Ariadna GonzÃ¡lez-Del Angel

List of Publications by Year
in descending order

34 papers	287 citations	933447 10 h-index	996975 15 g-index
36 all docs	36 docs citations	36 times ranked	484 citing authors

#	Article	IF	Citations
1	Analysis of the CTNS Gene in Nephropathic Cystinosis Mexican Patients: Report of Four Novel Mutations and Identification of a False Positive 57-kb Deletion Genotype with LDM-2/Exon 4 Multiplex PCR Assay. Genetic Testing and Molecular Biomarkers, 2008, 12, 409-414.	1.7	28
2	Effects of Fructans from Mexican Agave in Newborns Fed with Infant Formula: A Randomized Controlled Trial. Nutrients, 2015, 7, 8939-8951.	4.1	28
3	Expanding the clinical features of autoinflammation and phospholipase CÎ³2â€associated antibody deficiency and immune dysregulation by description of a novel patient. Journal of the European Academy of Dermatology and Venereology, 2019, 33, 2334-2339.	2.4	25
4	Delayed membranous cranial ossification in a mother and child. American Journal of Medical Genetics Part A, 1992, 44, 786-789.	2.4	17
5	5,10â€Methylenetetrahydrofolate reductase single nucleotide polymorphisms and geneâ€“environment interaction analysis in nonâ€syndromic cleft lip/palate. European Journal of Oral Sciences, 2014, 122, 109-113.	1.5	17
6	First comprehensive TSC1/TSC2 mutational analysis in Mexican patients with Tuberous Sclerosis Complex reveals numerous novel pathogenic variants. Scientific Reports, 2020, 10, 6589.	3.3	16
7	Association of interactions among the IRF6 gene, the 8q24 region, and maternal folic acid intake with nonâ€syndromic cleft lip/palate in Mexican Mestizos. American Journal of Medical Genetics, Part A, 2012, 158A, 3207-3210.	1.2	15
8	Gene Interactions Provide Evidence for Signaling Pathways Involved in Cleft Lip/Palate in Humans. Journal of Dental Research, 2016, 95, 1257-1264.	5.2	14
9	A Patient with Trisomy 13 Mosaicism with an Unusual Skin Pigmentary Pattern and Prolonged Survival. Pediatric Dermatology, 2014, 31, 580-583.	0.9	12
10	Expansion of the variable expression of Muenke syndrome: Hydrocephalus without craniosynostosis. American Journal of Medical Genetics, Part A, 2016, 170, 3189-3196.	1.2	12
11	Molecular diagnosis of the fragile X and FRAXE syndromes in patients with mental retardation of unknown cause in Mexico. Annales De GÃ©nÃ©tique, 2000, 43, 29-34.	0.4	9
12	Germline Mutations in NKX2-5, GATA4, and CRELD1 are Rare in a Mexican Sample of Down Syndrome Patients with Endocardial Cushion and Septal Heart Defects. Pediatric Cardiology, 2015, 36, 802-808.	1.3	9
13	Deletion of Exon 1 of the SLC16A2 Gene: A Common Occurrence in Patients with Allan-Herndon-Dudley Syndrome. Thyroid, 2015, 25, 361-367.	4.5	7
14	An Updated PAH Mutational Spectrum of Phenylketonuria in Mexican Patients Attending a Single Center: Biochemical, Clinical-Genotyping Correlations. Genes, 2021, 12, 1676.	2.4	7
15	Screening of Late-Onset Pompe Disease in a Sample of Mexican Patients With Myopathies of Unknown Etiology: Identification of a Novel Mutation in the Acid Î±-Glucosidase Gene. Journal of Child Neurology, 2010, 25, 1034-1037.	1.4	6
16	Molecular analysis of the PAX8 gene in a sample of Mexican patients with primary congenital hypothyroidism: identification of the recurrent p.Arg31His mutation. Clinical Endocrinology, 2012, 76, 148-150.	2.4	6
17	Predominance of Dystrophinopathy Genotypes in Mexican Male Patients Presenting as Muscular Dystrophy with A Normal Multiplex Polymerase Chain Reaction DMD Gene Result: A Study Including Targeted Next-Generation Sequencing. Genes, 2019, 10, 856.	2.4	6
18	Clinical characterization and identification of five novel FOXL2 pathogenic variants in a cohort of 12 Mexican subjects with the syndrome of blepharophimosis-ptosis-epicanthus inversus. Gene, 2019, 706, 62-68.	2.2	6

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19	TSC2/PKD1 contiguous gene syndrome, with emphasis on a case with an atypical mild polycystic kidney phenotype and a novel genetic variant. Nefrologia, 2020, 40, 91-98.	0.4	6
20	Genetic spectrum and clinical early natural history of glucose-6-phosphate dehydrogenase deficiency in Mexican children detected through newborn screening. Orphanet Journal of Rare Diseases, 2021, 16, 103.	2.7	6
21	Mutational spectrum of PTS gene and in silico pathological assessment of a novel variant in Mexico. Brain and Development, 2018, 40, 530-536.	1.1	5
22	An uncommon inheritance pattern in Niemann-Pick disease type C: identification of probable paternal germline mosaicism in a Mexican family. BMC Neurology, 2016, 16, 147.	1.8	4
23	Identification of a novel SLC12A6 pathogenic variant associated with hereditary motor and sensory neuropathy with agenesis of the corpus callosum (HMSN/ACC) in a non-French-Canadian family. Neurology India, 2018, 66, 1162.	0.4	4
24	Molecular Analysis Confirms that FKRP-Related Disorders are Underdiagnosed in Mexican Patients with Neuromuscular Diseases. Neuropediatrics, 2017, 48, 442-450.	0.6	3
25	Molecular characterization of Axenfeld-Rieger spectrum and other anterior segment dysgeneses in a sample of Mexican patients. Ophthalmic Genetics, 2018, 39, 728-734.	1.2	3
26	Kapurâ€“Toriello syndrome: Further delineation. American Journal of Medical Genetics, Part A, 2008, 146A, 2791-2793.	1.2	2
27	Unique association of hypochondroplasia with craniosynostosis and cleft palate in a Mexican family. American Journal of Medical Genetics, Part A, 2018, 176, 161-166.	1.2	2
28	Molecular analysis provides further evidence that Chitayat syndrome is caused by the recurrent p.(Tyr89Cys) pathogenic variant in the ERF gene. American Journal of Medical Genetics, Part A, 2019, 179, 118-122.	1.2	2
29	TSC2/PKD1 contiguous gene syndrome, with emphasis on a case with an atypical mild polycystic kidney phenotype and a novel genetic variant. Nefrologia, 2020, 40, 91-98.	0.4	2
30	Further Evidence That Defects in Main Thyroid Dysgenesis-Related Genes Are an Uncommon Etiology for Primary Congenital Hypothyroidism in Mexican Patients: Report of Rare Variants in FOXE1, NKX2-5 and TSHR. Children, 2021, 8, 457.	1.5	2
31	Proposed clinical approach and imaging studies in families with oculoâ€auriculoâ€vertebral spectrum to assess variable expressivity. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
32	Gene Variants in NKX2-1 Do Not Represent a Major Etiological Factor of Primary Congenital Hypothyroidism in Mexican Population. Journal of Pediatric Genetics, 2019, 08, 041-046.	0.7	1
33	Unusual Clinical Manifestations in a Mexican Patient with Sanfilippo B Syndrome. Diagnostics, 2022, 12, 1268.	2.6	1
34	Further delineation of achondroplasiaâ€“hypochondroplasia complex with longâ€term survival. American Journal of Medical Genetics, Part A, 2018, 176, 1225-1231.	1.2	0