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	Rett syndrome: Revised diagnostic criteria and nomenclature. Annals of Neurology, 2010, 68, 944-950.	5.3	1,045
2	Family income, parental education and brain structure in children and adolescents. Nature Neuroscience, 2015, 18, 773-778.	14.8	979
3	Advances in the Treatment of Fragile X Syndrome. Pediatrics, 2009, 123, 378-390.	2.1	513
4	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
5	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
6	The Pediatric Imaging, Neurocognition, and Genetics (PING) Data Repository. NeuroImage, 2016, 124, 1149-1154.	4.2	251
7	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
8	Diffusion tensor imaging of the developing mouse brain. Magnetic Resonance in Medicine, 2001, 46, 18-23.	3.0	237
9	Autism Spectrum Disorder in Fragile X Syndrome: Cooccurring Conditions and Current Treatment. Pediatrics, 2017, 139, S194-S206.	2.1	186
10	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
11	What Can We Learn about Autism from Studying Fragile X Syndrome?. Developmental Neuroscience, 2011, 33, 379-394.	2.0	154
12	Specificity of Cerebellar Vermian Abnormalities in Autism: A Quantitative Magnetic Resonance Imaging Study. Journal of Child Neurology, 2003, 18, 463-470.	1.4	153
13	Autism spectrum disorder in fragile X syndrome: A longitudinal evaluation. American Journal of Medical Genetics, Part A, 2009, 149A, 1125-1137.	1.2	150
14	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
15	Arbaclofen in fragile X syndrome: results of phase 3 trials. Journal of Neurodevelopmental Disorders, 2017, 9, 3.	3.1	135
16	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
17	Updated report on tools to measure outcomes of clinical trials in fragile X syndrome. Journal of Neurodevelopmental Disorders, 2017, 9, 14.	3.1	123
18	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

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19	Developmental delay in Rett syndrome: data from the natural history study. Journal of Neurodevelopmental Disorders, 2014, 6, 20.	3.1	118
20	In vivo visualization of human neural pathways by magnetic resonance imaging. Annals of Neurology, 2000, 47, 412-414.	5.3	109
21	The behavioral phenotype of FMR1 mutations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 469-476.	1.6	98
22	Visual evoked potentials detect cortical processing deficits in <scp>R</scp>ett syndrome. Annals of Neurology, 2015, 78, 775-786.	5.3	96
23	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
24	Towards a better diagnosis and treatment of Rett syndrome: a model synaptic disorder. Brain, 2019, 142, 239-248.	7.6	82
25	Longitudinal course of epilepsy in Rett syndrome and related disorders. Brain, 2017, 140, 306-318.	7.6	80
26	The Changing Face of Survival in Rett Syndrome andÂMECP2-Related Disorders. Pediatric Neurology, 2015, 53, 402-411.	2.1	79
27	Molecular and cellular genetics of fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 88, 11-24.	2.4	78
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	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
30	Parent Report of Community Psychiatric Comorbid Diagnoses in Autism Spectrum Disorders. Autism Research & Treatment, 2011, 2011, 1-10.	0.5	71
31	Anxiety-like behavior in Rett syndrome: characteristics and assessment by anxiety scales. Journal of Neurodevelopmental Disorders, 2015, 7, 30.	3.1	71
32	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
33	The course of awake breathing disturbances across the lifespan in Rett syndrome. Brain and Development, 2018, 40, 515-529.	1.1	60
34	The diagnosis of autism in a female: could it be Rett syndrome?. European Journal of Pediatrics, 2008, 167, 661-669.	2.7	58
35	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
36	Small molecule glutaminase inhibitors block glutamate release from stimulated microglia. Biochemical and Biophysical Research Communications, 2014, 443, 32-36.	2.1	54

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37	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
38	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
39	Autism spectrum disorder in Phelan-McDermid syndrome: initial characterization and genotype-phenotype correlations. Orphanet Journal of Rare Diseases, 2015, 10, 105.	2.7	53
40	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
41	Treatment of Epilepsy with Multiple Subpial Transections: An Acute Histologic Analysis in Human Subjects. Epilepsia, 1996, 37, 342-352.	5.1	43
42	Brain metabolism in rett syndrome: Age, clinical, and genotype correlations. Annals of Neurology, 2009, 65, 90-97.	5.3	43
43	Severity Assessment in CDKL5 Deficiency Disorder. Pediatric Neurology, 2019, 97, 38-42.	2.1	43
44	Behavioral profiles in Rett syndrome: Data from the natural history study. Brain and Development, 2019, 41, 123-134.	1.1	42
45	FORWARD: A Registry and Longitudinal Clinical Database to Study Fragile X Syndrome. Pediatrics, 2017, 139, S183-S193.	2.1	39
46	Characterizing the phenotypic effect of Xq28 duplication size in MECP2 duplication syndrome. Clinical Genetics, 2019, 95, 575-581.	2.0	37
47	Thalamic involvement in neurofibromatosis type 1: Evaluation with proton magnetic resonance spectroscopic imaging. Annals of Neurology, 2000, 47, 477-484.	5.3	34
48	Pubertal Development in Rett Syndrome Deviates From Typical Females. Pediatric Neurology, 2014, 51, 769-775.	2.1	32
49	Neurobiologically-based treatments in Rett syndrome: opportunities and challenges. Expert Opinion on Orphan Drugs, 2016, 4, 1043-1055.	0.8	31
50	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
51	FMR1 gene expression in olfactory neuroblasts from two males with fragile X syndrome. , 1999, 82, 25-30.		30
52	Hand stereotypies. Neurology, 2019, 92, e2594-e2603.	1.1	29
53	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
54	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

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55	Caretaker Quality of Life in Rett Syndrome: Disorder Features and Psychological Predictors. Pediatric Neurology, 2016, 58, 67-74.	2.1	25
56	Metabolic Signatures Differentiate Rett Syndrome From Unaffected Siblings. Frontiers in Integrative Neuroscience, 2020, 14, 7.	2.1	24
57	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
58	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
59	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
60	Response to Placebo in Fragile X Syndrome Clinical Trials: An Initial Analysis. Brain Sciences, 2020, 10, 629.	2.3	21
61	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
62	Seizures in Fragile X Syndrome: Associations and Longitudinal Analysis of a Large Clinic-Based Cohort. Frontiers in Pediatrics, 2021, 9, 736255.	1.9	21
63	Immunoblotting patterns of cytoskeletal dendritic protein expression in human neocortex. Molecular and Chemical Neuropathology, 1997, 31, 235-244.	1.0	19
64	Molecular phenotype of Fragile X syndrome: FMRP, FXRPs, and protein targets. Microscopy Research and Technique, 2002, 57, 135-144.	2.2	19
65	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
66	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
67	Ectopic Cerebellum Presenting as a Suprasellar Mass in Infancy: Implications for Cerebellar Development. Pediatric and Developmental Pathology, 2001, 4, 89-93.	1.0	15
68	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
69	Anxiety-like behavior and anxiolytic treatment in the Rett syndrome natural history study. Journal of Neurodevelopmental Disorders, 2022, 14, 31.	3.1	15
70	Defining Hand Stereotypies in Rett Syndrome: A Movement Disorders Perspective. Pediatric Neurology, 2017, 75, 91-95.	2.1	14
71	Cerebral visual impairment in CDKL5 deficiency disorder: vision as an outcome measure. Developmental Medicine and Child Neurology, 2021, 63, 1308-1315.	2.1	12
72	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

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73	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
74	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
75	Psychotropic Drug Treatment Patterns in Persons with Fragile X Syndrome. Journal of Child and Adolescent Psychopharmacology, 2021, 31, 659-669.	1.3	7
76	Neurogenetics in Child Neurology: Redefining a Discipline in the Twenty-first Century. Current Neurology and Neuroscience Reports, 2016, 16, 103.	4.2	6
77	Opiate and Cocaine Exposed Newborns: Growth Outcomes. Journal of Child and Adolescent Substance Abuse, 1999, 8, 1-16.	0.5	5
78	Functional Network Mapping Reveals State-Dependent Response to IGF1 Treatment in Rett Syndrome. Brain Sciences, 2020, 10, 515.	2.3	5
79	Autism Spectrum Disorder Versus Autism Spectrum Disorders: Terminology, Concepts, and Clinical Practice. Frontiers in Psychiatry, 2020, 11, 484.	2.6	4
80	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
81	Thalamic involvement in neurofibromatosis type 1: Evaluation with proton magnetic resonance spectroscopic imaging. Annals of Neurology, 2000, 47, 477-484.	5.3	3
82	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
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	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
85	In vivo visualization of human neural pathways by magnetic resonance imaging. , 2000, 47, 412.		1
86	Sequential neuromotor examination in children with intrauterine cocaine/ polydrug exposure. Developmental Medicine and Child Neurology, 1999, 41, 240-246.	2.1	0