Patrizia De-Marco

List of Publications by Year
in descending order

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64 papers	2,437 citations	201674 27 h-index	214800 47 g-index
68 all docs	68 docs citations	68 times ranked	2906 citing authors

#	Article	IF	Citations
1	Spinal involvement in pediatric familial cavernous malformation syndrome. Neuroradiology, 2022, , 1.	2.2	1
2	Dyslipidemia in Children Treated with a BRAF Inhibitor for Low-Grade Gliomas: A New Side Effect?. Cancers, 2022, 14, 2693.	3.7	2
3	Genomic Analysis Made It Possible to Identify Gene-Driver Alterations Covering the Time Window between Diagnosis of Neuroblastoma 4S and the Progression to Stage 4. International Journal of Molecular Sciences, 2022, 23, 6513.	4.1	6
4	Thymosin Alpha 1 Mitigates Cytokine Storm in Blood Cells From Coronavirus Disease 2019 Patients. Open Forum Infectious Diseases, 2021, 8, ofaa588.	0.9	27
5	RNF213 variant in a patient with Legius syndrome associated with moyamoya syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1669.	1.2	3
6	The first case of mosaic MNX1 mutation in an adult female with features of Currarino syndrome. Birth Defects Research, 2021, 113, 1161-1165.	1.5	0
7	L1CAM variants cause two distinct imaging phenotypes on fetal MRI. Annals of Clinical and Translational Neurology, 2021, 8, 2004-2012.	3.7	8
8	Role of diffusion weighted imaging for differentiating cerebral pilocytic astrocytoma and ganglioglioma BRAF V600E-mutant from wild type. Neuroradiology, 2020, 62, 71-80.	2.2	13
9	Functional Validation of CLDN Variants Identified in a Neural Tube Defect Cohort Demonstrates Their Contribution to Neural Tube Defects. Frontiers in Neuroscience, 2020, 14, 664.	2.8	5
10	Sinus pericranii, skull defects, and structural brain anomalies in TRAF7 â€related disorder. Birth Defects Research, 2020, 112, 1085-1092.	1.5	5
11	Moyamoya vasculopathy shows a genetic mutational gradient decreasing from East to West. Journal of Neurosurgical Sciences, 2020, 64, 165-172.	0.6	17
12	Nutritional characterisation of Zambian Moringa oleifera: acceptability and safety of short-term daily supplementation in a group of malnourished girls. International Journal of Food Sciences and Nutrition, 2019, 70, 107-115.	2.8	21
13	Chiari malformation type I: what information from the genetics?. Child's Nervous System, 2019, 35, 1665-1671.	1.1	12
14	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
15	Update on the Role of the Non-Canonical Wnt/Planar Cell Polarity Pathway in Neural Tube Defects. Cells, 2019, 8, 1198.	4.1	55
16	Migrating focal seizures in Autosomal Dominant Sleep-related Hypermotor Epilepsy with KCNT1 mutation. Seizure: the Journal of the British Epilepsy Association, 2019, 67, 57-60.	2.0	10
17	Whole exome sequencing identifies novel predisposing genes in neural tube defects. Molecular Genetics & Genomic Medicine, 2019, 7, e00467.	1.2	25
18	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86

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19	Rare deleterious variants in GRHL3 are associated with human spina bifida. Human Mutation, 2017, 38, 716-724.	2.5	28
20	Exome sequencing of two Italian pedigrees with non-isolated Chiari malformation type I reveals candidate genes for cranio-facial development. European Journal of Human Genetics, 2017, 25, 952-959.	2.8	18
21	Scribble1 plays an important role in the pathogenesis of neural tube defects through its mediating effect of Par-3 and Vangl1/2 localization. Human Molecular Genetics, 2017, 26, 2307-2320.	2.9	26
22	Spinal lipoma as a dysembryogenetic anomaly: Four unusual cases of ectopic iliac rib within the spinal lipoma. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 530-535.	1.6	1
23	Genetic Screening of Pediatric Cavernous Malformations. Journal of Molecular Neuroscience, 2016, 60, 232-238.	2.3	18
24	Sacral agenesis: a pilot whole exome sequencing and copy number study. BMC Medical Genetics, 2016, 17, 98.	2.1	15
25	Role of the planar cell polarity gene Protein tyrosine kinase 7 in neural tube defects in humans. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 1021-1027.	1.6	27
26	Expanding the mutational spectrum associated to neural tube defects: Literature revision and description of novel VANGL1 mutations. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 51-61.	1.6	28
27	Loss-of-function de novo mutations play an important role in severe human neural tube defects. Journal of Medical Genetics, 2015, 52, 493-497.	3.2	65
28	Cost effective assay choice for rare disease study designs. Orphanet Journal of Rare Diseases, 2015, 10, 10.	2.7	3
29	Genetic studies of ANKRD6 as a molecular switch between Wnt signaling pathways in human neural tube defects. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 20-26.	1.6	21
30	Novel mutations in Lrp6 orthologs in mouse and human neural tube defects affect a highly dosage-sensitive Wnt non-canonical planar cell polarity pathway. Human Molecular Genetics, 2014, 23, 4185-4185.	2.9	0
31	Planar cell polarity gene mutations contribute to the etiology of human neural tube defects in our population. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 633-641.	1.6	40
32	Novel mutations in Lrp6 orthologs in mouse and human neural tube defects affect a highly dosage-sensitive Wnt non-canonical planar cell polarity pathway. Human Molecular Genetics, 2014, 23, 1687-1699.	2.9	35
33	Constitutional chromosomal events at 22q11 and 15q26 in a child with a pilocytic astrocytoma of the spinal cord. Molecular Cytogenetics, 2014, 7, 31.	0.9	2
34	Identification of a novel mouse Dbl proto-oncogene splice variant: Evidence that SEC14 domain is involved in GEF activity regulation. Gene, 2014, 537, 220-229.	2.2	6
35	Genetic Analysis of Disheveled 2 and Disheveled 3 in Human Neural Tube Defects. Journal of Molecular Neuroscience, 2013, 49, 582-588.	2.3	40
36	Novel MNX1 mutations and clinical analysis of familial and sporadic Currarino cases. European Journal of Medical Genetics, 2013, 56, 648-654.	1.3	26

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37	Rare missense variants in DVL1, one of the human counterparts of the Drosophila dishevelled gene, do not confer increased risk for neural tube defects. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 452-455.	1.6	3
38	Role of the planar cell polarity gene CELSR1 in neural tube defects and caudal agenesis. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 176-181.	1.6	94
39	De Novo MGC4607 Gene Heterozygous Missense Variants in a Child with Multiple Cerebral Cavernous Malformations. Journal of Molecular Neuroscience, 2012, 47, 475-480.	2.3	8
40	FZD6is a novel gene for human neural tube defects. Human Mutation, 2012, 33, 384-390.	2.5	83
41	A de novo balanced translocation t(7;12)(p21.2;p12.3) in a patient with Saethreâ€“Chotzen-like phenotype downregulates TWIST and an osteoclastic protein-tyrosine phosphatase, PTP-oc. European Journal of Medical Genetics, 2011, 54, e478-e483.	1.3	5
42	Contribution of VANGL2 mutations to isolated neural tube defects. Clinical Genetics, 2011, 80, 76-82.	2.0	107
43	Human neural tube defects: Genetic causes and prevention. BioFactors, 2011, 37, 261-268.	5.4	37
44	Maternal periconceptional factors affect the risk of spina bifida-affected pregnancies: an Italian caseâ€“control study. Child's Nervous System, 2011, 27, 1073-1081.	1.1	52
45	Identification and characterization of novel rare mutations in the planar cell polarity genePRICKLE1in human neural tube defects. Human Mutation, 2011, 32, 1371-1375.	2.5	74
46	Mutations in the planar cell polarity gene, Fuzzy, are associated with neural tube defects in humans. Human Molecular Genetics, 2011, 20, 4324-4333.	2.9	93
47	VANGL1 rare variants associated with neural tube defects affect convergent extension in zebrafish. Mechanisms of Development, 2010, 127, 385-392.	1.7	49
48	Novel mutations inVANGL1in neural tube defects. Human Mutation, 2009, 30, E706-E715.	2.5	98
49	Craniosynostosis, hydrocephalus, Chiari I malformation and radioulnar synostosis: Probably a new syndrome. European Journal of Medical Genetics, 2009, 52, 17-22.	1.3	13
50	No major role for the EMX2 gene in schizencephaly. American Journal of Medical Genetics, Part A, 2008, 146A, 1142-1150.	1.2	51
51	Successful isolation and long-term establishment of a cell line with stem cell-like features from an anaplastic medulloblastoma. Neuropathology and Applied Neurobiology, 2008, 34, 306-315.	3.2	16
52	Mutations inVANGL1Associated with Neural-Tube Defects. New England Journal of Medicine, 2007, 356, 1432-1437.	27.0	261
53	Evaluation of a methylenetetrahydrofolate-dehydrogenase 1958G>A polymorphism for neural tube defect risk. Journal of Human Genetics, 2006, 51, 98-103.	2.3	66
54	Current perspectives on the genetic causes of neural tube defects. Neurogenetics, 2006, 7, 201-221.	1.4	47

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55	Mutational screening of theCYP26A1 gene in patients with caudal regression syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 86-95.	1.6	17
56	HLXB9 homeobox gene and caudal regression syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 205-209.	1.6	20
57	Molecular genetic analysis of human homologs ofCaenorhabditis elegans mab-21-like 1 gene in patients with neural tube defects. Birth Defects Research Part A: Clinical and Molecular Teratology, 2004, 70, 885-888.	1.6	5
58	Reduced folate carrier polymorphism (80Aât'G) and neural tube defects. European Journal of Human Genetics, 2003, 11, 245-252.	2.8	87
59	Study of MTHFR and MS polymorphisms as risk factors for NTD in the Italian population. Journal of Human Genetics, 2002, 47, 319-324.	2.3	105
60	Polymorphisms in Genes Involved in Folate Metabolism as Risk Factors for NTDs. European Journal of Pediatric Surgery, 2001, 11, S14-S17.	1.3	55
61	Folate pathway gene alterations in patients with neural tube defects. American Journal of Medical Genetics Part A, 2000, 95, 216-223.	2.4	53
62	Schizencephaly: Surgical Features and New Molecular Genetic Results. European Journal of Pediatric Surgery, 1996, 6, 27-29.	1.3	16
63	Thyroid hormone receptor Î² mRNA expression in Sertoli cells isolated from prepubertal testis. Journal of Molecular Endocrinology, 1995, 14, 131-134.	2.5	39
64	Thyroidal regulation of nuclear tri-iodothyronine receptors in the developing rat testis. Journal of Endocrinology, 1993, 136, 277-282.	2.6	30