

Monica H Wojcik

List of Publications by Year in descending order

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Version: 2024-02-01

44
papers

1,322
citations

516710

16
h-index

395702

33
g-index

45
all docs

45
docs citations

45
times ranked

3018
citing authors

#	ARTICLE	IF	CITATIONS
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
2	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
3	Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. Genetics in Medicine, 2020, 22, 736-744.	2.4	83
4	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	2.8	60
5	Genetic disorders and mortality in infancy and early childhood: delayed diagnoses and missed opportunities. Genetics in Medicine, 2018, 20, 1396-1404.	2.4	58
6	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
7	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
8	Infant mortality: the contribution of genetic disorders. Journal of Perinatology, 2019, 39, 1611-1619.	2.0	47
9	The influence of social determinants of health on the genetic diagnostic odyssey: who remains undiagnosed, why, and to what effect?. Pediatric Research, 2021, 89, 295-300.	2.3	47
10	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	2.4	47
11	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
12	Histone H3.3 beyond cancer: Germline mutations in <i>Histone 3 Family 3A and 3B</i> cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
13	A missense mutation in the catalytic domain of <i>O</i> -GlcNAc transferase links perturbations in protein <i>O</i> -GlcNAcylation to <i>X</i> -linked intellectual disability. FEBS Letters, 2020, 594, 717-727.	2.8	40
14	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35
15	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	2.4	24
16	A retrospective analysis of the prevalence of imprinting disorders in Estonia from 1998 to 2016. European Journal of Human Genetics, 2019, 27, 1649-1658.	2.8	21
17	<i>matchbox</i> : An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	2.5	20
18	De novo variant in KIF26B is associated with pontocerebellar hypoplasia with infantile spinal muscular atrophy. American Journal of Medical Genetics, Part A, 2018, 176, 2623-2629.	1.2	19

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19	Heterozygous Variants in KDM4B Lead to Global Developmental Delay and Neuroanatomical Defects. <i>American Journal of Human Genetics</i> , 2020, 107, 1170-1177.	6.2	13
20	Beta-Ketothiolase Deficiency Presenting with Metabolic Stroke After a Normal Newborn Screen in Two Individuals. <i>JIMD Reports</i> , 2017, 39, 45-54.	1.5	10
21	Genetic diagnosis in the fetus. <i>Journal of Perinatology</i> , 2020, 40, 997-1006.	2.0	10
22	Delayed diagnosis and racial bias in children with genetic conditions. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1118-1123.	1.2	10
23	Peri-mortem evaluation of infants who die without a diagnosis: focus on advances in genomic technology. <i>Journal of Perinatology</i> , 2018, 38, 1125-1134.	2.0	9
24	Updating the neurodevelopmental profile of Alazami syndrome: Illustrating the role of developmental assessment in rare genetic disorders. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1565-1569.	1.2	9
25	A new diagnosis of Williams-Beuren syndrome in a 49-year-old man with severe bullous emphysema. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2235-2239.	1.2	8
26	Expanding the phenotypic spectrum associated with OPHN1 variants. <i>European Journal of Medical Genetics</i> , 2019, 62, 137-143.	1.3	8
27	Medical and surgical interventions and outcomes for infants with trisomy 18 (T18) or trisomy 13 (T13) at children's hospitals neonatal intensive care units (NICUs). <i>Journal of Perinatology</i> , 2021, 41, 1745-1754.	2.0	8
28	Neuroimaging in Kabuki syndrome and another KMT2D-related disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3770-3783.	1.2	7
29	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. <i>JAMA Neurology</i> , 2022, 79, 405.	9.0	7
30	Genome Sequencing Identifies the Pathogenic Variant Missed by Prior Testing in an Infant with Marfan Syndrome. <i>Journal of Pediatrics</i> , 2019, 213, 235-240.	1.8	6
31	Perspectives of United States neonatologists on genetic testing practices. <i>Genetics in Medicine</i> , 2022, 24, 1372-1377.	2.4	6
32	A model to implement genomic medicine in the neonatal intensive care unit. <i>Journal of Perinatology</i> , 2023, 43, 248-252.	2.0	6
33	A 19-year-old at 37 weeks gestation with an acute acetylsalicylic acid overdose. <i>CKJ: Clinical Kidney Journal</i> , 2011, 4, 394-396.	2.9	5
34	Developmental Support for Infants With Genetic Disorders. <i>Pediatrics</i> , 2020, 145, e20190629.	2.1	5
35	Exome sequencing identifies novel missense and deletion variants in RTN4IP1 associated with optic atrophy, global developmental delay, epilepsy, ataxia, and choreoathetosis. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 203-207.	1.2	5
36	The Unrecognized Mortality Burden of Genetic Disorders in Infancy. <i>American Journal of Public Health</i> , 2021, 111, S156-S162.	2.7	5

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37	Deciphering congenital anomalies for the next generation. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a005504.	1.2	5
38	Delineation of a novel neurodevelopmental syndrome associated with <i>PAX5</i> haploinsufficiency. <i>Human Mutation</i> , 2022, 43, 461-470.	2.5	5
39	A novel missense mutation in <i>TFAP2B</i> associated with Char syndrome and central diabetes insipidus. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1299-1303.	1.2	4
40	PEHO syndrome caused by compound heterozygote variants in <i>ZNHIT3</i> gene. <i>European Journal of Medical Genetics</i> , 2020, 63, 103660.	1.3	3
41	Genomic Insights into Stillbirth. <i>New England Journal of Medicine</i> , 2020, 383, 1182-1183.	27.0	2
42	Mortality in the neonatal intensive care unit: improving the accuracy of death reporting. <i>Journal of Perinatology</i> , 2022, 42, 671-676.	2.0	2
43	Cover Image, Volume 173A, Number 8, August 2017. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, i.	1.2	0
44	Prenatal Diagnosis of a Ventral Abdominal Wall Defect. <i>NeoReviews</i> , 2020, 21, e286-e292.	0.8	0