Monica H Wojcik

List of Publications by Year
in descending order

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	A model to implement genomic medicine in the neonatal intensive care unit. Journal of Perinatology, 2023, 43, 248-252.	2.0	6
2	Mortality in the neonatal intensive care unit: improving the accuracy of death reporting. Journal of Perinatology, 2022, 42, 671-676.	2.0	2
3	Delayed diagnosis and racial bias in children with genetic conditions. American Journal of Medical Genetics, Part A, 2022, 188, 1118-1123.	1.2	10
4	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5
5	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
6	Perspectives of United States neonatologists on genetic testing practices. Genetics in Medicine, 2022, 24, 1372-1377.	2.4	6
7	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. JAMA Neurology, 2022, 79, 405.	9.0	7
8	Exome sequencing identifies novel missense and deletion variants in <scp>RTN4IP1</scp> associated with optic atrophy, global developmental delay, epilepsy, ataxia, and choreoathetosis. American Journal of Medical Genetics, Part A, 2021, 185, 203-207.	1.2	5
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	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
12	Medical and surgical interventions and outcomes for infants with trisomy 18 (T18) or trisomy 13 (T13) at childrenâ€™s hospitals neonatal intensive care units (NICUs). Journal of Perinatology, 2021, 41, 1745-1754.	2.0	8
13	The Unrecognized Mortality Burden of Genetic Disorders in Infancy. American Journal of Public Health, 2021, 111, S156-S162.	2.7	5
14	Neuroimaging in Kabuki syndrome and another <scp>KMT2D</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2021, 185, 3770-3783.	1.2	7
15	PEHO syndrome caused by compound heterozygote variants in ZNHIT3 gene. European Journal of Medical Genetics, 2020, 63, 103660.	1.3	3
16	A missense mutation in the catalytic domain of Oâ€GlcNAc transferase links perturbations in protein Oâ€GlcNAcylation to Xâ€linked intellectual disability. FEBS Letters, 2020, 594, 717-727.	2.8	40
17	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
18	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139

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19	Heterozygous Variants in KDM4B Lead to Global Developmental Delay and Neuroanatomical Defects. American Journal of Human Genetics, 2020, 107, 1170-1177.	6.2	13
20	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
21	Genomic Insights into Stillbirth. New England Journal of Medicine, 2020, 383, 1182-1183.	27.0	2
22	Genetic diagnosis in the fetus. Journal of Perinatology, 2020, 40, 997-1006.	2.0	10
23	Developmental Support for Infants With Genetic Disorders. Pediatrics, 2020, 145, e20190629.	2.1	5
24	Prenatal Diagnosis of a Ventral Abdominal Wall Defect. NeoReviews, 2020, 21, e286-e292.	0.8	0
25	Deciphering congenital anomalies for the next generation. Journal of Physical Education and Sports Management, 2020, 6, a005504.	1.2	5
26	Expanding the phenotypic spectrum associated with OPHN1 variants. European Journal of Medical Genetics, 2019, 62, 137-143.	1.3	8
27	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
28	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
29	Genome Sequencing Identifies the Pathogenic Variant Missed by Prior Testing in an Infant with Marfan Syndrome. Journal of Pediatrics, 2019, 213, 235-240.	1.8	6
30	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
31	Infant mortality: the contribution of genetic disorders. Journal of Perinatology, 2019, 39, 1611-1619.	2.0	47
32	Updating the neurodevelopmental profile of Alazami syndrome: Illustrating the role of developmental assessment in rare genetic disorders. American Journal of Medical Genetics, Part A, 2019, 179, 1565-1569.	1.2	9
33	A novel missense mutation in TFAP2B associated with Char syndrome and central diabetes insipidus. American Journal of Medical Genetics, Part A, 2019, 179, 1299-1303.	1.2	4
34	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	2.8	60
35	Genetic disorders and mortality in infancy and early childhood: delayed diagnoses and missed opportunities. Genetics in Medicine, 2018, 20, 1396-1404.	2.4	58
36	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

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37	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	2.5	20
38	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
39	Peri-mortem evaluation of infants who die without a diagnosis: focus on advances in genomic technology. Journal of Perinatology, 2018, 38, 1125-1134.	2.0	9
40	A new diagnosis of Williamsâ€“Beuren syndrome in a 49â€yearâ€old man with severe bullous emphysema. American Journal of Medical Genetics, Part A, 2017, 173, 2235-2239.	1.2	8
41	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
42	Beta-Ketothiolase Deficiency Presenting with Metabolic Stroke After a Normal Newborn Screen in Two Individuals. JIMD Reports, 2017, 39, 45-54.	1.5	10
43	Cover Image, Volume 173A, Number 8, August 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
44	A 19-year-old at 37 weeks gestation with an acute acetylsalicylic acid overdose. CKJ: Clinical Kidney Journal, 2011, 4, 394-396.	2.9	5