Flora Tassone

List of Publications by Year
in descending order

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306	20,560	8732 75	13338 130
papers	citations	h-index	g-index

310	310	310	10133
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Elevated Levels of FMR1 mRNA in Carrier Males: A New Mechanism of Involvement in the Fragile-X Syndrome. American Journal of Human Genetics, 2000, 66, 6-15.	2.6	756
2	Fragile X Premutation Tremor/Ataxia Syndrome: Molecular, Clinical, and Neuroimaging Correlates. American Journal of Human Genetics, 2003, 72, 869-878.	2.6	720
3	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
4	Fragile X syndrome. Nature Reviews Disease Primers, 2017, 3, 17065.	18.1	490
5	Mitochondrial Dysfunction in Autism. JAMA - Journal of the American Medical Association, 2010, 304, 2389.	3.8	380
6	Autism Profiles of Males With Fragile X Syndrome. American Journal on Intellectual and Developmental Disabilites, 2008, 113, 427-438.	2.7	357
7	Maternal periconceptional folic acid intake and risk of autism spectrum disorders and developmental delay in the CHARGE (CHildhood Autism Risks from Genetics and Environment) case-control study. American Journal of Clinical Nutrition, 2012, 96, 80-89.	2.2	336
8	Sam68 sequestration and partial loss of function are associated with splicing alterations in FXTAS patients. EMBO Journal, 2010, 29, 1248-1261.	3.5	326
9	A Rapid Polymerase Chain Reaction-Based Screening Method for Identification of All Expanded Alleles of the Fragile X (FMR1) Gene in Newborn and High-Risk Populations. Journal of Molecular Diagnostics, 2008, 10, 43-49.	1.2	323
10	FMRP expression as a potential prognostic indicator in fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 84, 250-261.	2.4	318
11	Fragile Xâ€associated tremor/ataxia syndrome: Clinical features, genetics, and testing guidelines. Movement Disorders, 2007, 22, 2018-2030.	2.2	306
12	Autism Spectrum Disorders and Attention-Deficit/Hyperactivity Disorder in Boys with the Fragile X Premutation. Journal of Developmental and Behavioral Pediatrics, 2006, 27, S137-S144.	0.6	292
13	Expanded clinical phenotype of women with the FMR1 premutation. American Journal of Medical Genetics, Part A, 2008, 146A, 1009-1016.	0.7	290
14	Fragile X premutation carriers: characteristic MR imaging findings of adult male patients with progressive cerebellar and cognitive dysfunction. American Journal of Neuroradiology, 2002, 23, 1757-66.	1.2	272
15	Fragile X syndrome: causes, diagnosis, mechanisms, and therapeutics. Journal of Clinical Investigation, 2012, 122, 4314-4322.	3.9	269
16	FMR1 CGG allele size and prevalence ascertained through newborn screening in the United States. Genome Medicine, 2012, 4, 100.	3.6	258
17	The FMR1 CGG repeat mouse displays ubiquitin-positive intranuclear neuronal inclusions; implications for the cerebellar tremor/ataxia syndrome. Human Molecular Genetics, 2003, 12, 949-959.	1.4	253
18	A Novel FMR1 PCR Method for the Routine Detection of Low Abundance Expanded Alleles and Full Mutations in Fragile X Syndrome. Clinical Chemistry, 2010, 56, 399-408.	1.5	250

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19	An antisense transcript spanning the CGG repeat region of FMR1 is upregulated in premutation carriers but silenced in full mutation individuals. Human Molecular Genetics, 2007, 16, 3174-3187.	1.4	241
20	FMR1 RNA within the Intranuclear Inclusions of Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). RNA Biology, 2004, 1, 103-105.	1.5	231
21	Sequestration of DROSHA and DGCR8 by Expanded CGG RNA Repeats Alters MicroRNA Processing in Fragile X-Associated Tremor/Ataxia Syndrome. Cell Reports, 2013, 3, 869-880.	2.9	216
22	Abnormal elevation ofFMR1 mRNA is associated with psychological symptoms in individuals with the fragile X premutation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 139B, 115-121.	1.1	215
23	A Randomized Double-Blind, Placebo-Controlled Trial of Minocycline in Children and Adolescents with Fragile X Syndrome. Journal of Developmental and Behavioral Pediatrics, 2013, 34, 147-155.	0.6	212
24	Translation of Expanded CGG Repeats into FMRpolyG Is Pathogenic and May Contribute to Fragile X Tremor Ataxia Syndrome. Neuron, 2017, 93, 331-347.	3.8	194
25	Transcription of the FMR1 gene in individuals with fragile X syndrome. American Journal of Medical Genetics Part A, 2000, 97, 195-203.	2.4	192
26	Sequencing the unsequenceable: Expanded CGG-repeat alleles of the fragile X gene. Genome Research, 2013, 23, 121-128.	2.4	191
27	Elevated FMR1 mRNA in premutation carriers is due to increased transcription. Rna, 2007, 13, 555-562.	1.6	176
28	Disease-Associated Short Tandem Repeats Co-localize with Chromatin Domain Boundaries. Cell, 2018, 175, 224-238.e15.	13.5	169
29	Cognitive profile of fragile X premutation carriers with and without fragile X-associated tremor/ataxia syndrome.. Neuropsychology, 2008, 22, 48-60.	1.0	167
30	An Information-Rich CGG Repeat Primed PCR That Detects the Full Range of Fragile X Expanded Alleles and Minimizes the Need for Southern Blot Analysis. Journal of Molecular Diagnostics, 2010, 12, 589-600.	1.2	166
31	Altered mTOR signaling and enhanced CYFIP2 expression levels in subjects with fragile X syndrome. Genes, Brain and Behavior, 2012, 11, 332-341.	1.1	164
32	The Fragile X Premutation Presenting as Essential Tremor. Archives of Neurology, 2003, 60, 117.	4.9	162
33	Psychiatric Phenotype of the Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS) in Males. Journal of Clinical Psychiatry, 2006, 67, 87-94.	1.1	158
34	A solution to limitations of cognitive testing in children with intellectual disabilities: the case of fragile X syndrome. Journal of Neurodevelopmental Disorders, 2009, 1, 33-45.	1.5	156
35	Fragile X males with unmethylated, full mutation trinucleotide repeat expansions have elevated levels ofFMR1 messenger RNA. American Journal of Medical Genetics Part A, 2000, 94, 232-236.	2.4	154
36	A Review of Fragile X Premutation Disorders. Journal of Clinical Psychiatry, 2009, 70, 852-862.	1.1	154

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37	Induction of inclusion formation and disruption of lamin A/C structure by premutation CGG-repeat RNA in human cultured neural cells. Human Molecular Genetics, 2005, 14, 3661-3671.	1.4	152
38	AGG interruptions within the maternal FMR1 gene reduce the risk of offspring with fragile X syndrome. Genetics in Medicine, 2012, 14, 729-736.	1.1	152
39	Expression profiling suggests underexpression of the GABAA receptor subunit Î´ in the fragile X knockout mouse model. Neurobiology of Disease, 2006, 21, 346-357.	2.1	151
40	Screening for Expanded Alleles of the FMR1 Gene in Blood Spots from Newborn Males in a Spanish Population. Journal of Molecular Diagnostics, 2009, 11, 324-329.	1.2	146
41	Murine hippocampal neurons expressing Fmr1 gene premutations show early developmental deficits and late degeneration. Human Molecular Genetics, 2010, 19, 196-208.	1.4	143
42	Reduced FMR1 mRNA translation efficiency in fragile X patients with premutations. Rna, 2002, 8, 1482-8.	1.6	143
43	High MMPâ€9 activity levels in fragile X syndrome are lowered by minocycline. American Journal of Medical Genetics, Part A, 2013, 161, 1897-1903.	0.7	140
44	The Prader-Willi Phenotype of Fragile X Syndrome. Journal of Developmental and Behavioral Pediatrics, 2007, 28, 133-138.	0.6	139
45	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
46	Fragile X Syndrome. Current Genomics, 2011, 12, 216-224.	0.7	136
47	Progression of tremor and ataxia in male carriers of theFMR1 premutation. Movement Disorders, 2007, 22, 203-206.	2.2	134
48	Molecular and cognitive predictors of the continuum of autistic behaviours in fragile X. Neuroscience and Biobehavioral Reviews, 2007, 31, 315-326.	2.9	130
49	Impairment in the cognitive functioning of men with fragile X-associated tremor/ataxia syndrome (FXTAS). Journal of the Neurological Sciences, 2006, 248, 227-233.	0.3	126
50	Amygdala dysfunction in men with the fragile X premutation. Brain, 2007, 130, 404-416.	3.7	125
51	The (CGG)n repeat element within the 5' untranslated region of the FMR1 message provides both positive and negative cis effects on in vivo translation of a downstream reporter. Human Molecular Genetics, 2003, 12, 3067-3074.	1.4	124
52	A neuropsychological investigation of male premutation carriers of fragile X syndrome. Neuropsychologia, 2004, 42, 1934-1947.	0.7	119
53	A Review of Fragile X Premutation Disorders. Journal of Clinical Psychiatry, 2009, 70, e1-e11.	1.1	119
54	Neuropathological, clinical and molecular pathology in female fragile X premutation carriers with and without FXTAS. Genes, Brain and Behavior, 2012, 11, 577-585.	1.1	117

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55	Autistic Spectrum Disorder and the Fragile X Premutation. Journal of Developmental and Behavioral Pediatrics, 2004, 25, 392-398.	0.6	116
56	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
57	Expansion of an FMR1 Grey-Zone Allele to a Full Mutation in Two Generations. Journal of Molecular Diagnostics, 2009, 11, 306-310.	1.2	109
58	Increased prevalence of seizures in boys who were probands with the FMR1 premutation and co-morbid autism spectrum disorder. Human Genetics, 2012, 131, 581-589.	1.8	108
59	Widespread non-central nervous system organ pathology in fragile X premutation carriers with fragile X-associated tremor/ataxia syndrome and CGG knock-in mice. Acta Neuropathologica, 2011, 122, 467-479.	3.9	102
60	An Enhanced Polymerase Chain Reaction Assay to Detect Pre- and Full Mutation Alleles of the Fragile X Mental Retardation 1 Gene. Journal of Molecular Diagnostics, 2005, 7, 605-612.	1.2	96
61	AGG interruptions and maternal age affect FMR1 CGG repeat allele stability during transmission. Journal of Neurodevelopmental Disorders, 2014, 6, 24.	1.5	94
62	Methylation of novel markers of fragile X alleles is inversely correlated with FMRP expression and FMR1 activation ratio. Human Molecular Genetics, 2010, 19, 1618-1632.	1.4	92
63	Clustered burst firing in FMR1 premutation hippocampal neurons: amelioration with allopregnanolone. Human Molecular Genetics, 2012, 21, 2923-2935.	1.4	92
64	Brief Report: Aggression and Stereotypic Behavior in Males with Fragile X Syndromeâ€"Moderating Secondary Genes in a â€œSingle Geneâ€•Disorder. Journal of Autism and Developmental Disorders, 2008, 38, 184-189.	1.7	89
65	Decreased Fragile X Mental Retardation Protein Expression Underlies Amygdala Dysfunction in Carriers of the Fragile X Premutation. Biological Psychiatry, 2011, 70, 859-865.	0.7	88
66	A voxel-based morphometry study of grey matter loss in fragile X-associated tremor/ataxia syndrome. Brain, 2011, 134, 863-878.	3.7	87
67	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	4.1	87
68	Immuneâ€mediated disorders among women carriers of fragile X premutation alleles. American Journal of Medical Genetics, Part A, 2012, 158A, 2473-2481.	0.7	86
69	Clinical and molecular implications of mosaicism in FMR1 full mutations. Frontiers in Genetics, 2014, 5, 318.	1.1	86
70	Impairment of executive cognitive functioning in males with fragile X-associated tremor/ataxia syndrome. Movement Disorders, 2007, 22, 645-650.	2.2	84
71	Variability in FMRP and Early Development in Males With Fragile X Syndrome. American Journal on Intellectual and Developmental Disabilites, 2001, 106, 16.	2.7	83
72	Cognitive, anxiety and mood disorders in the fragile X-associated tremor/ataxia syndrome. General Hospital Psychiatry, 2007, 29, 349-356.	1.2	83

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73	The primary cognitive deficit among males with fragile X-associated tremor/ataxia syndrome (FXTAS) is a dysexecutive syndrome. Journal of Clinical and Experimental Neuropsychology, 2008, 30, 853-869.	0.8	83
74	High-resolution methylation polymerase chain reaction for fragile X analysis: Evidence for novel FMR1 methylation patterns undetected in Southern blot analyses. Genetics in Medicine, 2011, 13, 528-538.	1.1	80
75	An fMRI study of the prefrontal activity during the performance of a working memory task in premutation carriers of the fragile X mental retardation 1 gene with and without fragile X-associated tremor/ataxia syndrome (FXTAS). Journal of Psychiatric Research, 2011, 45, 36-43.	1.5	80
76	Tissue heterogeneity of theFMR1 mutation in a high-functioning male with fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 84, 233-239.	2.4	79
77	Fragile X Mental Retardation Protein (FMRP) controls diacylglycerol kinase activity in neurons. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E3619-28.	3.3	79
78	CGG allele size somatic mosaicism and methylation inFMR1premutation alleles. Journal of Medical Genetics, 2014, 51, 309-318.	1.5	76
79	Abnormal Nerve Conduction Features in Fragile X Premutation Carriers. Archives of Neurology, 2008, 65, 495.	4.9	75
80	The effect of pre-mutation of X chromosome CGG trinucleotide repeats on brain anatomy. Brain, 2004, 127, 2672-2681.	3.7	74
81	Plasma cytokine profiles in Fragile X subjects: Is there a role for cytokines in the pathogenesis?. Brain, Behavior, and Immunity, 2010, 24, 898-902.	2.0	73
82	Young adult female fragile X premutation carriers show age- and genetically-modulated cognitive impairments. Brain and Cognition, 2011, 75, 255-260.	0.8	72
83	Diffusion tensor imaging in male premutation carriers of the fragile X mental retardation gene. Movement Disorders, 2011, 26, 1329-1336.	2.2	72
84	Fragile Xâ€“associated tremor ataxia syndrome in FMR1 gray zone allele carriers. Movement Disorders, 2012, 27, 297-301.	2.2	72
85	Redistribution of transcription start sites within the FMR1 promoter region with expansion of the downstream CGG-repeat element. Human Molecular Genetics, 2004, 13, 543-549.	1.4	71
86	Transcript levels of the intermediate size or grey zone fragile X mental retardation 1 alleles are raised, and correlate with the number of CGG repeats. Journal of Medical Genetics, 2006, 44, 200-204.	1.5	71
87	Brief Report: Sensorimotor Gating in Idiopathic Autism and Autism Associated with Fragile X Syndrome. Journal of Autism and Developmental Disorders, 2011, 41, 248-253.	1.7	71
88	Early mitochondrial abnormalities in hippocampal neurons cultured from Fmr1 preâ€mutation mouse model. Journal of Neurochemistry, 2012, 123, 613-621.	2.1	70
89	Secondary medical diagnosis in fragile X syndrome with and without autism spectrum disorder. American Journal of Medical Genetics, Part A, 2008, 146A, 1911-1916.	0.7	68
90	Clinical and Neuropathologic Findings in a Woman With the FMR1 Premutation and Multiple Sclerosis. Archives of Neurology, 2008, 65, 1114-6.	4.9	68

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91	Neuropathologic features in the hippocampus and cerebellum of three older men with fragile X syndrome. Molecular Autism, 2011, 2, 2.	2.6	68
92	CGG-repeat length threshold for FMR1 RNA pathogenesis in a cellular model for FXTAS. Human Molecular Genetics, 2011, 20, 2161-2170.	1.4	67
93	A randomized double-blind, placebo-controlled trial of ganaxolone in children and adolescents with fragile X syndrome. Journal of Neurodevelopmental Disorders, 2017, 9, 26.	1.5	67
94	Fibroblast phenotype in male carriers of FMR1 premutation alleles. Human Molecular Genetics, 2010, 19, 299-312.	1.4	66
95	CNS expression of murine fragile X protein (FMRP) as a function of CGG-repeat size. Human Molecular Genetics, 2014, 23, 3228-3238.	1.4	66
96	Rare FMR1 gene mutations causing fragile X syndrome: A review. American Journal of Medical Genetics, Part A, 2018, 176, 11-18.	0.7	65
97	Fragile Xâ€“Associated Tremor/Ataxia Syndrome. JAMA Neurology, 2013, 70, 1022.	4.5	64
98	Decreased DGCR8 Expression and miRNA Dysregulation in Individuals with 22q11.2 Deletion Syndrome. PLoS ONE, 2014, 9, e103884.	1.1	64
99	Magnetic resonance imaging study in older fragile X premutation male carriers. Annals of Neurology, 2005, 58, 326-330.	2.8	61
100	Neuropathy as a presenting feature in fragile Xâ€associated tremor/ataxia syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 2256-2260.	0.7	59
101	Aging in fragile X syndrome. Journal of Neurodevelopmental Disorders, 2010, 2, 70-76.	1.5	59
102	Adult Female Fragile X Premutation Carriers Exhibit Age- and CGG Repeat Length-Related Impairments on an Attentionally Based Enumeration Task. Frontiers in Human Neuroscience, 2011, 5, 63.	1.0	59
103	Dysregulated ADAM10-Mediated Processing of APP during a Critical Time Window Leads to Synaptic Deficits in Fragile X Syndrome. Neuron, 2015, 87, 382-398.	3.8	59
104	Global increases in both common and rare copy number load associated with autism. Human Molecular Genetics, 2013, 22, 2870-2880.	1.4	56
105	Mitochondrial Citrate Transporter-dependent Metabolic Signature in the 22q11.2 Deletion Syndrome. Journal of Biological Chemistry, 2015, 290, 23240-23253.	1.6	56
106	Altered Redox Mitochondrial Biology in the Neurodegenerative Disorder Fragile X-Tremor/Ataxia Syndrome: Use of Antioxidants in Precision Medicine. Molecular Medicine, 2016, 22, 548-559.	1.9	56
107	Expression of the FMR1 gene. Cytogenetic and Genome Research, 2003, 100, 124-128.	0.6	54
108	Reduced Hippocampal Activation During Recall is Associated with Elevated FMR1 mRNA and Psychiatric Symptoms in Men with the Fragile X Premutation. Brain Imaging and Behavior, 2008, 2, 105-116.	1.1	54

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109	A Quantitative ELISA Assay for the Fragile X Mental Retardation 1 Protein. Journal of Molecular Diagnostics, 2009, 11, 281-289.	1.2	52
110	Selected vitamin D metabolic gene variants and risk for autism spectrum disorder in the CHARGE Study. Early Human Development, 2015, 91, 483-489.	0.8	52
111	Molecular analyses of neurogenic defects in a human pluripotent stem cell model of fragile X syndrome. Brain, 2017, 140, aww357.	3.7	52
112	Strong similarities of theFMR1 mutation in multiple tissues: Postmortem studies of a male with a full mutation and a male carrier of a premutation. American Journal of Medical Genetics Part A, 1999, 84, 240-244.	2.4	51
113	Sleep apnea in fragile X premutation carriers with and without FXTAS. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 923-928.	1.1	51
114	Fibromyalgia in fragile X mental retardation 1 gene premutation carriers. Rheumatology, 2011, 50, 2233-2236.	0.9	51
115	Age-Dependent Structural Connectivity Effects in Fragile X Premutation. Archives of Neurology, 2012, 69, 482-9.	4.9	51
116	Protein synthesis levels are increased in a subset of individuals with fragile X syndrome. Human Molecular Genetics, 2018, 27, 2039-2051.	1.4	51
117	Fragile X syndrome. Colombia Medica, 2014, 45, 190-8.	0.7	48
118	Size and methylation mosaicism in males with Fragile X syndrome. Expert Review of Molecular Diagnostics, 2017, 17, 1023-1032.	1.5	47
119	Abnormal trajectories in cerebellum and brainstem volumes in carriers of the fragile X premutation. Neurobiology of Aging, 2017, 55, 11-19.	1.5	46
120	Differential usage of transcriptional start sites and polyadenylation sites in FMR1 premutation alleles. Nucleic Acids Research, 2011, 39, 6172-6185.	6.5	45
121	FMR1 grayâ€zone alleles: Association with Parkinson's disease in women?. Movement Disorders, 2011, 26, 1900-1906.	2.2	44
122	Controlled trial of lovastatin combined with an open-label treatment of a parent-implemented language intervention in youth with fragile X syndrome. Journal of Neurodevelopmental Disorders, 2020, 12, 12.	1.5	44
123	Memantine for Fragile Xâ€“Associated Tremor/Ataxia Syndrome. Journal of Clinical Psychiatry, 2014, 75, 264-271.	1.1	44
124	Cognitive Impairment in a 65-year-old Male With the Fragile X-associated Tremor-Ataxia Syndrome (FXTAS). Cognitive and Behavioral Neurology, 2006, 19, 165-171.	0.5	43
125	Enhanced Asynchronous Ca2+ Oscillations Associated with Impaired Glutamate Transport in Cortical Astrocytes Expressing Fmr1 Gene Premutation Expansion. Journal of Biological Chemistry, 2013, 288, 13831-13841.	1.6	43
126	MAOA, DBH, and SLC6A4 variants in CHARGE: a caseâ€“control study of autism spectrum disorders. Autism Research, 2011, 4, 250-261.	2.1	42

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127	Newborn Screening for Fragile X Syndrome. JAMA Neurology, 2014, 71, 355.	4.5	42
128	Parkinsonism in fragile X-associated tremor/ataxia syndrome (FXTAS): Revisited. Parkinsonism and Related Disorders, 2014, 20, 456-459.	1.1	42
129	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. American Journal of Human Genetics, 2020, 106, 26-40.	2.6	42
130	Dementia With Mood Symptoms in a Fragile X Premutation Carrier With the Fragile X-Associated Tremor/Ataxia Syndrome: Clinical Intervention With Donepezil and Venlafaxine. Journal of Neuropsychiatry and Clinical Neurosciences, 2006, 18, 171-177.	0.9	41
131	Hypertension in FMR1 premutation males with and without fragile Xâ€associated tremor/ataxia syndrome (FXTAS). American Journal of Medical Genetics, Part A, 2012, 158A, 1304-1309.	0.7	41
132	CDNA selection from 10 Mb of Chromosome 21 DNA: efficiency in transcriptional mapping and reflections of genome organization. Human Molecular Genetics, 1995, 4, 1509-1518.	1.4	40
133	Early onset of neurological symptoms in fragile X premutation carriers exposed to neurotoxins. NeuroToxicology, 2010, 31, 399-402.	1.4	40
134	Immortalized Parkinson's Disease lymphocytes have enhanced mitochondrial respiratory activity. DMM Disease Models and Mechanisms, 2016, 9, 1295-1305.	1.2	40
135	Newborn, Carrier, and Early Childhood Screening Recommendations for Fragile X. Pediatrics, 2012, 130, 1126-1135.	1.0	39
136	The development of cognitive control in children with chromosome 22q11.2 deletion syndrome. Frontiers in Psychology, 2014, 5, 566.	1.1	39
137	Metformin treatment in young children with fragile X syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e956.	0.6	39
138	Anxiety disorders in fragile X premutation carriers: Preliminary characterization of probands and non-probands. Intractable and Rare Diseases Research, 2015, 4, 123-130.	0.3	39
139	Plasma metabolic profile delineates roles for neurodegeneration, pro-inflammatory damage and mitochondrial dysfunction in the FMR1 premutation. Biochemical Journal, 2016, 473, 3871-3888.	1.7	38
140	Intranuclear inclusions in a fragile X mosaic male. Translational Neurodegeneration, 2013, 2, 10.	3.6	37
141	The multiple molecular facets of fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 23.	1.5	36
142	Screen for Excess FMR1 Premutation Alleles Among Males With Parkinsonism. Archives of Neurology, 2007, 64, 1002.	4.9	33
143	Rare Intranuclear Inclusions in the Brains of 3 Older Adult Males With Fragile X Syndrome: Implications for the Spectrum of Fragile X-Associated Disorders. Journal of Neuropathology and Experimental Neurology, 2011, 70, 462-469.	0.9	33
144	Warburg effect linked to cognitiveâ€executive deficits inFMR1premutation. FASEB Journal, 2016, 30, 3334-3351.	0.2	33

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145	Advanced technologies for the molecular diagnosis of fragile X syndrome. Expert Review of Molecular Diagnostics, 2015, 15, 1465-1473.	1.5	32
146	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. American Journal of Human Genetics, 2020, 107, 555-563.	2.6	32
147	Reduced excitatory amino acid transporter 1 and metabotropic glutamate receptor 5 expression in the cerebellum of fragile X mental retardation gene 1 premutation carriers with fragile X-associated tremor/ataxia syndrome. Neurobiology of Aging, 2014, 35, 1189-1197.	1.5	31
148	A Search for Genes from the Dark Band Regions of Human Chromosome 21. Genomics, 1995, 27, 1-8.	1.3	30
149	Tremor/Ataxia syndrome in fragile X carrier males. Movement Disorders, 2002, 17, 744-745.	2.2	30
150	Autoimmune disease in mothers with the FMR1 premutation is associated with seizures in their children with fragile X syndrome. Human Genetics, 2010, 128, 539-548.	1.8	30
151	Fear-Specific Amygdala Function in Children and Adolescents on the Fragile X Spectrum: A Dosage Response of the FMR1 Gene. Cerebral Cortex, 2014, 24, 600-613.	1.6	30
152	Differential increases of specificFMR1mRNA isoforms in premutation carriers. Journal of Medical Genetics, 2015, 52, 42-52.	1.5	29
153	FindingFMR1mosaicism in Fragile X syndrome. Expert Review of Molecular Diagnostics, 2016, 16, 501-507.	1.5	29
154	Molecular biomarkers predictive of sertraline treatment response in young children with fragile X syndrome. Brain and Development, 2017, 39, 483-492.	0.6	29
155	Enhanced Manual and Oral Motor Reaction Time in Young Adult Female Fragile X Premutation Carriers. Journal of the International Neuropsychological Society, 2011, 17, 746-750.	1.2	28
156	Neural Substrates of Executive Dysfunction in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS): a Brain Potential Study. Cerebral Cortex, 2013, 23, 2657-2666.	1.6	28
157	High functioning male with fragile X syndrome and fragile Xâ€associated tremor/ataxia syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2154-2161.	0.7	28
158	Altered expression of the FMR1 splicing variants landscape in premutation carriers. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2017, 1860, 1117-1126.	0.9	28
159	Allopregnanolone Treatment Improves Plasma Metabolomic Profile Associated with GABA Metabolism in Fragile X-Associated Tremor/Ataxia Syndrome: a Pilot Study. Molecular Neurobiology, 2019, 56, 3702-3713.	1.9	28
160	Testing the FMR1 Promoter for Mosaicism in DNA Methylation among CpG Sites, Strands, and Cells in FMR1-Expressing Males with Fragile X Syndrome. PLoS ONE, 2011, 6, e23648.	1.1	28
161	Translation of the FMR1 mRNA is not influenced by AGG interruptions. Nucleic Acids Research, 2009, 37, 6896-6904.	6.5	27
162	Broad Clinical Involvement in a Family Affected by the Fragile X Premutation. Journal of Developmental and Behavioral Pediatrics, 2009, 30, 544-551.	0.6	27

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163	Screening for the Presence of FMR1 Premutation Alleles in Women With Parkinsonism. Archives of Neurology, 2009, 66, 244-9.	4.9	27
164	Genetic cluster of fragile X syndrome in a Colombian district. Journal of Human Genetics, 2018, 63, 509-516.	1.1	27
165	A Quantitative Assessment of Tremor and Ataxia in FemaleFMR1Premutation Carriers Using CATSYS. Current Gerontology and Geriatrics Research, 2011, 2011, 1-7.	1.6	26
166	The Fragile X-Associated Tremor Ataxia Syndrome. Results and Problems in Cell Differentiation, 2012, 54, 337-357.	0.2	26
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