

Brian G Skotko

List of Publications by Year in descending order

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Version: 2024-02-01

95
papers

4,186
citations

172457

29
h-index

128289

60
g-index

105
all docs

105
docs citations

105
times ranked

3815
citing authors

#	ARTICLE	IF	CITATIONS
1	Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. <i>Genetics in Medicine</i> , 2016, 18, 1056-1065.	2.4	541
2	Down syndrome. <i>Nature Reviews Disease Primers</i> , 2020, 6, 9.	30.5	376
3	Down syndrome: Cognitive and behavioral functioning across the lifespan. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2015, 169, 135-149.	1.6	254
4	ACMG statement on noninvasive prenatal screening for fetal aneuploidy. <i>Genetics in Medicine</i> , 2013, 15, 395-398.	2.4	250
5	Estimation of the number of people with Down syndrome in the United States. <i>Genetics in Medicine</i> , 2017, 19, 439-447.	2.4	197
6	Estimates of the live births, natural losses, and elective terminations with Down syndrome in the United States. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 756-767.	1.2	191
7	Mothers of Children With Down Syndrome Reflect on Their Postnatal Support. <i>Pediatrics</i> , 2005, 115, 64-77.	2.1	166
8	Having a son or daughter with Down syndrome: Perspectives from mothers and fathers. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2335-2347.	1.2	135
9	Prenatally diagnosed Down syndrome: Mothers who continued their pregnancies evaluate their health care providers. <i>American Journal of Obstetrics and Gynecology</i> , 2005, 192, 670-677.	1.3	129
10	Self-perceptions from people with Down syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2360-2369.	1.2	100
11	Postnatal Diagnosis of Down Syndrome: Synthesis of the Evidence on How Best to Deliver the News. <i>Pediatrics</i> , 2009, 124, e751-e758.	2.1	91
12	Prenatal diagnosis of Down syndrome: How best to deliver the news. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2361-2367.	1.2	70
13	With new prenatal testing, will babies with Down syndrome slowly disappear?. <i>Archives of Disease in Childhood</i> , 2009, 94, 823-826.	1.9	68
14	Estimation of the number of people with Down syndrome in Europe. <i>European Journal of Human Genetics</i> , 2021, 29, 402-410.	2.8	65
15	Contributions of a specialty clinic for children and adolescents with Down syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 430-437.	1.2	61
16	Storybook-based communication intervention for girls with Rett syndrome and their mothers. <i>Disability and Rehabilitation</i> , 2001, 23, 149-159.	1.8	58
17	Having a brother or sister with Down syndrome: Perspectives from siblings. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2348-2359.	1.2	57
18	Estimation of live birth and population prevalence of Down syndrome in nine U.S. states. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2710-2719.	1.2	55

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19	Parent Reading Behaviors and Communication Outcomes in Girls with Rett Syndrome. <i>Exceptional Children</i> , 2004, 70, 145-166.	2.2	52
20	Parental Perceptions of Sleep Disturbances and Sleep-Disordered Breathing in Children With Down Syndrome. <i>Clinical Pediatrics</i> , 2011, 50, 121-125.	0.8	51
21	A predictive model for obstructive sleep apnea and Down syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 889-896.	1.2	51
22	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. <i>American Journal of Human Genetics</i> , 2019, 105, 493-508.	6.2	48
23	Puzzling Thoughts for H. M.: Can New Semantic Information Be Anchored to Old Semantic Memories?. <i>Neuropsychology</i> , 2004, 18, 756-769.	1.3	47
24	Supporting Communication of Girls with Rett Syndrome and their Mothers in Storybook Reading. <i>International Journal of Disability Development and Education</i> , 2001, 48, 395-410.	1.1	43
25	Postnatal Support for Mothers of Children With Down Syndrome. <i>Mental Retardation</i> , 2005, 43, 196-212.	1.0	40
26	Hypoglossal Nerve Stimulator Implantation in an Adolescent With Down Syndrome and Sleep Apnea. <i>Pediatrics</i> , 2016, 137, .	2.1	37
27	Pharmacological interventions to improve cognition and adaptive functioning in Down syndrome: Strides to date. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3029-3041.	1.2	37
28	Hypoglossal Nerve Stimulation in Adolescents With Down Syndrome and Obstructive Sleep Apnea. <i>JAMA Otolaryngology - Head and Neck Surgery</i> , 2018, 144, 37-42.	2.2	37
29	Unexplained regression in Down syndrome: 35 cases from an international Down syndrome database. <i>Genetics in Medicine</i> , 2020, 22, 767-776.	2.4	36
30	Down Syndrome Disintegrative Disorder: A Clinical Regression Syndrome of Increasing Importance. <i>Pediatrics</i> , 2020, 145, e20192939.	2.1	36
31	Thyroid dysfunction in patients with Down syndrome: Results from a multi-institutional registry study. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1539-1545.	1.2	34
32	What the other children are thinking: Brothers and sisters of persons with Down syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2006, 142C, 180-186.	1.6	30
33	Family perspectives about Down syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 930-941.	1.2	30
34	Adherence of cell-free DNA noninvasive prenatal screens to ACMG recommendations. <i>Genetics in Medicine</i> , 2019, 21, 2285-2292.	2.4	30
35	Use of Electronic Health Record Integration for Down Syndrome Guidelines. <i>Pediatrics</i> , 2018, 142, .	2.1	28
36	A Randomized, Double-Blind, Placebo-Controlled, Phase II Study of Oral ELND005 (scyllo-Inositol) in Young Adults with Down Syndrome without Dementia. <i>Journal of Alzheimer's Disease</i> , 2017, 58, 401-411.	2.6	27

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37	Parents' perceptions of functional abilities in people with Down syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 161-176.	1.2	27
38	Quantitative MRI Analyses of Regional Brain Growth in Living Fetuses with Down Syndrome. Cerebral Cortex, 2020, 30, 382-390.	2.9	24
39	Evaluation of Upper Airway Stimulation for Adolescents With Down Syndrome and Obstructive Sleep Apnea. JAMA Otolaryngology - Head and Neck Surgery, 2022, 148, 522.	2.2	24
40	Rapid clinical deterioration in an individual with Down syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1899-1902.	1.2	23
41	Out-of-pocket medical costs and third-party healthcare costs for children with Down syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 627-637.	1.2	23
42	Language and the medial temporal lobe: Evidence from H.M.'s spontaneous discourse. Journal of Memory and Language, 2005, 53, 397-415.	2.1	22
43	Live births, natural losses, and elective terminations with Down syndrome in Massachusetts. Genetics in Medicine, 2016, 18, 459-466.	2.4	22
44	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Genetics in Medicine, 2019, 21, 1058-1064.	2.4	22
45	Physical activity patterns in adults with Down Syndrome. Journal of Applied Research in Intellectual Disabilities, 2020, 33, 1457-1464.	2.0	20
46	Dynamics of plasma biomarkers in Down syndrome: the relative levels of A β 242 decrease with age, whereas NT1 tau and NfL increase. Alzheimer's Research and Therapy, 2020, 12, 27.	6.2	20
47	National down syndrome patient database: Insights from the development of a multi-center registry study. American Journal of Medical Genetics, Part A, 2015, 167, 2520-2526.	1.2	19
48	Craniofacial features as assessed by lateral cephalometric measurements in children with Down syndrome. Progress in Orthodontics, 2016, 17, 35.	3.5	19
49	Transition to virtual clinic: Experience in a multidisciplinary clinic for Down syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 70-82.	1.6	19
50	Assessment and Diagnosis of Down Syndrome Regression Disorder: International Expert Consensus. Frontiers in Neurology, 0, 13, .	2.4	18
51	The facial morphology in Down syndrome: A 3D comparison of patients with and without obstructive sleep apnea. American Journal of Medical Genetics, Part A, 2017, 173, 3013-3021.	1.2	16
52	<i>HIST1H1E</i> heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
53	A randomized controlled trial of an online health tool about Down syndrome. Genetics in Medicine, 2021, 23, 163-173.	2.4	16
54	Receiving the news of Down syndrome in the era of prenatal testing. American Journal of Medical Genetics, Part A, 2020, 182, 374-385.	1.2	15

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55	Epileptic spasms in individuals with Down syndrome: A review of the current literature. <i>Epilepsia Open</i> , 2020, 5, 344-353.	2.4	15
56	Cross-Sectional Exploration of Plasma Biomarkers of Alzheimer's Disease in Down Syndrome: Early Data from the Longitudinal Investigation for Enhancing Down Syndrome Research (LIFE-DSR) Study. <i>Journal of Clinical Medicine</i> , 2021, 10, 1907.	2.4	15
57	A randomized, double-blind, placebo-controlled phase II trial to explore the effects of a GABA _A receptor modulator (basmisanil) on intellectual disability associated with Down syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2022, 14, 10.	3.1	15
58	Detecting celiac disease in patients with Down syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3098-3105.	1.2	14
59	Urinary biomarkers and obstructive sleep apnea in patients with Down syndrome. <i>Sleep Medicine</i> , 2017, 34, 84-89.	1.6	13
60	Diminished Blood Pressure Profiles in Children With Down Syndrome. <i>Hypertension</i> , 2020, 75, 819-825.	2.7	13
61	Regional Alterations in Cortical Sulcal Depth in Living Fetuses with Down Syndrome. <i>Cerebral Cortex</i> , 2021, 31, 757-767.	2.9	13
62	Geospatial Analyses of Accessibility to Down Syndrome Specialty Care. <i>Journal of Pediatrics</i> , 2020, 218, 146-150.e1.	1.8	12
63	Redefining Success by Focusing on Failures After Pediatric Hypoglossal Stimulation in Down Syndrome. <i>Laryngoscope</i> , 2021, 131, 1663-1669.	2.0	12
64	Long-term stability of hypoglossal nerve stimulation for the treatment of obstructive sleep apnea in children with Down syndrome. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2021, 149, 110868.	1.0	12
65	Increased Autoimmunity in Individuals With Down Syndrome and Moyamoya Disease. <i>Frontiers in Neurology</i> , 2021, 12, 724969.	2.4	11
66	Safety, Tolerability, and Immunogenicity of the ACI-24 Vaccine in Adults With Down Syndrome. <i>JAMA Neurology</i> , 2022, 79, 565.	9.0	11
67	Detection of iron deficiency in children with Down syndrome. <i>Genetics in Medicine</i> , 2020, 22, 317-325.	2.4	10
68	H.M.'s personal crossword puzzles: Understanding memory and language. <i>Memory</i> , 2008, 16, 89-96.	1.7	9
69	Offering Prenatal Screening in the Age of Genomic Medicine: A Practical Guide. <i>Journal of Women's Health</i> , 2017, 26, 755-761.	3.3	9
70	Piloting the use of global health measures in a Down syndrome clinic. <i>Journal of Applied Research in Intellectual Disabilities</i> , 2021, 34, 1108-1117.	2.0	9
71	Pneumonia and respiratory infection in Down syndrome: A 10-year cohort analysis of inpatient and outpatient encounters across the lifespan. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2878-2887.	1.2	7
72	Preliminary Neurocognitive Results Post Hypoglossal Nerve Stimulation in Patients With Down Syndrome. <i>Laryngoscope</i> , 2021, 131, 2830-2833.	2.0	7

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73	Alzheimer's disease development in adults with Down syndrome: Caregivers' perspectives. American Journal of Medical Genetics, Part A, 2020, 182, 104-114.	1.2	6
74	The experiences and support needs of siblings of people with mucopolysaccharidosis. American Journal of Medical Genetics, Part A, 2021, 185, 3418-3426.	1.2	6
75	Assessment of Stakeholder Engagement in a Down Syndrome Research Study. Journal of Patient-centered Research and Reviews, 2021, 8, 64-67.	0.9	5
76	Words matter: The importance of nondirective language in first-trimester assessments for Down syndrome. American Journal of Obstetrics and Gynecology, 2006, 195, 625-626.	1.3	4
77	First- and Second-Trimester Evaluation of Risk for Down Syndrome. Obstetrics and Gynecology, 2007, 110, 1426.	2.4	3
78	Resources available for informed prenatal decisions. Genetics in Medicine, 2012, 14, 348-349.	2.4	3
79	Brief report: Caregiver perceived physical activity preferences of adults with Down syndrome. Journal of Applied Research in Intellectual Disabilities, 2022, 35, 910-915.	2.0	3
80	Characterization of two standard CMOS EEPROM designs. , 0, , .		2
81	ACLâ€24 vaccine in adults with Down syndrome. Alzheimer's and Dementia, 2020, 16, e038678.	0.8	2
82	Epileptic Spasms in Patients With Down Syndrome: Experiences From Caregivers. Journal of Child Neurology, 2020, 35, 813-819.	1.4	2
83	Using a Communication Passport within a Multidisciplinary Genetics Clinic. Pediatric Quality & Safety, 2021, 6, e472.	0.8	2
84	Comparing Three Screening Strategies for Combining First- and Second-Trimester Down Syndrome Markers. Obstetrics and Gynecology, 2006, 107, 1170.	2.4	1
85	New-Onset Abnormal Vocalizations in an Adult Woman With Down Syndrome. Psychosomatics, 2020, 61, 804-807.	2.5	1
86	Can a Christ child have Down syndrome?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 213-215.	1.6	1
87	A Surprising Postnatal Diagnosis. Obstetrics and Gynecology, 2006, 108, 1297.	2.4	0
88	Out-of-pocket medical costs for parents with Children with down Syndrome in the United States. Value in Health, 2015, 18, A258.	0.3	0
89	Income Growth Trajectory For Parents Of Children With Down Syndrome In The United States. Value in Health, 2016, 19, A17.	0.3	0
90	0769 Polysomnographic Analysis Of Post-stimulation Titration In Children With Down Syndrome And Hypoglossal Nerve Implant. Sleep, 2018, 41, A286-A286.	1.1	0

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91	Response to Johansen Taber et al.. Genetics in Medicine, 2019, 21, 2660-2661.	2.4	0
92	Comment on "The price of abandoning diagnostics testing for cell-free fetal DNA screening". Prenatal Diagnosis, 2019, 39, 130-130.	2.3	0
93	Response to Zhang et al.. Genetics in Medicine, 2020, 22, 662-662.	2.4	0
94	Adherence of Cell-Free DNA Noninvasive Prenatal Screens to ACMG Recommendations. Obstetrical and Gynecological Survey, 2020, 75, 224-226.	0.4	0
95	P2030: THE LIFE-DSR STUDY. Alzheimer's and Dementia, 2019, 15, .	0.8	0