MÃ³nica FernÃ¡ndez-Cancio

List of Publications by Year
in descending order

39 papers	945 citations	430442 18 h-index	476904 29 g-index
41 all docs	41 docs citations	41 times ranked	1334 citing authors

#	Article	IF	Citations
1	Sphingosine-1-phosphate lyase mutations cause primary adrenal insufficiency and steroid-resistant nephrotic syndrome. Journal of Clinical Investigation, 2017, 127, 942-953.	3.9	139
2	Rickets in the Middle East: Role of Environment and Genetic Predisposition. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 1743-1750.	1.8	107
3	Characterization of Novel StAR (Steroidogenic Acute Regulatory Protein) Mutations Causing Non-Classic Lipoid Adrenal Hyperplasia. PLoS ONE, 2011, 6, e20178.	1.1	75
4	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
5	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. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 147-153.	1.8	44
6	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
7	Compound Heterozygous Mutations in the SRD5A2 Gene Exon 4 in a Male Pseudohermaphrodite Patient of Chinese Origin. Journal of Andrology, 2004, 25, 412-416.	2.0	38
8	Longitudinal Pubertal Growth According to Age at Pubertal Growth Spurt Onset: Data from a Spanish Study Including 458 Children (223 Boys and 235 Girls). Journal of Pediatric Endocrinology and Metabolism, 2009, 22, 715-26.	0.4	31
9	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
10	Sources of interference in the use of 2,3-diaminonaphthalene for the fluorimetric determination of nitric oxide synthase activity in biological samples. Clinica Chimica Acta, 2001, 312, 205-212.	0.5	29
11	Familial short stature and intrauterine growth retardation associated with a novel mutation in the <scp>IGF</scp>â€<scp>I</scp> receptor (<scp>IGF1R</scp>) gene. Clinical Endocrinology, 2013, 78, 255-262.	1.2	26
12	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
13	Human growth hormone (GH1) gene polymorphism map in a normal-statured adult population. Clinical Endocrinology, 2007, 66, 258-268.	1.2	23
14	Latitudinal Clines of the Human Vitamin D Receptor and Skin Color Genes. G3: Genes, Genomes, Genetics, 2016, 6, 1251-1266.	0.8	23
15	Broad Phenotypes of Disorders/Differences of Sex Development in MAMLD1 Patients Through Oligogenic Disease. Frontiers in Genetics, 2019, 10, 746.	1.1	22
16	Human MAMLD1 Gene Variations Seem Not Sufficient to Explain a 46,XY DSD Phenotype. PLoS ONE, 2015, 10, e0142831.	1.1	19
17	Clinical, Biochemical and Morphologic Diagnostic Markers in an Infant Male Pseudohermaphrodite Patient with Compound Heterozygous Mutations (G115D/R246W) in SRD5A2 Gene. Hormone Research in Paediatrics, 2004, 62, 259-264.	0.8	16
18	<scp>STAR</scp> splicing mutations cause the severe phenotype of lipoid congenital adrenal hyperplasia: insights from a novel splice mutation and review of reported cases. Clinical Endocrinology, 2014, 80, 191-199.	1.2	15

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19	Incidence and Prevalence of Children's Diffuse Lung Disease in Spain. Archivos De Bronconeumologia, 2022, 58, 22-29.	0.4	15
20	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. PLoS ONE, 2014, 9, e104838.	1.1	12
21	Isolated pulmonary interstitial glycogenosis associated with alveolar growth abnormalities: A longâ€term followâ€up study. Pediatric Pulmonology, 2019, 54, 837-846.	1.0	11
22	Structural requirements of benzodiazepines for the inhibition of pig brain nitric oxide synthase. Molecular Brain Research, 2001, 96, 87-93.	2.5	10
23	Loss of the C Terminus of Melanocortin Receptor 2 (MC2R) Results in Impaired Cell Surface Expression and ACTH Insensitivity. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E65-E72.	1.8	10
24	Complete Androgen Insensitivity Syndrome Associated with Male Gender Identity or Female Precocious Puberty in the Same Family. Sexual Development, 2015, 9, 75-79.	1.1	10
25	Spectrophotometric Determination of Nitrites in Biological Samples Using 1,2-Diaminoanthraquinone. Potential Application to the Determination of Nitric Oxide Synthase Activity. Analytical Letters, 2003, 36, 2139-2149.	1.0	9
26	LRH-1 May Rescue SF-1 Deficiency for Steroidogenesis: An in vitro and in vivo Study. Sexual Development, 2015, 9, 144-154.	1.1	9
27	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
28	Contribution of human growth hormoneâ€releasing hormone receptor (<scp>GHRHR</scp>) gene sequence variation to isolated severe growth hormone deficiency (<scp>ISGHD</scp>) and normal adult height. Clinical Endocrinology, 2012, 77, 564-574.	1.2	7
29	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
30	Nutritional rickets: vitamin D, calcium, and the genetic make-up. Pediatric Research, 2017, 81, 356-363.	1.1	6
31	A Novel Homozygous AMRH2 Gene Mutation in a Patient with Persistent MÃ¼llerian Duct Syndrome. Sexual Development, 2019, 13, 87-91.	1.1	6
32	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
33	ImplementaciÃ³n de un panel de genes para el diagnÃ³stico genÃ©tico de la discinesia ciliar primaria. Archivos De Bronconeumologia, 2021, 57, 186-194.	0.4	5
34	Papel de la inmunofluorescencia y el diagnÃ³stico molecular en la caracterizaciÃ³n de la discinesia ciliar primaria. Archivos De Bronconeumologia, 2019, 55, 439-441.	0.4	5
35	Development of Laboratory Investigations in Disorders of Sex Development. Sexual Development, 2018, 12, 7-18.	1.1	4
36	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 NutriciÃ“n, 2021, 68, 612-620.	0.1	2

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37	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. EndocrinologÃa Diabetes Y NutriciÃ³n (English Ed), 2021, 68, 612-620.	0.1	2
38	Role of Immunofluorescence and Molecular Diagnosis in the Characterization of Primary Ciliary Dyskinesia. Archivos De Bronconeumologia, 2019, 55, 439-441.	0.4	1
39	SRD5A2 gene Q126R exon 2 point mutation in unrelated Spanish male pseudohermaphrodite patients.. International Journal on Disability and Human Development, 2005, 4, .	0.2	0