

Laurie Demmer

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5501056/publications.pdf>

Version: 2024-02-01

10
papers

299
citations

1684188

5
h-index

1474206

9
g-index

10
all docs

10
docs citations

10
times ranked

774
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|--|-----|-----------|
| 1 | Recommendations for the integration of genomics into clinical practice. <i>Genetics in Medicine</i> , 2016, 18, 1075-1084. | 2.4 | 125 |
| 2 | Systematic evidence-based review: outcomes from exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability. <i>Genetics in Medicine</i> , 2020, 22, 986-1004. | 2.4 | 53 |
| 3 | Spectrum of K _V 2.1 Dysfunction in <i>KCNB1</i> Associated Neurodevelopmental Disorders. <i>Annals of Neurology</i> , 2019, 86, 899-912. | 5.3 | 52 |
| 4 | Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 150-158. | 1.2 | 40 |
| 5 | Autosomal recessive <i>MFN2</i> -related Charcot-Marie-Tooth disease with diaphragmatic weakness: Case report and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1580-1584. | 1.2 | 17 |
| 6 | Defining the genotypic and phenotypic spectrum of X-linked <i>MSL3</i> -related disorder. <i>Genetics in Medicine</i> , 2021, 23, 384-395. | 2.4 | 4 |
| 7 | Recurrent <i>de novo</i> missense variants in <i>GNB2</i> can cause syndromic intellectual disability. <i>Journal of Medical Genetics</i> , 2022, 59, 511-516. | 3.2 | 4 |
| 8 | Development, Implementation, and Assessment of a Genetics Curriculum Across Institutions. <i>AJP Reports</i> , 2016, 06, e372-e377. | 0.7 | 3 |
| 9 | An unusual case of nephrotic syndrome in a microcephalic infant: Answers. <i>Pediatric Nephrology</i> , 2019, 34, 2327-2329. | 1.7 | 1 |
| 10 | An unusual case of nephrotic syndrome in a microcephalic infant: Questions. <i>Pediatric Nephrology</i> , 2019, 34, 2325-2326. | 1.7 | 0 |