

MÃ³nica FernÃ¡ndez-Cancio

List of Publications by Year in descending order

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docs citations

41
times ranked

1334
citing authors

#	ARTICLE	IF	CITATIONS
1	Incidence and Prevalence of Children's Diffuse Lung Disease in Spain. Archivos De Bronconeumologia, 2022, 58, 22-29.	0.4	15
2	Phenotypic Variability of Patients With PAX8 Variants Presenting With Congenital Hypothyroidism and Eutopic Thyroid. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e152-e170.	1.8	6
3	Implementaci3n de un panel de genes para el diagn3stico gen3tico de la discinesia ciliar primaria. Archivos De Bronconeumologia, 2021, 57, 186-194.	0.4	5
4	Seventy eight children born small for gestational age without catch-up growth treated with growth hormone from the prepubertal stage until adult height age. An evaluation of puberty and changes in the metabolic profile. Endocrinologia, Diabetes Y Nutrici3n, 2021, 68, 612-620.	0.1	2
5	Seventy eight children born small for gestational age without catch-up growth treated with growth hormone from the prepubertal stage until adult height age. An evaluation of puberty and changes in the metabolic profile. Endocrinolog3a Diabetes Y Nutrici3n (English Ed), 2021, 68, 612-620.	0.1	2
6	Immunofluorescence Analysis as a Diagnostic Tool in a Spanish Cohort of Patients with Suspected Primary Ciliary Dyskinesia. Journal of Clinical Medicine, 2020, 9, 3603.	1.0	7
7	Role of Immunofluorescence and Molecular Diagnosis in the Characterization of Primary Ciliary Dyskinesia. Archivos De Bronconeumologia, 2019, 55, 439-441.	0.4	1
8	Expanding the Clinical and Genetic Spectra of Primary Immunodeficiency-Related Disorders With Clinical Exome Sequencing: Expected and Unexpected Findings. Frontiers in Immunology, 2019, 10, 2325.	2.2	41
9	Broad Phenotypes of Disorders/Differences of Sex Development in MAMLD1 Patients Through Oligogenic Disease. Frontiers in Genetics, 2019, 10, 746.	1.1	22
10	Isolated pulmonary interstitial glycogenosis associated with alveolar growth abnormalities: A long-term follow-up study. Pediatric Pulmonology, 2019, 54, 837-846.	1.0	11
11	A Novel Homozygous <i>AMRH2</i> Gene Mutation in a Patient with Persistent M3/4llerian Duct Syndrome. Sexual Development, 2019, 13, 87-91.	1.1	6
12	Papel de la inmunofluorescencia y el diagn3stico molecular en la caracterizaci3n de la discinesia ciliar primaria. Archivos De Bronconeumologia, 2019, 55, 439-441.	0.4	5
13	Development of Laboratory Investigations in Disorders of Sex Development. Sexual Development, 2018, 12, 7-18.	1.1	4
14	GATA4 Variants in Individuals With a 46,XY Disorder of Sex Development (DSD) May or May Not Be Associated With Cardiac Defects Depending on Second Hits in Other DSD Genes. Frontiers in Endocrinology, 2018, 9, 142.	1.5	26
15	Mechanism of the Dual Activities of Human CYP17A1 and Binding to Anti-Prostate Cancer Drug Abiraterone Revealed by a Novel V366M Mutation Causing 17,20 Lyase Deficiency. Pharmaceuticals, 2018, 11, 37.	1.7	30
16	Broad phenotypes in heterozygous NR5A1 46,XY patients with a disorder of sex development: an oligogenic origin?. European Journal of Human Genetics, 2018, 26, 1329-1338.	1.4	47
17	Discordant Genotypic Sex and Phenotype Variations in Two Spanish Siblings with 173-Hydroxylase/17,20-Lyase Deficiency Carrying the Most Prevalent Mutated<i>CYP17A1</i> Alleles of Brazilian Patients. Sexual Development, 2017, 11, 70-77.	1.1	8
18	Nutritional rickets: vitamin D, calcium, and the genetic make-up. Pediatric Research, 2017, 81, 356-363.	1.1	6

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19	Sphingosine-1-phosphate lyase mutations cause primary adrenal insufficiency and steroid-resistant nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2017, 127, 942-953.	3.9	139
20	Latitudinal Clines of the Human Vitamin D Receptor and Skin Color Genes. <i>G3: Genes, Genomes, Genetics</i> , 2016, 6, 1251-1266.	0.8	23
21	LRH-1 May Rescue SF-1 Deficiency for Steroidogenesis: An in vitro and in vivo Study. <i>Sexual Development</i> , 2015, 9, 144-154.	1.1	9
22	Complete Androgen Insensitivity Syndrome Associated with Male Gender Identity or Female Precocious Puberty in the Same Family. <i>Sexual Development</i> , 2015, 9, 75-79.	1.1	10
23	Human MAMLD1 Gene Variations Seem Not Sufficient to Explain a 46,XY DSD Phenotype. <i>PLoS ONE</i> , 2015, 10, e0142831.	1.1	19
24	STAR splicing mutations cause the severe phenotype of lipid congenital adrenal hyperplasia: insights from a novel splice mutation and review of reported cases. <i>Clinical Endocrinology</i> , 2014, 80, 191-199.	1.2	15
25	Human NR5A1/SF-1 Mutations Show Decreased Activity on BDNF (Brain-Derived Neurotrophic Factor), an Important Regulator of Energy Balance: Testing Impact of Novel SF-1 Mutations Beyond Steroidogenesis. <i>PLoS ONE</i> , 2014, 9, e104838.	1.1	12
26	Familial short stature and intrauterine growth retardation associated with a novel mutation in the IGF1 receptor (IGF1R) gene. <i>Clinical Endocrinology</i> , 2013, 78, 255-262.	1.2	26
27	Contribution of human growth hormone-releasing hormone receptor (GHRHR) gene sequence variation to isolated severe growth hormone deficiency (ISGHD) and normal adult height. <i>Clinical Endocrinology</i> , 2012, 77, 564-574.	1.2	7
28	Characterization of Novel StAR (Steroidogenic Acute Regulatory Protein) Mutations Causing Non-Classic Lipoid Adrenal Hyperplasia. <i>PLoS ONE</i> , 2011, 6, e20178.	1.1	75
29	Loss of the C Terminus of Melanocortin Receptor 2 (MC2R) Results in Impaired Cell Surface Expression and ACTH Insensitivity. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011, 96, E65-E72.	1.8	10
30	Longitudinal Pubertal Growth According to Age at Pubertal Growth Spurt Onset: Data from a Spanish Study Including 458 Children (223 Boys and 235 Girls). <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2009, 22, 715-26.	0.4	31
31	Rickets in the Middle East: Role of Environment and Genetic Predisposition. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 1743-1750.	1.8	107
32	Growth Hormone (GH) Dose, But Not Exon 3-Deleted/Full-Length GH Receptor Polymorphism Genotypes, Influences Growth Response to Two-Year GH Therapy in Short Small-for-Gestational-Age Children. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 147-153.	1.8	44
33	Human growth hormone (GH1) gene polymorphism map in a normal-statured adult population. <i>Clinical Endocrinology</i> , 2007, 66, 258-268.	1.2	23
34	SRD5A2 gene Q126R exon 2 point mutation in unrelated Spanish male pseudohermaphrodite patients. <i>International Journal on Disability and Human Development</i> , 2005, 4, .	0.2	0
35	Clinical, Biochemical and Morphologic Diagnostic Markers in an Infant Male Pseudohermaphrodite Patient with Compound Heterozygous Mutations (G115D/R246W) in SRD5A2 Gene. <i>Hormone Research in Paediatrics</i> , 2004, 62, 259-264.	0.8	16
36	Compound Heterozygous Mutations in the SRD5A2 Gene Exon 4 in a Male Pseudohermaphrodite Patient of Chinese Origin. <i>Journal of Andrology</i> , 2004, 25, 412-416.	2.0	38

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37	Spectrophotometric Determination of Nitrites in Biological Samples Using 1,2-Diaminoanthraquinone. Potential Application to the Determination of Nitric Oxide Synthase Activity. <i>Analytical Letters</i> , 2003, 36, 2139-2149.	1.0	9
38	Structural requirements of benzodiazepines for the inhibition of pig brain nitric oxide synthase. <i>Molecular Brain Research</i> , 2001, 96, 87-93.	2.5	10
39	Sources of interference in the use of 2,3-diaminonaphthalene for the fluorimetric determination of nitric oxide synthase activity in biological samples. <i>Clinica Chimica Acta</i> , 2001, 312, 205-212.	0.5	29