Walter E Kaufmann

List of Publications by Year
in descending order

86 papers	8,073 citations	71102 41 h-index	56724 83 g-index
88 all docs	88 docs citations	88 times ranked	8339 citing authors

#	Article	IF	Citations
1	Sleep problems in fragile X syndrome: Crossâ€sectional analysis of a large clinicâ€based cohort. American Journal of Medical Genetics, Part A, 2022, 188, 1029-1039.	1.2	18
2	The association between mosaicism type and cognitive and behavioral functioning among males with fragile X syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 858-866.	1.2	16
3	Anxiety-like behavior and anxiolytic treatment in the Rett syndrome natural history study. Journal of Neurodevelopmental Disorders, 2022, 14, 31.	3.1	15
4	Brain cell signaling abnormalities are detected in blood in a murine model of Fragile X syndrome and corrected by Sigmaâ€1 receptor agonist Blarcamesine. American Journal of Medical Genetics, Part A, 2022, 188, 2497-2500.	1.2	2
5	Cerebral visual impairment in CDKL5 deficiency disorder: vision as an outcome measure. Developmental Medicine and Child Neurology, 2021, 63, 1308-1315.	2.1	12
6	Effects of the sigma-1 receptor agonist blarcamesine in a murine model of fragile X syndrome: neurobehavioral phenotypes and receptor occupancy. Scientific Reports, 2021, 11, 17150.	3.3	9
7	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
8	Psychotropic Drug Treatment Patterns in Persons with Fragile X Syndrome. Journal of Child and Adolescent Psychopharmacology, 2021, 31, 659-669.	1.3	7
9	Seizures in Fragile X Syndrome: Associations and Longitudinal Analysis of a Large Clinic-Based Cohort. Frontiers in Pediatrics, 2021, 9, 736255.	1.9	21
10	Improving the Diagnosis of Autism Spectrum Disorder in Fragile X Syndrome by Adapting the Social Communication Questionnaire and the Social Responsiveness Scale-2. Journal of Autism and Developmental Disorders, 2020, 50, 3276-3295.	2.7	26
11	A Genotype-Phenotype Study of High-Resolution FMR1 Nucleic Acid and Protein Analyses in Fragile X Patients with Neurobehavioral Assessments. Brain Sciences, 2020, 10, 694.	2.3	54
12	Assessment of a Clinical Trial Metric for Rett Syndrome: Critical Analysis of the Rett Syndrome Behaviour Questionnaire. Pediatric Neurology, 2020, 111, 4.	2.1	10
13	Functional Network Mapping Reveals State-Dependent Response to IGF1 Treatment in Rett Syndrome. Brain Sciences, 2020, 10, 515.	2.3	5
14	Response to Placebo in Fragile X Syndrome Clinical Trials: An Initial Analysis. Brain Sciences, 2020, 10, 629.	2.3	21
15	Autism Spectrum Disorder Versus Autism Spectrum Disorders: Terminology, Concepts, and Clinical Practice. Frontiers in Psychiatry, 2020, 11, 484.	2.6	4
16	Metabolic Signatures Differentiate Rett Syndrome From Unaffected Siblings. Frontiers in Integrative Neuroscience, 2020, 14, 7.	2.1	24
17	Long QT interval in Rett syndrome: expanding the knowledge of a poorly understood phenomenon. Developmental Medicine and Child Neurology, 2020, 62, 775-775.	2.1	1
18	A precision medicine framework using artificial intelligence for the identification and confirmation of genomic biomarkers of response to an Alzheimer's disease therapy: Analysis of the blarcamesine (ANAVEX2â€73) Phase 2a clinical study. Alzheimer's and Dementia: Translational Research and Clinical Interventions, 2020, 6, e12013.	3.7	31

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19	ANAVEXÂ®2-73 (blarcamesine), a Sigma-1 receptor agonist, ameliorates neurologic impairments in a mouse model of Rett syndrome. Pharmacology Biochemistry and Behavior, 2019, 187, 172796.	2.9	26
20	Pharmacologic Interventions for Irritability, Aggression, Agitation and Self-Injurious Behavior in Fragile X Syndrome: An Initial Cross-Sectional Analysis. Journal of Autism and Developmental Disorders, 2019, 49, 4595-4602.	2.7	23
21	Hand stereotypies. Neurology, 2019, 92, e2594-e2603.	1.1	29
22	Severity Assessment in CDKL5 Deficiency Disorder. Pediatric Neurology, 2019, 97, 38-42.	2.1	43
23	Characterizing the phenotypic effect of Xq28 duplication size in MECP2 duplication syndrome. Clinical Genetics, 2019, 95, 575-581.	2.0	37
24	Behavioral profiles in Rett syndrome: Data from the natural history study. Brain and Development, 2019, 41, 123-134.	1.1	42
25	Towards a better diagnosis and treatment of Rett syndrome: a model synaptic disorder. Brain, 2019, 142, 239-248.	7.6	82
26	The course of awake breathing disturbances across the lifespan in Rett syndrome. Brain and Development, 2018, 40, 515-529.	1.1	60
27	Placeboâ€controlled crossover assessment of mecasermin for the treatment of Rett syndrome. Annals of Clinical and Translational Neurology, 2018, 5, 323-332.	3.7	58
28	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genetics in Medicine, 2017, 19, 13-19.	2.4	74
29	Defining Hand Stereotypies in Rett Syndrome: A Movement Disorders Perspective. Pediatric Neurology, 2017, 75, 91-95.	2.1	14
30	Longitudinal course of epilepsy in Rett syndrome and related disorders. Brain, 2017, 140, 306-318.	7.6	80
31	Autism Spectrum Disorder in Fragile X Syndrome: Cooccurring Conditions and Current Treatment. Pediatrics, 2017, 139, S194-S206.	2.1	186
32	FORWARD: A Registry and Longitudinal Clinical Database to Study Fragile X Syndrome. Pediatrics, 2017, 139, S183-S193.	2.1	39
33	Arbaclofen in fragile X syndrome: results of phase 3 trials. Journal of Neurodevelopmental Disorders, 2017, 9, 3.	3.1	135
34	Updated report on tools to measure outcomes of clinical trials in fragile X syndrome. Journal of Neurodevelopmental Disorders, 2017, 9, 14.	3.1	123
35	Adapting the Mullen Scales of Early Learning for a Standardized Measure of Development in Children With Rett Syndrome. Intellectual and Developmental Disabilities, 2017, 55, 419-431.	1.1	22
36	Caretaker Quality of Life in Rett Syndrome: Disorder Features and Psychological Predictors. Pediatric Neurology, 2016, 58, 67-74.	2.1	25

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37	Neurobiologically-based treatments in Rett syndrome: opportunities and challenges. Expert Opinion on Orphan Drugs, 2016, 4, 1043-1055.	0.8	31
38	Neurogenetics in Child Neurology: Redefining a Discipline in the Twenty-first Century. Current Neurology and Neuroscience Reports, 2016, 16, 103.	4.2	6
39	The Pediatric Imaging, Neurocognition, and Genetics (PING) Data Repository. NeuroImage, 2016, 124, 1149-1154.	4.2	251
40	Anxiety-like behavior in Rett syndrome: characteristics and assessment by anxiety scales. Journal of Neurodevelopmental Disorders, 2015, 7, 30.	3.1	71
41	Autism spectrum disorder in Phelan-McDermid syndrome: initial characterization and genotype-phenotype correlations. Orphanet Journal of Rare Diseases, 2015, 10, 105.	2.7	53
42	Visual evoked potentials detect cortical processing deficits in <scp>R</scp>ett syndrome. Annals of Neurology, 2015, 78, 775-786.	5.3	96
43	Age of Diagnosis in Rett Syndrome: Patterns of Recognition Among Diagnosticians and Risk Factors for Late Diagnosis. Pediatric Neurology, 2015, 52, 585-591.e2.	2.1	44
44	The Changing Face of Survival in Rett Syndrome andÂMECP2-Related Disorders. Pediatric Neurology, 2015, 53, 402-411.	2.1	79
45	Mutations in epilepsy and intellectual disability genes in patients with features of Rett syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2017-2025.	1.2	72
46	Methyl CpG binding protein 2 deficiency enhances expression of inflammatory cytokines by sustaining NF-Î²B signaling in myeloid derived cells. Journal of Neuroimmunology, 2015, 283, 23-29.	2.3	54
47	Family income, parental education and brain structure in children and adolescents. Nature Neuroscience, 2015, 18, 773-778.	14.8	979
48	Treatment of cardiac arrhythmias in Rett Syndrome with sodium channel blocking antiepileptic drugs. DMM Disease Models and Mechanisms, 2015, 8, 363-71.	2.4	15
49	Pubertal Development in Rett Syndrome Deviates From Typical Females. Pediatric Neurology, 2014, 51, 769-775.	2.1	32
50	Safety, pharmacokinetics, and preliminary assessment of efficacy of mecasermin (recombinant human) Tj ETQq0 0 0 rgBT /Overlock 10 T United States of America, 2014, 111, 4596-4601.	7.1	178
51	Developmental delay in Rett syndrome: data from the natural history study. Journal of Neurodevelopmental Disorders, 2014, 6, 20.	3.1	118
52	Small molecule glutaminase inhibitors block glutamate release from stimulated microglia. Biochemical and Biophysical Research Communications, 2014, 443, 32-36.	2.1	54
53	Methyl-CpG-binding protein 2(MECP2) mutation type is associated with disease severity in Rett syndrome. Journal of Medical Genetics, 2014, 51, 152-158.	3.2	246
54	Psychometric Study of the Aberrant Behavior Checklist in Fragile X Syndrome and Implications for Targeted Treatment. Journal of Autism and Developmental Disorders, 2012, 42, 1377-1392.	2.7	148

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55	Parent Report of Community Psychiatric Comorbid Diagnoses in Autism Spectrum Disorders. Autism Research & Treatment, 2011, 2011, 1-10.	0.5	71
56	What Can We Learn about Autism from Studying Fragile X Syndrome?. Developmental Neuroscience, 2011, 33, 379-394.	2.0	154
57	The behavioral phenotype of FMR1 mutations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 469-476.	1.6	98
58	Rett syndrome: Revised diagnostic criteria and nomenclature. Annals of Neurology, 2010, 68, 944-950.	5.3	1,045
59	Advances in the Treatment of Fragile X Syndrome. Pediatrics, 2009, 123, 378-390.	2.1	513
60	Autism spectrum disorder in fragile X syndrome: A longitudinal evaluation. American Journal of Medical Genetics, Part A, 2009, 149A, 1125-1137.	1.2	150
61	Brain metabolism in rett syndrome: Age, clinical, and genotype correlations. Annals of Neurology, 2009, 65, 90-97.	5.3	43
62	The diagnosis of autism in a female: could it be Rett syndrome?. European Journal of Pediatrics, 2008, 167, 661-669.	2.7	58
63	Elongation Factor 2 and Fragile X Mental Retardation Protein Control the Dynamic Translation of Arc/Arg3.1 Essential for mGluR-LTD. Neuron, 2008, 59, 70-83.	8.1	471
64	MeCP2 expression and function during brain development: implications for Rett syndrome's pathogenesis and clinical evolution. Brain and Development, 2005, 27, S77-S87.	1.1	90
65	Social behavior profile in young males with fragile X syndrome: Characteristics and specificity. American Journal of Medical Genetics Part A, 2004, 126A, 9-17.	2.4	131
66	Autism spectrum disorder in fragile X syndrome: Communication, social interaction, and specific behaviors. American Journal of Medical Genetics Part A, 2004, 129A, 225-234.	2.4	359
67	Specificity of Cerebellar Vermian Abnormalities in Autism: A Quantitative Magnetic Resonance Imaging Study. Journal of Child Neurology, 2003, 18, 463-470.	1.4	153
68	Molecular phenotype of Fragile X syndrome: FMRP, FXRPs, and protein targets. Microscopy Research and Technique, 2002, 57, 135-144.	2.2	19
69	Ectopic Cerebellum Presenting as a Suprasellar Mass in Infancy: Implications for Cerebellar Development. Pediatric and Developmental Pathology, 2001, 4, 89-93.	1.0	15
70	Diffusion tensor imaging of the developing mouse brain. Magnetic Resonance in Medicine, 2001, 46, 18-23.	3.0	237
71	Annexin-1 is abnormally expressed in Fragile X syndrome: Two-dimensional electrophoresis study in lymphocytes. American Journal of Medical Genetics Part A, 2001, 103, 81-90.	2.4	23
72	In vivo visualization of human neural pathways by magnetic resonance imaging. Annals of Neurology, 2000, 47, 412-414.	5.3	109

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73	Thalamic involvement in neurofibromatosis type 1: Evaluation with proton magnetic resonance spectroscopic imaging. Annals of Neurology, 2000, 47, 477-484.	5.3	34
74	In vivo visualization of human neural pathways by magnetic resonance imaging. , 2000, 47, 412.		1
75	Thalamic involvement in neurofibromatosis type 1: Evaluation with proton magnetic resonance spectroscopic imaging. Annals of Neurology, 2000, 47, 477-484.	5.3	3
76	Opiate and Cocaine Exposed Newborns: Growth Outcomes. Journal of Child and Adolescent Substance Abuse, 1999, 8, 1-16.	0.5	5
77	FMR1 gene expression in olfactory neuroblasts from two males with fragile X syndrome. , 1999, 82, 25-30.		30
78	Molecular and cellular genetics of fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 88, 11-24.	2.4	78
79	Genotype, molecular phenotype, and cognitive phenotype: Correlations in fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 83, 286-295.	2.4	120
80	Neural activity and immediate early gene expression in the cerebral cortex. Mental Retardation and Developmental Disabilities Research Reviews, 1999, 5, 41-50.	3.6	8
81	Sequential neuromotor examination in children with intrauterine cocaine/ polydrug exposure. Developmental Medicine and Child Neurology, 1999, 41, 240-246.	2.1	0
82	Genotype, molecular phenotype, and cognitive phenotype: Correlations in fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 83, 286-295.	2.4	3
83	Sequential Neuromotor Examination of Children with Intrauterine Drug Exposurea. Annals of the New York Academy of Sciences, 1998, 846, 362-364.	3.8	1
84	Neuroanatomical and neurocognitive differences in a pair of monozygous twins discordant for strictly defined autism. Annals of Neurology, 1998, 43, 782-791.	5.3	67
85	Immunoblotting patterns of cytoskeletal dendritic protein expression in human neocortex. Molecular and Chemical Neuropathology, 1997, 31, 235-244.	1.0	19
86	Treatment of Epilepsy with Multiple Subpial Transections: An Acute Histologic Analysis in Human Subjects. Epilepsia, 1996, 37, 342-352.	5.1	43