Hugo H Abarca-Barriga

List of Publications by Year
in descending order

24 papers	290 citations	1477746 6 h-index	887659 17 g-index
27 all docs	27 docs citations	27 times ranked	640 citing authors

#	Article	IF	Citations
1	Microduplications encompassing the Sonic hedgehog limb enhancer <scp>ZRS</scp> are associated with Haasâ€type polysyndactyly and Laurinâ€Sandrow syndrome. Clinical Genetics, 2014, 86, 318-325.	1.0	72
2	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	0.7	68
3	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	0.7	55
4	A novel ASPH variant extends the phenotype of Shawafâ€Traboulsi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2494-2500.	0.7	17
5	Genetics and genomics in Peru: Clinical and research perspective. Molecular Genetics & Genomic Medicine, 2018, 6, 873-886.	0.6	12
6	Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	1.1	8
7	Ocular pterygiumâ€“Digital keloid dysplasia. American Journal of Medical Genetics, Part A, 2014, 164, 2901-2907.	0.7	7
8	A Peruvian Child with 18p-/18q+ Syndrome and Persistent Microscopic Hematuria. Journal of Pediatric Genetics, 2017, 06, 258-266.	0.3	7
9	Novel contiguous gene deletion in peruvian girl with Trichothiodystrophy type 4 and glutaric aciduria type 3. European Journal of Medical Genetics, 2018, 61, 388-392.	0.7	7
10	MLPA followed by targetâ€NGS to detect mutations in the dystrophin gene of Peruvian patients suspected of DMD/DMB. Molecular Genetics & Genomic Medicine, 2021, 9, e1759.	0.6	5
11	Factores de riesgo en las enfermedades genÃ©ticas. Acta Medica Peruana, 2018, 35, 43-50.	0.3	4
12	Homozygous Deletion of the CFTR Gene Caused by Interstitial Maternal Isodisomy in a Peruvian Child with Cystic Fibrosis. Journal of Pediatric Genetics, 2019, 08, 147-152.	0.3	2
13	GATAD2B Gene Microdeletion Causing Intellectual Disability Autosomal Dominant Type 18: Case Report and Review of the Literature. Molecular Syndromology, 2019, 10, 186-194.	0.3	2
14	Peruvian Newborn Male with 3p13 Deletion Syndrome Encompassing the FOXP1 Gene: Review of the Literature. Journal of Pediatric Genetics, 2020, 09, 270-278.	0.3	2
15	Management of genetic diseases: Present and future. Revista De La Facultad De Medicina Humana, 2021, 21, 399-416.	0.1	2
16	High prevalence of congenital generalized lipodystrophy in Piura, Peru. Intractable and Rare Diseases Research, 2020, 9, 58-60.	0.3	1
17	Avances genÃ³micos de la Ãºltima dÃ©cada y su influencia en el enfoque diagnÃ³stico de la discapacidad intelectual.. Revista De Neuro-psiquiatria, 2021, 84, 33-50.	0.0	1
18	Variantes en el nÃºmero de copias en trastornos del neurodesarrollo, sÃndrome malformativo y talla baja en PerÃº. Acta Medica Peruana, 2020, 37, .	0.3	1

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19	AmpliaciÃ³n del tamizaje de errores innatos del metabolismo en PerÃº: reporte de caso con trastorno del metabolismo de cobalamina. Acta Medica Peruana, 2020, 37, .	0.3	1
20	Variantes en el nÃºmero de copias y consanguinidad parental en neonatos de altura con anomalÃas congÃ©nitas en PerÃº. Revista De La Facultad De Ciencias Medicas De Cordoba, 2022, 79, 132-140.	0.1	1
21	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
22	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
23	Importancia de la determinaciÃ³n de variantes en el nÃºmero de copias en neonatos con aneuploidÃas autosÃ³micas. Biomedica, 2021, 41, 282-292.	0.3	0
24	Genetic basis of pulmonary arterial hypertension. Revista De La Facultad De Medicina Humana, 2020, 20, 670-681.	0.1	0