# 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 | 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 |
| 2 | In vitro dexamethasone treatment does not induce alternative ATM transcripts in cells from Ataxiaâ€“Telangiectasia patients. Scientific Reports, 2020, 10, 20182. | 3.3 | 3 |
| 3 | 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 |
| 4 | 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 |
| 5 | 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 |
| 6 | ATXN2 intermediate repeat expansions influence the clinical phenotype in frontotemporal dementia. Neurobiology of Aging, 2019, 73, 231.e7-231.e9. | 3.1 | 21 |
| 7 | Mitochondrial stress response triggered by defects in protein synthesis quality control. Life Science Alliance, 2019, 2, e201800219. | 2.8 | 26 |
| 8 | Spinocerebellar Ataxia Tethering PCR. Journal of Molecular Diagnostics, 2018, 20, 289-297. | 2.8 | 16 |
| 9 | 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 |
| 10 | Updated genetic testing of Italian patients referred with a clinical diagnosis of primary hyperoxaluria. Journal of Nephrology, 2017, 30, 219-225. | 2.0 | 9 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | 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 |
| 15 | Cover Image, Volume 170A, Number 7, July 2016. , 2016, 170, i-i. | | 0 |
| 16 | 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 |
| 17 | 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 |
| 18 | Ribosomal <scp>RNA</scp> analysis in the diagnosis of Diamondâ€Blackfan Anaemia. British Journal of Haematology, 2016, 172, 782-785. | 2.5 | 24 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 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 | 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 |
| 22 | Blood metal levels and related antioxidant enzyme activities in patients with ataxia telangiectasia. Neurobiology of Disease, 2015, 81, 162-167. | 4.4 | 13 |
| 23 | Large cryptic genomic rearrangements with apparently normal karyotypes detected by array-CGH. Molecular Cytogenetics, 2014, 7, 82. | 0.9 | 25 |
| 24 | ELOVL5 Mutations Cause Spinocerebellar Ataxia 38. American Journal of Human Genetics, 2014, 95, 209-217. | 6.2 | 107 |
| 25 | 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 |
| 26 | 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 |
| 27 | 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 |
| 28 | Missense mutations in the AFG3L2 proteolytic domain account for âˆ¼1.5% of European autosomal dominant cerebellar ataxias. Human Mutation, 2010, 31, 1117-1124. | 2.5 | 81 |