Elizabeth M Berry-Kravis

List of Publications by Year
in descending order

170 papers	10,538 citations	53939 47 h-index	42259 96 g-index
180 all docs	180 docs citations	180 times ranked	7654 citing authors

#	Article	IF	Citations
1	Characterization of the Cerebrospinal Fluid Proteome in Patients with Fragile X-Associated Tremor/Ataxia Syndrome. Cerebellum, 2022, 21, 86-98.	1.4	5
2	Strong evidence for genotypeâ€“phenotype correlations in Phelan-McDermid syndrome: results from the developmental synaptopathies consortium. Human Molecular Genetics, 2022, 31, 625-637.	1.4	32
3	Design and outcome measures of LAVENDER, a phase 3 study of trofinetide for Rett syndrome. Contemporary Clinical Trials, 2022, 114, 106704.	0.8	30
4	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.	0.7	18
5	Fragile X Syndrome: Supportive Treatment, Unmet Needs, and Paths to Novel Interventions and Disease-Targeted Therapies. American Journal on Intellectual and Developmental Disabilities, 2022, 127, 90-94.	0.8	2
6	Fragile X Mental Retardation Protein and Cerebral Expression of Metabotropic Glutamate Receptor Subtype 5 in Men with Fragile X Syndrome: A Pilot Study. Brain Sciences, 2022, 12, 314.	1.1	7
7	Independent evaluation of the harvard automated processing pipeline for Electroencephalography 1.0 using multi-site EEG data from children with Fragile X Syndrome. Journal of Neuroscience Methods, 2022, 371, 109501.	1.3	0
8	Verbal inhibition declines among older women with high FMR1 premutation expansions: A prospective study. Brain and Cognition, 2022, 159, 105851.	0.8	3
9	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.	0.7	16
10	Analysis of a Repetitive Language Coding System: Comparisons between Fragile X Syndrome, Autism, and Down Syndrome. Brain Sciences, 2022, 12, 575.	1.1	4
11	Gene-based therapeutics for rare genetic neurodevelopmental psychiatric disorders. Molecular Therapy, 2022, 30, 2416-2428.	3.7	9
12	Disease-Targeted Treatment Translation in Fragile X Syndrome as a Model for Neurodevelopmental Disorders. Journal of Child Neurology, 2022, 37, 797-812.	0.7	6
13	The Effects of Dual Task Cognitive Interference and Fast-Paced Walking on Gait, Turns, and Falls in Men and Women with FXTAS. Cerebellum, 2021, 20, 212-221.	1.4	8
14	A Unique Visual Attention Profile Associated With the FMR1 Premutation. Frontiers in Genetics, 2021, 12, 591211.	1.1	1
15	Diagnostic profile of the AmplideX Fragile X Dx and Carrier Screen Kit for diagnosis and screening of fragile X syndrome and other FMR1-related disorders. Expert Review of Molecular Diagnostics, 2021, 21, 255-267.	1.5	3
16	Shifted phase of EEG cross-frequency coupling in individuals with Phelan-McDermid syndrome. Molecular Autism, 2021, 12, 29.	2.6	9
17	Niemann-Pick Disease, Type C: Diagnosis, Management and Disease-Targeted Therapies in Development. Seminars in Pediatric Neurology, 2021, 37, 100879.	1.0	9
18	Spoken language outcome measures for treatment studies in Down syndrome: feasibility, practice effects, test-retest reliability, and construct validity of variables generated from expressive language sampling. Journal of Neurodevelopmental Disorders, 2021, 13, 13.	1.5	18

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19	Cell-type-specific profiling of human cellular models of fragile X syndrome reveal PI3K-dependent defects in translation and neurogenesis. Cell Reports, 2021, 35, 108991.	2.9	36
20	Inhibition of phosphodiesterase-4D in adults with fragile X syndrome: a randomized, placebo-controlled, phase 2 clinical trial. Nature Medicine, 2021, 27, 862-870.	15.2	57
21	Paired-like homeobox gene (PHOX2B) nonpolyalanine repeat expansion mutations (NPARMs): genotypeâ€“phenotype correlation in congenital central hypoventilation syndrome (CCHS). Genetics in Medicine, 2021, 23, 1656-1663.	1.1	16
22	Stress and genetics influence hair cortisol in FMR1 premutation carrier mothers of children with fragile X syndrome. Psychoneuroendocrinology, 2021, 129, 105266.	1.3	3
23	Is FMR1 CGG Repeat Number Polymorphism Associated With Phenotypic Variation in the General Population? Report From a Cohort of 5,499 Adults. Frontiers in Psychiatry, 2021, 12, 727085.	1.3	2
24	A novel measure of matching categories for early development: Item creation and pilot feasibility study. Research in Developmental Disabilities, 2021, 115, 103993.	1.2	2
25	The Phenotypic Profile Associated With the FMR1 Premutation in Women: An Investigation of Clinical-Behavioral, Social-Cognitive, and Executive Abilities. Frontiers in Psychiatry, 2021, 12, 718485.	1.3	8
26	Identifying susceptibility genes for primary ovarian insufficiency on the high-risk genetic background of a fragile X premutation. Fertility and Sterility, 2021, 116, 843-854.	0.5	5
27	Symptoms of Autism Spectrum Disorder in Individuals with Down Syndrome. Brain Sciences, 2021, 11, 1278.	1.1	9
28	Normative database of postural sway measures using inertial sensors in typically developing children and young adults. Gait and Posture, 2021, 90, 112-119.	0.6	8
29	Gaboxadol in Fragile X Syndrome: A 12-Week Randomized, Double-Blind, Parallel-Group, Phase 2a Study. Frontiers in Pharmacology, 2021, 12, 757825.	1.6	9
30	Parent-reported measure of repetitive behavior in Phelan-McDermid syndrome. Journal of Neurodevelopmental Disorders, 2021, 13, 53.	1.5	6
31	International consensus on clinical severity scale use in evaluating Niemannâ€“Pick disease Type C in paediatric and adult patients: results from a Delphi Study. Orphanet Journal of Rare Diseases, 2021, 16, 482.	1.2	8
32	Consistently high agreement between independent raters of Niemann-Pick type C1 clinical severity scale in phase 2/3 trial. Pediatric Neurology, 2021, 127, 32-38.	1.0	1
33	Psychotropic Drug Treatment Patterns in Persons with Fragile X Syndrome. Journal of Child and Adolescent Psychopharmacology, 2021, 31, 659-669.	0.7	7
34	The CD22-IGF2R interaction is a therapeutic target for microglial lysosome dysfunction in Niemann-Pick type C. Science Translational Medicine, 2021, 13, eabg2919.	5.8	18
35	Seizures in Fragile X Syndrome: Associations and Longitudinal Analysis of a Large Clinic-Based Cohort. Frontiers in Pediatrics, 2021, 9, 736255.	0.9	21
36	De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. Npj Genomic Medicine, 2021, 6, 104.	1.7	7

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37	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.	1.7	26
38	Inhibition deficits are modulated by age and CGG repeat length in carriers of the FMR1 premutation allele who are mothers of children with fragile X syndrome. Brain and Cognition, 2020, 139, 105511.	0.8	22
39	Physiological regulation and social-emotional processing in female carriers of the FMR1 premutation. Physiology and Behavior, 2020, 214, 112746.	1.0	7
40	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.	1.1	54
41	Neurodevelopmental Characterization of Young Children Diagnosed with Niemann-Pick Disease, Type C1. Journal of Developmental and Behavioral Pediatrics, 2020, 41, 388-396.	0.6	6
42	Application of N-palmitoyl-O-phosphocholineserine for diagnosis and assessment of response to treatment in Niemann-Pick type C disease. Molecular Genetics and Metabolism, 2020, 129, 292-302.	0.5	24
43	Optimization of Protocols for Detection of De Novo Protein Synthesis in Whole Blood Samples via Azideâ€“Alkyne Cycloaddition. Journal of Proteome Research, 2020, 19, 3856-3866.	1.8	4
44	Response to Placebo in Fragile X Syndrome Clinical Trials: An Initial Analysis. Brain Sciences, 2020, 10, 629.	1.1	21
45	Reduced Expression of Cerebral Metabotropic Glutamate Receptor Subtype 5 in Men with Fragile X Syndrome. Brain Sciences, 2020, 10, 899.	1.1	15
46	TECPR2 mutationâ€“associated respiratory dysregulation: more than central apnea. Journal of Clinical Sleep Medicine, 2020, 16, 977-982.	1.4	8
47	Soy-Based Infant Formula is Associated with an Increased Prevalence of Comorbidities in Fragile X Syndrome. Nutrients, 2020, 12, 3136.	1.7	13
48	Intracerebroventricular Cerliponase Alfa for Neuronal Ceroid Lipofuscinosis Type 2 Disease: Clinical Practice Considerations From US Clinics. Pediatric Neurology, 2020, 110, 64-70.	1.0	12
49	Psychometric Study of the Social Responsiveness Scale in Phelanâ€“McDermid Syndrome. Autism Research, 2020, 13, 1383-1396.	2.1	14
50	Validation of the NIH Toolbox Cognitive Battery in intellectual disability. Neurology, 2020, 94, e1229-e1240.	1.5	44
51	Fragile X Gray Zone Alleles Are Associated With Signs of Parkinsonism and Earlier Death. Movement Disorders, 2020, 35, 1448-1456.	2.2	16
52	Expressive language sampling as a source of outcome measures for treatment studies in fragile X syndrome: feasibility, practice effects, test-retest reliability, and construct validity. Journal of Neurodevelopmental Disorders, 2020, 12, 10.	1.5	32
53	Tremorography in fragile X-associated tremor/ataxia syndrome, Parkinson's disease and essential tremor. Clinical Parkinsonism & Related Disorders, 2020, 3, 100040.	0.5	3
54	Diffusion Tensor Imaging Abnormalities in the Uncinate Fasciculus and Inferior Longitudinal Fasciculus in Phelan-McDermid Syndrome. Pediatric Neurology, 2020, 106, 24-31.	1.0	9

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55	Anesthetic management of pediatric patients with Niemannâ€Pick disease type C for intrathecal 2â€hydroxypropylâ€Îˆâ€cyclodextrin injection. Paediatric Anaesthesia, 2020, 30, 766-772.	0.6	3
56	Delineating Repetitive Behavior Profiles across the Lifespan in Fragile X Syndrome. Brain Sciences, 2020, 10, 239.	1.1	16
57	Language across the Lifespan in Fragile X Syndrome: Characteristics and Considerations for Assessment. Brain Sciences, 2020, 10, 212.	1.1	8
58	Examination of Correlates to Health-Related Quality of Life in Individuals with Fragile X Syndrome. Brain Sciences, 2020, 10, 213.	1.1	6
59	Normative database of spatiotemporal gait parameters using inertial sensors in typically developing children and young adults. Gait and Posture, 2020, 80, 206-213.	0.6	35
60	A Double-Blind, Randomized, Placebo-Controlled Clinical Study of Trofinetide in the Treatment of Fragile X Syndrome. Pediatric Neurology, 2020, 110, 30-41.	1.0	50
61	Application of a glycinated bile acid biomarker for diagnosis and assessment of response to treatment in Niemann-pick disease type C1. Molecular Genetics and Metabolism, 2020, 131, 405-417.	0.5	11
62	Development of Neural Response to Novel Sounds in Fragile X Syndrome: Potential Biomarkers. American Journal on Intellectual and Developmental Disabilities, 2020, 125, 449-464.	0.8	13
63	Language processing skills linked to FMR1 variation: A study of gaze-language coordination during rapid automatized naming among women with the FMR1 premutation. PLoS ONE, 2019, 14, e0219924.	1.1	11
64	Data-driven phenotype discovery of FMR1 premutation carriers in a population-based sample. Science Advances, 2019, 5, eaaw7195.	4.7	33
65	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.	1.7	23
66	Effects of mavoglurant on visual attention and pupil reactivity while viewing photographs of faces in Fragile X Syndrome. PLoS ONE, 2019, 14, e0209984.	1.1	22
67	Congenital central hypoventilation syndrome: Severe disease caused by coâ€occurrence of two PHOX2B variants inherited separately from asymptomatic family members. American Journal of Medical Genetics, Part A, 2019, 179, 503-506.	0.7	10
68	N-acyl-O-phosphocholineserines: structures of a novel class of lipids that are biomarkers for Niemann-Pick C1 disease. Journal of Lipid Research, 2019, 60, 1410-1424.	2.0	31
69	Preventive care services and health behaviors in children with fragile X syndrome. Disability and Health Journal, 2019, 12, 564-573.	1.6	4
70	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	1.1	38
71	Expression and Characterization of Human Fragile X Mental Retardation Protein Isoforms and Interacting Proteins in Human Cells. Proteomics Insights, 2019, 10, 117864181882526.	2.0	5
72	Altered steady state and activity-dependent de novo protein expression in fragile X syndrome. Nature Communications, 2019, 10, 1710.	5.8	27

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73	Double-blind, randomized, placebo-controlled study of trofinetide in pediatric Rett syndrome. Neurology, 2019, 92, e1912-e1925.	1.5	106
74	Voice of People with Fragile X Syndrome and Their Families: Reports from a Survey on Treatment Priorities. Brain Sciences, 2019, 9, 18.	1.1	30
75	Vocabulary comprehension in adults with fragile X syndrome (FXS). Journal of Neurodevelopmental Disorders, 2019, 11, 25.	1.5	5
76	Emerging pharmacological therapies in fragile X syndrome and autism. Current Opinion in Neurology, 2019, 32, 635-640.	1.8	23
77	Toilet Training in Fragile X Syndrome. Journal of Developmental and Behavioral Pediatrics, 2019, 40, 751-761.	0.6	6
78	Neuropsychiatric decompensation in adolescents and adults with Phelan-McDermid syndrome: a systematic review of the literature. Molecular Autism, 2019, 10, 50.	2.6	47
79	Diagnosis of niemann-pick C1 by measurement of bile acid biomarkers in archived newborn dried blood spots. Molecular Genetics and Metabolism, 2019, 126, 183-187.	0.5	21
80	Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome. Pediatric Neurology, 2019, 90, 37-43.	1.0	19
81	Fragile X syndrome and fragile X-associated tremor ataxia syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 377-391.	1.0	26
82	Long-Term Treatment of Niemann-Pick Type C1 Disease With Intrathecal 2-Hydroxypropyl-Î²-Cyclodextrin. Pediatric Neurology, 2018, 80, 24-34.	1.0	60
83	Effect of the mGluR5-NAM Basimglurant on Behavior in Adolescents and Adults with Fragile X Syndrome in a Randomized, Double-Blind, Placebo-Controlled Trial: FragXis Phase 2 Results. Neuropsychopharmacology, 2018, 43, 503-512.	2.8	102
84	Drug development for neurodevelopmental disorders: lessons learned from fragile X syndrome. Nature Reviews Drug Discovery, 2018, 17, 280-299.	21.5	247
85	Clinical Development of Targeted Fragile X Syndrome Treatments: An Industry Perspective. Brain Sciences, 2018, 8, 214.	1.1	29
86	Best Practices in Fragile X Syndrome Treatment Development. Brain Sciences, 2018, 8, 224.	1.1	37
87	Cognitive function impacts gait, functional mobility and falls in fragile X-associated tremor/ataxia syndrome. Gait and Posture, 2018, 66, 288-293.	0.6	24
88	ASFMR1 splice variant. Neurology: Genetics, 2018, 4, e246.	0.9	13
89	Health Profiles of Mosaic Versus Non-mosaic FMR1 Premutation Carrier Mothers of Children With Fragile X Syndrome. Frontiers in Genetics, 2018, 9, 173.	1.1	18
90	Utility of the Hebbâ€“Williams Maze Paradigm for Translational Research in Fragile X Syndrome: A Direct Comparison of Mice and Humans. Frontiers in Molecular Neuroscience, 2018, 11, 99.	1.4	9

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91	Fragile X Newborn Screening: Lessons Learned From a Multisite Screening Study. Pediatrics, 2017, 139, S216-S225.	1.0	22
92	Fragile X syndrome. Nature Reviews Disease Primers, 2017, 3, 17065.	18.1	490
93	Mutations in GPAA1 , Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. American Journal of Human Genetics, 2017, 101, 856-865.	2.6	49
94	Autism Spectrum Disorder in Fragile X Syndrome: Cooccurring Conditions and Current Treatment. Pediatrics, 2017, 139, S194-S206.	1.0	186
95	The Future of Fragile X Syndrome: CDC Stakeholder Meeting Summary. Pediatrics, 2017, 139, S147-S152.	1.0	14
96	FORWARD: A Registry and Longitudinal Clinical Database to Study Fragile X Syndrome. Pediatrics, 2017, 139, S183-S193.	1.0	39
97	Intrathecal 2-hydroxypropyl-Î²-cyclodextrin decreases neurological disease progression in Niemann-Pick disease, type C1: a non-randomised, open-label, phase 1â€“2 trial. Lancet, The, 2017, 390, 1758-1768.	6.3	275
98	Arbaclofen in fragile X syndrome: results of phase 3 trials. Journal of Neurodevelopmental Disorders, 2017, 9, 3.	1.5	135
99	Fragile X targeted pharmacotherapy: lessons learned and future directions. Journal of Neurodevelopmental Disorders, 2017, 9, 7.	1.5	99
100	Updated report on tools to measure outcomes of clinical trials in fragile X syndrome. Journal of Neurodevelopmental Disorders, 2017, 9, 14.	1.5	123
101	Clinical Phenotype of Adult Fragile X Gray Zone Allele Carriers: a Case Series. Cerebellum, 2016, 15, 623-631.	1.4	20
102	A developmental, longitudinal investigation of autism phenotypic profiles in fragile X syndrome. Journal of Neurodevelopmental Disorders, 2016, 8, 47.	1.5	52
103	A methylation PCR method determines FMR1 activation ratios and differentiates premutation allele mosaicism in carrier siblings. Clinical Epigenetics, 2016, 8, 130.	1.8	14
104	Congenital Central Hypoventilation Syndrome. Chest, 2016, 149, 809-815.	0.4	44
105	Cliniciansâ€™ experiences with the fragile X clinical and research consortium. American Journal of Medical Genetics, Part A, 2016, 170, 3138-3143.	0.7	6
106	Treatment of Neurogenetic Developmental Conditions: From 2016 into the Future. Pediatric Neurology, 2016, 65, 1-13.	1.0	34
107	Developmental profiles of infants with an FMR1 premutation. Journal of Neurodevelopmental Disorders, 2016, 8, 40.	1.5	21
108	Importance of a specialty clinic for individuals with fragile X syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3144-3149.	0.7	7

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109	Development of a bile acidâ€“based newborn screen for Niemann-Pick disease type C. Science Translational Medicine, 2016, 8, 337ra63.	5.8	89
110	The NIH Toolbox Cognitive Battery for intellectual disabilities: three preliminary studies and future directions. Journal of Neurodevelopmental Disorders, 2016, 8, 35.	1.5	96
111	Cognitive trajectories in rare neurogenetic diseases: minding the gaps and filling the potholes. Developmental Medicine and Child Neurology, 2016, 58, 221-222.	1.1	4
112	Mavoglurant in fragile X syndrome: Results of two randomized, double-blind, placebo-controlled trials. Science Translational Medicine, 2016, 8, 321ra5.	5.8	210
113	X-inactivation in the clinical phenotype of fragile X premutation carrier sisters. Neurology: Genetics, 2016, 2, e45.	0.9	25
114	Absence of mutations in HCRT , HCRTR1 and HCRTR2 in patients with ROHHAD. Respiratory Physiology and Neurobiology, 2016, 221, 59-63.	0.7	19
115	Gait and Functional Mobility Deficits in Fragile X-Associated Tremor/Ataxia Syndrome. Cerebellum, 2016, 15, 475-482.	1.4	27
116	Motor stereotypies in fragile X syndrome. Journal of Pediatric Neurology, 2015, 12, 029-034.	0.0	1
117	Rapid-Onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD): exome sequencing of trios, monozygotic twins and tumours. Orphanet Journal of Rare Diseases, 2015, 10, 103.	1.2	45
118	Activation of Peroxisome Proliferator-activated Receptor Î± Induces Lysosomal Biogenesis in Brain Cells. Journal of Biological Chemistry, 2015, 290, 10309-10324.	1.6	108
119	Therapeutic Strategies in Fragile X Syndrome: From Bench to Bedside and Back. Neurotherapeutics, 2015, 12, 584-608.	2.1	88
120	White matter disease and cognitive impairment in FMR1 premutation carriers. Neurology, 2015, 84, 2146-2152.	1.5	52
121	Cholesterol levels in Fragile X syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 379-384.	0.7	51
122	Maternal Consequences of the Detection of Fragile X Carriers in Newborn Screening. Pediatrics, 2015, 136, e433-e440.	1.0	14
123	Characterization and Early Detection of Balance Deficits in Fragile X Premutation Carriers With and Without Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). Cerebellum, 2015, 14, 650-662.	1.4	31
124	Distribution of AGG interruption patterns within nine world populations. Intractable and Rare Diseases Research, 2014, 3, 153-161.	0.3	20
125	New observations in the fragile X-associated tremor/ataxia syndrome (FXTAS) phenotype. Frontiers in Genetics, 2014, 5, 365.	1.1	29
126	A Pilot Study of Social Information Processing Skills in Girls With Fragile X Syndrome. Journal of Mental Health Research in Intellectual Disabilities, 2014, 7, 143-168.	1.3	5

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127	Improving IQ measurement in intellectual disabilities using true deviation from population norms. Journal of Neurodevelopmental Disorders, 2014, 6, 16.	1.5	111
128	Autism and Fragile X Syndrome. Seminars in Neurology, 2014, 34, 258-265.	0.5	43
129	Climbing the Branches of a Family Tree: Diagnosis of Fragile X Syndrome. Journal of Pediatrics, 2014, 164, 1292-1295.	0.9	13
130	Mechanism-Based Treatments in Neurodevelopmental Disorders: Fragile X Syndrome. Pediatric Neurology, 2014, 50, 297-302.	1.0	68
131	AGG interruptions and maternal age affect FMR1 CGG repeat allele stability during transmission. Journal of Neurodevelopmental Disorders, 2014, 6, 24.	1.5	94
132	Associated features in females with an FMR1 premutation. Journal of Neurodevelopmental Disorders, 2014, 6, 30.	1.5	116
133	Emerging topics in FXTAS. Journal of Neurodevelopmental Disorders, 2014, 6, 31.	1.5	76
134	Fragile X Syndrome: A Review of Associated Medical Problems. Pediatrics, 2014, 134, 995-1005.	1.0	194
135	Fragile X gene expansions are not associated with dementia. Neurobiology of Aging, 2014, 35, 2637-2638.	1.5	9
136	Implementation of a markerless motion analysis method to quantify hyperkinesis in males with fragile X syndrome. Gait and Posture, 2014, 39, 827-830.	0.6	7
137	Fragile X AGG analysis provides new risk predictions for 45â€“69 repeat alleles. American Journal of Medical Genetics, Part A, 2013, 161, 771-778.	0.7	110
138	Development of an Expressive Language Sampling Procedure in Fragile X Syndrome. Journal of Developmental and Behavioral Pediatrics, 2013, 34, 245-251.	0.6	53
139	Outcome Measures for Clinical Trials in Fragile X Syndrome. Journal of Developmental and Behavioral Pediatrics, 2013, 34, 508-522.	0.6	136
140	Therapeutic Strategies in Fragile X Syndrome: Dysregulated mGluR Signaling and Beyond. Neuropsychopharmacology, 2012, 37, 178-195.	2.8	97
141	Clinic-Based Retrospective Analysis of Psychopharmacology for Behavior in Fragile X Syndrome. International Journal of Pediatrics (United Kingdom), 2012, 2012, 1-11.	0.2	37
142	Effects of STX209 (Arbaclofen) on Neurobehavioral Function in Children and Adults with Fragile X Syndrome: A Randomized, Controlled, Phase 2 Trial. Science Translational Medicine, 2012, 4, 152ra127.	5.8	289
143	Feasibility, reliability, and clinical validity of the Test of Attentional Performance for Children (KiTAP) in Fragile X syndrome (FXS). Journal of Neurodevelopmental Disorders, 2012, 4, 2.	1.5	47
144	FMR1 CGG allele size and prevalence ascertained through newborn screening in the United States. Genome Medicine, 2012, 4, 100.	3.6	258

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145	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.	1.7	148
146	Epigenetic Modification of the FMR1 Gene in Fragile X Syndrome Is Associated with Differential Response to the mGluR5 Antagonist AFQ056. Science Translational Medicine, 2011, 3, 64ra1.	5.8	344
147	Targeted treatments for fragile X syndrome. Journal of Neurodevelopmental Disorders, 2011, 3, 193-210.	1.5	91
148	FMR1 grayâ€zone alleles: Association with Parkinson's disease in women?. Movement Disorders, 2011, 26, 1900-1906.	2.2	44
149	Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation: Analysis of Hypothalamic and Autonomic Candidate Genes. Pediatric Research, 2011, 70, 375-378.	1.1	66
150	Monozygotic Twins Discordant for ROHHAD Phenotype. Pediatrics, 2011, 128, e711-e715.	1.0	47
151	Seizures in Fragile X Syndrome: Characteristics and Comorbid Diagnoses. American Journal on Intellectual and Developmental Disabilities, 2010, 115, 461-472.	0.8	134
152	An Official ATS Clinical Policy Statement: Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2010, 181, 626-644.	2.5	433
153	Advances in the Treatment of Fragile X Syndrome. Pediatrics, 2009, 123, 378-390.	1.0	513
154	A Review of Fragile X Premutation Disorders. Journal of Clinical Psychiatry, 2009, 70, e1-e11.	1.1	119
155	A Review of Fragile X Premutation Disorders. Journal of Clinical Psychiatry, 2009, 70, 852-862.	1.1	154
156	Characterization of Potential Outcome Measures for Future Clinical Trials in Fragile X Syndrome. Journal of Autism and Developmental Disorders, 2008, 38, 1751-1757.	1.7	20
157	Open-Label Treatment Trial of Lithium to Target the Underlying Defect in Fragile X Syndrome. Journal of Developmental and Behavioral Pediatrics, 2008, 29, 293-302.	0.6	203
158	Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation Presenting in Childhood. Pediatrics, 2007, 120, e179-e188.	1.0	175
159	Neuropathic features in fragile X premutation carriers. American Journal of Medical Genetics, Part A, 2007, 143A, 19-26.	0.7	89
160	CGG repeat length correlates with age of onset of motor signs of the fragile X-associated tremor/ataxia syndrome (FXTAS). American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 566-569.	1.1	138
161	Fragile Xâ€associated tremor/ataxia syndrome: Clinical features, genetics, and testing guidelines. Movement Disorders, 2007, 22, 2018-2030.	2.2	306
162	Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2006, 174, 1139-1144.	2.5	238

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163	Effect of CX516, an AMPA-Modulating Compound, on Cognition and Behavior in Fragile X Syndrome: AControlled Trial. Journal of Child and Adolescent Psychopharmacology, 2006, 16, 525-540.	0.7	136
164	Fragile X-associated tremor/ataxia syndrome in sisters related to X-inactivation. Annals of Neurology, 2005, 57, 144-147.	2.8	124
165	Penetrance of the Fragile Xâ€"Associated Tremor/Ataxia Syndrome in a Premutation Carrier Population. JAMA - Journal of the American Medical Association, 2004, 291, 460.	3.8	571
166	Overexpression of fragile X gene (FMR-1) transcripts increases cAMP production in neural cells. , 1998, 51, 41.		40
167	Reduced Cyclic AMP Production in Fragile X Syndrome: Cytogenetic and Molecular Correlations. Pediatric Research, 1995, 38, 638-643.	1.1	51
168	Demonstration of abnormal cyclic AMP production in platelets from patients with fragile X syndrome. American Journal of Medical Genetics Part A, 1993, 45, 81-87.	2.4	48
169	Cyclic AMP metabolism in fragile X syndrome. Annals of Neurology, 1992, 31, 22-26.	2.8	72
170	Specific Receptor-Mediated Inhibition of Cyclic AMP Synthesis by Dopamine in a Neuroblastoma Ã— Brain Hybrid Cell Line NCB-20. Journal of Neurochemistry, 1984, 43, 413-420.	2.1	21