

# Brian G Skotko

## List of Publications by Year in descending order

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Version: 2024-02-01

95  
papers

4,186  
citations

196777

29  
h-index

145109

60  
g-index

105  
all docs

105  
docs citations

105  
times ranked

4033  
citing authors

#	ARTICLE	IF	CITATIONS
1	Brief report: Caregiver perceived physical activity preferences of adults with Down syndrome. <i>Journal of Applied Research in Intellectual Disabilities</i> , 2022, 35, 910-915.	1.3	3
2	A randomized, double-blind, placebo-controlled phase II trial to explore the effects of a GABAA- $\alpha$ 5 NAM (basmisanil) on intellectual disability associated with Down syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2022, 14, 10.	1.5	15
3	Evaluation of Upper Airway Stimulation for Adolescents With Down Syndrome and Obstructive Sleep Apnea. <i>JAMA Otolaryngology - Head and Neck Surgery</i> , 2022, 148, 522.	1.2	24
4	Safety, Tolerability, and Immunogenicity of the ACI-24 Vaccine in Adults With Down Syndrome. <i>JAMA Neurology</i> , 2022, 79, 565.	4.5	11
5	Redefining Success by Focusing on Failures After Pediatric Hypoglossal Stimulation in Down Syndrome. <i>Laryngoscope</i> , 2021, 131, 1663-1669.	1.1	12
6	Estimation of the number of people with Down syndrome in Europe. <i>European Journal of Human Genetics</i> , 2021, 29, 402-410.	1.4	65
7	Regional Alterations in Cortical Sulcal Depth in Living Fetuses with Down Syndrome. <i>Cerebral Cortex</i> , 2021, 31, 757-767.	1.6	13
8	A randomized controlled trial of an online health tool about Down syndrome. <i>Genetics in Medicine</i> , 2021, 23, 163-173.	1.1	16
9	Transition to virtual clinic: Experience in a multidisciplinary clinic for Down syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 70-82.	0.7	19
10	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.	1.3	9
11	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.	1.0	15
12	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.	0.7	7
13	Can a Christ child have Down syndrome?. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 213-215.	0.7	1
14	The experiences and support needs of siblings of people with mucopolysaccharidosis. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3418-3426.	0.7	6
15	Preliminary Neurocognitive Results Post Hypoglossal Nerve Stimulation in Patients With Down Syndrome. <i>Laryngoscope</i> , 2021, 131, 2830-2833.	1.1	7
16	Increased Autoimmunity in Individuals With Down Syndrome and Moyamoya Disease. <i>Frontiers in Neurology</i> , 2021, 12, 724969.	1.1	11
17	Using a Communication Passport within a Multidisciplinary Genetics Clinic. <i>Pediatric Quality &amp; Safety</i> , 2021, 6, e472.	0.4	2
18	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.	0.4	12

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19	Assessment of Stakeholder Engagement in a Down Syndrome Research Study. <i>Journal of Patient-centered Research and Reviews</i> , 2021, 8, 64-67.	0.6	5
20	Detection of iron deficiency in children with Down syndrome. <i>Genetics in Medicine</i> , 2020, 22, 317-325.	1.1	10
21	Quantitative MRI Analyses of Regional Brain Growth in Living Fetuses with Down Syndrome. <i>Cerebral Cortex</i> , 2020, 30, 382-390.	1.6	24
22	Response to Zhang et al.. <i>Genetics in Medicine</i> , 2020, 22, 662-662.	1.1	0
23	Alzheimer's disease development in adults with Down syndrome: Caregivers' perspectives. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 104-114.	0.7	6
24	Geospatial Analyses of Accessibility to Down Syndrome Specialty Care. <i>Journal of Pediatrics</i> , 2020, 218, 146-150.e1.	0.9	12
25	Unexplained regression in Down syndrome: 35 cases from an international Down syndrome database. <i>Genetics in Medicine</i> , 2020, 22, 767-776.	1.1	36
26	Receiving the news of Down syndrome in the era of prenatal testing. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 374-385.	0.7	15
27	Physical activity patterns in adults with Down Syndrome. <i>Journal of Applied Research in Intellectual Disabilities</i> , 2020, 33, 1457-1464.	1.3	20
28	ACÎ²4 vaccine in adults with Down syndrome. <i>Alzheimer's and Dementia</i> , 2020, 16, e038678.	0.4	2
29	Down Syndrome Disintegrative Disorder: A Clinical Regression Syndrome of Increasing Importance. <i>Pediatrics</i> , 2020, 145, e20192939.	1.0	36
30	Epileptic spasms in individuals with Down syndrome: A review of the current literature. <i>Epilepsia Open</i> , 2020, 5, 344-353.	1.3	15
31	Adherence of Cell-Free DNA Noninvasive Prenatal Screens to ACMG Recommendations. <i>Obstetrical and Gynecological Survey</i> , 2020, 75, 224-226.	0.2	0
32	Dynamics of plasma biomarkers in Down syndrome: the relative levels of AÎ²42 decrease with age, whereas NT1 tau and NfL increase. <i>Alzheimer's Research and Therapy</i> , 2020, 12, 27.	3.0	20
33	Epileptic Spasms in Patients With Down Syndrome: Experiences From Caregivers. <i>Journal of Child Neurology</i> , 2020, 35, 813-819.	0.7	2
34	Down syndrome. <i>Nature Reviews Disease Primers</i> , 2020, 6, 9.	18.1	376
35	Diminished Blood Pressure Profiles in Children With Down Syndrome. <i>Hypertension</i> , 2020, 75, 819-825.	1.3	13
36	New-Onset Abnormal Vocalizations in an Adult Woman With Down Syndrome. <i>Psychosomatics</i> , 2020, 61, 804-807.	2.5	1

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37	<i>HIST1H1E</i> heterozygous proteinâ€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.	0.7	16
38	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508.	2.6	48
39	Response to Johansen Taber et al.. Genetics in Medicine, 2019, 21, 2660-2661.	1.1	0
40	Adherence of cell-free DNA noninvasive prenatal screens to ACMG recommendations. Genetics in Medicine, 2019, 21, 2285-2292.	1.1	30
41	Parents' perceptions of functional abilities in people with Down syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 161-176.	0.7	27
42	Comment on â€œThe price of abandoning diagnostics testing for cellâ€free fetal DNA screeningâ€• Prenatal Diagnosis, 2019, 39, 130-130.	1.1	0
43	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Genetics in Medicine, 2019, 21, 1058-1064.	1.1	22
44	P2â€030: THE LIFEâ€DSR STUDY. Alzheimer's and Dementia, 2019, 15, .	0.4	0
45	Hypoglossal Nerve Stimulation in Adolescents With Down Syndrome and Obstructive Sleep Apnea. JAMA Otolaryngology - Head and Neck Surgery, 2018, 144, 37-42.	1.2	37
46	0769 Polysomnographic Analysis Of Post-stimulation Titration In Children With Down Syndrome And Hypoglossal Nerve Implant. Sleep, 2018, 41, A286-A286.	0.6	0
47	Use of Electronic Health Record Integration for Down Syndrome Guidelines. Pediatrics, 2018, 142, .	1.0	28
48	A predictive model for obstructive sleep apnea and Down syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 889-896.	0.7	51
49	A Randomized, Double-Blind, Placebo-Controlled, Phase II Study of Oral ELND005 (scyllo-Inositol) in Young Adults with Down Syndrome without Dementia. Journal of Alzheimer's Disease, 2017, 58, 401-411.	1.2	27
50	Thyroid dysfunction in patients with Down syndrome: Results from a multiâ€institutional registry study. American Journal of Medical Genetics, Part A, 2017, 173, 1539-1545.	0.7	34
51	Offering Prenatal Screening in the Age of Genomic Medicine: A Practical Guide. Journal of Women's Health, 2017, 26, 755-761.	1.5	9
52	Urinary biomarkers and obstructive sleep apnea in patients with Down syndrome. Sleep Medicine, 2017, 34, 84-89.	0.8	13
53	Pharmacological interventions to improve cognition and adaptive functioning in Down syndrome: Strides to date. American Journal of Medical Genetics, Part A, 2017, 173, 3029-3041.	0.7	37
54	Outâ€ofâ€pocket medical costs and thirdâ€party healthcare costs for children with Down syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 627-637.	0.7	23

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55	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.	0.7	16
56	Estimation of live birth and population prevalence of Down syndrome in nine U.S. states. American Journal of Medical Genetics, Part A, 2017, 173, 2710-2719.	0.7	55
57	Estimation of the number of people with Down syndrome in the United States. Genetics in Medicine, 2017, 19, 439-447.	1.1	197
58	Craniofacial features as assessed by lateral cephalometric measurements in children with Down syndrome. Progress in Orthodontics, 2016, 17, 35.	1.3	19
59	Live births, natural losses, and elective terminations with Down syndrome in Massachusetts. Genetics in Medicine, 2016, 18, 459-466.	1.1	22
60	Hypoglossal Nerve Stimulator Implantation in an Adolescent With Down Syndrome and Sleep Apnea. Pediatrics, 2016, 137, .	1.0	37
61	Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. Genetics in Medicine, 2016, 18, 1056-1065.	1.1	541
62	Family perspectives about Down syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 930-941.	0.7	30
63	Rapid clinical deterioration in an individual with Down syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1899-1902.	0.7	23
64	Detecting celiac disease in patients with Down syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3098-3105.	0.7	14
65	Income Growth Trajectory For Parents Of Children With Down Syndrome In The United States. Value in Health, 2016, 19, A17.	0.1	0
66	Estimates of the live births, natural losses, and elective terminations with Down syndrome in the United States. American Journal of Medical Genetics, Part A, 2015, 167, 756-767.	0.7	191
67	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.	0.7	19
68	Down syndrome: Cognitive and behavioral functioning across the lifespan. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 135-149.	0.7	254
69	Out-of-pocket medical costs for parents with Children with down Syndrome in the United States. Value in Health, 2015, 18, A258.	0.1	0
70	ACMG statement on noninvasive prenatal screening for fetal aneuploidy. Genetics in Medicine, 2013, 15, 395-398.	1.1	250
71	Contributions of a specialty clinic for children and adolescents with Down syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 430-437.	0.7	61
72	Resources available for informed prenatal decisions. Genetics in Medicine, 2012, 14, 348-349.	1.1	3

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73	Having a brother or sister with Down syndrome: Perspectives from siblings. American Journal of Medical Genetics, Part A, 2011, 155, 2348-2359.	0.7	57
74	Self-perceptions from people with Down syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 2360-2369.	0.7	100
75	Having a son or daughter with Down syndrome: Perspectives from mothers and fathers. American Journal of Medical Genetics, Part A, 2011, 155, 2335-2347.	0.7	135
76	Parental Perceptions of Sleep Disturbances and Sleep-Disordered Breathing in Children With Down Syndrome. Clinical Pediatrics, 2011, 50, 121-125.	0.4	51
77	With new prenatal testing, will babies with Down syndrome slowly disappear?. Archives of Disease in Childhood, 2009, 94, 823-826.	1.0	68
78	Postnatal Diagnosis of Down Syndrome: Synthesis of the Evidence on How Best to Deliver the News. Pediatrics, 2009, 124, e751-e758.	1.0	91
79	Prenatal diagnosis of Down syndrome: How best to deliver the news. American Journal of Medical Genetics, Part A, 2009, 149A, 2361-2367.	0.7	70
80	H.M.'s personal crossword puzzles: Understanding memory and language. Memory, 2008, 16, 89-96.	0.9	9
81	First- and Second-Trimester Evaluation of Risk for Down Syndrome. Obstetrics and Gynecology, 2007, 110, 1426.	1.2	3
82	Comparing Three Screening Strategies for Combining First- and Second-Trimester Down Syndrome Markers. Obstetrics and Gynecology, 2006, 107, 1170.	1.2	1
83	A Surprising Postnatal Diagnosis. Obstetrics and Gynecology, 2006, 108, 1297.	1.2	0
84	Words matter: The importance of nondirective language in first-trimester assessments for Down syndrome. American Journal of Obstetrics and Gynecology, 2006, 195, 625-626.	0.7	4
85	What the other children are thinking: Brothers and sisters of persons with Down syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2006, 142C, 180-186.	0.7	30
86	Language and the medial temporal lobe: Evidence from H.M.'s spontaneous discourse. Journal of Memory and Language, 2005, 53, 397-415.	1.1	22
87	Prenatally diagnosed Down syndrome: Mothers who continued their pregnancies evaluate their health care providers. American Journal of Obstetrics and Gynecology, 2005, 192, 670-677.	0.7	129
88	Mothers of Children With Down Syndrome Reflect on Their Postnatal Support. Pediatrics, 2005, 115, 64-77.	1.0	166
89	Postnatal Support for Mothers of Children With Down Syndrome. Mental Retardation, 2005, 43, 196-212.	1.1	40
90	Parent Reading Behaviors and Communication Outcomes in Girls with Rett Syndrome. Exceptional Children, 2004, 70, 145-166.	1.4	52

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91	Puzzling Thoughts for H. M.: Can New Semantic Information Be Anchored to Old Semantic Memories?. Neuropsychology, 2004, 18, 756-769.	1.0	47
92	Storybook-based communication intervention for girls with Rett syndrome and their mothers. Disability and Rehabilitation, 2001, 23, 149-159.	0.9	58
93	Supporting Communication of Girls with Rett Syndrome and their Mothers in Storybook Reading. International Journal of Disability Development and Education, 2001, 48, 395-410.	0.6	43
94	Characterization of two standard CMOS EEPROM designs. , 0, , .		2
95	Assessment and Diagnosis of Down Syndrome Regression Disorder: International Expert Consensus. Frontiers in Neurology, 0, 13, .	1.1	18