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	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	27.8	805
2	Germline KRAS mutations cause Noonan syndrome. Nature Genetics, 2006, 38, 331-336.	21.4	670
3	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
4	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	21.4	139
5	A balanced chromosomal translocation disruptingARHGEF9is associated with epilepsy, anxiety, aggression, and mental retardation. Human Mutation, 2009, 30, 61-68.	2.5	131
6	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	7.9	131
7	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
8	Genetics of recessive cognitive disorders. Trends in Genetics, 2014, 30, 32-39.	6.7	120
9	Homozygous YME1L1 mutation causes mitochondriopathy with optic atrophy and mitochondrial network fragmentation. ELife, 2016, 5, .	6.0	88
10	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
11	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
12	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
13	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
14	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
15	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
16	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
17	Characterization of a murine gene homologous to the bovine CaCC chloride channel. Gene, 1999, 228, 181-188.	2.2	47
18	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

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19	Autocrine Regulation of Volume-sensitive Anion Channels in Airway Epithelial Cells by Adenosine. Journal of Biological Chemistry, 1999, 274, 11701-11707.	3.4	41
20	NDST1 missense mutations in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2753-2763.	1.2	34
21	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
22	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. Human Mutation, 2014, 35, 1427-1435.	2.5	31
23	In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells. Molecular Psychiatry, 2015, 20, 459-471.	7.9	31
24	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. Epilepsia, 2020, 61, 2474-2485.	5.1	31
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	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
27	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
28	Homozygous ARHGEF2 mutation causes intellectual disability and midbrain-hindbrain malformation. PLoS Genetics, 2017, 13, e1006746.	3.5	27
29	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
30	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
31	Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. Human Molecular Genetics, 2018, 27, 3177-3188.	2.9	19
32	Common pathological mutations inPQBP1induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. Human Mutation, 2010, 31, 90-98.	2.5	18
33	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
34	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
35	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
36	Expanding Phenotype of Poirierâ€“Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes, 2022, 13, 276.	2.4	10

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37	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
38	Previously reported new type of autosomal recessive primary microcephaly is caused by compound heterozygousASPMgene mutations. Cell Cycle, 2014, 13, 1650-1651.	2.6	8
39	The Genetic Diagnosis of Ultrarare DEEs: An Ongoing Challenge. Genes, 2022, 13, 500.	2.4	7
40	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
41	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
42	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
43	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
44	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
45	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
46	Encefalopatie epilettiche e dello sviluppo: dalla pratica alla genetica, andata e ritorno. Medico E Bambino, 2022, 41, 33-40.	0.1	0