Emanuele Panza

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3514903/publications.pdf

Version: 2024-02-01

26 papers	1,008 citations	567281 15 h-index	580821 25 g-index
26 all docs	26 docs citations	26 times ranked	1365 citing authors

#	Article	IF	Citations
1	MYH9-Related Disease. Medicine (United States), 2003, 82, 203-215.	1.0	255
2	Position of nonmuscle myosin heavy chain IIA (NMMHC-IIA) mutations predicts the natural history ofMYH9-related disease. Human Mutation, 2008, 29, 409-417.	2.5	172
3	New perspectives in the diagnosis and management of enteric neuropathies. Nature Reviews Gastroenterology and Hepatology, 2013, 10, 206-218.	17.8	97
4	Heavy chain myosin 9-related disease (MYH9-RD): Neutrophil inclusions of myosin-9 as a pathognomonic sign of the disorder. Thrombosis and Haemostasis, 2010, 103, 826-832.	3.4	81
5	ALDH18A1gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism. Brain, 2016, 139, e3-e3.	7.6	42
6	A family with autosomal dominant leukodystrophy linked to 5q23.2â€“q23.3 without lamin B1 mutations. European Journal of Neurology, 2010, 17, 541-549.	3.3	36
7	Genetics of human enteric neuropathies. Progress in Neurobiology, 2012, 96, 176-189.	5.7	36
8	Genetic Predisposition to Familial Neuroblastoma: Identification of Two Novel Genomic Regions at 2p and 12p. Human Heredity, 2007, 63, 205-211.	0.8	34
9	MYH9 related disease: four novel mutations of the tail domain of myosinâ€9 correlating with a mild clinical phenotype. European Journal of Haematology, 2010, 84, 291-297.	2.2	32
10	Title is missing!. Medicine (United States), 2003, 82, 203-215.	1.0	30
11	A novel locus for syndromic chronic idiopathic intestinal pseudo-obstruction maps to chromosome 8q23â€“q24. European Journal of Human Genetics, 2007, 15, 889-897.	2.8	29
12	Mutations responsible for MYH9-related thrombocytopenia impair SDF-1-driven migration of megakaryoblastic cells. Thrombosis and Haemostasis, 2011, 106, 693-704.	3.4	24
13	Hereditary Spastic Paraplegia Is a Common Phenotypic Finding in ARG1 Deficiency, P5CS Deficiency and HHH Syndrome: Three Inborn Errors of Metabolism Caused by Alteration of an Interconnected Pathway of Glutamate and Urea Cycle Metabolism. Frontiers in Neurology, 2019, 10, 131.	2.4	24
14	Association of hereditary thrombocythemia and distal limb defects with a thrombopoietin gene mutation. Blood, 2009, 114, 1655-1657.	1.4	21
15	Î”¹â€Pyrrolineâ€5â€carboxylate synthetase deficiency: An emergent multifaceted urea cycleâ€related disorder. Journal of Inherited Metabolic Disease, 2020, 43, 657-670.	3.6	20
16	The clear cell sarcoma functional genomic landscape. Journal of Clinical Investigation, 2021, 131, .	8.2	15
17	P5CS expression study in a new family with ALDH18A1â€associated hereditary spastic paraplegia SPG9. Annals of Clinical and Translational Neurology, 2019, 6, 1533-1540.	3.7	14
18	Autosomal recessive hereditary spastic paraplegia with thin corpus callosum: a novel mutation in the SPG11 gene and further evidence for genetic heterogeneity. European Journal of Neurology, 2009, 16, 121-126.	3.3	13

#	Article	IF	Citations
19	Refinement of the SPG9 locus on chromosome 10q23.3â€24.2 and exclusion of candidate genes. European Journal of Neurology, 2008, 15, 520-524.	3.3	8
20	A heritable cause of cleft lip and palateâ€"Van der Woude syndrome caused by a novel IRF6 mutation. Review of the literature and of the differential diagnosis. European Journal of Pediatrics, 2010, 169, 223-228.	2.7	7
21	Transfection of the mutant MYH9 cDNA reproduces the most typical cellular phenotype of MYH9-related disease in different cell lines. PathoGenetics, 2008, 1, 5.	5.7	5
22	The breakpoint identified in a balanced de novo translocation t(7;9)(p14.1;q31.3) disrupts the A-kinase (PRKA) anchor protein 2 gene (AKAP2) on chromosome 9 in a patient with Kallmann syndrome and bone anomalies. International Journal of Molecular Medicine, 2007, 19, 429.	4.0	4
23	Delineation of the 9q31 deletion syndrome: Genomic microarray characterization of two patients with overlapping deletions. American Journal of Medical Genetics, Part A, 2018, 176, 2901-2906.	1.2	4
24	Benign albeit glycolytic: MCT4 expression and lactate release in giant cell tumour of bone. Bone, 2020, 134, 115302.	2.9	4
25	Application of a Fluorescent PCR Method for Molecular Diagnosis of Posttransplant Lymphoproliferative Disorders on Routine Tissue Sections. Diagnostic Molecular Pathology, 2005, 14, 170-176.	2.1	1
26	Seizures and Cardiomyopathy in a Patient with Pallister-Killian Syndrome due to Hexasomy 12p Mosaicism. Molecular Syndromology, 2020, 11, 125-129.	0.8	0