Luciana Musante

List of Publications by Year
in descending order

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46	3,374	236925	206112
papers	citations	25 h-index	48 g-index

50	50	50	6571
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Encefalopatie epilettiche e dello sviluppo: dalla pratica alla genetica, andata e ritorno. Medico E Bambino, 2022, 41, 33-40.	0.1	0
2	Expanding Phenotype of Poirierâ€“Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes, 2022, 13, 276.	2.4	10
3	The Genetic Diagnosis of Ultrarare DEEs: An Ongoing Challenge. Genes, 2022, 13, 500.	2.4	7
4	TTC5 syndrome: Clinical and molecular spectrum of a severe and recognizable condition. American Journal of Medical Genetics, Part A, 2022, 188, 2652-2665.	1.2	4
5	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
6	Just Expect It: Compound Heterozygous Variants of POMT1 in a Consanguineous Familyâ€”The Role of Next Generation Sequencing in Neuromuscular Disorders. Neuropediatrics, 2020, 51, 072-075.	0.6	1
7	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. Epilepsia, 2020, 61, 2474-2485.	5.1	31
8	Description of a peculiar alternating ictal electroclinical pattern in a young boy with a novel SPATA5 mutation[*]. Epileptic Disorders, 2020, 22, 659-663.	1.3	9
9	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	7.9	131
10	De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. Molecular Psychiatry, 2018, 23, 222-230.	7.9	45
11	Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. Human Molecular Genetics, 2018, 27, 3177-3188.	2.9	19
12	Mutations of the aminoacyl-tRNA-synthetases SARS and WARS2 are implicated in the etiology of autosomal recessive intellectual disability. Human Mutation, 2017, 38, 621-636.	2.5	54
13	KlÃ¼verâ€“Bucy syndrome associated with a recessive variant in HGSNAT in two siblings with Mucopolysaccharidosis type IIIC (Sanfilippo C). European Journal of Human Genetics, 2017, 25, 253-256.	2.8	5
14	Homozygous ARHGEF2 mutation causes intellectual disability and midbrain-hindbrain malformation. PLoS Genetics, 2017, 13, e1006746.	3.5	27
15	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. European Journal of Human Genetics, 2016, 24, 392-399.	2.8	17
16	Homozygous YME1L1 mutation causes mitochondriopathy with optic atrophy and mitochondrial network fragmentation. ELife, 2016, 5, .	6.0	88
17	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. American Journal of Human Genetics, 2015, 96, 386-396.	6.2	27
18	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53

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19	Mutations in the histamineN-methyltransferase gene,HNMT, are associated with nonsyndromic autosomal recessive intellectual disability. Human Molecular Genetics, 2015, 24, 5697-5710.	2.9	27
20	In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells. Molecular Psychiatry, 2015, 20, 459-471.	7.9	31
21	A defect in the CLIP1 gene (CLIP-170) can cause autosomal recessive intellectual disability. European Journal of Human Genetics, 2015, 23, 331-336.	2.8	22
22	The Role of a Novel TRMT1 Gene Mutation and Rare GRM1 Gene Defect in Intellectual Disability in Two Azeri Families. PLoS ONE, 2015, 10, e0129631.	2.5	56
23	New evidence for the role of calpain 10 in autosomal recessive intellectual disability: identification of two novel nonsense variants by exome sequencing in Iranian families. Archives of Iranian Medicine, 2015, 18, 179-84.	0.6	6
24	Exome Sequencing and Linkage Analysis Identified Novel Candidate Genes in Recessive Intellectual Disability Associated with Ataxia. Archives of Iranian Medicine, 2015, 18, 670-82.	0.6	4
25	Mutations in PTRH2 cause novel infantileâ€onset multisystem disease with intellectual disability, microcephaly, progressive ataxia, and muscle weakness. Annals of Clinical and Translational Neurology, 2014, 1, 1024-1035.	3.7	29
26	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. Human Mutation, 2014, 35, 1427-1435.	2.5	31
27	Previously reported new type of autosomal recessive primary microcephaly is caused by compound heterozygousASPMgene mutations. Cell Cycle, 2014, 13, 1650-1651.	2.6	8
28	Genetics of recessive cognitive disorders. Trends in Genetics, 2014, 30, 32-39.	6.7	120
29	NDST1 missense mutations in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2753-2763.	1.2	34
30	De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. Genome Medicine, 2013, 5, 11.	8.2	128
31	A novel ALDH5A1 mutation is associated with succinic semialdehyde dehydrogenase deficiency and severe intellectual disability in an Iranian family. American Journal of Medical Genetics, Part A, 2013, 161, 1915-1922.	1.2	16
32	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	27.8	805
33	Hybridisation-based resequencing of 17 X-linked intellectual disability genes in 135 patients reveals novel mutations in ATRX, SLC6A8 and PQBP1. European Journal of Human Genetics, 2011, 19, 717-720.	2.8	21
34	The X-chromosome-linked intellectual disability protein PQBP1 is a component of neuronal RNA granules and regulates the appearance of stress granules. Human Molecular Genetics, 2011, 20, 4916-4931.	2.9	52
35	Common pathological mutations inPQBP1induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. Human Mutation, 2010, 31, 90-98.	2.5	18
36	A balanced chromosomal translocation disruptingARHGEF9is associated with epilepsy, anxiety, aggression, and mental retardation. Human Mutation, 2009, 30, 61-68.	2.5	131

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37	Multiple giant cell lesions in patients with Noonan syndrome and cardio-facio-cutaneous syndrome. European Journal of Human Genetics, 2009, 17, 420-425.	2.8	79
38	Disruption of the TCF4 gene in a girl with mental retardation but without the classical Pittâ€“Hopkins syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2053-2059.	1.2	68
39	Germline KRAS mutations cause Noonan syndrome. Nature Genetics, 2006, 38, 331-336.	21.4	670
40	cDNA cloning and characterization of the human THRAP2 gene which maps to chromosome 12q24, and its mouse ortholog Thrap2. Gene, 2004, 332, 119-127.	2.2	14
41	Spectrum of mutations in PTPN11 and genotypeâ€“phenotype correlation in 96 patients with Noonan syndrome and five patients with cardio-facio-cutaneous syndrome. European Journal of Human Genetics, 2003, 11, 201-206.	2.8	148
42	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	21.4	139
43	Autocrine Regulation of Volume-sensitive Anion Channels in Airway Epithelial Cells by Adenosine. Journal of Biological Chemistry, 1999, 274, 11701-11707.	3.4	41
44	Characterization of a murine gene homologous to the bovine CaCC chloride channel. Gene, 1999, 228, 181-188.	2.2	47
45	Characterization of the human gene coding for the swelling-dependent chloride channel ICln at position 11q13.5â€“14.1 (CLNS1A) and further characterization of the chromosome 6 (CLNS1B) localization. Gene, 1998, 209, 59-63.	2.2	5
46	An electrogenic amino acid transporter in the apical membrane of cultured human bronchial epithelial cells. American Journal of Physiology - Lung Cellular and Molecular Physiology, 1998, 275, L917-L923.	2.9	31