

Nicole J Boczek

List of Publications by Year in descending order

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Version: 2024-02-01

30
papers

940
citations

623734

14
h-index

526287

27
g-index

30
all docs

30
docs citations

30
times ranked

2041
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|--|-----|-----------|
| 1 | Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. <i>Human Mutation</i> , 2018, 39, 1593-1613. | 2.5 | 312 |
| 2 | Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 696-705. | 6.2 | 105 |
| 3 | Identification and Functional Characterization of a Novel <i>CACNA1C</i> -Mediated Cardiac Disorder Characterized by Prolonged QT Intervals With Hypertrophic Cardiomyopathy, Congenital Heart Defects, and Sudden Cardiac Death. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2015, 8, 1122-1132. | 4.8 | 76 |
| 4 | Characterization of SEMA3A -Encoded Semaphorin as a Naturally Occurring K v 4.3 Protein Inhibitor and its Contribution to Brugada Syndrome. <i>Circulation Research</i> , 2014, 115, 460-469. | 4.5 | 54 |
| 5 | Best practices for the interpretation and reporting of clinical whole genome sequencing. <i>Npj Genomic Medicine</i> , 2022, 7, 27. | 3.8 | 48 |
| 6 | Novel long QT syndrome-associated missense mutation, L762F, in <i>CACNA1C</i> -encoded L-type calcium channel imparts a slower inactivation tau and increased sustained and window current. <i>International Journal of Cardiology</i> , 2016, 220, 290-298. | 1.7 | 40 |
| 7 | Developmental delay and failure to thrive associated with a loss-of-function variant in <i>WHSC1 (NSD2)</i> . <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2798-2802. | 1.2 | 33 |
| 8 | Pharmacogenomic findings from clinical whole exome sequencing of diagnostic odyssey patients. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 269-279. | 1.2 | 30 |
| 9 | Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2018, 38, 307-319. | 3.8 | 29 |
| 10 | De Novo <i>DNM1L</i> Variant in a Teenager With Progressive Paroxysmal Dystonia and Lethal Super-refractory Myoclonic Status Epilepticus. <i>Journal of Child Neurology</i> , 2018, 33, 651-658. | 1.4 | 25 |
| 11 | Impact of integrated translational research on clinical exome sequencing. <i>Genetics in Medicine</i> , 2021, 23, 498-507. | 2.4 | 24 |
| 12 | Characterization of three ciliopathy pedigrees expands the phenotype associated with biallelic <i>C2CD3</i> variants. <i>European Journal of Human Genetics</i> , 2018, 26, 1797-1809. | 2.8 | 19 |
| 13 | Whole Exome Sequencing and Heterologous Cellular Electrophysiology Studies Elucidate a Novel Loss-of-Function Mutation in the <i>CACNA1A</i> -Encoded Neuronal P/Q-Type Calcium Channel in a Child With Congenital Hypotonia and Developmental Delay. <i>Pediatric Neurology</i> , 2016, 55, 46-51. | 2.1 | 18 |
| 14 | Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. <i>Genetics in Medicine</i> , 2021, 23, 2208-2212. | 2.4 | 18 |
| 15 | The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. <i>Molecular Genetics and Metabolism Reports</i> , 2017, 13, 46-51. | 1.1 | 17 |
| 16 | Type 8 long QT syndrome: pathogenic variants in <i>CACNA1C</i> -encoded Cav1.2 cluster in STAC protein binding site. <i>Europace</i> , 2019, 21, 1725-1732. | 1.7 | 15 |
| 17 | A novel <i>ANO3</i> variant identified in a 53-year-old woman presenting with hyperkinetic dysarthria, blepharospasm, hyperkinesias, and complex motor Åtics. <i>BMC Medical Genetics</i> , 2016, 17, 93. | 2.1 | 14 |
| 18 | Multigenerational pedigree with STAR syndrome: A novel <i>FAM58A</i> variant and expansion of the phenotype. , 2017, 173, 1328-1333. | | 11 |

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|----|---|-----|-----------|
| 19 | Familial Creutzfeldt-Jakob Disease: Case report and role of genetic counseling in post mortem testing. Prion, 2016, 10, 502-506. | 1.8 | 9 |
| 20 | Functional validation reveals the novel missense V419L variant in <i>TGFBR2</i> associated with Loey's-Dietz syndrome (LDS) impairs canonical TGF- β ² signaling. Journal of Physical Education and Sports Management, 2017, 3, a001727. | 1.2 | 7 |
| 21 | Early-onset limb-girdle muscular dystrophy-2L in a female athlete. Muscle and Nerve, 2017, 55, E19-E21. | 2.2 | 7 |
| 22 | Co-occurrence of a maternally inherited DNMT3A duplication and a paternally inherited pathogenic variant in EZH2 in a child with growth retardation and severe short stature: atypical Weaver syndrome or evidence of a DNMT3A dosage effect?. Journal of Physical Education and Sports Management, 2018, 4, a002899. | 1.2 | 7 |
| 23 | Whole exome sequencing of a patient with suspected mitochondrial myopathy reveals novel compound heterozygous variants in <i>RYR1</i> . Molecular Genetics & Genomic Medicine, 2017, 5, 295-302. | 1.2 | 6 |
| 24 | Novel Pathogenic Variant in TGFBR2 Confirmed by Molecular Modeling Is a Rare Cause of Loey's-Dietz Syndrome. Case Reports in Genetics, 2017, 2017, 1-4. | 0.2 | 4 |
| 25 | Expansion of <i>PURA</i> -Related Phenotypes and Discovery of a Novel PURA Variant: A Case Report. Child Neurology Open, 2020, 7, 2329048X2095500. | 1.1 | 4 |
| 26 | Development and Validation of a Next-Generation Sequencing Panel for Syndromic and Nonsyndromic Hearing Loss. journal of applied laboratory medicine, The, 2020, 5, 467-479. | 1.3 | 4 |
| 27 | First Report of Bilateral External Auditory Canal Cochlin Aggregates (Cochlinomas) with Multifocal Amyloid-Like Deposits, Associated with Sensorineural Hearing Loss and a Novel Genetic Variant in COCH Encoding Cochlin. Head and Neck Pathology, 2020, 14, 808-816. | 2.6 | 2 |
| 28 | Limited diagnostic impact of duplications <1 Mb of uncertain clinical significance: a 10-year retrospective analysis of reporting practices at the Mayo Clinic. Genetics in Medicine, 2020, 22, 2120-2124. | 2.4 | 2 |
| 29 | Whole Exome Sequencing Leading to the Diagnosis of Dysferlinopathy with a Novel Missense Mutation (c.959G>C). Case Reports in Genetics, 2016, 2016, 1-4. | 0.2 | 0 |
| 30 | Unconventional Diagnosis Based on Somatic Findings through Germ Line Whole-Exome Sequencing. Clinical Chemistry, 2020, 66, 48-51. | 3.2 | 0 |