Flora Tassone

List of Publications by Citations

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	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	11	659
302	Fragile X premutation tremor/ataxia syndrome: molecular, clinical, and neuroimaging correlates. *American Journal of Human Genetics*, **2003**, 72, 869-78	11	624
301	Penetrance of the fragile X-associated tremor/ataxia syndrome in a premutation carrier population. *JAMA - Journal of the American Medical Association*, **2004**, 291, 460-9	27.4	462
300	Autism profiles of males with fragile X syndrome. *American Journal on Intellectual and Developmental Disabilites*, **2008**, 113, 427-38		308
299	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, 43-9	5.1	280
298	Sam68 sequestration and partial loss of function are associated with splicing alterations in FXTAS patients. *EMBO Journal*, **2010**, 29, 1248-61	13	275
297	FMRP expression as a potential prognostic indicator in fragile X syndrome. *American Journal of Medical Genetics Part A*, **1999**, 84, 250-261		274
296	Mitochondrial dysfunction in autism. *JAMA - Journal of the American Medical Association*, **2010**, 304, 2389-96	2.4	265
295	Fragile X-associated tremor/ataxia syndrome: clinical features, genetics, and testing guidelines. *Movement Disorders*, **2007**, 22, 2018-30, quiz 2140	7	261
294	Fragile X syndrome. *Nature Reviews Disease Primers*, **2017**, 3, 17065	51.1	257
293	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, 80-9	7	255
292	Autism spectrum disorders and attention-deficit/hyperactivity disorder in boys with the fragile X premutation. *Journal of Developmental and Behavioral Pediatrics*, **2006**, 27, S137-44	2.4	254
291	Expanded clinical phenotype of women with the FMR1 premutation. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1009-16	2.5	252
290	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	4.4	248
289	The FMR1 CGG repeat mouse displays ubiquitin-positive intranuclear neuronal inclusions; implications for the cerebellar tremor/ataxia syndrome. *Human Molecular Genetics*, **2003**, 12, 949-59	5.6	224
288	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, 3174-87	5.6	218
287	FMR1 RNA within the intranuclear inclusions of fragile X-associated tremor/ataxia syndrome (FXTAS). *RNA Biology*, **2004**, 1, 103-5	4.8	206

286	Fragile X syndrome: causes, diagnosis, mechanisms, and therapeutics. *Journal of Clinical Investigation*, **2012**, 122, 4314-22	15.9	203
285	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	5.5	203
284	FMR1 CGG allele size and prevalence ascertained through newborn screening in the United States. *Genome Medicine*, **2012**, 4, 100	14.4	198
283	Abnormal elevation of FMR1 mRNA is associated with psychological symptoms in individuals with the fragile X premutation. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2005**, 139B, 115-21	3.5	189
282	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
281	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
280	Transcription of the FMR1 gene in individuals with fragile X syndrome. *American Journal of Medical Genetics Part A*, **2000**, 97, 195-203		160
279	Elevated FMR1 mRNA in premutation carriers is due to increased transcription. *Rna*, **2007**, 13, 555-62	5.8	151
278	Sequencing the unsequenceable: expanded CGG-repeat alleles of the fragile X gene. *Genome Research*, **2013**, 23, 121-8	9.7	148
277	Cognitive profile of fragile X premutation carriers with and without fragile X-associated tremor/ataxia syndrome. *Neuropsychology*, **2008**, 22, 48-60	3.8	148
276	The fragile X premutation presenting as essential tremor. *Archives of Neurology*, **2003**, 60, 117-21		145
275	A Review of Fragile X Premutation Disorders. *Journal of Clinical Psychiatry*, **2009**, 70, 852-862	4.6	140
274	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, 3661-71	5.6	139
273	Fragile X males with unmethylated, full mutation trinucleotide repeat expansions have elevated levels of FMR1 messenger RNA. *American Journal of Medical Genetics Part A*, **2000**, 94, 232-6		139
272	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
271	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	5.1	138
270	Reduced FMR1 mRNA translation efficiency in fragile X patients with premutations. *Rna*, **2002**, 8, 1482-85.8		135
269	Expression profiling suggests underexpression of the GABA(A) receptor subunit delta in the fragile X knockout mouse model. *Neurobiology of Disease*, **2006**, 21, 346-57	7.5	133

268	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	4.6	132
267	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
266	Screening for expanded alleles of the FMR1 gene in blood spots from newborn males in a Spanish population. *Journal of Molecular Diagnostics*, **2009**, 11, 324-9	5.1	131
265	Psychiatric phenotype of the fragile X-associated tremor/ataxia syndrome (FXTAS) in males: newly described fronto-subcortical dementia. *Journal of Clinical Psychiatry*, **2006**, 67, 87-94	4.6	130
264	Murine hippocampal neurons expressing Fmr1 gene premutations show early developmental deficits and late degeneration. *Human Molecular Genetics*, **2010**, 19, 196-208	5.6	127
263	The Prader-Willi phenotype of fragile X syndrome. *Journal of Developmental and Behavioral Pediatrics*, **2007**, 28, 133-8	2.4	127
262	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
261	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, 566-9	3.5	118
260	Molecular and cognitive predictors of the continuum of autistic behaviours in fragile X. *Neuroscience and Biobehavioral Reviews*, **2007**, 31, 315-26	9	118
259	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
258	Progression of tremor and ataxia in male carriers of the FMR1 premutation. *Movement Disorders*, **2007**, 22, 203-6	7	113
257	Amygdala dysfunction in men with the fragile X premutation. *Brain*, **2007**, 130, 404-16	11.2	112
256	Fragile x syndrome. *Current Genomics*, **2011**, 12, 216-24	2.6	109
255	Impairment in the cognitive functioning of men with fragile X-associated tremor/ataxia syndrome (FXTAS). *Journal of the Neurological Sciences*, **2006**, 248, 227-33	3.2	107
254	Autistic spectrum disorder and the fragile X premutation. *Journal of Developmental and Behavioral Pediatrics*, **2004**, 25, 392-8	2.4	107
253	A neuropsychological investigation of male premutation carriers of fragile X syndrome. *Neuropsychologia*, **2004**, 42, 1934-47	3.2	105
252	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
251	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, 3067-74	5.6	102

250	A review of fragile X premutation disorders: expanding the psychiatric perspective. *Journal of Clinical Psychiatry*, **2009**, 70, 852-62	4.6	102
249	Expansion of an FMR1 grey-zone allele to a full mutation in two generations. *Journal of Molecular Diagnostics*, **2009**, 11, 306-10	5.1	99
248	Disease-Associated Short Tandem Repeats Co-localize with Chromatin Domain Boundaries. *Cell*, **2018**, 175, 224-238.e15	56.2	99
247	Increased prevalence of seizures in boys who were probands with the FMR1 premutation and co-morbid autism spectrum disorder. *Human Genetics*, **2012**, 131, 581-9	6.3	92
246	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
245	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, 467-79	14.3	91
244	Decreased fragile X mental retardation protein expression underlies amygdala dysfunction in carriers of the fragile X premutation. *Biological Psychiatry*, **2011**, 70, 859-65	7.9	84
243	Methylation of novel markers of fragile X alleles is inversely correlated with FMRP expression and FMR1 activation ratio. *Human Molecular Genetics*, **2010**, 19, 1618-32	5.6	81
242	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, 605-12	5.1	81
241	Variability in FMRP and early development in males with fragile X syndrome. *American Journal on Intellectual and Developmental Disabilites*, **2001**, 106, 16-27		80
240	Brief report: aggression and stereotypic behavior in males with fragile X syndrome--moderating secondary genes in a "single gene" disorder. *Journal of Autism and Developmental Disorders*, **2008**, 38, 184-9	4.6	79
239	Clustered burst firing in FMR1 premutation hippocampal neurons: amelioration with allopregnanolone. *Human Molecular Genetics*, **2012**, 21, 2923-35	5.6	77
238	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
237	A voxel-based morphometry study of grey matter loss in fragile X-associated tremor/ataxia syndrome. *Brain*, **2011**, 134, 863-78	11.2	74
236	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, 853-69	2.1	74
235	Impairment of executive cognitive functioning in males with fragile X-associated tremor/ataxia syndrome. *Movement Disorders*, **2007**, 22, 645-50	7	73
234	AGG interruptions and maternal age affect FMR1 CGG repeat allele stability during transmission. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 24	4.6	72
233	Cognitive, anxiety and mood disorders in the fragile X-associated tremor/ataxia syndrome. *General Hospital Psychiatry*, **2007**, 29, 349-56	5.6	70

232	Tissue heterogeneity of the FMR1 mutation in a high-functioning male with fragile X syndrome. *American Journal of Medical Genetics Part A*, **1999**, 84, 233-239		70
231	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	5.2	69
230	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*, **2007**, 44, 200-4	5.8	67
229	Young adult female fragile X premutation carriers show age- and genetically-modulated cognitive impairments. *Brain and Cognition*, **2011**, 75, 255-60	2.7	66
228	Diffusion tensor imaging in male premutation carriers of the fragile X mental retardation gene. *Movement Disorders*, **2011**, 26, 1329-36	7	66
227	Clinical and molecular implications of mosaicism in FMR1 full mutations. *Frontiers in Genetics*, **2014**, 5, 318	4.5	65
226	Abnormal nerve conduction features in fragile X premutation carriers. *Archives of Neurology*, **2008**, 65, 495-8		65
225	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	8.1	63
224	Early mitochondrial abnormalities in hippocampal neurons cultured from Fmr1 pre-mutation mouse model. *Journal of Neurochemistry*, **2012**, 123, 613-21	6	62
223	Clinical and neuropathologic findings in a woman with the FMR1 premutation and multiple sclerosis. *Archives of Neurology*, **2008**, 65, 1114-6		62
222	Secondary medical diagnosis in fragile X syndrome with and without autism spectrum disorder. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1911-6	2.5	62
221	Redistribution of transcription start sites within the FMR1 promoter region with expansion of the downstream CGG-repeat element. *Human Molecular Genetics*, **2004**, 13, 543-9	5.6	62
220	Plasma cytokine profiles in Fragile X subjects: is there a role for cytokines in the pathogenesis?. *Brain, Behavior, and Immunity*, **2010**, 24, 898-902	16.6	61
219	CGG allele size somatic mosaicism and methylation in FMR1 premutation alleles. *Journal of Medical Genetics*, **2014**, 51, 309-18	5.8	60
218	Fragile X-associated tremor ataxia syndrome in FMR1 gray zone allele carriers. *Movement Disorders*, **2012**, 27, 296-300	7	60
217	Fibroblast phenotype in male carriers of FMR1 premutation alleles. *Human Molecular Genetics*, **2010**, 19, 299-312	5.6	60
216	The effect of pre-mutation of X chromosome CGG trinucleotide repeats on brain anatomy. *Brain*, **2004**, 127, 2672-81	11.2	60
215	Brief report: Sensorimotor gating in idiopathic autism and autism associated with fragile X syndrome. *Journal of Autism and Developmental Disorders*, **2011**, 41, 248-53	4.6	59

214	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
213	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	3.3	57
212	CGG-repeat length threshold for FMR1 RNA pathogenesis in a cellular model for FXTAS. *Human Molecular Genetics*, **2011**, 20, 2161-70	5.6	56
211	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
210	Neuropathologic features in the hippocampus and cerebellum of three older men with fragile X syndrome. *Molecular Autism*, **2011**, 2, 2	6.5	54
209	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
208	A quantitative ELISA assay for the fragile x mental retardation 1 protein. *Journal of Molecular Diagnostics*, **2009**, 11, 281-9	5.1	52
207	Expression of the FMR1 gene. *Cytogenetic and Genome Research*, **2003**, 100, 124-8	1.9	52
206	Aging in fragile X syndrome. *Journal of Neurodevelopmental Disorders*, **2010**, 2, 70-76	4.6	51
205	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	4.1	51
204	Global increases in both common and rare copy number load associated with autism. *Human Molecular Genetics*, **2013**, 22, 2870-80	5.6	50
203	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
202	Magnetic resonance imaging study in older fragile X premutation male carriers. *Annals of Neurology* , **2005**, 58, 326-30	9.4	49
201	Age-dependent structural connectivity effects in fragile x premutation. *Archives of Neurology*, **2012**, 69, 482-9		48
200	Strong similarities of the FMR1 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		48
199	Decreased DGCR8 expression and miRNA dysregulation in individuals with 22q11.2 deletion syndrome. *PLoS ONE*, **2014**, 9, e103884	3.7	48
198	Fibromyalgia in fragile X mental retardation 1 gene premutation carriers. *Rheumatology*, **2011**, 50, 2233-6	5.9	47
197	Neuropathy as a presenting feature in fragile X-associated tremor/ataxia syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2256-60	2.5	45

196	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
195	Fragile X syndrome. *Colombia Medica*, **2014**, 45, 190-8	1.5	43
194	Sleep apnea in fragile X premutation carriers with and without FXTAS. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156B, 923-8	3.5	42
193	Molecular analyses of neurogenic defects in a human pluripotent stem cell model of fragile X syndrome. *Brain*, **2017**, 140, 582-598	11.2	41
192	Rare FMR1 gene mutations causing fragile X syndrome: A review. *American Journal of Medical Genetics, Part A*, **2018**, 176, 11-18	2.5	41
191	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
190	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
189	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. *Molecular Psychiatry*, **2021**, 26, 4496-4510	15.1	39
188	Hypertension in FMR1 premutation males with and without fragile X-associated tremor/ataxia syndrome (FXTAS). *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1304-9	2.5	38
187	MAOA, DBH, and SLC6A4 variants in CHARGE: a case-control study of autism spectrum disorders. *Autism Research*, **2011**, 4, 250-61	5.1	38
186	Early onset of neurological symptoms in fragile X premutation carriers exposed to neurotoxins. *NeuroToxicology*, **2010**, 31, 399-402	4.4	38
185	Differential usage of transcriptional start sites and polyadenylation sites in FMR1 premutation alleles. *Nucleic Acids Research*, **2011**, 39, 6172-85	20.1	38
184	cDNA selection from 10 Mb of chromosome 21 DNA: efficiency in transcriptional mapping and reflections of genome organization. *Human Molecular Genetics*, **1995**, 4, 1509-18	5.6	38
183	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
182	FMR1 gray-zone alleles: association with Parkinson's disease in women?. *Movement Disorders*, **2011**, 26, 1900-6	7	37
181	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, 171-7	2.7	37
180	Cognitive impairment in a 65-year-old male with the fragile X-associated tremor-ataxia syndrome (FXTAS). *Cognitive and Behavioral Neurology*, **2006**, 19, 165-71	1.6	35
179	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

178	Mitochondrial Citrate Transporter-dependent Metabolic Signature in the 22q11.2 Deletion Syndrome. *Journal of Biological Chemistry*, **2015**, 290, 23240-53	5.4	34
177	The development of cognitive control in children with chromosome 22q11.2 deletion syndrome. *Frontiers in Psychology*, **2014**, 5, 566	3.4	34
176	Abnormal trajectories in cerebellum and brainstem volumes in carriers of the fragile X premutation. *Neurobiology of Aging*, **2017**, 55, 11-19	5.6	32
175	Parkinsonism in fragile X-associated tremor/ataxia syndrome (FXTAS): revisited. *Parkinsonism and Related Disorders*, **2014**, 20, 456-9	3.6	32
174	Protein synthesis levels are increased in a subset of individuals with fragile X syndrome. *Human Molecular Genetics*, **2018**, 27, 2039-2051	5.6	31
173	Intranuclear inclusions in a fragile X mosaic male. *Translational Neurodegeneration*, **2013**, 2, 10	10.3	31
172	Newborn, carrier, and early childhood screening recommendations for fragile X. *Pediatrics*, **2012**, 130, 1126-35	7.4	31
171	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
170	The multiple molecular facets of fragile X-associated tremor/ataxia syndrome. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 23	4.6	30
169	Newborn screening for fragile X syndrome. *JAMA Neurology*, **2014**, 71, 355-9	17.2	30
168	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
167	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, 462-9	3.1	29
166	Autoimmune disease in mothers with the FMR1 premutation is associated with seizures in their children with fragile X syndrome. *Human Genetics*, **2010**, 128, 539-48	6.3	29
165	Tremor/Ataxia syndrome in fragile X carrier males. *Movement Disorders*, **2002**, 17, 744-745	7	29
164	A search for genes from the dark band regions of human chromosome 21. *Genomics*, **1995**, 27, 1-8	4.3	29
163	Enhanced manual and oral motor reaction time in young adult female fragile X premutation carriers. *Journal of the International Neuropsychological Society*, **2011**, 17, 746-50	3.1	28
162	Screen for excess FMR1 premutation alleles among males with parkinsonism. *Archives of Neurology*, **2007**, 64, 1002-6		28
161	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

160	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
159	Immortalized Parkinson's disease lymphocytes have enhanced mitochondrial respiratory activity. *DMM Disease Models and Mechanisms*, **2016**, 9, 1295-1305	4.1	26
158	Broad clinical involvement in a family affected by the fragile X premutation. *Journal of Developmental and Behavioral Pediatrics*, **2009**, 30, 544-51	2.4	25
157	Screening for the presence of FMR1 premutation alleles in women with parkinsonism. *Archives of Neurology*, **2009**, 66, 244-9		24
156	GRAND ROUNDS: an atypical progressive dementia in a male carrier of the fragile X premutation: an example of fragile X-associated tremor/ataxia syndrome. *Applied Neuropsychology*, **2005**, 12, 169-78		24
155	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
154	Size and methylation mosaicism in males with Fragile X syndrome. *Expert Review of Molecular Diagnostics*, **2017**, 17, 1023-1032	3.8	23
153	Translation of the FMR1 mRNA is not influenced by AGG interruptions. *Nucleic Acids Research*, **2009**, 37, 6896-904	20.1	23
152	Warburg effect linked to cognitive-executive deficits in FMR1 premutation. *FASEB Journal*, **2016**, 30, 3334-3351	0.9	23
151	Differential increases of specific FMR1 mRNA isoforms in premutation carriers. *Journal of Medical Genetics*, **2015**, 52, 42-52	5.8	22
150	Screening newborn blood spots for 22q11.2 deletion syndrome using multiplex droplet digital PCR. *Clinical Chemistry*, **2015**, 61, 182-90	5.5	22
149	Phenotypes of hypofrontality in older female fragile X premutation carriers. *Annals of Neurology*, **2013**, 74, 275-83	9.4	22
148	Altered expression of the FMR1 splicing variants landscape in premutation carriers. *Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms*, **2017**, 1860, 1117-1126	6	22
147	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
146	A Quantitative Assessment of Tremor and Ataxia in Female FMR1 Premutation Carriers Using CATSYS. *Current Gerontology and Geriatrics Research*, **2011**, 2011, 484713	2.9	22
145	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	3.7	22
144	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
143	Advanced technologies for the molecular diagnosis of fragile X syndrome. *Expert Review of Molecular Diagnostics*, **2015**, 15, 1465-73	3.8	21

142	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
141	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
140	The fragile X-associated tremor ataxia syndrome. *Results and Problems in Cell Differentiation*, **2012**, 54, 337-57	1.4	21
139	Linking the FMR1 alleles with small CGG expansions with neurodevelopmental disorders: preliminary data suggest an involvement of epigenetic mechanisms. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2306-10	2.5	21
138	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
137	Molecular biomarkers predictive of sertraline treatment response in young children with fragile X syndrome. *Brain and Development*, **2017**, 39, 483-492	2.2	20
136	Metformin treatment in young children with fragile X syndrome. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e956	2.3	20
135	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
134	Mapping the deletion endpoints in individuals with 22q11.2 deletion syndrome by droplet digital PCR. *BMC Medical Genetics*, **2014**, 15, 106	2.1	20
133	Abnormal N400 word repetition effects in fragile X-associated tremor/ataxia syndrome. *Brain*, **2010** , 133, 1438-50	11.2	20
132	De novo microduplication of the FMR1 gene in a patient with developmental delay, epilepsy and hyperactivity. *European Journal of Human Genetics*, **2012**, 20, 1197-200	5.3	20
131	Newborn screening and cascade testing for FMR1 mutations. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 59-69	2.5	19
130	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
129	Male carriers of the FMR1 premutation show altered hippocampal-prefrontal function during memory encoding. *Frontiers in Human Neuroscience*, **2012**, 6, 297	3.3	19
128	Map location, genomic organization and expression patterns of the human RED1 RNA editase. *Somatic Cell and Molecular Genetics*, **1997**, 23, 135-45		19
127	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
126	Genetic cluster of fragile X syndrome in a Colombian district. *Journal of Human Genetics*, **2018**, 63, 509-516	4.3	18
125	Genomic studies in fragile X premutation carriers. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 27	4.6	18

124	The role of AGG interruptions in the transcription of FMR1 premutation alleles. *PLoS ONE*, **2011**, 6, e21738	3.7	18
123	Reduced telomere length in older men with premutation alleles of the fragile X mental retardation 1 gene. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1543-6	2.5	18
122	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
121	Fragile X Newborn Screening: Lessons Learned From a Multisite Screening Study. *Pediatrics*, **2017**, 139, S216-S225	7.4	17
120	Tissue-specific methylation differences in a fragile X premutation carrier. *Clinical Genetics*, **1999**, 55, 346-51	4	17
119	Plasma Biomarkers for Monitoring Brain Pathophysiology in FMR1 Premutation Carriers. *Frontiers in Molecular Neuroscience*, **2016**, 9, 71	6.1	17
118	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
117	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
116	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
115	Prepulse inhibition in patients with fragile X-associated tremor ataxia syndrome. *Neurobiology of Aging*, **2012**, 33, 1045-53	5.6	16
114	Distribution of AGG interruption patterns within nine world populations. *Intractable and Rare Diseases Research*, **2014**, 3, 153-61	1.4	15
113	Two boys with fragile x syndrome and hepatic tumors. *Journal of Pediatric Hematology/Oncology*, **2008**, 30, 239-41	1.2	15
112	Clinical Phenotype of Adult Fragile X Gray Zone Allele Carriers: a Case Series. *Cerebellum*, **2016**, 15, 623-31	4.3	15
111	Phenobarbital use and neurological problems in FMR1 premutation carriers. *NeuroToxicology*, **2016**, 53, 141-147	4.4	14
110	Finding FMR1 mosaicism in Fragile X syndrome. *Expert Review of Molecular Diagnostics*, **2016**, 16, 501-7	3.8	14
109	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
108	Clinical phenotypes of a juvenile sibling pair carrying the fragile X premutation. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 519-25	2.5	14
107	Developmental profiles of infants with an premutation. *Journal of Neurodevelopmental Disorders*, **2016**, 8, 40	4.6	13

106	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
105	Construction of a 2.5-Mb integrated physical and gene map of distal 21q22.3. *Genomics*, **1998**, 49, 1-13	4.3	13
104	Microglial cell activation and senescence are characteristic of the pathology FXTAS. *Movement Disorders*, **2018**, 33, 1887-1894	7	13
103	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
102	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
101	Fragile x premutation. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 22	4.6	12
100	Polymerase chain reaction, nuclease digestion, and mass spectrometry based assay for the trinucleotide repeat status of the fragile X mental retardation 1 gene. *Analytical Chemistry*, **2009**, 81, 5533-40	7.8	12
99	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
98	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
97	Molecular Biomarkers in Fragile X Syndrome. *Brain Sciences*, **2019**, 9,	3.4	11
96	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
95	A Randomized Controlled Trial of Sertraline in Young Children With Autism Spectrum Disorder. *Frontiers in Psychiatry*, **2019**, 10, 810	5	11
94	Molecular and cytogenetic characterization of a Chinese hamster/human hybrid cell line containing a der (21)t(Ypter-->cenY::cen21-->21qter) chromosome. *Genomics*, **1993**, 15, 177-9	4.3	11
93	Impact of Premutation on Neurobehavior and Bioenergetics in Young Monozygotic Twins. *Frontiers in Genetics*, **2018**, 9, 338	4.5	11
92	Presence of Middle Cerebellar Peduncle Sign in Premutation Carriers Without Tremor and Ataxia. *Frontiers in Neurology*, **2018**, 9, 695	4.1	11
91	Repeat Instability in the Fragile X-Related Disorders: Lessons from a Mouse Model. *Brain Sciences*, **2019**, 9,	3.4	10
90	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
89	Middle Cerebellar Peduncle Width-A Novel MRI Biomarker for FXTAS?. *Frontiers in Neuroscience*, **2018**, 12, 379	5.1	10

88	Eye movements reveal impaired inhibitory control in adult male fragile X premutation carriers asymptomatic for FXTAS. *Neuropsychology*, **2014**, 28, 571-584	3.8	10
87	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
86	Group I metabotropic glutamate receptor mediated dynamic immune dysfunction in children with fragile X syndrome. *Journal of Neuroinflammation*, **2014**, 11, 110	10.1	10
85	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
84	Structures, sequence characteristics, and synteny relationships of the transcription factor E4TF1, the splicing factor U2AF35 and the cystathionine beta synthetase genes from Fugu rubripes. *Gene*, **1999**, 226, 211-23	3.8	10
83	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
82	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
81	Children With Fragile X Syndrome Display Threat-Specific Biases Toward Emotion. *Biological Psychiatry: Cognitive Neuroscience and Neuroimaging*, **2017**, 2, 487-492	3.4	9
80	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
79	Parkinsonism and cognitive decline in a fragile X mosaic male. *Movement Disorders*, **2010**, 25, 1523-4	7	9
78	Compound heterozygous female with fragile X syndrome. *American Journal of Medical Genetics Part A*, **1999**, 83, 318-321		9
77	Role of p53, Mitochondrial DNA Deletions, and Paternal Age in Autism: A Case-Control Study. *Pediatrics*, **2016**, 137,	7.4	9
76	Maternal Consequences of the Detection of Fragile X Carriers in Newborn Screening. *Pediatrics*, **2015**, 136, e433-40	7.4	8
75	Parkinsonism Versus Concomitant Parkinson's Disease in Fragile X-Associated Tremor/Ataxia Syndrome. *Movement Disorders Clinical Practice*, **2020**, 7, 413-418	2.2	8
74	Women with Fragile X-associated Tremor/Ataxia Syndrome. *Movement Disorders Clinical Practice*, **2020**, 7, 910-919	2.2	8
73	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
72	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
71	Clinical and molecular correlates in fragile X premutation females. *ENeurologicalSci*, **2017**, 7, 49-56	2.1	7

70	Aging in Fragile X Premutation Carriers. *Cerebellum*, **2016**, 15, 587-94	4.3	7
69	Association between macroorchidism and intelligence in FMR1 premutation carriers. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2206-11	2.5	7
68	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
67	Reduced telomere length in individuals with FMR1 premutations and full mutations. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1060-5	2.5	7
66	A fragile X sibship from a consanguineous family with a compound heterozygous female and partially methylated full mutation male. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1221-4	2.5	7
65	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
64	Alcohol use dependence in fragile X syndrome. *Intractable and Rare Diseases Research*, **2016**, 5, 207-13	1.4	7
63	FMRP expression as a potential prognostic indicator in fragile X syndrome **1999**, 84, 250		7
62	Nested Inversion Polymorphisms Predispose Chromosome 22q11.2 to Meiotic Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 616-622	11	6
61	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
60	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
59	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
58	Investigation of amygdala volume in men with the fragile X premutation. *Brain Imaging and Behavior*, **2011**, 5, 285-94	4.1	6
57	Covariate adjusted correlation analysis with application to FMR1 premutation female carrier data. *Biometrics*, **2009**, 65, 781-92	1.8	6
56	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. *American Journal of Human Genetics*, **2020**, 107, 555-563	11	6
55	Fragile X- associated Neuropsychiatric Disorders: A Case Report. *Future Neurology*, **2019**, 14,	1.5	5
54	Parent-delivered touchscreen intervention for children with fragile X syndrome. *Intractable and Rare Diseases Research*, **2014**, 3, 166-77	1.4	5
53	High-risk fragile x screening in Guatemala: use of a new blood spot polymerase chain reaction technique. *Genetic Testing and Molecular Biomarkers*, **2009**, 13, 855-9	1.6	5

52	The emerging molecular mechanisms for mitochondrial dysfunctions in FXTAS. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2020**, 1866, 165918	6.9	5
51	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
50	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
49	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
48	Tissue heterogeneity of the FMR1 mutation in a high-functioning male with fragile X syndrome **1999**, 84, 233		5
47	Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). *Methods in Molecular Biology*, **2019**, 1942, 173-189	1.4	4
46	Developmental aspects of FXAND in a man with the FMR1 premutation. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1050	2.3	4
45	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
44	Cerebral Microbleeds in Fragile X-Associated Tremor/Ataxia Syndrome. *Movement Disorders*, **2021**, 36, 1935-1943	7	4
43	Blood-Based Biomarkers Predictive of Metformin Target Engagement in Fragile X Syndrome. *Brain Sciences*, **2020**, 10,	3.4	3
42	Rapidly Progressing Neurocognitive Disorder in a Male with FXTAS and Alzheimer's Disease. *Clinical Interventions in Aging*, **2020**, 15, 285-292	4	3
41	FMR1 locus isoforms: potential biomarker candidates in fragile X-associated tremor/ataxia syndrome (FXTAS). *Scientific Reports*, **2020**, 10, 11099	4.9	3
40	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
39	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
38	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
37	Identification of a male with fragile X syndrome through newborn screening. *Intractable and Rare Diseases Research*, **2015**, 4, 198-202	1.4	3
36	A family with two female siblings with compound heterozygous FMR1 premutation alleles. *Clinical Genetics*, **2014**, 85, 458-63	4	3
35	Fentanyl overdose in a female with the premutation and FXTAS **2018**, 1,		3

34	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
33	Hypermobile Ehlers-Danlos syndrome (hEDS) phenotype in fragile X premutation carriers: case series. *Journal of Medical Genetics*, **2021**,	5.8	3
32	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
31	Highly polymorphic repeat marker within the beta-amyloid precursor protein gene. *Human Genetics* , **1994**, 93, 85-6	6.3	2
30	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
29	Cortical gyrification and its relationships with molecular measures and cognition in children with the FMR1 premutation. *Scientific Reports*, **2020**, 10, 16059	4.9	2
28	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
27	Inequities in diagnosis of Fragile X syndrome in Colombia. *Journal of Applied Research in Intellectual Disabilities*, **2021**, 34, 830-839	2.2	2
26	Assessment of Molecular Measures in Non-FXTAS Male Premutation Carriers. *Frontiers in Genetics*, **2018**, 9, 302	4.5	2
25	Case Report: Coexistence of Alzheimer-Type Neuropathology in Fragile X-Associated Tremor Ataxia Syndrome. *Frontiers in Neuroscience*, **2021**, 15, 720253	5.1	2
24	Molecular Biomarkers Predictive of Sertraline Treatment Response in Young Children With Autism Spectrum Disorder. *Frontiers in Genetics*, **2020**, 11, 308	4.5	1
23	Methylation analysis in newborn screening for fragile X syndrome--reply. *JAMA Neurology*, **2014**, 71, 800-1	17.2	1
22	Increased Pain Symptomatology Among Females vs. Males With Fragile X-Associated Tremor/Ataxia Syndrome.. *Frontiers in Psychiatry*, **2021**, 12, 762915	5	1
21	EEG Signal Complexity Is Reduced During Resting-State in Fragile X Syndrome. *Frontiers in Psychiatry*, **2021**, 12, 716707	5	1
20	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
19	Clinical and Molecular Assessment in a Female with Fragile X Syndrome and Tuberous Sclerosis **2016**, 5,		1
18	Genotype/Phenotype Relationships in FXTAS **2016**, 129-160		1
17	Urine-Derived Epithelial Cell Lines: A New Tool to Model Fragile X Syndrome (FXS). *Cells*, **2020**, 9,	7.9	1

16	'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
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	3.8	1
14	Mosaicism in Fragile X syndrome: A family case series. *Journal of Intellectual Disabilities*, **2021**, 17446295219953146	2.8	1
13	FMRP Levels in Human Peripheral Blood Leukocytes Correlates with Intellectual Disability. *Diagnostics*, **2021**, 11,	3.8	1
12	Genotype/Phenotype Relationships in FXTAS **2010**, 95-122		1
11	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
10	Molecular Diagnostics and Genetic Counseling in Fragile X Syndrome and FMR1-Associated Disorders **2017**, 41-55		0
9	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
8	The Molecular Biology of Premutation Expanded Alleles **2016**, 101-127		
7	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	
6	Response to letter: No evidence of paternal transmission of fragile X syndrome by Doris and Peter Steinbach. *American Journal of Medical Genetics, Part A*, **2005**, 136A, 109-110	2.5	
5	Delineating the Relationships Between Motor, Cognitive-Executive and Psychiatric Symptoms in Female Premutation Carriers.. *Frontiers in Psychiatry*, **2021**, 12, 742929	5	
4	The Molecular Biology of FXTAS **2010**, 77-93		
3	Cognitive Deficits and Associated ERP N400 Abnormalities in FXTAS With Parkinsonism. *Frontiers in Genetics*, **2018**, 9, 327	4.5	
2	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	
1	Prosaccade and Antisaccade Behavior in Fragile X-Associated Tremor/Ataxia Syndrome Progression.. *Movement Disorders Clinical Practice*, **2022**, 9, 473-478	2.2	