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	Mutations inVANGL1Associated with Neural-Tube Defects. New England Journal of Medicine, 2007, 356, 1432-1437.	27.0	261
2	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
3	Contribution of VANGL2 mutations to isolated neural tube defects. Clinical Genetics, 2011, 80, 76-82.	2.0	107
4	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
5	Novel mutations inVANGL1in neural tube defects. Human Mutation, 2009, 30, E706-E715.	2.5	98
6	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
7	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
8	Reduced folate carrier polymorphism (80Aât'G) and neural tube defects. European Journal of Human Genetics, 2003, 11, 245-252.	2.8	87
9	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
10	FZD6is a novel gene for human neural tube defects. Human Mutation, 2012, 33, 384-390.	2.5	83
11	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
12	Evaluation of a methylenetetrahydrofolate-dehydrogenase 1958G>A polymorphism for neural tube defect risk. Journal of Human Genetics, 2006, 51, 98-103.	2.3	66
13	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
14	Polymorphisms in Genes Involved in Folate Metabolism as Risk Factors for NTDs. European Journal of Pediatric Surgery, 2001, 11, S14-S17.	1.3	55
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	Folate pathway gene alterations in patients with neural tube defects. American Journal of Medical Genetics Part A, 2000, 95, 216-223.	2.4	53
17	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
18	No major role for the EMX2 gene in schizencephaly. American Journal of Medical Genetics, Part A, 2008, 146A, 1142-1150.	1.2	51

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19	VANGL1 rare variants associated with neural tube defects affect convergent extension in zebrafish. Mechanisms of Development, 2010, 127, 385-392.	1.7	49
20	Current perspectives on the genetic causes of neural tube defects. Neurogenetics, 2006, 7, 201-221.	1.4	47
21	Genetic Analysis of Disheveled 2 and Disheveled 3 in Human Neural Tube Defects. Journal of Molecular Neuroscience, 2013, 49, 582-588.	2.3	40
22	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
23	Thyroid hormone receptor Î² mRNA expression in Sertoli cells isolated from prepubertal testis. Journal of Molecular Endocrinology, 1995, 14, 131-134.	2.5	39
24	Human neural tube defects: Genetic causes and prevention. BioFactors, 2011, 37, 261-268.	5.4	37
25	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
26	Thyroidal regulation of nuclear tri-iodothyronine receptors in the developing rat testis. Journal of Endocrinology, 1993, 136, 277-282.	2.6	30
27	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
28	Rare deleterious variants in GRHL3 are associated with human spina bifida. Human Mutation, 2017, 38, 716-724.	2.5	28
29	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
30	Thymosin Alpha 1 Mitigates Cytokine Storm in Blood Cells From Coronavirus Disease 2019 Patients. Open Forum Infectious Diseases, 2021, 8, ofaa588.	0.9	27
31	Novel MNX1 mutations and clinical analysis of familial and sporadic Currarino cases. European Journal of Medical Genetics, 2013, 56, 648-654.	1.3	26
32	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
33	Whole exome sequencing identifies novel predisposing genes in neural tube defects. Molecular Genetics & Genomic Medicine, 2019, 7, e00467.	1.2	25
34	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
35	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
36	HLXB9 homeobox gene and caudal regression syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 205-209.	1.6	20

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37	Genetic Screening of Pediatric Cavernous Malformations. Journal of Molecular Neuroscience, 2016, 60, 232-238.	2.3	18
38	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
39	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
40	Moyamoya vasculopathy shows a genetic mutational gradient decreasing from East to West. Journal of Neurosurgical Sciences, 2020, 64, 165-172.	0.6	17
41	Schizencephaly: Surgical Features and New Molecular Genetic Results. European Journal of Pediatric Surgery, 1996, 6, 27-29.	1.3	16
42	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
43	Sacral agenesis: a pilot whole exome sequencing and copy number study. BMC Medical Genetics, 2016, 17, 98.	2.1	15
44	Craniosynostosis, hydrocephalus, Chiari I malformation and radioulnar synostosis: Probably a new syndrome. European Journal of Medical Genetics, 2009, 52, 17-22.	1.3	13
45	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
46	Chiari malformation type I: what information from the genetics?. Child's Nervous System, 2019, 35, 1665-1671.	1.1	12
47	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
48	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
49	L1CAM variants cause two distinct imaging phenotypes on fetal MRI. Annals of Clinical and Translational Neurology, 2021, 8, 2004-2012.	3.7	8
50	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
51	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
52	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
53	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
54	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

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55	Sinus pericranii, skull defects, and structural brain anomalies in TRAF7 â€related disorder. Birth Defects Research, 2020, 112, 1085-1092.	1.5	5
56	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
57	Cost effective assay choice for rare disease study designs. Orphanet Journal of Rare Diseases, 2015, 10, 10.	2.7	3
58	RNF213 variant in a patient with Legius syndrome associated with moyamoya syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1669.	1.2	3
59	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
60	Dyslipidemia in Children Treated with a BRAF Inhibitor for Low-Grade Gliomas: A New Side Effect?. Cancers, 2022, 14, 2693.	3.7	2
61	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
62	Spinal involvement in pediatric familial cavernous malformation syndrome. Neuroradiology, 2022, , 1.	2.2	1
63	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
64	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