Mnica Fernndez-Cancio

List of Publications by Year in Descending Order

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	Incidence and Prevalence of Children's Diffuse Lung Disease in Spain.. *Archivos De Bronconeumologia*, **2022**, 58, 22-29	0.7	1
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.. *Endocrinología Diabetes Y Nutrición (English Ed)*, **2021**, 68, 612-620	0.1	
36	Implementation of a Gene Panel for Genetic Diagnosis of Primary Ciliary Dyskinesia. *Archivos De Bronconeumologia*, **2021**, 57, 186-194	0.7	4
35	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 Nutrición*, **2021**,	1.3	1
34	Phenotypic Variability of Patients With PAX8 Variants Presenting With Congenital Hypothyroidism and Eutopic Thyroid. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, e152-e170	5.6	1
33	Immunofluorescence Analysis as a Diagnostic Tool in a Spanish Cohort of Patients with Suspected Primary Ciliary Dyskinesia. *Journal of Clinical Medicine*, **2020**, 9,	5.1	1
32	Broad Phenotypes of Disorders/Differences of Sex Development in Patients Through Oligogenic Disease. *Frontiers in Genetics*, **2019**, 10, 746	4.5	10
31	Isolated pulmonary interstitial glycogenosis associated with alveolar growth abnormalities: A long-term follow-up study. *Pediatric Pulmonology*, **2019**, 54, 837-846	3.5	7
30	A Novel Homozygous AMRH2 Gene Mutation in a Patient with Persistent Müllerian Duct Syndrome. *Sexual Development*, **2019**, 13, 87-91	1.6	3
29	Role of Immunofluorescence and Molecular Diagnosis in the Characterization of Primary Ciliary Dyskinesia. *Archivos De Bronconeumologia*, **2019**, 55, 439-441	0.7	1
28	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	8.4	20
27	Role of Immunofluorescence and Molecular Diagnosis in the Characterization of Primary Ciliary Dyskinesia. *Archivos De Bronconeumologia*, **2019**, 55, 439-441	0.7	5
26	Development of Laboratory Investigations in Disorders of Sex Development. *Sexual Development*, **2018**, 12, 7-18	1.6	2
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. *Frontiers in Endocrinology*, **2018**, 9, 142	5.7	14
24	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,	5.2	18
23	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	5.3	27
22	Discordant Genotypic Sex and Phenotype Variations in Two Spanish Siblings with 17α-Hydroxylase/17,20-Lyase Deficiency Carrying the Most Prevalent Mutated CYP17A1 Alleles of Brazilian Patients. *Sexual Development*, **2017**, 11, 70-77	1.6	5

21	Nutritional rickets: vitamin D, calcium, and the genetic make-up. *Pediatric Research*, **2017**, 81, 356-363	3.2	2
20	Sphingosine-1-phosphate lyase mutations cause primary adrenal insufficiency and steroid-resistant nephrotic syndrome. *Journal of Clinical Investigation*, **2017**, 127, 942-953	15.9	89
19	Latitudinal Clines of the Human Vitamin D Receptor and Skin Color Genes. *G3: Genes, Genomes, Genetics*, **2016**, 6, 1251-66	3.2	16
18	LRH-1 May Rescue SF-1 Deficiency for Steroidogenesis: An in vitro and in vivo Study. *Sexual Development*, **2015**, 9, 144-54	1.6	6
17	Complete androgen insensitivity syndrome associated with male gender identity or female precocious puberty in the same family. *Sexual Development*, **2015**, 9, 75-9	1.6	10
16	Human MAMLD1 Gene Variations Seem Not Sufficient to Explain a 46,XY DSD Phenotype. *PLoS ONE* , **2015**, 10, e0142831	3.7	14
15	STAR 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, 191-9	3.4	13
14	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	3.7	9
13	Familial short stature and intrauterine growth retardation associated with a novel mutation in the IGF-I receptor (IGF1R) gene. *Clinical Endocrinology*, **2013**, 78, 255-62	3.4	23
12	Contribution of human growth hormone-releasing hormone receptor (GHRHR) gene sequence variation to isolated severe growth hormone deficiency (ISGHD) and normal adult height. *Clinical Endocrinology*, **2012**, 77, 564-74	3.4	6
11	Characterization of novel StAR (steroidogenic acute regulatory protein) mutations causing non-classic lipoid adrenal hyperplasia. *PLoS ONE*, **2011**, 6, e20178	3.7	57
10	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, E65-72	5.6	10
9	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	1.6	20
8	Rickets in the Middle East: role of environment and genetic predisposition. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 1743-50	5.6	89
7	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, 147-53	5.6	39
6	Human growth hormone (GH1) gene polymorphism map in a normal-statured adult population. *Clinical Endocrinology*, **2007**, 66, 258-68	3.4	19
5	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, 259-64	3.3	9
4	Compound heterozygous mutations in the SRD5A2 gene exon 4 in a male pseudohermaphrodite patient of Chinese origin. *Journal of Andrology*, **2004**, 25, 412-6		31

3	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	2.2	8
2	Structural requirements of benzodiazepines for the inhibition of pig brain nitric oxide synthase. *Molecular Brain Research*, **2001**, 96, 87-93		9
1	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, 205-12	6.2	26