Liana Veneziano

List of Publications by Year
in descending order

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35 papers	1,675 citations	471061 17 h-index	414034 32 g-index
38 all docs	38 docs citations	38 times ranked	2423 citing authors

#	Article	IF	Citations
1	Mutations in the mitochondrial protease gene AFG3L2 cause dominant hereditary ataxia SCA28. Nature Genetics, 2010, 42, 313-321.	9.4	291
2	Episodic Ataxia Type 2 (EA2) and Spinocerebellar Ataxia Type 6 (SCA6) Due to CAG Repeat Expansion in the CACNA1A Gene on Chromosome 19p. Human Molecular Genetics, 1997, 6, 1973-1978.	1.4	264
3	Complete Loss of P/Q Calcium Channel Activity Caused by a CACNA1A Missense Mutation Carried by Patients with Episodic Ataxia Type 2. American Journal of Human Genetics, 2001, 68, 759-764.	2.6	147
4	A G301R Na+/K+-ATPase mutation causes familial hemiplegic migraine type 2 with cerebellar signs. Neurogenetics, 2004, 5, 177-185.	0.7	117
5	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
6	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	2.6	96
7	A Novel De Novo Mutation of the TITF1/NKX2-1 Gene Causing Ataxia, Benign Hereditary Chorea, Hypothyroidism and a Pituitary Mass in a UK Family and Review of the Literature. Cerebellum, 2014, 13, 588-595.	1.4	93
8	The role of the SCA2 trinucleotide repeat expansion in 89 autosomal dominant cerebellar ataxia families. Frequency, clinical and genetic correlates. Brain, 1998, 121, 459-467.	3.7	84
9	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. Parkinsonism and Related Disorders, 2017, 41, 37-43.	1.1	67
10	Identification of novel and recurrent CACNA1A gene mutations in fifteen patients with episodic ataxia type 2. Journal of the Neurological Sciences, 2010, 291, 30-36.	0.3	63
11	Spinocerebellar ataxia type 6 and episodic ataxia type 2: differences and similarities between two allelic disorders. Cytogenetic and Genome Research, 2003, 100, 147-153.	0.6	45
12	Clusters of non-truncating mutations of P/Q type Ca2+ channel subunit Cav2.1 causing episodic ataxia 2. Journal of Medical Genetics, 2004, 41, e82-e82.	1.5	40
13	Functional characterization of a novel mutation in TITF-1 in a patient with benign hereditary chorea. Journal of the Neurological Sciences, 2008, 264, 56-62.	0.3	35
14	Acetazolamide-responsive episodic ataxia in an Italian family refines gene mapping on chromosome 19p13. Brain, 1997, 120, 805-812.	3.7	24
15	Leukocyte telomere shortening in Huntington's disease. Journal of the Neurological Sciences, 2019, 396, 25-29.	0.3	24
16	Molecular mechanism of Spinocerebellar Ataxia type 6: glutamine repeat disorder, channelopathy and transcriptional dysregulation. The multifaceted aspects of a single mutation. Frontiers in Cellular Neuroscience, 2015, 9, 36.	1.8	23
17	Genetic fitness in Huntington's Disease and Spinocerebellar Ataxia 1: a population genetics model for CAG repeat expansions. Annals of Human Genetics, 1996, 60, 423-435.	0.3	22
18	Complexity of the Genetics and Clinical Presentation of Spinocerebellar Ataxia 17. Frontiers in Cellular Neuroscience, 2018, 12, 429.	1.8	21

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19	Characterization of human frataxin missense variants in cancer tissues. Human Mutation, 2019, 40, 1400-1413.	1.1	16
20	A fine physical map of the CACNA1A gene region on 19p13.1â€“p13.2 chromosome. Gene, 2000, 241, 45-50.	1.0	15
21	A channelopathy mutation in the voltage-sensor discloses contributions of a conserved phenylalanine to gating properties of Kv1.1 channels and ataxia. Scientific Reports, 2017, 7, 4583.	1.6	15
22	Newly characterised 5â€² and 3â€² regions of CACNA1A gene harbour mutations associated with Familial Hemiplegic Migraine and Episodic Ataxia. Journal of the Neurological Sciences, 2009, 276, 31-37.	0.3	14
23	Analyzing the Effects of a G137V Mutation in the FXN Gene. Frontiers in Molecular Neuroscience, 2015, 8, 66.	1.4	14
24	A multistep process for the dispersal of a Y chromosomal lineage in the Mediterranean area. Annals of Human Genetics, 2001, 65, 339-49.	0.3	14
25	A shared haplotype for dentatorubropallidoluysian atrophy (DRPLA) in Italian families testifies of the recent introduction of the mutation. Journal of Human Genetics, 2014, 59, 153-157.	1.1	6
26	Restless Legs Syndrome in NKX2-1-related chorea: An expansion of the disease spectrum. Brain and Development, 2019, 41, 250-256.	0.6	6
27	Dramatically different levels of cacna1a gene expression between pre-weaning wild type and leaner mice. Journal of the Neurological Sciences, 2011, 305, 71-74.	0.3	5
28	Functional characterization of two novel mutations in TTF-1/NKX2.1 homeodomain in patients with benign hereditary chorea. Journal of the Neurological Sciences, 2016, 360, 78-83.	0.3	5
29	Forensic DNA Challenges: Replacing Numbers with Names of Fosse Ardeatineâ€™s Victims*. Journal of Forensic Sciences, 2009, 54, 905-908.	0.9	4
30	Construction and preliminary characterization of human recombinant proNGF-A variant. Neurochemistry International, 2020, 140, 104812.	1.9	2
31	Altered pituitary morphology as a sign of benign hereditary chorea caused by TITF1/NKX2.1 mutations. Neurogenetics, 2022, 23, 91.	0.7	2
32	DNA Markers in Diagnosis of Adult Dominant Polycystic Kidney Disease. European Urology, 1992, 21, 57-59.	0.9	0
33	Ordering of 44 Genetic Markers in the 6p22 Cytogenetic Band. DNA Sequence, 1996, 7, 51-52.	0.7	0
34	Early onset progressive ataxia associated with the first CACNA1A mutation identified within the Iâ€“II loop. Journal of the Neurological Sciences, 2007, 263, 226.	0.3	0
35	NOVEL DE NOVO MUTATION CAUSING BENIGN HEREDITARY CHOREA WITH HYPOTHRYOIDISM AND A PITUITARY MASS. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, A11.1-A11.	0.9	0