Francisco J GuzmÃ¡n-Vega

List of Publications by Year
in descending order

10 papers	258 citations	1307594 7 h-index	1372567 10 g-index
12 all docs	12 docs citations	12 times ranked	589 citing authors

#	Article	IF	Citations
1	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
2	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	7.6	70
3	The genome of the zoonotic malaria parasite Plasmodium simium reveals adaptations to host switching. BMC Biology, 2021, 19, 219.	3.8	21
4	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
5	Novel Missense Variant in Heterozygous State in the BRPF1 Gene Leading to Intellectual Developmental Disorder With Dysmorphic Facies and Ptosis. Frontiers in Genetics, 2020, 11, 368.	2.3	13
6	Interleukin-26 activates macrophages and facilitates killing of Mycobacterium tuberculosis. Scientific Reports, 2020, 10, 17178.	3.3	12
7	A Novel Biallelic STING1 Gene Variant Causing SAVI in Two Siblings. Frontiers in Immunology, 2020, 11, 599564.	4.8	12
8	<scp>MYH1</scp> is a candidate gene for recurrent rhabdomyolysis in humans. American Journal of Medical Genetics, Part A, 2021, 185, 2131-2135.	1.2	8
9	Clinical and Genetic Characterization of Craniosynostosis in Saudi Arabia. Frontiers in Pediatrics, 2021, 9, 582816.	1.9	5
10	Truncating biallelic variant in DNAJA1, encoding the co-chaperone Hsp40, is associated with intellectual disability and seizures. Neurogenetics, 2019, 20, 109-115.	1.4	3