Daniela Di Bella

List of Publications by Year in descending order

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218662 189881 2,769 61 26 50 citations h-index g-index papers 61 61 61 3717 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Digenic inheritance of STUB1 variants and TBP polyglutamine expansions explains the incomplete penetrance of SCA17 and SCA48. Genetics in Medicine, 2022, 24, 29-40.	2.4	24
2	Hypomyelinating leukodystrophies in adults: Clinical and genetic features. European Journal of Neurology, 2021, 28, 934-944.	3.3	14
3	Multifaceted and Age-Dependent Phenotypes Associated With Biallelic PNPLA6 Gene Variants: Eight Novel Cases and Review of the Literature. Frontiers in Neurology, 2021, 12, 793547.	2.4	6
4	<i>RARS1</i> à€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	3.7	18
5	Parkinsonism and Nigrostriatal Damage Secondary to <scp><i>CSF1R</i></scp> â€Related Primary Microgliopathy. Movement Disorders, 2020, 35, 2360-2362.	3.9	6
6	Severe worsening of adult-onset Alexander disease after minor head trauma: Report of two patients and review of the literature. Journal of Clinical Neuroscience, 2020, 75, 221-223.	1.5	6
7	From congenital microcephaly to adult onset cerebellar ataxia: Distinct and overlapping phenotypes in patients with <i>PNKP</i> gene mutations. American Journal of Medical Genetics, Part A, 2019, 179, 2277-2283.	1.2	18
8	Spasmodic dysphonia as a presenting symptom of spinocerebellar ataxia type 12. Neurogenetics, 2019, 20, 161-164.	1.4	3
9	Neuropsychological features of adult form of Alexander disease. Journal of the Neurological Sciences, 2019, 401, 87-89.	0.6	7
10	Expanding the central nervous system disease spectrum associated with <i>FLNC</i> mutation. Muscle and Nerve, 2019, 59, E33-E37.	2.2	3
11	Saposin B deficiency as a cause of adult-onset metachromatic leukodystrophy. Neurology, 2019, 93, 310-312.	1.1	6
12	ANO10 mutational screening in recessive ataxia: genetic findings and refinement of the clinical phenotype. Journal of Neurology, 2019, 266, 378-385.	3.6	22
13	Clinical and genetic characteristics of late-onset Huntington's disease. Parkinsonism and Related Disorders, 2019, 61, 101-105.	2.2	17
14	Concurrent <i>AFG3L2</i> and <i>SPG7</i> mutations associated with syndromic parkinsonism and optic atrophy with aberrant OPA1 processing and mitochondrial network fragmentation. Human Mutation, 2018, 39, 2060-2071.	2.5	32
15	Hereditary gelsolin amyloidosis (<scp>HGA</scp>): a neglected cause of bilateral progressive or recurrent facial palsy. Journal of the Peripheral Nervous System, 2017, 22, 59-63.	3.1	8
16	SPG5 siblings with different phenotypes showing reduction of 27-hydroxycholesterol after simvastatin-ezetimibe treatment. Journal of the Neurological Sciences, 2017, 383, 39-41.	0.6	3
17	MRI Evidence of Cerebellar and Extraocular Muscle Atrophy Differently Contributing to Eye Movement Abnormalities in SCA2 and SCA28 Diseases. , 2016, 57, 2714.		11
18	Multisystemic <i>SYNE1</i> ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. Brain, 2016, 139, e46-e46.	7.6	40

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19	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	7.6	87
20	Early-onset progressive spastic paraplegia caused by a novel TUBB4A mutation: brain MRI and FDG-PET findings. Journal of Neurology, 2016, 263, 591-593.	3.6	17
21	Mutational mechanisms in <i><scp>MFN2</scp></i> i>â€related neuropathy: compound heterozygosity for recessive and semidominant mutations. Journal of the Peripheral Nervous System, 2015, 20, 380-386.	3.1	23
22	Somatosensory Conduction Pathway in Spastic Paraplegia Type 5. Journal of Clinical Neurology		

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37	Case–control and combined family trios analysis of three polymorphisms in the ghrelin gene in European patients with anorexia and bulimia nervosa. Psychiatric Genetics, 2006, 16, 51-52.	1.1	40
38	SCA28, a novel form of autosomal dominant cerebellar ataxia on chromosome 18p11.22–q11.2. Brain, 2006, 129, 235-242.	7.6	122
39	Association of BDNF with restricting anorexia nervosa and minimum body mass index: a family-based association study of eight European populations. European Journal of Human Genetics, 2005, 13, 428-434.	2.8	131
40	Antipanic Efficacy of Paroxetine and Polymorphism within the Promoter of the Serotonin Transporter Gene. Neuropsychopharmacology, 2005, 30, 2230-2235.	5.4	93
41	Association of BDNF with anorexia, bulimia and age of onset of weight loss in six European populations. Human Molecular Genetics, 2004, 13, 1205-1212.	2.9	193
42	Lack of relationship between CO2 reactivity and serotonin transporter gene regulatory region polymorphism in panic disorder. American Journal of Medical Genetics Part A, 2004, 129B, 41-43.	2.4	18
43	Bulimia Nervosa, 5-HTTLPR Polymorphism and Treatment Response to Four SSRIs. Journal of Clinical Psychopharmacology, 2004, 24, 680-682.	1.4	9
44	Obsessive-Compulsive Disorder, 5-HTTLPR polymorphism and treatment response. Pharmacogenomics Journal, 2002, 2, 176-181.	2.0	68
45	No Association Between Obsessive-Compulsive Disorder and the 5-HT _{1Dβ} Receptor Gene. American Journal of Psychiatry, 2002, 159, 1783-1785.	7.2	45
46	Exploratory factor analysis of obsessiveâ€compulsive patients and association with 5â€HTTLPR polymorphism. American Journal of Medical Genetics Part A, 2002, 114, 347-353.	2.4	119
47	Novel 5′-regulatory region polymorphisms of the 5-HT2C receptor gene: association study with panic disorder. International Journal of Neuropsychopharmacology, 2000, 3, 321-325.	2.1	32
48	Polymorphic MAO-A and 5-HT-Transporter Genes: Analysis of Interactions in Panic Disorder. World Journal of Biological Psychiatry, 2000, 1, 147-150.	2.6	19
49	An association study between 5-HTTLPR polymorphism, COMT polymorphism, and Tourette's syndrome. Psychiatry Research, 2000, 97, 93-100.	3.3	53
50	Efficacy of Paroxetine in Depression Is Influenced by a Functional Polymorphism Within the Promoter of the Serotonin Transporter Gene. Journal of Clinical Psychopharmacology, 2000, 20, 105-107.	1.4	290
51	Patients Requesting Psychiatric Hospitalization. American Journal of Psychiatry, 2000, 157, 1886-1886.	7.2	0
52	Dr. Benedetti and Colleagues Reply. American Journal of Psychiatry, 2000, 157, 1887-1888.	7.2	0
53	No interaction of GABAA alpha-1 subunit and dopamine receptor D4 exon 3 genes in symptomatology of major psychoses. , 1999, 88, 44-49.		18
54	Dopamine receptorD4 gene is not associated with major psychoses., 1999, 88, 486-491.		24

Daniela Di Bella

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55	Dopamine receptor D2 and D4 genes, GABAA alpha-1 subunit gene and response to lithium prophylaxis in mood disorders. Psychiatry Research, 1999, 87, 7-19.	3.3	66
56	5HT2C CYS23/SER23 polymorphism is not associated with obsessive–compulsive disorder. Psychiatry Research, 1998, 77, 97-104.	3.3	46
57	Self-esteem in remitted patients with mood disorders is not associated with the dopamine receptor D4 and the serotonin transporter genes. Psychiatry Research, 1998, 80, 137-144.	3.3	7
58	CO ₂ -Induced Panic Attacks: A Twin Study. American Journal of Psychiatry, 1998, 155, 1184-1188.	7.2	86
59	Intragenic tetranucleotide repeat polymorphism at the human histidase (HAL) locus. Clinical Genetics, 1997, 52, 194-195.	2.0	1
60	Systematic screening for mutations in the coding region of the human serotonin transporter (5-HTT) gene using PCR and DGGE., 1996, 67, 541-545.		53
61	Lack of association between obsessive-compulsive disorder and the dopamine D3 receptor gene: Some preliminary considerations. American Journal of Medical Genetics Part A, 1994, 54, 253-255.	2.4	38