Mirian Yumie Nishi

List of Publications by Year
in descending order

81	2,066	236612	264894
papers	citations	25 h-index	42 g-index

87	87	87	2496
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	WT1 Pathogenic Variants are Associated with a Broad Spectrum of Differences in Sex Development Phenotypes and Heterogeneous Progression of Renal Disease. Sexual Development, 2022, 16, 46-54.	1.1	5
2	Variants in 46,XY DSD-Related Genes in Syndromic and Non-Syndromic Small for Gestational Age Children with Hypospadias. Sexual Development, 2022, 16, 27-33.	1.1	3
3	A Small Supernumerary Xp Marker Chromosome Including Genes <i>NR0B1</i> and <i>MAGEB</i> Causing Partial Gonadal Dysgenesis and Gonadoblastoma. Sexual Development, 2022, 16, 55-63.	1.1	1
4	Contribution of Clinical and Genetic Approaches for Diagnosing 209 Index Cases With 46,XY Differences of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1797-e1806.	1.8	11
5	High-throughput Sequencing to Identify Monogenic Etiologies in a Preselected Polycystic Ovary Syndrome Cohort. Journal of the Endocrine Society, 2022, 6, .	0.1	4
6	Performance of mutation pathogenicity prediction tools on missense variants associated with 46,XY differences of sex development. Clinics, 2021, 76, e2052.	0.6	10
7	Allelic Variants in Established Hypopituitarism Genes Expand Our Knowledge of the Phenotypic Spectrum. Genes, 2021, 12, 1128.	1.0	0
8	A New Insight into the Surgical Treatment of Primary Macronodular Adrenal Hyperplasia. Journal of the Endocrine Society, 2020, 4, bvaa083.	0.1	14
9	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, .	0.1	0
10	High Prevalence of Alterations in DNA Mismatch Repair Genes of Lynch Syndrome in Pediatric Patients with Adrenocortical Tumors Carrying a Germline Mutation on TP53. Cancers, 2020, 12, 621.	1.7	4
11	Allelic Variants of ARMC5 in Patients With Adrenal Incidentalomas and in Patients With Cushing's Syndrome Associated With Bilateral Adrenal Nodules. Frontiers in Endocrinology, 2020, 11, 36.	1.5	7
12	Sterol O-Acyl Transferase 1 as a Prognostic Marker of Adrenocortical Carcinoma. Cancers, 2020, 12, 247.	1.7	22
13	SELAdb: A database of exonic variants in a Brazilian population referred to a quaternary medical center in SÃ£o Paulo. Clinics, 2020, 75, e1913.	0.6	15
14	SUN-709 MiR-200c Expression Profiles in Plasma of 46,XY DSD Patients of Unknown Etiology. Journal of the Endocrine Society, 2020, 4, .	0.1	0
15	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. PLoS ONE, 2020, 15, e0240795.	1.1	21
16	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
17	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
18	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0

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19	Screening of targeted panel genes in Brazilian patients with primary ovarian insufficiency. , 2020, 15, e0240795.		0
20	Two rare loss-of-function variants in the STAG3 gene leading to primary ovarian insufficiency. European Journal of Medical Genetics, 2019, 62, 186-189.	0.7	30
21	Combined pituitary hormone deficiency caused by PROP1 mutations: update 20 years post-discovery. Archives of Endocrinology and Metabolism, 2019, 63, 167-174.	0.3	23
22	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.	1.8	26
23	Genetic Disorders in Prenatal Onset Syndromic Short Stature Identified by Exome Sequencing. Journal of Pediatrics, 2019, 215, 192-198.	0.9	36
24	Evaluation of SHOX defects in the era of nextâ€generation sequencing. Clinical Genetics, 2019, 96, 261-265.	1.0	9
25	Exome Sequencing Reveals the POLR3H Gene as a Novel Cause of Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2827-2841.	1.8	28
26	Mobile DNA in Endocrinology: LINE-1 Retrotransposon Causing Partial Androgen Insensitivity Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 6385-6390.	1.8	10
27	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.	1.4	21
28	A 46,XX testicular disorder of sex development caused by a Wilms' tumour Factorâ€1 (WT1) pathogenic variant. Clinical Genetics, 2019, 95, 172-176.	1.0	24
29	ESR1 polymorphism (rs2234693) influences femoral bone mass in patients with Turner syndrome. Endocrine Connections, 2019, 8, 1513-1519.	0.8	12
30	Longâ€term outcomes and molecular analysis of a large cohort of patients with 46,<scp>XY</scp> disorder of sex development due to partial gonadal dysgenesis. Clinical Endocrinology, 2018, 89, 164-177.	1.2	13
31	Partial androgen insensitivity syndrome due to somatic mosaicism of the androgen receptor. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 223-228.	0.4	9
32	The role of ARMC5 in human cell cultures from nodules of primary macronodular adrenocortical hyperplasia (PMAH). Molecular and Cellular Endocrinology, 2018, 460, 36-46.	1.6	38
33	Identification of the first homozygous 1â€bp deletion in GDF9 gene leading to primary ovarian insufficiency by using targeted massively parallel sequencing. Clinical Genetics, 2018, 93, 408-411.	1.0	29
34	Androgen receptor mRNA analysis from whole blood: a lowâ€cost strategy for detection of androgen receptor gene splicing defects. Clinical Genetics, 2018, 94, 489-490.	1.0	2
35	Androgen insensitivity syndrome: a review. Archives of Endocrinology and Metabolism, 2018, 62, 227-235.	0.3	100
36	46,XY disorder of sex development (DSD) due to 17Î²-hydroxysteroid dehydrogenase type 3 deficiency. Journal of Steroid Biochemistry and Molecular Biology, 2017, 165, 79-85.	1.2	66

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37	Discordant Genotypic Sex and Phenotype Variations in Two Spanish Siblings with 17Î±-Hydroxylase/17,20-Lyase Deficiency Carrying the Most Prevalent Mutated CYP17A1 Alleles of Brazilian Patients. Sexual Development, 2017, 11, 70-77.	1.1	8
38	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.	1.1	8
39	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.	1.1	26
40	A novel homozygous 1-bp deletion in the NOBOX gene in two Brazilian sisters with primary ovarian failure. Endocrine, 2017, 58, 442-447.	1.1	17
41	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.	1.2	16
42	Molecular analysis of brazilian patients with combined pituitary hormone deficiency and orthotopic posterior pituitary lobe reveals eight different <scp>PROP</scp>1 alterations with three novel mutations. Clinical Endocrinology, 2017, 87, 725-732.	1.2	13
43	Malignant testicular germ cell tumors in postpubertal individuals with androgen insensitivity: prevalence, pathology and relevance of single nucleotide polymorphism-based susceptibility profiling. Human Reproduction, 2017, 32, 2561-2573.	0.4	50
44	Good response to longâ€term therapy with growth hormone in a patient with 9p trisomy syndrome: A case report and review of the literature. American Journal of Medical Genetics, Part A, 2016, 170, 1046-1049.	0.7	5
45	Effects of Type 1 Insulin-Like Growth Factor Receptor Silencing in a Human Adrenocortical Cell Line. Hormone and Metabolic Research, 2016, 48, 484-488.	0.7	3
46	Wide spectrum of NR5A1â€related phenotypes in 46,XY and 46,XX individuals. Birth Defects Research Part C: Embryo Today Reviews, 2016, 108, 309-320.	3.6	76
47	Mutation analysis of NANOS3 in Brazilian women with primary ovarian failure. Clinics, 2016, 71, 695-698.	0.6	6
48	Pitfalls in hormonal diagnosis of 17-beta hydroxysteroid dehydrogenase III deficiency. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 623-8.	0.4	26
49	Disorders of sex development: effect of molecular diagnostics. Nature Reviews Endocrinology, 2015, 11, 478-488.	4.3	81
50	FGFR1 and PROKR2 rare variants found in patients with combined pituitary hormone deficiencies. Endocrine Connections, 2015, 4, 100-107.	0.8	34
51	Amplification of theInsulin-Like Growth Factor 1 ReceptorGene Is a Rare Event in Adrenocortical Adenocarcinomas: Searching for Potential Mechanisms of Overexpression. BioMed Research International, 2014, 2014, 1-7.	0.9	11
52	Homozygous Inactivating Mutation inNANOS3in Two Sisters with Primary Ovarian Insufficiency. BioMed Research International, 2014, 2014, 1-8.	0.9	36
53	46,XY DSD due to 17Î²-Hydroxysteroid Dehydrogenase 3 Deficiency. , 2014, , 191-197.		0
54	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, 180-186.	0.5	5

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55	ARMC5Mutations Are a Frequent Cause of Primary Macronodular Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1501-E1509.	1.8	120
56	Heterozygous Mutations in Natriuretic Peptide Receptor-B (NPR2) Gene as a Cause of Short Stature in Patients Initially Classified as Idiopathic Short Stature. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1636-E1644.	1.8	111
57	The Sitting Height/Height Ratio for Age in Healthy and Short Individuals and Its Potential Role in Selecting Short Children for <i>SHOX</i> Analysis. Hormone Research in Paediatrics, 2013, 80, 449-456.	0.8	45
58	Y chromosome aberration in a patient with cloacal-bladder exstrophy-epispadias complex: an unusual finding. Arquivos Brasileiros De Endocrinologia E Metabologia, 2013, 57, 148-152.	1.3	0
59	Post-receptor IGF1 insensitivity restricted to the MAPK pathway in a Silverâ€“Russell syndrome patient with hypomethylation at the imprinting control region on chromosome 11. European Journal of Endocrinology, 2012, 166, 543-550.	1.9	7
60	GH-Releasing Hormone Receptor Gene: A Novel Splice-Disrupting Mutation and Study of Founder Effects. Hormone Research in Paediatrics, 2012, 78, 165-172.	0.8	18
61	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, 690-694.	0.7	9
62	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
63	A novel DAX1/NR0B1 mutation in a patient with adrenal hypoplasia congenita and hypogonadotropic hypogonadism. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 496-500.	1.3	11
64	A novel WT1 heterozygous nonsense mutation (p.K248X) causing a mild and slightly progressive nephropathy in a 46,XY patient with Denysâ€“Drash syndrome. Pediatric Nephrology, 2011, 26, 1311-1315.	0.9	8
65	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.	0.8	6
66	Effectiveness of the Combined Recombinant Human Growth Hormone and Gonadotropin-Releasing Hormone Analog Therapy in Pubertal Patients with Short Stature due toSHOXDeficiency. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 328-332.	1.8	35
67	Steroidogenic Factor 1 Overexpression and Gene Amplification Are More Frequent in Adrenocortical Tumors from Children than from Adults. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 1458-1462.	1.8	66
68	Usefulness of MLPA in the detection of SHOX deletions. European Journal of Medical Genetics, 2010, 53, 234-238.	0.7	23
69	Expression profiles of the glucose-dependent insulinotropic peptide receptor and LHCGR in sporadic adrenocortical tumors. Journal of Endocrinology, 2009, 200, 167-175.	1.2	5
70	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, 326-331.	1.3	5
71	Expression of Insulin-Like Growth Factor-II and Its Receptor in Pediatric and Adult Adrenocortical Tumors. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3524-3531.	1.8	149
72	Cryptic intragenic deletion of the SHOX gene in a family with LÃ©ri-Weill dyschondrosteosis detected by Multiplex Ligation-Dependent Probe Amplification (MLPA). Arquivos Brasileiros De Endocrinologia E Metabologia, 2008, 52, 1382-1387.	1.3	9

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73	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, 1282-1287.	1.3	9
74	Combined pituitary hormone deficiency (CPHD) due to a complete PROP1 deletion. Clinical Endocrinology, 2006, 65, 294-300.	1.2	35
75	SHOX mutations in idiopathic short stature and Leri-Weill dyschondrosteosis: frequency and phenotypic variability. Clinical Endocrinology, 2006, 66, 061031010617004-???.	1.2	75
76	Mutations in the SRY, DAX1, SF1 and WNT4 genes in Brazilian sex-reversed patients. Brazilian Journal of Medical and Biological Research, 2004, 37, 145-150.	0.7	53
77	Familial Hyperestrogenism in Both Sexes: Clinical, Hormonal, and Molecular Studies of Two Siblings. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3027-3034.	1.8	52
78	Detection of Y-specific sequences in 122 patients with Turner syndrome: Nested PCR is not a reliable method. American Journal of Medical Genetics Part A, 2002, 107, 299-305.	2.4	45
79	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.	2.4	2
80	A Novel Missense Mutation, GLY424SER, in Brazilian Patients with 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 2870-2872.	1.8	31
81	A novel missense mutation (S18N) in the 5â€² non-HMG box region of the SRY gene in a patient with partial gonadal dysgenesis and his normal male relatives. Human Genetics, 1998, 102, 213-215.	1.8	65