M Chiara Manzini

List of Publications by Year
in descending order

46	2,139	279798	233421
papers	citations	23 h-index	45 g-index

52	52	52	3356
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A Human Homologue of the Drosophila melanogaster diaphanous Gene Is Disrupted in a Patient with Premature Ovarian Failure: Evidence for Conserved Function in Oogenesis and Implications for Human Sterility. American Journal of Human Genetics, 1998, 62, 533-541.	6.2	248
2	Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 92, 354-365.	6.2	172
3	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. American Journal of Human Genetics, 2012, 91, 541-547.	6.2	167
4	What disorders of cortical development tell us about the cortex: one plus one does not always make two. Current Opinion in Genetics and Development, 2011, 21, 333-339.	3.3	151
5	COL4A1 Mutations Cause Ocular Dysgenesis, Neuronal Localization Defects, and Myopathy in Mice and Walker-Warburg Syndrome in Humans. PLoS Genetics, 2011, 7, e1002062.	3.5	121
6	Microsampling Capillary Electrophoresis Mass Spectrometry Enables Single-Cell Proteomics in Complex Tissues: Developing Cell Clones in Live Xenopus laevis and Zebrafish Embryos. Analytical Chemistry, 2019, 91, 4797-4805.	6.5	97
7	Mutation analysis of two candidate genes for premature ovarian failure, DACH2 and POF1B. Human Reproduction, 2004, 19, 2759-2766.	0.9	82
8	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. Human Molecular Genetics, 2014, 23, 5781-5792.	2.9	72
9	Kainate Receptors Expressed by a Subpopulation of Developing Nociceptors Rapidly Switch from High to Low Ca²⁺Permeability. Journal of Neuroscience, 2001, 21, 4572-4581.	3.6	70
10	Isolation and Culture of Post-Natal Mouse Cerebellar Granule Neuron Progenitor Cells and Neurons. Journal of Visualized Experiments, 2009, , .	0.3	69
11	Ethnically diverse causes of Walker-Warburg syndrome (WWS):FCMDmutations are a more common cause of WWS outside of the Middle East. Human Mutation, 2008, 29, E231-E241.	2.5	67
12	Mutations in INPP5K Cause a Form of Congenital Muscular Dystrophy Overlapping Marinesco-SjÃ¶gren Syndrome and Dystroglycanopathy. American Journal of Human Genetics, 2017, 100, 537-545.	6.2	67
13	CC2D1A Regulates Human Intellectual and Social Function as well as NF-Î°B Signaling Homeostasis. Cell Reports, 2014, 8, 647-655.	6.4	60
14	Tapered-Tip Capillary Electrophoresis Nano-Electrospray Ionization Mass Spectrometry for Ultrasensitive Proteomics: the Mouse Cortex. Journal of the American Society for Mass Spectrometry, 2017, 28, 597-607.	2.8	53
15	A Splice Site Mutation in Laminin-Î±2 Results in a Severe Muscular Dystrophy and Growth Abnormalities in Zebrafish. PLoS ONE, 2012, 7, e43794.	2.5	48
16	The Stop Signal Revised: Immature Cerebellar Granule Neurons in the External Germinal Layer Arrest Pontine Mossy Fiber Growth. Journal of Neuroscience, 2006, 26, 6040-6051.	3.6	46
17	Current Perspectives in Autism Spectrum Disorder: From Genes to Therapy. Journal of Neuroscience, 2016, 36, 11402-11410.	3.6	44
18	Cc2d1a Loss of Function Disrupts Functional and Morphological Development in Forebrain Neurons Leading to Cognitive and Social Deficits. Cerebral Cortex, 2017, 27, 1670-1685.	2.9	36

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19	Abrogated Freud-1/Cc2d1a Repression of 5-HT1A Autoreceptors Induces Fluoxetine-Resistant Anxiety/Depression-Like Behavior. Journal of Neuroscience, 2017, 37, 11967-11978.	3.6	35
20	Enhanced Peptide Detection Toward Single-Neuron Proteomics by Reversed-Phase Fractionation Capillary Electrophoresis Mass Spectrometry. Journal of the American Society for Mass Spectrometry, 2018, 29, 913-922.	2.8	34
21	Structure of the polyisoprenyl-phosphate glycosyltransferase GtrB and insights into the mechanism of catalysis. Nature Communications, 2016, 7, 10175.	12.8	33
22	Developmental and degenerative features in a complicated spastic paraplegia. Annals of Neurology, 2010, 67, 516-525.	5.3	31
23	Male-Specific cAMP Signaling in the Hippocampus Controls Spatial Memory Deficits in a Mouse Model of Autism and Intellectual Disability. Biological Psychiatry, 2019, 85, 760-768.	1.3	28
24	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
25	A novel form of lethal microcephaly with simplified gyral pattern and brain stem hypoplasia. American Journal of Medical Genetics, Part A, 2007, 143A, 2761-2767.	1.2	20
26	SOBP Is Mutated in Syndromic and Nonsyndromic Intellectual Disability and Is Highly Expressed in the Brain Limbic System. American Journal of Human Genetics, 2010, 87, 694-700.	6.2	20
27	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. Genetics in Medicine, 2019, 21, 2059-2069.	2.4	20
28	Digging behavior discrimination test to probe burrowing and exploratory digging in male and female mice. Journal of Neuroscience Research, 2021, 99, 2046-2058.	2.9	20
29	Molecular causes of sexâ€specific deficits in rodent models of neurodevelopmental disorders. Journal of Neuroscience Research, 2021, 99, 37-56.	2.9	19
30	Severe muscleâ€“eyeâ€“brain disease is associated with a homozygous mutation in the POMGnT1 gene. European Journal of Paediatric Neurology, 2008, 12, 133-136.	1.6	18
31	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	2.4	18
32	Data-Dependent Acquisition Ladder for Capillary Electrophoresis Mass Spectrometry-Based Ultrasensitive (Neuro)Proteomics. Analytical Chemistry, 2021, 93, 15964-15972.	6.5	18
33	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. American Journal of Human Genetics, 2019, 105, 844-853.	6.2	17
34	Overlap of polymicrogyria, hydrocephalus, and Joubert syndrome in a family with novel truncating mutations in ADGRG1/GPR56 and KIAA0556. Neurogenetics, 2019, 20, 91-98.	1.4	17
35	Loss of the Intellectual Disability and Autism Gene Cc2d1a and Its Homolog Cc2d1b Differentially Affect Spatial Memory, Anxiety, and Hyperactivity. Frontiers in Genetics, 2018, 9, 65.	2.3	16
36	Balancing Act: Maintaining Amino Acid Levels in the Autistic Brain. Neuron, 2017, 93, 476-479.	8.1	15

#	Article	IF	Citations
37	Capillary Electrophoresis Mass Spectrometry for Scalable Single-Cell Proteomics. Frontiers in Chemistry, 2022, 10, 863979.	3.6	15
38	Variable disease severity in Saudi Arabian and Sudanese families with c.3924 + 2 T > C mutation of LAMA2. BMC Research Notes, 2011, 4, 534.	1.4	13
39	Expanding the spectrum of rearrangements involving chromosome 19: A mild phenotype associated with a 19p13.12â€“p13.13 deletion. American Journal of Medical Genetics, Part A, 2012, 158A, 888-893.	1.2	8
40	Translating genetic and preclinical findings into autism therapies. Dialogues in Clinical Neuroscience, 2017, 19, 335-343.	3.7	6
41	Unraveling the mysteries of MYT1L: From reprogramming factor to multifaceted regulator of neuronal differentiation. Neuron, 2021, 109, 3713-3715.	8.1	6
42	Differential effects of AMPA receptor activation on survival and neurite integrity during neuronal development. Molecular and Cellular Neurosciences, 2007, 35, 328-338.	2.2	4
43	Cc2d1b Contributes to the Regulation of Developmental Myelination in the Central Nervous System. Frontiers in Molecular Neuroscience, 2022, 15, 881571.	2.9	4
44	Novel mutation identification and copy number variant detection via exome sequencing in congenital muscular dystrophy. Molecular Genetics & Genomic Medicine, 2020, 8, e1387.	1.2	3
45	Myopathic changes associated with psychomotor delay and seizures caused by a novel homozygous mutation in TBCK. Muscle and Nerve, 2020, 62, 266-271.	2.2	3
46	Male-Specific Intracellular Signaling in Sex-Bias in Neurodevelopmental Disorders. Biological Psychiatry, 2020, 87, S65-S66.	1.3	0