Lorena Travaglini

List of Publications by Year in descending order

48 papers	3,054 citations	218662 26 h-index	206102 48 g-index
49 all docs	49 docs citations	49 times ranked	4857 citing authors

#	Article	IF	Citations
1	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	21.4	383
2	Epigenetic Silencing of the Myelopoiesis Regulator microRNA-223 by the AML1/ETO Oncoprotein. Cancer Cell, 2007, 12, 457-466.	16.8	373
3	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
4	Polycombs and microRNA-223 regulate human granulopoiesis by transcriptional control of target gene expression. Blood, 2012, 119, 4034-4046.	1.4	139
5	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	6.2	137
6	MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. Brain, 2013, 136, 872-881.	7.6	130
7	Sequential Valproic Acid/All-trans Retinoic Acid Treatment Reprograms Differentiation in Refractory and High-Risk Acute Myeloid Leukemia. Cancer Research, 2006, 66, 8903-8911.	0.9	125
8	Exposure of normal and transformed cells to nevirapine, a reverse transcriptase inhibitor, reduces cell growth and promotes differentiation. Oncogene, 2003, 22, 2750-2761.	5.9	105
9	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	2.5	96
10	Retinoic acid targets DNA-methyltransferases and histone deacetylases during APL blast differentiation in vitro and in vivo. Oncogene, 2005, 24, 1820-1830.	5.9	83
11	Heterochromatic gene repression of the retinoic acid pathway in acute myeloid leukemia. Blood, 2007, 109, 4432-4440.	1.4	82
12	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
13	Frataxin Deficiency Leads to Reduced Expression and Impaired Translocation of NF-E2-Related Factor (Nrf2) in Cultured Motor Neurons. International Journal of Molecular Sciences, 2013, 14, 7853-7865.	4.1	75
14	Oxidative stress in Duchenne muscular dystrophy: focus on the NRF2 redox pathway. Human Molecular Genetics, 2017, 26, 2781-2790.	2.9	71
15	RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170.	2.0	64
16	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
17	Phenomenology and clinical course of movement disorder in GNAO1 variants: Results from an analytical review. Parkinsonism and Related Disorders, 2019, 61, 19-25.	2.2	64
18	LMNA -associated myopathies. Neurology, 2014, 83, 1634-1644.	1.1	57

#	Article	IF	Citations
19	The impact of next-generation sequencing on the diagnosis of pediatric-onset hereditary spastic paraplegias: new genotype-phenotype correlations for rare HSP-related genes. Neurogenetics, 2018, 19, 111-121.	1.4	52
20	Epigenetic reprogramming of breast cancer cells by valproic acid occurs regardless of estrogen receptor status. International Journal of Biochemistry and Cell Biology, 2009, 41, 225-234.	2.8	48
21	Targeting of the Nâ€terminal coiled coil oligomerization interface by a helixâ€2 peptide inhibits unmutated and imatinibâ€resistant BCR/ABL. International Journal of Cancer, 2008, 122, 2744-2752.	5.1	38
22	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
23	Missense mutations of CACNA1A are a frequent cause of autosomal dominant nonprogressive congenital ataxia. European Journal of Paediatric Neurology, 2017, 21, 450-456.	1.6	37
24	Clinical and Genetic Overview of Paroxysmal Movement Disorders and Episodic Ataxias. International Journal of Molecular Sciences, 2020, 21, 3603.	4.1	36
25	Diagnostic Yield of a Targeted Next-Generation Sequencing Gene Panel for Pediatric-Onset Movement Disorders: A 3-Year Cohort Study. Frontiers in Genetics, 2019, 10, 1026.	2.3	33
26	Dynamic and reversibility of heterochromatic gene silencing in human disease. Cell Research, 2005, 15, 679-690.	12.0	32
27	AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661.	3.7	27
28	Childhood Rapid-Onset Ataxia: Expanding the Phenotypic Spectrum of ATP1A3 Mutations. Cerebellum, 2018, 17, 489-493.	2.5	24
29	ATP1A3 -related epileptic encephalopathy responding to ketogenic diet. Brain and Development, 2018, 40, 433-438.	1.1	23
30	FGF17, a gene involved in cerebellar development, is downregulated in a patient with Dandyâ€“Walker malformation carrying a de novo 8p deletion. Neurogenetics, 2011, 12, 241-245.	1.4	22
31	PRRT2 is mutated in familial and non-familial benign infantile seizures. European Journal of Paediatric Neurology, 2013, 17, 77-81.	1.6	22
32	Frataxin Silencing Inactivates Mitochondrial Complex I in NSC34 Motoneuronal Cells and Alters Glutathione Homeostasis. International Journal of Molecular Sciences, 2014, 15, 5789-5806.	4.1	22
33	A novel KCTD17 mutation is associated with childhood early-onset hyperkinetic movement disorder. Parkinsonism and Related Disorders, 2019, 61, 4-6.	2.2	22
34	Expanding the clinical phenotype of CAPN1- associated mutations: A new case with congenital-onset pure spastic paraplegia. Journal of the Neurological Sciences, 2017, 378, 210-212.	0.6	21
35	Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions. Orphanet Journal of Rare Diseases, 2013, 8, 75.	2.7	19
36	Molecular and clinical characterization of a series of patients with childhood-onset lysosomal acid lipase deficiency. Retrospective investigations, follow-up and detection of two novel LIPA pathogenic variants. Atherosclerosis, 2017, 265, 124-132.	0.8	19

#	Article	IF	Citations
37	Defining the clinical-genetic and neuroradiological features in SPG54: description of eight additional cases and nine novel DDHD2 variants. Journal of Neurology, 2019, 266, 2657-2664.	3.6	19
38	Childhood-onset ATP1A3-related conditions: Report of two new cases of phenotypic spectrum. Parkinsonism and Related Disorders, 2016, 30, 81-82.	2.2	18
39	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. Brain, 2017, 140, 2550-2556.	7.6	18
40	Hereditary spastic paraplegia is a novel phenotype for germline de novo ATP1A1 mutation. Clinical Genetics, 2020, 97, 521-526.	2.0	14
41	Heart transplant and 2-year follow up in a child with generalized arterial calcification of infancy. European Journal of Pediatrics, 2014, 173, 1735-1740.	2.7	13
42	SLC2A1 mutations are a rare cause of pediatric-onset hereditary spastic paraplegia. European Journal of Paediatric Neurology, 2019, 23, 329-332.	1.6	11
43	Novel homozygous GBA2 mutation in a patient with complicated spastic paraplegia. Clinical Neurology and Neurosurgery, 2018, 168, 60-63.	1.4	9
44	Clinical variability at the mild end of BRAT1 â€related spectrum: Evidence from two families with genotypeâ€“phenotype discordance. Human Mutation, 2022, 43, 67-73.	2.5	9
45	Congenital-onset spastic paraplegia in a patient with TUBB4A mutation and mild hypomyelination. Journal of the Neurological Sciences, 2016, 368, 145-146.	0.6	8
46	Longitudinal follow up of a boy affected by Pol III-related leukodystrophy: a detailed phenotype description. BMC Medical Genetics, 2015, 16, 53.	2.1	5
47	Uniparental disomy of chromosome 1 unmasks recessive mutations of PPT1 in a boy with neuronal ceroid lipofuscinosis type 1. Brain and Development, 2017, 39, 182-183.	1.1	5
48	â€œAtypicalâ€•Krabbe disease in two siblings harboring biallelic GALC mutations including a deep intronic variant. European Journal of Human Genetics, 2022, , .	2.8	4