Flora Tassone

List of Publications by Year in Descending Order

303 papers	16,809 citations	70 h-index	120 g-index
310 ext. papers	18,775 ext. citations	5.5 avg, IF	6.26 L-index

#	Paper	IF	Citations
303	Clinical and Molecular Correlates of Abnormal Changes in the Cerebellum and Globus Pallidus in Fragile X Premutation.. *Frontiers in Neurology*, **2022**, 13, 797649	4.1	0
302	Neuropsychological changes in FMR1 premutation carriers and onset of fragile X-associated tremor/ataxia syndrome.. *Journal of Neurodevelopmental Disorders*, **2022**, 14, 23	4.6	1
301	Prosaccade and Antisaccade Behavior in Fragile X-Associated Tremor/Ataxia Syndrome Progression.. *Movement Disorders Clinical Practice*, **2022**, 9, 473-478	2.2	
300	Increased Pain Symptomatology Among Females vs. Males With Fragile X-Associated Tremor/Ataxia Syndrome.. *Frontiers in Psychiatry*, **2021**, 12, 762915	5	1
299	EEG Signal Complexity Is Reduced During Resting-State in Fragile X Syndrome. *Frontiers in Psychiatry*, **2021**, 12, 716707	5	1
298	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	5	1
297	Delineating the Relationships Between Motor, Cognitive-Executive and Psychiatric Symptoms in Female Premutation Carriers.. *Frontiers in Psychiatry*, **2021**, 12, 742929	5	
296	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. *Molecular Psychiatry*, **2021**, 26, 4496-4510	15.1	39
295	Cerebral Microbleeds in Fragile X-Associated Tremor/Ataxia Syndrome. *Movement Disorders*, **2021**, 36, 1935-1943	7	4
294	'Essential Tremor' Phenotype in FMR1 Premutation/Gray Zone Sibling Series: Exploring Possible Genetic Modifiers. *Twin Research and Human Genetics*, **2021**, 24, 95-102	2.2	1
293	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	3.8	1
292	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	7.8	3
291	Mosaicism in Fragile X syndrome: A family case series. *Journal of Intellectual Disabilities*, **2021**, 17446295218995346	2.8	
290	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	1.7	2
289	Hypermobile Ehlers-Danlos syndrome (hEDS) phenotype in fragile X premutation carriers: case series. *Journal of Medical Genetics*, **2021**,	5.8	3
288	Inequities in diagnosis of Fragile X syndrome in Colombia. *Journal of Applied Research in Intellectual Disabilities*, **2021**, 34, 830-839	2.2	2
287	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	5	

286	Case Report: Coexistence of Alzheimer-Type Neuropathology in Fragile X-Associated Tremor Ataxia Syndrome. *Frontiers in Neuroscience*, **2021**, 15, 720253	5.1	2
285	FMRP Levels in Human Peripheral Blood Leukocytes Correlates with Intellectual Disability. *Diagnostics*, **2021**, 11,	3.8	1
284	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	3.1	2
283	Metabolic profiling reveals dysregulated lipid metabolism and potential biomarkers associated with the development and progression of Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). *FASEB Journal*, **2020**, 34, 16676-16692	0.9	6
282	Blood-Based Biomarkers Predictive of Metformin Target Engagement in Fragile X Syndrome. *Brain Sciences*, **2020**, 10,	3.4	3
281	Parkinsonism Versus Concomitant Parkinson's Disease in Fragile X-Associated Tremor/Ataxia Syndrome. *Movement Disorders Clinical Practice*, **2020**, 7, 413-418	2.2	8
280	Rapidly Progressing Neurocognitive Disorder in a Male with FXTAS and Alzheimer's Disease. *Clinical Interventions in Aging*, **2020**, 15, 285-292	4	3
279	Elevated FMR1-mRNA and lowered FMRP - A double-hit mechanism for psychiatric features in men with FMR1 premutations. *Translational Psychiatry*, **2020**, 10, 205	8.6	10
278	FMR1 locus isoforms: potential biomarker candidates in fragile X-associated tremor/ataxia syndrome (FXTAS). *Scientific Reports*, **2020**, 10, 11099	4.9	3
277	Developmental aspects of FXAND in a man with the FMR1 premutation. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1050	2.3	4
276	Molecular Biomarkers Predictive of Sertraline Treatment Response in Young Children With Autism Spectrum Disorder. *Frontiers in Genetics*, **2020**, 11, 308	4.5	1
275	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	2.4	2
274	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	11	24
273	Urine-Derived Epithelial Cell Lines: A New Tool to Model Fragile X Syndrome (FXS). *Cells*, **2020**, 9,	7.9	1
272	The emerging molecular mechanisms for mitochondrial dysfunctions in FXTAS. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2020**, 1866, 165918	6.9	5
271	Characterization of the Metabolic, Clinical and Neuropsychological Phenotype of Female Carriers of the Premutation in the X-Linked Gene. *Frontiers in Molecular Biosciences*, **2020**, 7, 578640	5.6	7
270	Cortical gyrification and its relationships with molecular measures and cognition in children with the FMR1 premutation. *Scientific Reports*, **2020**, 10, 16059	4.9	2
269	Women with Fragile X-associated Tremor/Ataxia Syndrome. *Movement Disorders Clinical Practice*, **2020**, 7, 910-919	2.2	8

268	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. *American Journal of Human Genetics*, **2020**, 107, 555-563	11	6
267	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	5.6	5
266	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	4.6	22
265	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	5.6	8
264	Metformin treatment in young children with fragile X syndrome. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e956	2.3	20
263	Total and Regional White Matter Lesions Are Correlated With Motor and Cognitive Impairments in Carriers of the Premutation. *Frontiers in Neurology*, **2019**, 10, 832	4.1	9
262	Cognitive and behavioral improvement in adults with fragile X syndrome treated with metformin-two cases. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00745	2.3	11
261	Molecular Biomarkers in Fragile X Syndrome. *Brain Sciences*, **2019**, 9,	3.4	11
260	Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). *Methods in Molecular Biology*, **2019**, 1942, 173-189	1.4	4
259	Repeat Instability in the Fragile X-Related Disorders: Lessons from a Mouse Model. *Brain Sciences*, **2019**, 9,	3.4	10
258	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	6.9	16
257	Fragile X- associated Neuropsychiatric Disorders: A Case Report. *Future Neurology*, **2019**, 14,	1.5	5
256	A Randomized Controlled Trial of Sertraline in Young Children With Autism Spectrum Disorder. *Frontiers in Psychiatry*, **2019**, 10, 810	5	11
255	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	2.3	6
254	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	6.2	18
253	Increased severity of fragile X spectrum disorders in the agricultural community of Ricaurte, Colombia. *International Journal of Developmental Neuroscience*, **2019**, 72, 1-5	2.7	5
252	Genetic cluster of fragile X syndrome in a Colombian district. *Journal of Human Genetics*, **2018**, 63, 509-516	4.3	18
251	Age- and CGG repeat-related slowing of manual movement in fragile X carriers: A prodrome of fragile X-associated tremor ataxia syndrome?. *Movement Disorders*, **2018**, 33, 628-636	7	11

250	Protein synthesis levels are increased in a subset of individuals with fragile X syndrome. *Human Molecular Genetics*, **2018**, 27, 2039-2051	5.6	31
249	Middle Cerebellar Peduncle Width-A Novel MRI Biomarker for FXTAS?. *Frontiers in Neuroscience*, **2018**, 12, 379	5.1	10
248	Fentanyl overdose in a female with the premutation and FXTAS **2018**, 1,		3
247	Rare FMR1 gene mutations causing fragile X syndrome: A review. *American Journal of Medical Genetics, Part A*, **2018**, 176, 11-18	2.5	41
246	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	2.6	12
245	The Spectrum of Neurological and White Matter Changes and Premutation Status Categories of Older Male Carriers of the Alleles Are Linked to Genetic (CGG and FMR1 mRNA) and Cellular Stress (AMPK) Markers. *Frontiers in Genetics*, **2018**, 9, 531	4.5	5
244	Microglial cell activation and senescence are characteristic of the pathology FXTAS. *Movement Disorders*, **2018**, 33, 1887-1894	7	13
243	Cognitive Deficits and Associated ERP N400 Abnormalities in FXTAS With Parkinsonism. *Frontiers in Genetics*, **2018**, 9, 327	4.5	
242	Assessment of Molecular Measures in Non-FXTAS Male Premutation Carriers. *Frontiers in Genetics*, **2018**, 9, 302	4.5	2
241	Impact of Premutation on Neurobehavior and Bioenergetics in Young Monozygotic Twins. *Frontiers in Genetics*, **2018**, 9, 338	4.5	11
240	Presence of Middle Cerebellar Peduncle Sign in Premutation Carriers Without Tremor and Ataxia. *Frontiers in Neurology*, **2018**, 9, 695	4.1	11
239	Curvilinear Association Between Language Disfluency and CGG Repeat Size Across the Normal, Intermediate, and Premutation Range. *Frontiers in Genetics*, **2018**, 9, 344	4.5	17
238	Evidence for the role of FMR1 gray zone alleles as a risk factor for parkinsonism in females. *Movement Disorders*, **2018**, 33, 1178-1181	7	8
237	Disease-Associated Short Tandem Repeats Co-localize with Chromatin Domain Boundaries. *Cell*, **2018**, 175, 224-238.e15	56.2	99
236	Translation of Expanded CGG Repeats into FMRpolyG Is Pathogenic and May Contribute to Fragile X Tremor Ataxia Syndrome. *Neuron*, **2017**, 93, 331-347	13.9	131
235	Molecular biomarkers predictive of sertraline treatment response in young children with fragile X syndrome. *Brain and Development*, **2017**, 39, 483-492	2.2	20
234	Clinical and molecular correlates in fragile X premutation females. *ENeurologicalSci*, **2017**, 7, 49-56	2.1	7
233	Fragile X Newborn Screening: Lessons Learned From a Multisite Screening Study. *Pediatrics*, **2017**, 139, S216-S225	7.4	17

232	Molecular analyses of neurogenic defects in a human pluripotent stem cell model of fragile X syndrome. *Brain*, **2017**, 140, 582-598	11.2	41
231	Abnormal trajectories in cerebellum and brainstem volumes in carriers of the fragile X premutation. *Neurobiology of Aging*, **2017**, 55, 11-19	5.6	32
230	Fragile X syndrome. *Nature Reviews Disease Primers*, **2017**, 3, 17065	51.1	257
229	Nested Inversion Polymorphisms Predispose Chromosome 22q11.2 to Meiotic Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 616-622	11	6
228	A randomized double-blind, placebo-controlled trial of ganaxolone in children and adolescents with fragile X syndrome. *Journal of Neurodevelopmental Disorders*, **2017**, 9, 26	4.6	44
227	Concomitant occurrence of FXTAS and clinically defined sporadic inclusion body myositis: report of two cases. *Croatian Medical Journal*, **2017**, 58, 310-315	1.6	3
226	Altered expression of the FMR1 splicing variants landscape in premutation carriers. *Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms*, **2017**, 1860, 1117-1126	6	22
225	Size and methylation mosaicism in males with Fragile X syndrome. *Expert Review of Molecular Diagnostics*, **2017**, 17, 1023-1032	3.8	23
224	Reduced vagal tone in women with the premutation is associated with mRNA but not depression or anxiety. *Journal of Neurodevelopmental Disorders*, **2017**, 9, 16	4.6	10
223	Children With Fragile X Syndrome Display Threat-Specific Biases Toward Emotion. *Biological Psychiatry: Cognitive Neuroscience and Neuroimaging*, **2017**, 2, 487-492	3.4	9
222	Down Syndrome and Fragile X Syndrome in a Colombian Woman: Case Report. *Journal of Applied Research in Intellectual Disabilities*, **2017**, 30, 970-974	2.2	3
221	Novel Blood Biomarkers Are Associated with White Matter Lesions in Fragile X- Associated Tremor/Ataxia Syndrome. *Neurodegenerative Diseases*, **2017**, 17, 22-30	2.3	13
220	Molecular Diagnostics and Genetic Counseling in Fragile X Syndrome and FMR1-Associated Disorders **2017**, 41-55		0
219	Prenatal Diagnosis of Fragile X: Can a Full Mutation Allele in the Gene Contract to a Normal Size?. *Frontiers in Genetics*, **2017**, 8, 158	4.5	6
218	Developmental profiles of infants with an premutation. *Journal of Neurodevelopmental Disorders*, **2016**, 8, 40	4.6	13
217	The Molecular Biology of Premutation Expanded Alleles **2016**, 101-127		
216	Psychiatric disorders among women with the fragile X premutation without children affected by fragile X syndrome. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2016**, 171, 1139-1147	3.5	16
215	Plasma metabolic profile delineates roles for neurodegeneration, pro-inflammatory damage and mitochondrial dysfunction in the FMR1 premutation. *Biochemical Journal*, **2016**, 473, 3871-3888	3.8	26

214	Aging in Fragile X Premutation Carriers. *Cerebellum*, **2016**, 15, 587-94	4.3	7
213	Phenobarbital use and neurological problems in FMR1 premutation carriers. *NeuroToxicology*, **2016**, 53, 141-147	4.4	14
212	Finding FMR1 mosaicism in Fragile X syndrome. *Expert Review of Molecular Diagnostics*, **2016**, 16, 501-7	3.8	14
211	Clinical and Molecular Assessment in a Female with Fragile X Syndrome and Tuberous Sclerosis **2016**, 5,		1
210	Genotype/Phenotype Relationships in FXTAS **2016**, 129-160		1
209	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	6.2	40
208	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	5.1	21
207	Plasma Biomarkers for Monitoring Brain Pathophysiology in FMR1 Premutation Carriers. *Frontiers in Molecular Neuroscience*, **2016**, 9, 71	6.1	17
206	Alcohol use dependence in fragile X syndrome. *Intractable and Rare Diseases Research*, **2016**, 5, 207-13	1.4	7
205	Clinical Phenotype of Adult Fragile X Gray Zone Allele Carriers: a Case Series. *Cerebellum*, **2016**, 15, 623-31	4.3	15
204	Warburg effect linked to cognitive-executive deficits in FMR1 premutation. *FASEB Journal*, **2016**, 30, 3334-3351	0.9	23
203	Immortalized Parkinson's disease lymphocytes have enhanced mitochondrial respiratory activity. *DMM Disease Models and Mechanisms*, **2016**, 9, 1295-1305	4.1	26
202	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	11.5	59
201	Role of p53, Mitochondrial DNA Deletions, and Paternal Age in Autism: A Case-Control Study. *Pediatrics*, **2016**, 137,	7.4	9
200	Mitochondrial Citrate Transporter-dependent Metabolic Signature in the 22q11.2 Deletion Syndrome. *Journal of Biological Chemistry*, **2015**, 290, 23240-53	5.4	34
199	Dysregulated ADAM10-Mediated Processing of APP during a Critical Time Window Leads to Synaptic Deficits in Fragile X Syndrome. *Neuron*, **2015**, 87, 382-98	13.9	49
198	Maternal Consequences of the Detection of Fragile X Carriers in Newborn Screening. *Pediatrics*, **2015**, 136, e433-40	7.4	8
197	Differential increases of specific FMR1 mRNA isoforms in premutation carriers. *Journal of Medical Genetics*, **2015**, 52, 42-52	5.8	22

196	Advanced technologies for the molecular diagnosis of fragile X syndrome. *Expert Review of Molecular Diagnostics*, **2015**, 15, 1465-73	3.8	21
195	Immune mediated disorders in women with a fragile X expansion and FXTAS. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 190-7	2.5	18
194	Screening newborn blood spots for 22q11.2 deletion syndrome using multiplex droplet digital PCR. *Clinical Chemistry*, **2015**, 61, 182-90	5.5	22
193	Methadone use in a male with the FMRI premutation and FXTAS. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1354-9	2.5	19
192	Axonal neuropathy in female carriers of the fragile X premutation with fragile x-associated tremor ataxia syndrome. *Muscle and Nerve*, **2015**, 52, 234-9	3.4	4
191	High functioning male with fragile X syndrome and fragile X-associated tremor/ataxia syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2154-61	2.5	21
190	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	3.3	3
189	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	3.7	7
188	Identification of a male with fragile X syndrome through newborn screening. *Intractable and Rare Diseases Research*, **2015**, 4, 198-202	1.4	3
187	Selected vitamin D metabolic gene variants and risk for autism spectrum disorder in the CHARGE Study. *Early Human Development*, **2015**, 91, 483-9	2.2	39
186	Anxiety disorders in fragile X premutation carriers: Preliminary characterization of probands and non-probands. *Intractable and Rare Diseases Research*, **2015**, 4, 123-30	1.4	30
185	Parkinsonism in fragile X-associated tremor/ataxia syndrome (FXTAS): revisited. *Parkinsonism and Related Disorders*, **2014**, 20, 456-9	3.6	32
184	CGG allele size somatic mosaicism and methylation in FMR1 premutation alleles. *Journal of Medical Genetics*, **2014**, 51, 309-18	5.8	60
183	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, 1275-86	6.9	21
182	AGG interruptions and maternal age affect FMR1 CGG repeat allele stability during transmission. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 24	4.6	72
181	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, 1189-97	5.6	30
180	Altered neural activity in the 'when' pathway during temporal processing in fragile X premutation carriers. *Behavioural Brain Research*, **2014**, 261, 240-8	3.4	12
179	Association between macroorchidism and intelligence in FMR1 premutation carriers. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2206-11	2.5	7

178	Eye movements reveal impaired inhibitory control in adult male fragile X premutation carriers asymptomatic for FXTAS. *Neuropsychology*, **2014**, 28, 571-584	3.8	10
177	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	3.7	20
176	Parent-delivered touchscreen intervention for children with fragile X syndrome. *Intractable and Rare Diseases Research*, **2014**, 3, 166-77	1.4	5
175	Distribution of AGG interruption patterns within nine world populations. *Intractable and Rare Diseases Research*, **2014**, 3, 153-61	1.4	15
174	CNS expression of murine fragile X protein (FMRP) as a function of CGG-repeat size. *Human Molecular Genetics*, **2014**, 23, 3228-38	5.6	53
173	Clinical and molecular implications of mosaicism in FMR1 full mutations. *Frontiers in Genetics*, **2014**, 5, 318	4.5	65
172	The development of cognitive control in children with chromosome 22q11.2 deletion syndrome. *Frontiers in Psychology*, **2014**, 5, 566	3.4	34
171	The multiple molecular facets of fragile X-associated tremor/ataxia syndrome. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 23	4.6	30
170	Group I metabotropic glutamate receptor mediated dynamic immune dysfunction in children with fragile X syndrome. *Journal of Neuroinflammation*, **2014**, 11, 110	10.1	10
169	Mapping the deletion endpoints in individuals with 22q11.2 deletion syndrome by droplet digital PCR. *BMC Medical Genetics*, **2014**, 15, 106	2.1	20
168	Newborn screening for fragile X syndrome. *JAMA Neurology*, **2014**, 71, 355-9	17.2	30
167	Methylation analysis in newborn screening for fragile X syndrome--reply. *JAMA Neurology*, **2014**, 71, 800-1	17.2	1
166	Fragile x premutation. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 22	4.6	12
165	Genomic studies in fragile X premutation carriers. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 27	4.6	18
164	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	4.6	
163	Fear-specific amygdala function in children and adolescents on the fragile x spectrum: a dosage response of the FMR1 gene. *Cerebral Cortex*, **2014**, 24, 600-13	5.1	27
162	A family with two female siblings with compound heterozygous FMR1 premutation alleles. *Clinical Genetics*, **2014**, 85, 458-63	4	3
161	Decreased DGCR8 expression and miRNA dysregulation in individuals with 22q11.2 deletion syndrome. *PLoS ONE*, **2014**, 9, e103884	3.7	48

160	Fragile X syndrome. *Colombia Medica*, **2014**, 45, 190-8	1.5	43
159	Memantine for fragile X-associated tremor/ataxia syndrome: a randomized, double-blind, placebo-controlled trial. *Journal of Clinical Psychiatry*, **2014**, 75, 264-71	4.6	35
158	Fragile X-associated tremor/ataxia syndrome: influence of the FMR1 gene on motor fiber tracts in males with normal and premutation alleles. *JAMA Neurology*, **2013**, 70, 1022-9	17.2	55
157	High MMP-9 activity levels in fragile X syndrome are lowered by minocycline. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1897-903	2.5	113
156	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, 530-9	4.6	9
155	Intranuclear inclusions in a fragile X mosaic male. *Translational Neurodegeneration*, **2013**, 2, 10	10.3	31
154	Sequestration of DROSHA and DGCR8 by expanded CGG RNA repeats alters microRNA processing in fragile X-associated tremor/ataxia syndrome. *Cell Reports*, **2013**, 3, 869-80	10.6	185
153	Phenotypes of hypofrontality in older female fragile X premutation carriers. *Annals of Neurology*, **2013**, 74, 275-83	9.4	22
152	Newborn screening and cascade testing for FMR1 mutations. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 59-69	2.5	19
151	Altered neural activity of magnitude estimation processing in adults with the fragile X premutation. *Journal of Psychiatric Research*, **2013**, 47, 1909-16	5.2	12
150	Sequencing the unsequenceable: expanded CGG-repeat alleles of the fragile X gene. *Genome Research*, **2013**, 23, 121-8	9.7	148
149	Fragile X AGG analysis provides new risk predictions for 45-69 repeat alleles. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 771-8	2.5	91
148	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, 553-9	4.3	10
147	Global increases in both common and rare copy number load associated with autism. *Human Molecular Genetics*, **2013**, 22, 2870-80	5.6	50
146	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, 147-55	2.4	183
145	Neural substrates of executive dysfunction in fragile X-associated tremor/ataxia syndrome (FXTAS): a brain potential study. *Cerebral Cortex*, **2013**, 23, 2657-66	5.1	22
144	Enhanced asynchronous Ca(2+) oscillations associated with impaired glutamate transport in cortical astrocytes expressing Fmr1 gene premutation expansion. *Journal of Biological Chemistry*, **2013**, 288, 13831-41	5.4	37
143	Altered mTOR signaling and enhanced CYFIP2 expression levels in subjects with fragile X syndrome. *Genes, Brain and Behavior*, **2012**, 11, 332-41	3.6	138

142	Neuropathological, clinical and molecular pathology in female fragile X premutation carriers with and without FXTAS. *Genes, Brain and Behavior*, **2012**, 11, 577-85	3.6	104
141	Fragile X-associated tremor ataxia syndrome in FMR1 gray zone allele carriers. *Movement Disorders*, **2012**, 27, 296-300	7	60
140	Identification of expanded alleles of the FMR1 gene among high-risk population in Indonesia by using blood spot screening. *Genetic Testing and Molecular Biomarkers*, **2012**, 16, 162-6	1.6	9
139	Prepulse inhibition in patients with fragile X-associated tremor ataxia syndrome. *Neurobiology of Aging*, **2012**, 33, 1045-53	5.6	16
138	AGG interruptions within the maternal FMR1 gene reduce the risk of offspring with fragile X syndrome. *Genetics in Medicine*, **2012**, 14, 729-36	8.1	126
137	The fragile X-associated tremor ataxia syndrome. *Results and Problems in Cell Differentiation*, **2012**, 54, 337-57	1.4	21
136	Immune-mediated disorders among women carriers of fragile X premutation alleles. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2473-81	2.5	74
135	Young adult male carriers of the fragile X premutation exhibit genetically modulated impairments in visuospatial tasks controlled for psychomotor speed. *Journal of Neurodevelopmental Disorders*, **2012**, 4, 26	4.6	14
134	Newborn, carrier, and early childhood screening recommendations for fragile X. *Pediatrics*, **2012**, 130, 1126-35	7.4	31
133	Early mitochondrial abnormalities in hippocampal neurons cultured from Fmr1 pre-mutation mouse model. *Journal of Neurochemistry*, **2012**, 123, 613-21	6	62
132	FMR1 CGG allele size and prevalence ascertained through newborn screening in the United States. *Genome Medicine*, **2012**, 4, 100	14.4	198
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