

Monica Fernandez-Cancio

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

38
papers

655
citations

14
h-index

24
g-index

41
ext. papers

814
ext. citations

3.3
avg, IF

3.23
L-index

#	Paper	IF	Citations
38	Sphingosine-1-phosphate lyase mutations cause primary adrenal insufficiency and steroid-resistant nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2017 , 127, 942-953	15.9	89
37	Rickets in the Middle East: role of environment and genetic predisposition. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008 , 93, 1743-50	5.6	89
36	Characterization of novel StAR (steroidogenic acute regulatory protein) mutations causing non-classic lipid adrenal hyperplasia. <i>PLoS ONE</i> , 2011 , 6, e20178	3.7	57
35	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, 147-53	5.6	39
34	Compound heterozygous mutations in the SRD5A2 gene exon 4 in a male pseudohermaphrodite patient of Chinese origin. <i>Journal of Andrology</i> , 2004 , 25, 412-6		31
33	Broad phenotypes in heterozygous NR5A1 46,XY patients with a disorder of sex development: an oligogenic origin?. <i>European Journal of Human Genetics</i> , 2018 , 26, 1329-1338	5.3	27
32	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, 205-12	6.2	26
31	Familial short stature and intrauterine growth retardation associated with a novel mutation in the IGF-I receptor (IGF1R) gene. <i>Clinical Endocrinology</i> , 2013 , 78, 255-62	3.4	23
30	Expanding the Clinical and Genetic Spectra of Primary Immunodeficiency-Related Disorders With Clinical Exome Sequencing: Expected and Unexpected Findings. <i>Frontiers in Immunology</i> , 2019 , 10, 2325	8.4	20
29	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	1.6	20
28	Human growth hormone (GH1) gene polymorphism map in a normal-statured adult population. <i>Clinical Endocrinology</i> , 2007 , 66, 258-68	3.4	19
27	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. <i>Pharmaceuticals</i> , 2018 , 11,	5.2	18
26	Latitudinal Clines of the Human Vitamin D Receptor and Skin Color Genes. <i>G3: Genes, Genomes, Genetics</i> , 2016 , 6, 1251-66	3.2	16
25	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. <i>Frontiers in Endocrinology</i> , 2018 , 9, 142	5.7	14
24	Human MAMLD1 Gene Variations Seem Not Sufficient to Explain a 46,XY DSD Phenotype. <i>PLoS ONE</i> , 2015 , 10, e0142831	3.7	14
23	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, 191-94	3.4	13
22	Broad Phenotypes of Disorders/Differences of Sex Development in Patients Through Oligogenic Disease. <i>Frontiers in Genetics</i> , 2019 , 10, 746	4.5	10

21	Complete androgen insensitivity syndrome associated with male gender identity or female precocious puberty in the same family. <i>Sexual Development</i> , 2015 , 9, 75-9	1.6	10
20	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, E65-72	5.6	10
19	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, 259-64	3.3	9
18	Structural requirements of benzodiazepines for the inhibition of pig brain nitric oxide synthase. <i>Molecular Brain Research</i> , 2001 , 96, 87-93		9
17	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	3.7	9
16	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	2.2	8
15	Isolated pulmonary interstitial glycogenosis associated with alveolar growth abnormalities: A long-term follow-up study. <i>Pediatric Pulmonology</i> , 2019 , 54, 837-846	3.5	7
14	LRH-1 May Rescue SF-1 Deficiency for Steroidogenesis: An in vitro and in vivo Study. <i>Sexual Development</i> , 2015 , 9, 144-54	1.6	6
13	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, 564-74	3.4	6
12	Discordant Genotypic Sex and Phenotype Variations in Two Spanish Siblings with 17 β -Hydroxylase/17,20-Lyase Deficiency Carrying the Most Prevalent Mutated CYP17A1 Alleles of Brazilian Patients. <i>Sexual Development</i> , 2017 , 11, 70-77	1.6	5
11	Role of Immunofluorescence and Molecular Diagnosis in the Characterization of Primary Ciliary Dyskinesia. <i>Archivos De Bronconeumologia</i> , 2019 , 55, 439-441	0.7	5
10	Implementation of a Gene Panel for Genetic Diagnosis of Primary Ciliary Dyskinesia. <i>Archivos De Bronconeumologia</i> , 2021 , 57, 186-194	0.7	4
9	A Novel Homozygous AMRH2 Gene Mutation in a Patient with Persistent Müllerian Duct Syndrome. <i>Sexual Development</i> , 2019 , 13, 87-91	1.6	3
8	Nutritional rickets: vitamin D, calcium, and the genetic make-up. <i>Pediatric Research</i> , 2017 , 81, 356-363	3.2	2
7	Development of Laboratory Investigations in Disorders of Sex Development. <i>Sexual Development</i> , 2018 , 12, 7-18	1.6	2
6	Role of Immunofluorescence and Molecular Diagnosis in the Characterization of Primary Ciliary Dyskinesia. <i>Archivos De Bronconeumologia</i> , 2019 , 55, 439-441	0.7	1
5	Immunofluorescence Analysis as a Diagnostic Tool in a Spanish Cohort of Patients with Suspected Primary Ciliary Dyskinesia. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	1
4	Incidence and Prevalence of Children's Diffuse Lung Disease in Spain.. <i>Archivos De Bronconeumologia</i> , 2022 , 58, 22-29	0.7	1

3	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. <i>Endocrinología, Diabetes Y Nutrición</i> , 2021 ,	1.3	1
2	Phenotypic Variability of Patients With PAX8 Variants Presenting With Congenital Hypothyroidism and Eutopic Thyroid. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 , 106, e152-e170	5.6	1
1	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.. <i>Endocrinología Diabetes Y Nutrición (English Ed)</i> , 2021 , 68, 612-620	0.1	