Nicole J Boczek

List of Publications by Year
in descending order

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	Best practices for the interpretation and reporting of clinical whole genome sequencing. Npj Genomic Medicine, 2022, 7, 27.	3.8	48
2	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	2.4	24
3	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
4	First Report of Bilateral External Auditory Canal Cochlin Aggregates (â€œCochlinomasâ€) 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
5	Expansion of PURA-Related Phenotypes and Discovery of a Novel PURA Variant: A Case Report. Child Neurology Open, 2020, 7, 2329048X2095500.	1.1	4
6	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
7	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
8	Unconventional Diagnosis Based on Somatic Findings through Germ Line Whole-Exome Sequencing. Clinical Chemistry, 2020, 66, 48-51.	3.2	0
9	Type 8 long QT syndrome: pathogenic variants in CACNA1C-encoded Cav1.2 cluster in STAC protein binding site. Europace, 2019, 21, 1725-1732.	1.7	15
10	Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. Journal of Clinical Immunology, 2018, 38, 307-319.	3.8	29
11	Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. American Journal of Human Genetics, 2018, 102, 696-705.	6.2	105
12	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	2.5	312
13	Developmental delay and failure to thrive associated with a lossâ€ofâ€function variant in WHSC1 (NSD2). American Journal of Medical Genetics, Part A, 2018, 176, 2798-2802.	1.2	33
14	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
15	Characterization of three ciliopathy pedigrees expands the phenotype associated with biallelic C2CD3 variants. European Journal of Human Genetics, 2018, 26, 1797-1809.	2.8	19
16	De Novo DNM1L Variant in a Teenager With Progressive Paroxysmal Dystonia and Lethal Super-refractory Myoclonic Status Epilepticus. Journal of Child Neurology, 2018, 33, 651-658.	1.4	25
17	Whole exome sequencing of a patient with suspected mitochondrial myopathy reveals novel compound heterozygous variants in <scp>RYR</scp>1. Molecular Genetics & Genomic Medicine, 2017, 5, 295-302.	1.2	6
18	Multigenerational pedigree with STAR syndrome: A novel FAM58A variant and expansion of the phenotype. , 2017, 173, 1328-1333.		11

#	Article	IF	Citations
19	Pharmacogenomic findings from clinical whole exome sequencing of diagnostic odyssey patients. Molecular Genetics & Genomic Medicine, 2017, 5, 269-279.	1.2	30
20	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
21	Functional validation reveals the novel missense V419L variant in TGFBR2 associated with Loeysâ€“Dietz syndrome (LDS) impairs canonical TGF-Î² signaling. Journal of Physical Education and Sports Management, 2017, 3, a001727.	1.2	7
22	Earlyâ€onset limbâ€girdle muscular dystrophyâ€2L in a female athlete. Muscle and Nerve, 2017, 55, E19-E21.	2.2	7
23	Novel Pathogenic Variant in TGFBR2 Confirmed by Molecular Modeling Is a Rare Cause of Loeys-Dietz Syndrome. Case Reports in Genetics, 2017, 2017, 1-4.	0.2	4
24	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
25	Novel long QT syndrome-associated missense mutation, L762F, in CACNA1C-encoded L-type calcium channel imparts a slower inactivation tau and increased sustained and window current. International Journal of Cardiology, 2016, 220, 290-298.	1.7	40
26	Familial Creutzfeldt-Jakob Disease: Case report and role of genetic counseling in post mortem testing. Prion, 2016, 10, 502-506.	1.8	9
27	A novel ANO3 variant identified in a 53-year-old woman presenting with hyperkinetic dysarthria, blepharospasm, hyperkinesias, and complex motorÂtics. BMC Medical Genetics, 2016, 17, 93.	2.1	14
28	Whole Exome Sequencing and Heterologous Cellular Electrophysiology Studies Elucidate a Novel Loss-of-Function Mutation in the CACNA1A-Encoded Neuronal P/Q-Type Calcium Channel in a Child With Congenital Hypotonia and Developmental Delay. Pediatric Neurology, 2016, 55, 46-51.	2.1	18
29	Identification and Functional Characterization of a Novel CACNA1C -Mediated Cardiac Disorder Characterized by Prolonged QT Intervals With Hypertrophic Cardiomyopathy, Congenital Heart Defects, and Sudden Cardiac Death. Circulation: Arrhythmia and Electrophysiology, 2015, 8, 1122-1132.	4.8	76
30	Characterization of SEMA3A -Encoded Semaphorin as a Naturally Occurring K v 4.3 Protein Inhibitor and its Contribution to Brugada Syndrome. Circulation Research, 2014, 115, 460-469.	4.5	54