Bruce R Korf

List of Publications by Year
in descending order

113	13,595	38742 50	23533 111
papers	citations	h-index	g-index

123	123	123	16343
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Analysis of patientâ€specific NF1 variants leads to functional insights for Ras signaling that can impact personalized medicine. Human Mutation, 2022, 43, 30-41.	2.5	6
2	Genome sequencing as a first-line diagnostic test for hospitalized infants. Genetics in Medicine, 2022, 24, 851-861.	2.4	22
3	An interview on rare and genetic diseases with Dr Bruce Korf, Associate Dean for Genomic Medicine at the University of Alabama at Birmingham. Current Medical Research and Opinion, 2022, 38, 161-163.	1.9	0
4	The seventh international <scp>RASopathies</scp> symposium: Pathways to a cureâ€"expanding knowledge, enhancing research, and therapeutic discovery. American Journal of Medical Genetics, Part A, 2022, 188, 1915-1927.	1.2	10
5	Education and Training of Non-Genetics Providers on the Return of Genome Sequencing Results in a NICU Setting. Journal of Personalized Medicine, 2022, 12, 405.	2.5	13
6	Targeted exon skipping of NF1 exon 17 as a therapeutic for neurofibromatosis type I. Molecular Therapy - Nucleic Acids, 2022, 28, 261-278.	5.1	7
7	Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls. Genetics in Medicine, 2021, 23, 280-288.	2.4	9
8	A state-based approach to genomics for rare disease and population screening. Genetics in Medicine, 2021, 23, 777-781.	2.4	19
9	Visual outcomes following everolimus targeted therapy for neurofibromatosis type 1â€associated optic pathway gliomas in children. Pediatric Blood and Cancer, 2021, 68, e28833.	1.5	9
10	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	2.5	10
11	Comparison of family health history in surveys vs electronic health record data mapped to the observational medical outcomes partnership data model in the All of Us Research Program. Journal of the American Medical Informatics Association: JAMIA, 2021, 28, 695-703.	4.4	11
12	Cabozantinib for neurofibromatosis type 1â€"related plexiform neurofibromas: a phase 2 trial. Nature Medicine, 2021, 27, 165-173.	30.7	46
13	NF106: A Neurofibromatosis Clinical Trials Consortium Phase II Trial of the MEK Inhibitor Mirdametinib (PD-0325901) in Adolescents and Adults With NF1-Related Plexiform Neurofibromas. Journal of Clinical Oncology, 2021, 39, 797-806.	1.6	54
14	Ensuring best practice in genomics education and evaluation: reporting item standards for education and its evaluation in genomics (RISE2 Genomics). Genetics in Medicine, 2021, 23, 1356-1365.	2.4	17
15	Pitfalls and challenges in genetic test interpretation: An exploration of genetic professionals experience with interpretation of results. Clinical Genetics, 2021, 99, 638-649.	2.0	15
16	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
17	An evaluation of selumetinib for the treatment of neurofibromatosis type 1-associated symptomatic, inoperable plexiform neurofibromas. Expert Review of Precision Medicine and Drug Development, 2021, 6, 239-246.	0.7	1
18	Status and Recommendations for Incorporating Biomarkers for Cutaneous Neurofibromas Into Clinical Research. Neurology, 2021, 97, S42-S49.	1.1	2

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19	Restoration of Normal NF1 Function with Antisense Morpholino Treatment of Recurrent Pathogenic Patient-Specific Variant c.1466A>G; p.Y489C. Journal of Personalized Medicine, 2021, 11, 1320.	2.5	3
20	Return of raw data in genomic testing and research: ownership, partnership, and riskâ€"benefit. Genetics in Medicine, 2020, 22, 12-14.	2.4	2
21	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€"phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
22	An Update on Neurofibromatosis Type 1-Associated Gliomas. Cancers, 2020, 12, 114.	3.7	50
23	Fibulin-5 mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis. Neurology: Genetics, 2020, 6, e476.	1.9	0
24	Mutation-Directed Therapeutics for Neurofibromatosis Type I. Molecular Therapy - Nucleic Acids, 2020, 20, 739-753.	5.1	16
25	Two novel cases further expand the phenotype of TOR1AIP1-associated nuclear envelopathies. Human Genetics, 2020, 139, 483-498.	3.8	11
26	Recruiting diversity where it exists: The Alabama Genomic Health Initiative. Journal of Genetic Counseling, 2020, 29, 471-478.	1.6	11
27	A phase II study of continuous oral mTOR inhibitor everolimus for recurrent, radiographic-progressive neurofibromatosis type 1â€"associated pediatric low-grade glioma: a Neurofibromatosis Clinical Trials Consortium study. Neuro-Oncology, 2020, 22, 1527-1535.	1.2	45
28	Affinity Purification of NF1 Proteinâ€"Protein Interactors Identifies Keratins and Neurofibromin Itself as Binding Partners. Genes, 2019, 10, 650.	2.4	11
29	The Genomic Medicine Integrative Research Framework: A Conceptual Framework for Conducting Genomic Medicine Research. American Journal of Human Genetics, 2019, 104, 1088-1096.	6.2	35
30	A YWHAZ Variant Associated With Cardiofaciocutaneous Syndrome Activates the RAF-ERK Pathway. Frontiers in Physiology, 2019, 10, 388.	2.8	23
31	Multiâ€Omics Profiling for NF1 Target Discovery in Neurofibromin (NF1) Deficient Cells. Proteomics, 2019, 19, e1800334.	2.2	5
32	Child Neurology: Spastic paraparesis and dystonia with a novel ADCY5 mutation. Neurology, 2019, 93, 510-514.	1.1	7
33	Reproducibility of cognitive endpoints in clinical trials: lessons from neurofibromatosis type 1. Annals of Clinical and Translational Neurology, 2019, 6, 2555-2565.	3.7	24
34	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€"phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
35	Neurofibromin (NF1) genetic variant structure-function analyses using a full-length mouse cDNA. Human Mutation, 2018, 39, 816-821.	2.5	15
36	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€"848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144

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37	Care of adults with neurofibromatosis type 1: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 671-682.	2.4	128
38	Breast cancer risk and germline genomic profiling of women with neurofibromatosis type 1 who developed breast cancer. Genes Chromosomes and Cancer, 2018, 57, 19-27.	2.8	22
39	Unusual presentation of hereditary leiomyomatosis mimicking neurofibromatosis. JAAD Case Reports, 2018, 4, 440-441.	0.8	0
40	Cutaneous neurofibromas. Neurology, 2018, 91, S5-S13.	1.1	79
41	Clinical trial design for cutaneous neurofibromas. Neurology, 2018, 91, S31-S37.	1.1	11
42	Germline and Somatic NF1 Alterations Are Linked to Increased HER2 Expression in Breast Cancer. Cancer Prevention Research, 2018, 11, 655-664.	1.5	4
43	Neurofibromatosis type 1. Nature Reviews Disease Primers, 2017, 3, 17004.	30.5	498
44	Patterns of Disease Monitoring and Treatment Among Patients With Tuberous Sclerosis Complex-related Angiomyolipomas. Urology, 2017, 104, 110-114.	1.0	7
45	Overview of Genetic Diagnosis in Cancer. Current Protocols in Human Genetics, 2017, 93, 10.1.1-10.1.9.	3.5	2
46	Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. Genetics in Medicine, 2017, 19, 249-255.	2.4	1,398
47	Clinical relevance of small copy-number variants in chromosomal microarray clinical testing. Genetics in Medicine, 2017, 19, 377-385.	2.4	24
48	Characterization and utilization of an international neurofibromatosis web-based, patientâ€“entered registry: An observational study. PLoS ONE, 2017, 12, e0178639.	2.5	24
49	Mice with missense and nonsense NF1 mutations display divergent phenotypes compared to NF1 patients. DMM Disease Models and Mechanisms, 2016, 9, 759-67.	2.4	23
50	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
51	Randomized placebo-controlled study of lovastatin in children with neurofibromatosis type 1. Neurology, 2016, 87, 2575-2584.	1.1	76
52	Overview of Clinical Cytogenetics. Current Protocols in Human Genetics, 2016, 89, 8.1.1-8.1.13.	3.5	7
53	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
54	Global implementation of genomic medicine: We are not alone. Science Translational Medicine, 2015, 7, 290ps13.	12.4	146

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55	Sirolimus for progressive neurofibromatosis type 1-associated plexiform neurofibromas: a Neurofibromatosis Clinical Trials Consortium phase II study. Neuro-Oncology, 2015, 17, 596-603.	1.2	118
56	How to know when physicians are ready for genomic medicine. Science Translational Medicine, 2015, 7, 287fs19.	12.4	54
57	Pushing the envelope in genomics education. Genetics in Medicine, 2015, 17, 857-858.	2.4	4
58	Spinal neurofibromatosis and phenotypic heterogeneity in NF1. Clinical Genetics, 2015, 87, 399-400.	2.0	6
59	Clinical response to bevacizumab in schwannomatosis. Neurology, 2014, 83, 1986-1987.	1.1	33
60	Sirolimus for nonâ€progressive NF1â€associated plexiform neurofibromas: An NF clinical trials consortium phase II study. Pediatric Blood and Cancer, 2014, 61, 982-986.	1.5	73
61	Phase 2 randomized, flexible crossover, double-blinded, placebo-controlled trial of the farnesyltransferase inhibitor tipifarnib in children and young adults with neurofibromatosis type 1 and progressive plexiform neurofibromas. Neuro-Oncology, 2014, 16, 707-718.	1.2	93
62	Germline loss-of-function mutations in LZTR1 predispose to an inherited disorder of multiple schwannomas. Nature Genetics, 2014, 46, 182-187.	21.4	242
63	Framework for development of physician competencies in genomic medicine: report of the Competencies Working Group of the Inter-Society Coordinating Committee for Physician Education in Genomics. Genetics in Medicine, 2014, 16, 804-809.	2.4	123
64	The effect of everolimus on renal angiomyolipoma in patients with tuberous sclerosis complex being treated for subependymal giant cell astrocytoma: subgroup results from the randomized, placebo-controlled, Phase 3 trial EXIST-1. Nephrology Dialysis Transplantation, 2014, 29, 1203-1210.	0.7	79
65	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genetics in Medicine, 2013, 15, 565-574.	2.4	2,186
66	New Approaches to Molecular Diagnosis. JAMA - Journal of the American Medical Association, 2013, 309, 1511.	7.4	116
67	Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 243-254.	2.1	1,185
68	Tuberous Sclerosis Complex Surveillance and Management: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 255-265.	2.1	693
69	Neurofibromatosis. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 333-340.	1.8	77
70	Implementing genomic medicine in the clinic: the future is here. Genetics in Medicine, 2013, 15, 258-267.	2.4	472
71	Optimizing biologically targeted clinical trials for neurofibromatosis. Expert Opinion on Investigational Drugs, 2013, 22, 443-462.	4.1	77
72	Phase I trial and pharmacokinetic study of sorafenib in children with neurofibromatosis type I and plexiform neurofibromas. Pediatric Blood and Cancer, 2013, 60, 396-401.	1.5	67

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73	Genomic Privacy in the Information Age. Clinical Chemistry, 2013, 59, 1148-1150.	3.2	4
74	Genomic medicine: educational challenges. Molecular Genetics & Genomic Medicine, 2013, 1, 119-122.	1.2	14
75	Integration of genomics into medical practice. Discovery Medicine, 2013, 16, 241-8.	0.5	9
76	Exploring concordance and discordance for return of incidental findings from clinical sequencing. Genetics in Medicine, 2012, 14, 405-410.	2.4	149
77	Genetic and Genomic Competency in Medical Practice. AMA Journal of Ethics, 2012, 14, 622-626.	0.7	5
78	Clinically relevant single gene or intragenic deletions encompassing critical neurodevelopmental genes in patients with developmental delay, mental retardation, and/or autism spectrum disorders. American Journal of Medical Genetics, Part A, 2011, 155, 2386-2396.	1.2	159
79	Genetics and genomics education: The next generation. Genetics in Medicine, 2011, 13, 201-202.	2.4	19
80	Future Health Applications of Genomics. American Journal of Preventive Medicine, 2010, 38, 556-565.	3.0	136
81	Consensus Recommendations to Accelerate Clinical Trials for Neurofibromatosis Type 2. Clinical Cancer Research, 2009, 15, 5032-5039.	7.0	74
82	Clinical and Mutational Spectrum of Neurofibromatosis Type 1â€“like Syndrome. JAMA - Journal of the American Medical Association, 2009, 302, 2111.	7.4	160
83	Neurofibromatosis Type 1 Revisited. Pediatrics, 2009, 123, 124-133.	2.1	562
84	Neurofibromatosis type 1. Journal of the American Academy of Dermatology, 2009, 61, 1-14.	1.2	443
85	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. American Journal of Human Genetics, 2008, 82, 712-722.	6.2	95
86	Statins, bone, and neurofibromatosis type 1. BMC Medicine, 2008, 6, 22.	5.5	6
87	Report of the Banbury Summit Meeting on the evolving role of the medical geneticist, February 12â€“14, 2006. Genetics in Medicine, 2008, 10, 502-507.	2.4	19
88	Genetic Testing in Cardiovascular Disease. Journal of the American College of Cardiology, 2007, 50, 727-737.	2.8	59
89	Pathophysiology of Neurofibromatosis Type 1. Annals of Internal Medicine, 2006, 144, 842.	3.9	121
90	Genetics training in the genomic era. Current Opinion in Pediatrics, 2005, 17, 747-750.	2.0	3

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91	Superficial Neurofibroma: A Lesion with Unique MRI Characteristics in Patients with Neurofibromatosis Type 1. American Journal of Roentgenology, 2005, 184, 962-968.	2.2	75
92	Case 13-2005. New England Journal of Medicine, 2005, 352, 1800-1808.	27.0	17
93	The case for strategic international alliances to harness nutritional genomics for public and personal health. British Journal of Nutrition, 2005, 94, 623-632.	2.3	137
94	The phakomatoses. Clinics in Dermatology, 2005, 23, 78-84.	1.6	25
95	Psychiatric Genetics: A Survey of Psychiatrists' Knowledge, Opinions, and Practice Patterns. Journal of Clinical Psychiatry, 2005, 66, 821-830.	2.2	67
96	Basic genetics. Primary Care - Clinics in Office Practice, 2004, 31, 461-478.	1.6	0
97	The phakomatoses. Neuroimaging Clinics of North America, 2004, 14, 139-148.	1.0	7
98	Integration of genetics into medical practice. Growth Hormone and IGF Research, 2004, 14, 146-149.	1.1	2
99	What's new in Neurogenetics? Amish microcephaly. European Journal of Paediatric Neurology, 2003, 7, 393-394.	1.6	1
100	Review Article : Clinical Features and Pathobiology of Neurofibromatosis 1. Journal of Child Neurology, 2002, 17, 573-577.	1.4	57
101	Cardiovascular disease in neurofibromatosis 1: Report of the NF1 Cardiovascular Task Force. Genetics in Medicine, 2002, 4, 105-111.	2.4	330
102	Effectiveness of sequencing connexin 26 (GJB2) in cases of familial or sporadic childhood deafness referred for molecular diagnostic testing. Genetics in Medicine, 2002, 4, 279-288.	2.4	56
103	American College of Medical Genetics Consensus Statement on Factor V Leiden Mutation Testing. Genetics in Medicine, 2001, 3, 139-148.	2.4	166
104	Diagnosis and management of neurofibromatosis type 1. Current Neurology and Neuroscience Reports, 2001, 1, 162-167.	4.2	45
105	Malignancy in Neurofibromatosis Type 1. Oncologist, 2000, 5, 477-485.	3.7	292
106	NF1 Microdeletion Syndrome: Refined FISH Characterization of Sporadic and Familial Deletions with Locus-Specific Probes. American Journal of Human Genetics, 2000, 66, 100-109.	6.2	105
107	Genetic Heterogeneity of Saethre-Chotzen Syndrome, Due to TWIST and FGFR Mutations. American Journal of Human Genetics, 1998, 62, 1370-1380.	6.2	202
108	Identification of a Novel Genetic Locus for Familial Cardiac Myxomas and Carney Complex. Circulation, 1998, 98, 2560-2566.	1.6	209

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109	Genetic Heterogeneity of Familial Atrial Myxoma Syndromes (Carney Complex). American Journal of Cardiology, 1997, 79, 994-995.	1.6	48
110	Patterns of Seizures Observed in Association with Neurofibromatosis 1. Epilepsia, 1993, 34, 616-620.	5.1	48
111	Consistent cytogenetic aberrations in hepatoblastoma: A common pathway of genetic alterations in embryonal liver and skeletal muscle malignancies?. Genes Chromosomes and Cancer, 1991, 3, 37-43.	2.8	62
112	Stage IV neuroblastoma in infants. Long-term survival. Cancer, 1991, 67, 1493-1497.	4.1	50
113	A Proposed Approach for Implementing Genomics-Based Screening Programs for Healthy Adults. NAM Perspectives, 0, , .	2.9	30