Emanuele Panza

List of Publications by Year
in descending order

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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	The clear cell sarcoma functional genomic landscape. Journal of Clinical Investigation, 2021, 131, .	8.2	15
2	Benign albeit glycolytic: MCT4 expression and lactate release in giant cell tumour of bone. Bone, 2020, 134, 115302.	2.9	4
3	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
4	Seizures and Cardiomyopathy in a Patient with Pallister-Killian Syndrome due to Hexasomy 12p Mosaicism. Molecular Syndromology, 2020, 11, 125-129.	0.8	0
5	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
6	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
7	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
8	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
9	New perspectives in the diagnosis and management of enteric neuropathies. Nature Reviews Gastroenterology and Hepatology, 2013, 10, 206-218.	17.8	97
10	Genetics of human enteric neuropathies. Progress in Neurobiology, 2012, 96, 176-189.	5.7	36
11	Mutations responsible for MYH9-related thrombocytopenia impair SDF-1-driven migration of megakaryoblastic cells. Thrombosis and Haemostasis, 2011, 106, 693-704.	3.4	24
12	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
13	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
14	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
15	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
16	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
17	Association of hereditary thrombocythemia and distal limb defects with a thrombopoietin gene mutation. Blood, 2009, 114, 1655-1657.	1.4	21
18	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

#	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	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
21	Genetic Predisposition to Familial Neuroblastoma: Identification of Two Novel Genomic Regions at 2p and 12p. Human Heredity, 2007, 63, 205-211.	0.8	34
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	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
24	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
25	Title is missing!. Medicine (United States), 2003, 82, 203-215.	1.0	30
26	MYH9-Related Disease. Medicine (United States), 2003, 82, 203-215.	1.0	255