 2017, 1-6.	0.9	2
33	408 Cases of Genital Ambiguity Followed by Single Multidisciplinary Team during 23 Years: Etiologic Diagnosis and Sex of Rearing. <i>International Journal of Endocrinology</i> , 2016, 2016, 1-9.	0.6	23
34	<i>NPHS1</i> gene mutations confirm congenital nephrotic syndrome in four Brazilian cases: A novel mutation is described. <i>Nephrology</i> , 2016, 21, 753-757.	0.7	10
35	<i>NR5A1</i> Loss-of-Function Mutations Lead to 46,XY Partial Gonadal Dysgenesis Phenotype: Report of Three Novel Mutations. <i>Sexual Development</i> , 2016, 10, 191-199.	1.1	18
36	Lipomatous nevus and urethral caruncle mistaken for ambiguous genitalia in a female infant. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 233-6.	0.4	1

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37	New approach to phenotypic variability and karyotype-phenotype correlation in Turner syndrome. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 475-9.	0.4	23
38	NPHS2 mutations account for only 15% of nephrotic syndrome cases. <i>BMC Medical Genetics</i> , 2015, 16, 88.	2.1	18
39	Ovotesticular disorder of sex development with unusual karyotype: patient report. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2015, 28, 677-80.	0.4	7
40	Parents'™ experiences of having a baby with ambiguous genitalia. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2015, 28, 833-8.	0.4	13
41	A importância da interdisciplinaridade no atendimento dos distúrbios da diferenciação do sexo em hospital universitário. <i>Serviço Social E Saúde</i> , 2015, 2, 55.	0.1	0
42	Prader-Willi syndrome: a case report with atypical developmental features. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014, 27, 983-8.	0.4	1
43	Long-Term Follow-Up of Patients with 46,XY Partial Gonadal Dysgenesis Reared as Males. <i>International Journal of Endocrinology</i> , 2014, 2014, 1-8.	0.6	21
44	Homozygous Inactivating Mutation in <i>NANOS3</i> in Two Sisters with Primary Ovarian Insufficiency. <i>BioMed Research International</i> , 2014, 2014, 1-8.	0.9	36
45	Preserved Fertility in a Patient with Gynecomastia Associated with the p.Pro695Ser Mutation in the Androgen Receptor. <i>Sexual Development</i> , 2014, 8, 350-355.	1.1	8
46	The novel p.Cys65Tyr mutation in <i>NR5A1</i> gene in three 46,XY siblings with normal testosterone levels and their mother with primary ovarian insufficiency. <i>BMC Medical Genetics</i> , 2014, 15, 7.	2.1	25
47	Screening of Y chromosome microdeletions in 46,XY partial gonadal dysgenesis and in patients with a 45,X/46,XY karyotype or its variants. <i>BMC Medical Genetics</i> , 2013, 14, 115.	2.1	17
48	Two distinct <i>WT1</i> mutations identified in patients and relatives with isolated nephrotic proteinuria. <i>Biochemical and Biophysical Research Communications</i> , 2013, 441, 371-376.	1.0	12
49	Clinical and Laboratorial Features That May Differentiate 46,XY DSD due to Partial Androgen Insensitivity and <i>5α-Reductase Type 2</i> Deficiency. <i>International Journal of Endocrinology</i> , 2012, 2012, 1-7.	0.6	25
50	Analysis of anti-Müllerian hormone (AMH) and its receptor (<i>AMHR2</i>) genes in patients with persistent Müllerian duct syndrome. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , 2012, 56, 473-478.	1.3	21
51	Clinical and molecular spectrum of patients with <i>17β-hydroxysteroid dehydrogenase type 3 (17β-HSD3)</i> deficiency. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , 2012, 56, 533-539.	1.3	29
52	Frasier syndrome: four new cases with unusual presentations. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , 2012, 56, 525-532.	1.3	14
53	Klinefelter syndrome: an unusual diagnosis in pediatric patients. <i>Jornal De Pediatria</i> , 2012, 88, 323-7.	0.9	9
54	46,XX DSD and Antley-Bixler syndrome due to novel mutations in the cytochrome P450 oxidoreductase gene. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , 2012, 56, 578-585.	1.3	13

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55	Sexual difference, identification and object choice in individuals with sex differentiation disorders. Revista Latinoamericana De Psicopatologia Fundamental, 2012, 15, 464-481.	0.0	1
56	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
57	Turner syndrome and metabolic derangements: Another example of fetal programming. Early Human Development, 2012, 88, 99-102.	0.8	13
58	Spontaneous puberty in girls with early diagnosis of Turner syndrome. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 653-657.	1.3	17
59	Prevalence of genital abnormalities in neonates. Jornal De Pediatria, 2012, 88, 489-95.	0.9	10
60	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.	0.5	42
61	Multifunctional role of steroidogenic factor 1 and disorders of sex development. Arquivos Brasileiros De Endocrinologia E Metabologia, 2011, 55, 607-612.	1.3	14
62	Social skills in women with Turner Syndrome. Scandinavian Journal of Psychology, 2011, 52, 440-447.	0.8	13
63	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.	0.6	11
64	Growth hormone effect on body composition in Turner syndrome. Endocrine, 2011, 40, 486-491.	1.1	10
65	Molecular Diagnosis of 5 α -Reductase Type II Deficiency in Brazilian Siblings with 46,XY Disorder of Sex Development. International Journal of Molecular Sciences, 2011, 12, 9471-9480.	1.8	3
66	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.	0.7	3
67	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
68	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
69	Six new cases confirm the clinical molecular profile of complete combined 17 α -hydroxylase/ 17,20-lyase deficiency in Brazil. Arquivos Brasileiros De Endocrinologia E Metabologia, 2010, 54, 711-716.	1.3	12
70	Síndrome de Turner: um diagnóstico pediátrico frequentemente realizado por não pediatras. Jornal De Pediatria, 2010, 86, 121-125.	0.9	1
71	Leber's hereditary optic neuropathy: Clinical and molecular profile of a Brazilian sample. Ophthalmic Genetics, 2010, 31, 126-128.	0.5	5
72	Turner syndrome: a pediatric diagnosis frequently made by non-pediatricians. Jornal De Pediatria, 2010, 86, 121-125.	0.9	11

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73	Turner's Syndrome and Subclinical Autoimmune Thyroid Disease: A Two-Year Follow-up Study. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2009, 22, 109-18.	0.4	4
74	Mutations in NR5A1 Associated with Ovarian Insufficiency. <i>New England Journal of Medicine</i> , 2009, 360, 1200-1210.	13.9	339
75	Mutations in NR5A1 Associated With Ovarian Insufficiency. <i>Obstetrical and Gynecological Survey</i> , 2009, 64, 665-666.	0.2	3
76	XX Maleness and XX True Hermaphroditism in SRY-Negative Monozygotic Twins: Additional Evidence for a Common Origin. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 339-343.	1.8	44
77	Interaction between audiology and genetics in the study of a family: the complexity of molecular diagnosis and genetic counseling. <i>Brazilian Journal of Otorhinolaryngology</i> , 2008, 74, 698-702.	0.4	2
78	Clinical and genetic findings of five patients with WT1-related disorders. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , 2008, 52, 1236-1243.	1.3	16
79	An illustrative case of Léri-Weill dyschondrosteosis. <i>Genetics and Molecular Biology</i> , 2008, 31, 839-842.	0.6	1
80	Spontaneous Final Height in Turner's Syndrome in Brazil. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2007, 20, 1207-14.	0.4	11
81	O pediatra frente a uma criança com ambigüidade genital. <i>Jornal De Pediatria</i> , 2007, 83, S184-S191.	0.9	15
82	Infertility and marker chromosomes: Application of molecular cytogenetic techniques in a case of inv dup(15). <i>Journal of Applied Genetics</i> , 2006, 47, 89-91.	1.0	1
83	Female counterpart of shawl scrotum in Klinefelter syndrome. <i>International Braz J Urol: Official Journal of the Brazilian Society of Urology</i> , 2006, 32, 459-461.	0.7	3
84	Idiopathic male pseudohermaphroditism is associated with prenatal growth retardation. <i>European Journal of Pediatrics</i> , 2005, 164, 287-291.	1.3	13
85	Mutations in SRY and WT1 genes required for gonadal development are not responsible for XY partial gonadal dysgenesis. <i>Brazilian Journal of Medical and Biological Research</i> , 2005, 38, 17-25.	0.7	17
86	Diagnosis of 5 α -Reductase Type 2 Deficiency: Contribution of Anti-Müllerian Hormone Evaluation. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2005, 18, 1383-9.	0.4	13
87	Intersexo: entre o gene e o gênero. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , 2005, 49, 1-3.	1.3	3
88	Classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency: a cross-sectional study of factors involved in bone mineral density. <i>Journal of Bone and Mineral Metabolism</i> , 2003, 21, 396-401.	1.3	46
89	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. <i>American Journal of Human Genetics</i> , 2003, 73, 1452-1458.	2.6	269
90	Identification of a neocentromere in a rearranged Y chromosome with no detectable DYZ3 centromeric sequence. <i>American Journal of Medical Genetics Part A</i> , 2002, 113, 263-267.	2.4	8

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91	Bone mineralization in Turner syndrome: a transverse study of the determinant factors in 58 patients. <i>Journal of Bone and Mineral Metabolism</i> , 2002, 20, 294-297.	1.3	32
92	Deafness resulting from mutations in the GJB2 (connexin 26) gene in Brazilian patients. <i>Clinical Genetics</i> , 2002, 61, 354-358.	1.0	35
93	Densidade Mineral ssea: Estudo Transversal em Pacientes com Sndrome de Turner. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , 2002, 46, 143-149.	1.3	1
94	Morphometry and Histology of Gonads From 13 Children With Dysgenetic Male Pseudohermaphroditism. <i>Archives of Pathology and Laboratory Medicine</i> , 2001, 125, 652-656.	1.2	25
95	A boy with mental retardation, blepharophimosis and hypothyroidism: a diagnostic dilemma between Young-Simpson and Ohdo syndrome. <i>Clinical Dysmorphology</i> , 2000, 9, 199-204.	0.1	6
96	Determination of the frequency of the 35delG allele in Brazilian neonates. <i>Clinical Genetics</i> , 2000, 58, 339-340.	1.0	29
97	Turner's Syndrome and Thyroid Disease: A Transverse Study of Pediatric Patients in Brazil. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2000, 13, 357-62.	0.4	25
98	Mutations of androgen receptor gene in Brazilian patients with male pseudohermaphroditism. <i>Brazilian Journal of Medical and Biological Research</i> , 1998, 31, 775-778.	0.7	8
99	True gonadism: Report of a case analyzed with Y-specific DNA probes. <i>American Journal of Medical Genetics Part A</i> , 1991, 41, 444-445.	2.4	23