Flora Tassone

List of Publications by Year in descending order

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Version: 2024-02-01

306 papers	20,560 citations	10070 75 h-index	15253 130 g-index
310 all docs	310 docs citations	310 times ranked	11132 citing authors

#	Article	IF	Citations
1	Mosaicism in Fragile X syndrome: A family case series. Journal of Intellectual Disabilities, 2022, 26, 800-807.	1.0	4
2	Hypermobile Ehlers-Danlos syndrome (hEDS) phenotype in fragile X premutation carriers: case series. Journal of Medical Genetics, 2022, 59, 687-690.	1.5	5
3	Clinical and Molecular Correlates of Abnormal Changes in the Cerebellum and Globus Pallidus in Fragile X Premutation. Frontiers in Neurology, 2022, 13, 797649.	1.1	7
4	Neuropsychological changes in FMR1 premutation carriers and onset of fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2022, 14, 23.	1.5	8
5	Prosaccade and Antisaccade Behavior in Fragile Xâ€associated Tremor/Ataxia Syndrome Progression. Movement Disorders Clinical Practice, 2022, 9, 473-478.	0.8	1
6	De Novo Large Deletion Leading to Fragile X Syndrome. Frontiers in Genetics, 2022, 13, .	1.1	1
7	Both cis and trans-acting genetic factors drive somatic instability in female carriers of the FMR1 premutation. Scientific Reports, 2022, 12, .	1.6	11
8	Inequities in diagnosis of Fragile X syndrome in Colombia. Journal of Applied Research in Intellectual Disabilities, 2021, 34, 830-839.	1.3	3
9	Cerebral Microbleeds in Fragile Xâ€“Associated Tremor/Ataxia Syndrome. Movement Disorders, 2021, 36, 1935-1943.	2.2	17
10	â€˜Essential Tremorâ€™ Phenotype in FMR1 Premutation/Gray Zone Sibling Series: Exploring Possible Genetic Modifiers. Twin Research and Human Genetics, 2021, 24, 95-102.	0.3	3
11	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
12	Expression of expanded FMR1-CGG repeats alters mitochondrial miRNAs and modulates mitochondrial functions and cell death in cellular model of FXTAS. Free Radical Biology and Medicine, 2021, 165, 100-110.	1.3	9
13	Relationships between motor scores and cognitive functioning in FMR1 female premutation X carriers indicate early involvement of cerebello-cerebral pathways. Cerebellum and Ataxias, 2021, 8, 15.	1.9	12
14	Metabolomic Biomarkers Are Associated With Area of the Pons in Fragile X Premutation Carriers at Risk for Developing FXTAS. Frontiers in Psychiatry, 2021, 12, 691717.	1.3	2
15	Case Report: Coexistence of Alzheimer-Type Neuropathology in Fragile X-Associated Tremor Ataxia Syndrome. Frontiers in Neuroscience, 2021, 15, 720253.	1.4	8
16	FMRP Levels in Human Peripheral Blood Leukocytes Correlates with Intellectual Disability. Diagnostics, 2021, 11, 1780.	1.3	6
17	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	4.1	87
18	EEG Signal Complexity Is Reduced During Resting-State in Fragile X Syndrome. Frontiers in Psychiatry, 2021, 12, 716707.	1.3	5

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19	Cellular Bioenergetics and AMPK and TORC1 Signalling in Blood Lymphoblasts Are Biomarkers of Clinical Status in FMR1 Premutation Carriers. Frontiers in Psychiatry, 2021, 12, 747268.	1.3	4
20	Delineating the Relationships Between Motor, Cognitive-Executive and Psychiatric Symptoms in Female FMR1 Premutation Carriers. Frontiers in Psychiatry, 2021, 12, 742929.	1.3	1
21	Increased Pain Symptomatology Among Females vs. Males With Fragile X-Associated Tremor/Ataxia Syndrome. Frontiers in Psychiatry, 2021, 12, 762915.	1.3	6
22	Interaction between ventricular expansion and structural changes in the corpus callosum and putamen in males with FMR1 normal and premutation alleles. Neurobiology of Aging, 2020, 86, 27-38.	1.5	10
23	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
24	Urine-Derived Epithelial Cell Lines: A New Tool to Model Fragile X Syndrome (FXS). Cells, 2020, 9, 2240.	1.8	2
25	The emerging molecular mechanisms for mitochondrial dysfunctions in FXTAS. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165918.	1.8	8
26	Characterization of the Metabolic, Clinical and Neuropsychological Phenotype of Female Carriers of the Premutation in the X-Linked FMR1 Gene. Frontiers in Molecular Biosciences, 2020, 7, 578640.	1.6	12
27	Cortical gyrification and its relationships with molecular measures and cognition in children with the FMR1 premutation. Scientific Reports, 2020, 10, 16059.	1.6	3
28	Women with Fragile Xâ€“associated Tremor/Ataxia Syndrome. Movement Disorders Clinical Practice, 2020, 7, 910-919.	0.8	13
29	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. American Journal of Human Genetics, 2020, 107, 555-563.	2.6	32
30	Experiences with offering pro bono medical genetics services in the West Indies: Benefits to patients, physicians, and the community. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 1030-1041.	0.7	8
31	Metabolic profiling reveals dysregulated lipid metabolism and potential biomarkers associated with the development and progression of Fragile Xâ€Associated Tremor/Ataxia Syndrome (FXTAS). FASEB Journal, 2020, 34, 16676-16692.	0.2	11
32	Blood-Based Biomarkers Predictive of Metformin Target Engagement in Fragile X Syndrome. Brain Sciences, 2020, 10, 361.	1.1	3
33	Parkinsonism Versus Concomitant Parkinson's Disease in Fragile Xâ€“Associated Tremor/Ataxia Syndrome. Movement Disorders Clinical Practice, 2020, 7, 413-418.	0.8	13
34	Rapidly Progressing Neurocognitive Disorder in a Male with FXTAS and Alzheimerâ€™s Disease . Clinical Interventions in Aging, 2020, Volume 15, 285-292.	1.3	7
35	Elevated FMR1-mRNA and lowered FMRP â€“ A double-hit mechanism for psychiatric features in men with FMR1 premutations. Translational Psychiatry, 2020, 10, 205.	2.4	20
36	FMR1 locus isoforms: potential biomarker candidates in fragile X-associated tremor/ataxia syndrome (FXTAS). Scientific Reports, 2020, 10, 11099.	1.6	11

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37	Developmental aspects of FXAND in a man with the FMR1 premutation. Molecular Genetics & Genomic Medicine, 2020, 8, e1050.	0.6	5
38	Molecular Biomarkers Predictive of Sertraline Treatment Response in Young Children With Autism Spectrum Disorder. Frontiers in Genetics, 2020, 11, 308.	1.1	3
39	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
40	Differential Progression of Motor Dysfunction Between Male and Female Fragile X Premutation Carriers Reveals Novel Aspects of Sex-Specific Neural Involvement. Frontiers in Molecular Biosciences, 2020, 7, 577246.	1.6	13
41	Double Genetic Hit: Fragile X Syndrome and Partial Deletion of Protein Patched Homolog 1 Antisense as Cause of Severe Autism Spectrum Disorder. Journal of Developmental and Behavioral Pediatrics, 2020, 41, 724-728.	0.6	7
42	Fragile X-associated neuropsychiatric disorders: a case report. Future Neurology, 2019, 14, FNL14.	0.9	5
43	A Randomized Controlled Trial of Sertraline in Young Children With Autism Spectrum Disorder. Frontiers in Psychiatry, 2019, 10, 810.	1.3	22
44	The role of AGG interruptions in the FMR1 gene stability: A survey in ethnic groups with low and high rate of consanguinity. Molecular Genetics & Genomic Medicine, 2019, 7, e00946.	0.6	8
45	Metformin treatment in young children with fragile X syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e956.	0.6	39
46	Total and Regional White Matter Lesions Are Correlated With Motor and Cognitive Impairments in Carriers of the FMR1 Premutation. Frontiers in Neurology, 2019, 10, 832.	1.1	13
47	Cognitive and behavioral improvement in adults with fragile X syndrome treated with metforminâ€two cases. Molecular Genetics & Genomic Medicine, 2019, 7, e00745.	0.6	21
48	Molecular Biomarkers in Fragile X Syndrome. Brain Sciences, 2019, 9, 96.	1.1	21
49	Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). Methods in Molecular Biology, 2019, 1942, 173-189.	0.4	6
50	Repeat Instability in the Fragile X-Related Disorders: Lessons from a Mouse Model. Brain Sciences, 2019, 9, 52.	1.1	19
51	FMRpolyG alters mitochondrial transcripts level and respiratory chain complex assembly in Fragile X associated tremor/ataxia syndrome [FXTAS]. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 1379-1388.	1.8	25
52	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
53	Increased severity of fragile X spectrum disorders in the agricultural community of Ricaurte, Colombia. International Journal of Developmental Neuroscience, 2019, 72, 1-5.	0.7	10
54	Genetic cluster of fragile X syndrome in a Colombian district. Journal of Human Genetics, 2018, 63, 509-516.	1.1	27

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55	Ageâ€•and CGG repeatâ€‐related slowing of manual movement in fragile X carriers: A prodrome of fragile Xâ€‐associated tremor ataxia syndrome?. Movement Disorders, 2018, 33, 628-636.	2.2	13
56	Protein synthesis levels are increased in a subset of individuals with fragile X syndrome. Human Molecular Genetics, 2018, 27, 2039-2051.	1.4	51
57	Rare FMR1 gene mutations causing fragile X syndrome: A review. American Journal of Medical Genetics, Part A, 2018, 176, 11-18.	0.7	65
58	A higher rare CNV burden in the genetic background potentially contributes to intellectual disability phenotypes in 22q11.2 deletion syndrome. European Journal of Medical Genetics, 2018, 61, 209-212.	0.7	17
59	The Spectrum of Neurological and White Matter Changes and Premutation Status Categories of Older Male Carriers of the FMR1 Alleles Are Linked to Genetic (CGG and FMR1 mRNA) and Cellular Stress (AMPK) Markers. Frontiers in Genetics, 2018, 9, 531.	1.1	7
60	Microglial cell activation and senescence are characteristic of the pathology FXTAS. Movement Disorders, 2018, 33, 1887-1894.	2.2	19
61	Cognitive Deficits and Associated ERP N400 Abnormalities in FXTAS With Parkinsonism. Frontiers in Genetics, 2018, 9, 327.	1.1	0
62	Assessment of Molecular Measures in Non-FXTAS Male Premutation Carriers. Frontiers in Genetics, 2018, 9, 302.	1.1	4
63	Impact of FMR1 Premutation on Neurobehavior and Bioenergetics in Young Monozygotic Twins. Frontiers in Genetics, 2018, 9, 338.	1.1	17
64	Presence of Middle Cerebellar Peduncle Sign in FMR1 Premutation Carriers Without Tremor and Ataxia. Frontiers in Neurology, 2018, 9, 695.	1.1	13
65	Curvilinear Association Between Language Disfluency and FMR1 CGG Repeat Size Across the Normal, Intermediate, and Premutation Range. Frontiers in Genetics, 2018, 9, 344.	1.1	22
66	Evidence for the role of FMR1 gray zone alleles as a risk factor for parkinsonism in females. Movement Disorders, 2018, 33, 1178-1181.	2.2	20
67	Disease-Associated Short Tandem Repeats Co-localize with Chromatin Domain Boundaries. Cell, 2018, 175, 224-238.e15.	13.5	169
68	Middle Cerebellar Peduncle Widthâ€”A Novel MRI Biomarker for FXTAS?. Frontiers in Neuroscience, 2018, 12, 379.	1.4	16
69	Fentanyl overdose in a female with the premutation and FXTAS. , 2018, 1, .		3
70	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
71	Molecular biomarkers predictive of sertraline treatment response in young children with fragile X syndrome. Brain and Development, 2017, 39, 483-492.	0.6	29
72	Clinical and molecular correlates in fragile X premutation females. ENeurologicalSci, 2017, 7, 49-56.	0.5	13

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73	Fragile X Newborn Screening: Lessons Learned From a Multisite Screening Study. Pediatrics, 2017, 139, S216-S225.	1.0	22
74	Molecular analyses of neurogenic defects in a human pluripotent stem cell model of fragile X syndrome. Brain, 2017, 140, aww357.	3.7	52
75	Abnormal trajectories in cerebellum and brainstem volumes in carriers of the fragile X premutation. Neurobiology of Aging, 2017, 55, 11-19.	1.5	46
76	Fragile X syndrome. Nature Reviews Disease Primers, 2017, 3, 17065.	18.1	490
77	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
78	Size and methylation mosaicism in males with Fragile X syndrome. Expert Review of Molecular Diagnostics, 2017, 17, 1023-1032.	1.5	47
79	Reduced vagal tone in women with the FMR1 premutation is associated with FMR1 mRNA but not depression or anxiety. Journal of Neurodevelopmental Disorders, 2017, 9, 16.	1.5	12
80	Children With Fragile X Syndrome Display Threat-Specific Biases Toward Emotion. Biological Psychiatry: Cognitive Neuroscience and Neuroimaging, 2017, 2, 487-492.	1.1	10
81	Down Syndrome and Fragile X Syndrome in a Colombian Woman: Case Report. Journal of Applied Research in Intellectual Disabilities, 2017, 30, 970-974.	1.3	6
82	Novel Blood Biomarkers Are Associated with White Matter Lesions in Fragile X- Associated Tremor/Ataxia Syndrome. Neurodegenerative Diseases, 2017, 17, 22-30.	0.8	19
83	Molecular Diagnostics and Genetic Counseling in Fragile X Syndrome andÂ FMR1 -Associated Disorders. , 2017, , 41-55.		1
84	Prenatal Diagnosis of Fragile X: Can a Full Mutation Allele in the FMR1 Gene Contract to a Normal Size?. Frontiers in Genetics, 2017, 8, 158.	1.1	8
85	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
86	Concomitant occurrence of FXTAS and clinically defined sporadic inclusion body myositis: report of two cases. Croatian Medical Journal, 2017, 58, 310-315.	0.2	4
87	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
88	Premutation in the Fragile X Mental Retardation 1 (FMR1) Gene Affects Maternal Zn-milk and Perinatal Brain Bioenergetics and Scaffolding. Frontiers in Neuroscience, 2016, 10, 159.	1.4	24
89	Plasma Biomarkers for Monitoring Brain Pathophysiology in FMR1 Premutation Carriers. Frontiers in Molecular Neuroscience, 2016, 9, 71.	1.4	24
90	Alcohol use dependence in fragile X syndrome. Intractable and Rare Diseases Research, 2016, 5, 207-213.	0.3	8

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91	Clinical Phenotype of Adult Fragile X Gray Zone Allele Carriers: a Case Series. Cerebellum, 2016, 15, 623-631.	1.4	20
92	Warburg effect linked to cognitiveâ€executive deficits inFMR1premutation. FASEB Journal, 2016, 30, 3334-3351.	0.2	33
93	Immortalized Parkinson's Disease lymphocytes have enhanced mitochondrial respiratory activity. DMM Disease Models and Mechanisms, 2016, 9, 1295-1305.	1.2	40
94	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
95	Role of p53, Mitochondrial DNA Deletions, and Paternal Age in Autism: A Case-Control Study. Pediatrics, 2016, 137, .	1.0	18
96	Developmental profiles of infants with an FMR1 premutation. Journal of Neurodevelopmental Disorders, 2016, 8, 40.	1.5	21
97	The Molecular Biology of Premutation Expanded Alleles. , 2016, , 101-127.		0
98	Psychiatric disorders among women with theâ€‰fragile X premutation without children affected by fragile X syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1139-1147.	1.1	21
99	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
100	Aging in Fragile X Premutation Carriers. Cerebellum, 2016, 15, 587-594.	1.4	14
101	Phenobarbital use and neurological problems in FMR1 premutation carriers. NeuroToxicology, 2016, 53, 141-147.	1.4	20
102	FindingFMR1mosaicism in Fragile X syndrome. Expert Review of Molecular Diagnostics, 2016, 16, 501-507.	1.5	29
103	Genotype/Phenotype Relationships in FXTAS. , 2016, , 129-160.		1
104	Clinical and Molecular Assessment in a Female with Fragile X Syndrome and Tuberous Sclerosis. Journal of Genetic Disorders & Genetic Reports, 2016, 5, .	0.1	1
105	Methadone use in a male with the FMRI premutation and FXTAS. American Journal of Medical Genetics, Part A, 2015, 167, 1354-1359.	0.7	24
106	Axonal neuropathy in female carriers of the fragile X premutation with fragile xâ€“associated tremor ataxia syndrome. Muscle and Nerve, 2015, 52, 234-239.	1.0	5
107	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
108	Temporal dynamics of attentional selection in adult male carriers of the fragile X premutation allele and adult controls. Frontiers in Human Neuroscience, 2015, 9, 37.	1.0	4

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109	Molecular Diagnosis of Fragile X Syndrome in Subjects with Intellectual Disability of Unknown Origin: Implications of Its Prevalence in Regional Pakistan. PLoS ONE, 2015, 10, e0122213.	1.1	8
110	Identification of a male with fragile X syndrome through newborn screening. Intractable and Rare Diseases Research, 2015, 4, 198-202.	0.3	3
111	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
112	Mitochondrial Citrate Transporter-dependent Metabolic Signature in the 22q11.2 Deletion Syndrome. Journal of Biological Chemistry, 2015, 290, 23240-23253.	1.6	56
113	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
114	Maternal Consequences of the Detection of Fragile X Carriers in Newborn Screening. Pediatrics, 2015, 136, e433-e440.	1.0	14
115	Differential increases of specificFMR1mRNA isoforms in premutation carriers. Journal of Medical Genetics, 2015, 52, 42-52.	1.5	29
116	Advanced technologies for the molecular diagnosis of fragile X syndrome. Expert Review of Molecular Diagnostics, 2015, 15, 1465-1473.	1.5	32
117	Immune mediated disorders in women with a fragile X expansion and FXTAS. American Journal of Medical Genetics, Part A, 2015, 167, 190-197.	0.7	25
118	Screening Newborn Blood Spots for 22q11.2 Deletion Syndrome Using Multiplex Droplet Digital PCR. Clinical Chemistry, 2015, 61, 182-190.	1.5	26
119	Contraction of a Maternal Fragile X Mental Retardation 1 Premutation Allele. Journal of Medical Cases, 2015, 6, 547-553.	0.4	3
120	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
121	Immune Dysregulation as a Cause of Autoinflammation in Fragile X Premutation Carriers: Link between FMRI CGG Repeat Number and Decreased Cytokine Responses. PLoS ONE, 2014, 9, e94475.	1.1	26
122	Parent-delivered touchscreen intervention for children with fragile X syndrome. Intractable and Rare Diseases Research, 2014, 3, 166-177.	0.3	8
123	Distribution of AGG interruption patterns within nine world populations. Intractable and Rare Diseases Research, 2014, 3, 153-161.	0.3	20
124	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
125	Clinical and molecular implications of mosaicism in FMR1 full mutations. Frontiers in Genetics, 2014, 5, 318.	1.1	86
126	The development of cognitive control in children with chromosome 22q11.2 deletion syndrome. Frontiers in Psychology, 2014, 5, 566.	1.1	39

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127	The multiple molecular facets of fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 23.	1.5	36
128	Group I metabotropic glutamate receptor mediated dynamic immune dysfunction in children with fragile X syndrome. Journal of Neuroinflammation, 2014, 11, 110.	3.1	14
129	Mapping the deletion endpoints in individuals with 22q11.2 Deletion Syndrome by droplet digital PCR. BMC Medical Genetics, 2014, 15, 106.	2.1	25
130	Newborn Screening for Fragile X Syndrome. JAMA Neurology, 2014, 71, 355.	4.5	42
131	Methylation Analysis in Newborn Screening for Fragile X Syndromeâ€"Reply. JAMA Neurology, 2014, 71, 800.	4.5	1
132	Fragile X Premutation. Journal of Neurodevelopmental Disorders, 2014, 6, 22.	1.5	20
133	Genomic studies in fragile X premutation carriers. Journal of Neurodevelopmental Disorders, 2014, 6, 27.	1.5	24
134	A cross-sectional analysis of orienting of visuospatial attention in child and adult carriers of the fragile X premutation. Journal of Neurodevelopmental Disorders, 2014, 6, 45.	1.5	0
135	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
136	A family with two female siblings with compound heterozygous <scp>FMR1</scp> premutation alleles. Clinical Genetics, 2014, 85, 458-463.	1.0	3
137	Parkinsonism in fragile X-associated tremor/ataxia syndrome (FXTAS): Revisited. Parkinsonism and Related Disorders, 2014, 20, 456-459.	1.1	42
138	CGG allele size somatic mosaicism and methylation inFMR1premutation alleles. Journal of Medical Genetics, 2014, 51, 309-318.	1.5	76
139	The Autism Spectrum Disorders Stem Cell Resource at Children's Hospital of Orange County: Implications for Disease Modeling and Drug Discovery. Stem Cells Translational Medicine, 2014, 3, 1275-1286.	1.6	24
140	AGG interruptions and maternal age affect FMR1 CGG repeat allele stability during transmission. Journal of Neurodevelopmental Disorders, 2014, 6, 24.	1.5	94
141	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
142	Altered neural activity in the â€˜whenâ€™ pathway during temporal processing in fragile X premutation carriers. Behavioural Brain Research, 2014, 261, 240-248.	1.2	13
143	Association between macroorchidism and intelligence in FMR1 premutation carriers. American Journal of Medical Genetics, Part A, 2014, 164, 2206-2211.	0.7	8
144	Eye movements reveal impaired inhibitory control in adult male fragile X premutation carriers asymptomatic for FXTAS.. Neuropsychology, 2014, 28, 571-584.	1.0	14

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145	Decreased DGCR8 Expression and miRNA Dysregulation in Individuals with 22q11.2 Deletion Syndrome. PLoS ONE, 2014, 9, e103884.	1.1	64
146	Memantine for Fragile Xâ€“Associated Tremor/Ataxia Syndrome. Journal of Clinical Psychiatry, 2014, 75, 264-271.	1.1	44
147	Fragile X syndrome. Colombia Medica, 2014, 45, 190-8.	0.7	48
148	Fragile Xâ€“Associated Tremor/Ataxia Syndrome. JAMA Neurology, 2013, 70, 1022.	4.5	64
149	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
150	Identification of Expanded Alleles of the FMR1 Gene in the CHildhood Autism Risks from Genes and Environment (CHARGE) Study. Journal of Autism and Developmental Disorders, 2013, 43, 530-539.	1.7	12
151	Intranuclear inclusions in a fragile X mosaic male. Translational Neurodegeneration, 2013, 2, 10.	3.6	37
152	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
153	Phenotypes of hypofrontality in older female fragile x premutation carriers. Annals of Neurology, 2013, 74, n/a-n/a.	2.8	25
154	Newborn screening and cascade testing for FMR1 mutations. American Journal of Medical Genetics, Part A, 2013, 161, 59-69.	0.7	24
155	Altered neural activity of magnitude estimation processing in adults with the fragile X premutation. Journal of Psychiatric Research, 2013, 47, 1909-1916.	1.5	13
156	Sequencing the unsequenceable: Expanded CGG-repeat alleles of the fragile X gene. Genome Research, 2013, 23, 121-128.	2.4	191
157	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
158	Transmission of an FMR1 premutation allele in a large family identified through newborn screening: the role of AGG interruptions. Journal of Human Genetics, 2013, 58, 553-559.	1.1	13
159	Global increases in both common and rare copy number load associated with autism. Human Molecular Genetics, 2013, 22, 2870-2880.	1.4	56
160	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
161	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
162	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

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163	De novo microduplication of the FMR1 gene in a patient with developmental delay, epilepsy and hyperactivity. European Journal of Human Genetics, 2012, 20, 1197-1200.	1.4	25
164	Clustered burst firing in FMR1 premutation hippocampal neurons: amelioration with allopregnanolone. Human Molecular Genetics, 2012, 21, 2923-2935.	1.4	92
165	Age-Dependent Structural Connectivity Effects in Fragile X Premutation. Archives of Neurology, 2012, 69, 482-9.	4.9	51
166	Identification of Expanded Alleles of theFMR1Gene Among High-Risk Population in Indonesia by Using Blood Spot Screening. Genetic Testing and Molecular Biomarkers, 2012, 16, 162-166.	0.3	9
167	Prepulse inhibition in patients with fragile X-associated tremor ataxia syndrome. Neurobiology of Aging, 2012, 33, 1045-1053.	1.5	17
168	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
169	The Fragile X-Associated Tremor Ataxia Syndrome. Results and Problems in Cell Differentiation, 2012, 54, 337-357.	0.2	26
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