Cecilia Mancini

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7794636/publications.pdf

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28	609	567281	610901
papers	citations	15 h-index	24 g-index

28	28	28	1361
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	ELOVL5 Mutations Cause Spinocerebellar Ataxia 38. American Journal of Human Genetics, 2014, 95, 209-217.	6.2	107
2	Missense mutations in the AFG3L2 proteolytic domain account for â^¼1.5% of European autosomal dominant cerebellar ataxias. Human Mutation, 2010, 31, 1117-1124.	2.5	81
3	A novel 3q29 deletion associated with autism, intellectual disability, psychiatric disorders, and obesity. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 290-299.	1.7	34
4	Adult-onset autosomal recessive ataxia associated with neuronal ceroid lipofuscinosis type 5 gene (CLN5) mutations. Journal of Neurology, 2015, 262, 173-178.	3.6	29
5	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proofâ€ofâ€concept examples. American Journal of Medical Genetics, Part A, 2016, 170, 1772-1779.	1.2	26
6	Mitochondrial stress response triggered by defects in protein synthesis quality control. Life Science Alliance, 2019, 2, e201800219.	2.8	26
7	Large cryptic genomic rearrangements with apparently normal karyotypes detected by array-CGH. Molecular Cytogenetics, 2014, 7, 82.	0.9	25
8	Ribosomal <scp>RNA</scp> analysis in the diagnosis of Diamondâ€Blackfan Anaemia. British Journal of Haematology, 2016, 172, 782-785.	2.5	24
9	Spontaneous remission in a Diamondâ€Blackfan anaemia patient due to a revertant uniparental disomy ablating a de novo RPS19 mutation. British Journal of Haematology, 2019, 185, 994-998.	2.5	24
10	Mice harbouring a SCA28 patient mutation in AFG3L2 develop late-onset ataxia associated with enhanced mitochondrial proteotoxicity. Neurobiology of Disease, 2019, 124, 14-28.	4.4	23
11	Megalencephalic leukoencephalopathy with subcortical cysts type 1 (MLC1) due to a homozygous deep intronic splicing mutation (c.895-226T>G) abrogated in vitro using an antisense morpholino oligonucleotide. Neurogenetics, 2012, 13, 205-214.	1.4	21
12	Two families with novel missense mutations in COL4A1: When diagnosis can be missed. Journal of the Neurological Sciences, 2015, 352, 99-104.	0.6	21
13	ATXN2 intermediate repeat expansions influence the clinical phenotype in frontotemporal dementia. Neurobiology of Aging, 2019, 73, 231.e7-231.e9.	3.1	21
14	A novel homozygous change ofCLCN2(p.His590Pro) is associated with a subclinical form of leukoencephalopathy with ataxia (LKPAT). Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 894-896.	1.9	20
15	Spinocerebellar Ataxia Tethering PCR. Journal of Molecular Diagnostics, 2018, 20, 289-297.	2.8	16
16	Genome-wide expression profiling and functional characterization of SCA28 lymphoblastoid cell lines reveal impairment in cell growth and activation of apoptotic pathways. BMC Medical Genomics, 2013, 6, 22.	1.5	14
17	Exome sequencing in children of women with skewed X-inactivation identifies atypical cases and complex phenotypes. European Journal of Paediatric Neurology, 2017, 21, 475-484.	1.6	14
18	Blood metal levels and related antioxidant enzyme activities in patients with ataxia telangiectasia. Neurobiology of Disease, 2015, 81, 162-167.	4.4	13

#	Article	IF	Citations
19	Gene-targeted embryonic stem cells: real-time PCR assay for estimation of the number of neomycin selection cassettes. Biological Procedures Online, 2011, 13, 10.	2.9	12
20	An atypical form of AOA2 with myoclonus associated with mutations in SETX and AFG3L2. BMC Medical Genetics, 2015, 16, 16.	2.1	12
21	A case of Feingold type 2 syndrome associated with keratoconus refines keratoconus type 7 locus on chromosome 13q. European Journal of Medical Genetics, 2017, 60, 224-227.	1.3	10
22	Human canonical CD157/Bst1 is an alternatively spliced isoform masking a previously unidentified primate-specific exon included in a novel transcript. Scientific Reports, 2017, 7, 15923.	3.3	10
23	Updated genetic testing of Italian patients referred with a clinical diagnosis of primary hyperoxaluria. Journal of Nephrology, 2017, 30, 219-225.	2.0	9
24	Altered homeostasis of trace elements in the blood of SCA2 patients. Journal of Trace Elements in Medicine and Biology, 2018, 47, 111-114.	3.0	7
25	A fetal case of microphthalmia and limb anomalies with abnormal neuronal migration associated with SMOC1 biallelic variants. European Journal of Medical Genetics, 2019, 62, 103578.	1.3	4
26	In vitro dexamethasone treatment does not induce alternative ATM transcripts in cells from Ataxiaâ€“Telangiectasia patients. Scientific Reports, 2020, 10, 20182.	3.3	3
27	KCNK18 Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. International Journal of Molecular Sciences, 2021, 22, 6064.	4.1	3
28	Cover Image, Volume 170A, Number 7, July 2016. , 2016, 170, i-i.		0