Jaime Vengoechea

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5406919/publications.pdf

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8 papers	94 citations	1937685 4 h-index	1720034 7 g-index
8 all docs	8 docs citations	8 times ranked	253 citing authors

#	Article	IF	Citations
1	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood, 2021, 137, 493-499.	1.4	26
2	Two females with distinct de novo missense pathogenic variants in <scp>MED12</scp> and vastly differing phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 2582-2585.	1.2	0
3	Segregation of two variants suggests the presence of autosomal dominant and recessive forms of WFS1-related disease within the same family: expanding the phenotypic spectrum of Wolfram Syndrome. Journal of Medical Genetics, 2020, 57, 121-123.	3.2	6
4	Combined occurrence of deleterious TOR1A and ANO3 variants in isolated generalized dystonia. Parkinsonism and Related Disorders, 2020, 73, 55-56.	2.2	1
5	A case of Singletonâ€“Merten syndrome without cardiac involvement harboring a novel <scp>IFIH1</scp> variant. American Journal of Medical Genetics, Part A, 2020, 182, 1535-1536.	1.2	4
6	In reply to â€œMast Cell Disorders in Ehlersâ€“Danlos Syndromeâ€• American Journal of Medical Genetics, Part A, 2018, 176, 250-250.	1.2	2
7	A splice site mutation in ATP6AP2 causes X-linked intellectual disability, epilepsy, and parkinsonism. Parkinsonism and Related Disorders, 2015, 21, 1473-1475.	2.2	30
8	Methylation markers of malignant potential in meningiomas. Journal of Neurosurgery, 2013, 119, 899-906.	1.6	25