# Brendan C Lanpher

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6653955/publications.pdf

Version: 2024-02-01

| 17 | 507 | 933447 | 940533 |
|---|---|---|---|
| papers | citations | 10 | 16 |
| | | h-index | g-index |

| 17 | 17 | 17 | 1205 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975. | 3.2 | 13 |
| 2 | Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507. | 2.4 | 24 |
| 3 | Bilateral subdural hematomas and retinal hemorrhages mimicking nonaccidental trauma in a patient with Dâ€2â€hydroxyglutaric aciduria. JIMD Reports, 2021, 58, 21-28. | 1.5 | 1 |
| 4 | A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856. | 6.2 | 15 |
| 5 | <scp>TSPEAR</scp> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. American Journal of Medical Genetics, Part A, 2021, 185, 2417-2433. | 1.2 | 10 |
| 6 | Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021. | 21.4 | 44 |
| 7 | Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. Brain Sciences, 2021, 11, 931. | 2.3 | 7 |
| 8 | Novel loss-of-function variants in TRIO are associated with neurodevelopmental disorder: case report. BMC Medical Genetics, 2020, 21, 219. | 2.1 | 6 |
| 9 | A novel missense variant and multiexon deletion causing a delayed presentation of xeroderma pigmentosum, group C. Journal of Physical Education and Sports Management, 2020, 6, a005165. | 1.2 | 1 |
| 10 | De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782. | 2.8 | 27 |
| 11 | RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121. | 6.2 | 39 |
| 12 | Kabuki syndrome: international consensus diagnostic criteria. Journal of Medical Genetics, 2019, 56, 89-95. | 3.2 | 146 |
| 13 | Diagnosis of Attenuated Mucopolysaccharidosis VI: Clinical, Biochemical, and Genetic Pitfalls. Pediatrics, 2018, 142, e20180658. | 2.1 | 6 |
| 14 | Multigenerational pedigree with STAR syndrome: A novel FAM58A variant and expansion of the phenotype. , 2017, 173, 1328-1333. | | 11 |
| 15 | Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536. | 21.4 | 113 |
| 16 | The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. Molecular Genetics and Metabolism Reports, 2017, 13, 46-51. | 1.1 | 17 |
| 17 | Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. JIMD Reports, 2017, 40, 63-69. | 1.5 | 27 |