

Expanded clinical phenotype of women with the *FMR1*

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Testing for Fragile X Gene Mutations Throughout the Life Span. JAMA - Journal of the American Medical Association, 2008, 300, 2419.	3.8	33
2	Population Screening for Reproductive Risk for Single Gene Disorders in Australia: Now and the Future. Twin Research and Human Genetics, 2008, 11, 422-430.	0.3	18
3	The Fragile X Family of Disorders: A Model for Autism and Targeted Treatments. Current Pediatric Reviews, 2008, 4, 40-52.	0.4	83
4	Clinical and Neuropathologic Findings in a Woman With the FMR1 Premutation and Multiple Sclerosis. Archives of Neurology, 2008, 65, 1114-6.	4.9	68
5	RETIRED: Fragile X Testing in Obstetrics and Gynaecology in Canada. Journal of Obstetrics and Gynaecology Canada, 2008, 30, 837-841.	0.3	18
6	D'Épistage du X fragile en obstétrique-gynécologie au Canada. Journal of Obstetrics and Gynaecology Canada, 2008, 30, 842-846.	0.3	1
7	Treatment of fragile X-associated tremor ataxia syndrome (FXTAS) and related neurological problems. Clinical Interventions in Aging, 2008, Volume 3, 251-262.	1.3	122
8	Advances in the Treatment of Fragile X Syndrome. Pediatrics, 2009, 123, 378-390.	1.0	513
9	Evidence for RNA-mediated toxicity in the fragile X-associated tremor/ataxia syndrome. Future Neurology, 2009, 4, 785-798.	0.9	48
10	The quadruplex r(CGG)n destabilizing cationic porphyrin TMPyP4 cooperates with hnRNPs to increase the translation efficiency of fragile X premutation mRNA. Nucleic Acids Research, 2009, 37, 2712-2722.	6.5	69
11	Conversion disorder in women with the <i>FMR1</i> premutation. American Journal of Medical Genetics, Part A, 2009, 149A, 2501-2506.	0.7	15
12	Psychological symptoms correlate with reduced hippocampal volume in fragile X premutation carriers. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 775-785.	1.1	51
14	Penetrance of FMR1 premutation associated pathologies in fragile X syndrome families. European Journal of Human Genetics, 2009, 17, 1359-1362.	1.4	254
15	Fragile X: A Family of Disorders. Advances in Pediatrics, 2009, 56, 165-186.	0.5	92
16	Primary ovarian insufficiency: X chromosome defects and autoimmunity. Journal of Autoimmunity, 2009, 33, 35-41.	3.0	100
17	Broad Clinical Involvement in a Family Affected by the Fragile X Premutation. Journal of Developmental and Behavioral Pediatrics, 2009, 30, 544-551.	0.6	27
18	Fragile X-Associated Tremor/Ataxia Syndrome: Clinical Phenotype, Diagnosis, and Treatment. Journal of Investigative Medicine, 2009, 57, 830-836.	0.7	150
19	Expanded Clinical Phenotype of Women With the FMR1 Premutation. Yearbook of Obstetrics Gynecology and Women S Health, 2009, 2009, 5-6.	0.0	0

#	ARTICLE	IF	CITATIONS
20	Screening for the Presence of FMR1 Premutation Alleles in Women With Parkinsonism. Archives of Neurology, 2009, 66, 244-9.	4.9	27
21	Molecular Pathogenesis of Fragile X-Associated Tremor/Ataxia Syndrome. Journal of Investigative Medicine, 2009, 57, 825-829.	0.7	22
22	FRAGILE X SYNDROME AND ASSOCIATED DISORDERS IN ADULTHOOD. CONTINUUM Lifelong Learning in Neurology, 2009, 15, 32-49.	0.4	1
23	Improving Fragile X-Associated Tremor/Ataxia Syndrome Symptoms With Memantine and Venlafaxine. Journal of Clinical Psychopharmacology, 2010, 30, 642-644.	0.7	26
24	The FMR1 Gene as Regulator of Ovarian Recruitment and Ovarian Reserve. Obstetrical and Gynecological Survey, 2010, 65, 523-530.	0.2	35
26	Fragile X: Leading the Way for Targeted Treatments in Autism. Neurotherapeutics, 2010, 7, 264-274.	2.1	157
27	Autoimmune disease in mothers with the FMR1 premutation is associated with seizures in their children with fragile X syndrome. Human Genetics, 2010, 128, 539-548.	1.8	30
28	Aging in fragile X syndrome. Journal of Neurodevelopmental Disorders, 2010, 2, 70-76.	1.5	59
29	Tandem repeat polymorphisms: modulators of disease susceptibility and candidates for "missing heritability". Trends in Genetics, 2010, 26, 59-65.	2.9	137
30	The behavioral phenotype of <i>FMR1</i> mutations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 469-476.	0.7	98
31	Hyperintensity in the basis pontis: Atypical neuroradiological findings in a woman with FXTAS. Movement Disorders, 2010, 25, 649-650.	2.2	8
32	Fragile X and autism: Intertwined at the molecular level leading to targeted treatments. Molecular Autism, 2010, 1, 12.	2.6	204
33	Co-occurring diagnoses among <i>FMR1</i> premutation allele carriers. Clinical Genetics, 2010, 77, 374-381.	1.0	80
34	Clinical involvement in daughters of men with fragile X-associated tremor ataxia syndrome. Clinical Genetics, 2010, 78, 38-46.	1.0	54
35	Intergenerational effects of mutations in the fragile X mental retardation 1 gene. Fragile X: A model of X-linked mental retardation and neurodegeneration. , 0, , 3-18.		1
36	The fragile x-associated tremor and ataxia syndrome (FXTAS). Arquivos De Neuro-Psiquiatria, 2010, 68, 791-798.	0.3	14
37	Motor and mental dysfunction in mother-daughter transmitted FXTAS. Neurology, 2010, 75, 1370-1376.	1.5	26
38	Fibroblast phenotype in male carriers of FMR1 premutation alleles. Human Molecular Genetics, 2010, 19, 299-312.	1.4	66

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39	Murine hippocampal neurons expressing Fmr1 gene premutations show early developmental deficits and late degeneration. <i>Human Molecular Genetics</i> , 2010, 19, 196-208.	1.4	143
40	Assessment of patient and caregiver needs in fragile X-associated tremor/ataxia syndrome by utilizing Q-sort methodology. <i>Aging and Mental Health</i> , 2010, 14, 1000-1007.	1.5	12
41	Prenatal Carrier Testing for Fragile X: Counseling Issues and Challenges. <i>Obstetrics and Gynecology Clinics of North America</i> , 2010, 37, 61-70.	0.7	20
42	Early onset of neurological symptoms in fragile X premutation carriers exposed to neurotoxins. <i>NeuroToxicology</i> , 2010, 31, 399-402.	1.4	40
43	Plasma cytokine profiles in Fragile X subjects: Is there a role for cytokines in the pathogenesis?. <i>Brain, Behavior, and Immunity</i> , 2010, 24, 898-902.	2.0	73
44	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. <i>Journal of Molecular Diagnostics</i> , 2010, 12, 589-600.	1.2	166
45	Advances in understanding the molecular basis of FXTAS. <i>Human Molecular Genetics</i> , 2010, 19, R83-R89.	1.4	119
46	The Fragile X-associated Disorders: Time to Order Fragile X DNA Testing. <i>Biological Psychiatry</i> , 2011, 70, 802-803.	0.7	1
47	The Role of AGG Interruptions in the Transcription of FMR1 Premutation Alleles. <i>PLoS ONE</i> , 2011, 6, e21728.	1.1	24
48	Monoclonal Gammopathy of Undetermined Significance (MGUS) in a Man with Fragile X-associated Tremor/Ataxia Syndrome. <i>Case Reports in Genetics</i> , 2011, 2011, 1-5.	0.1	2
49	One world, one woman. <i>Menopause</i> , 2011, 18, 480-487.	0.8	4
50	Rare Intranuclear Inclusions in the Brains of 3 Older Adult Males With Fragile X Syndrome: Implications for the Spectrum of Fragile X-Associated Disorders. <i>Journal of Neuropathology and Experimental Neurology</i> , 2011, 70, 462-469.	0.9	33
51	Pain in adults with intellectual disabilities. <i>Pain</i> , 2011, 152, 971-974.	2.0	27
52	Fragile X syndrome: an overview. <i>The Obstetrician and Gynaecologist</i> , 2011, 13, 92-97.	0.2	1
53	Widespread non-central nervous system organ pathology in fragile X premutation carriers with fragile X-associated tremor/ataxia syndrome and CGG knock-in mice. <i>Acta Neuropathologica</i> , 2011, 122, 467-479.	3.9	102
54	FMR1 premutation and full mutation molecular mechanisms related to autism. <i>Journal of Neurodevelopmental Disorders</i> , 2011, 3, 211-224.	1.5	74
55	Fragile X analysis of 1112 prenatal samples from 1991 to 2010. <i>Prenatal Diagnosis</i> , 2011, 31, 925-931.	1.1	86
56	Clinical phenotypes of a juvenile sibling pair carrying the fragile X premutation. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 519-525.	0.7	15

#	ARTICLE	IF	CITATIONS
57	Terminal deletions of the long arm of chromosome X that include the FMR1 gene in female patients: A case series. , 2011, 155, 870-874.		16
58	The FRAXopathies: Definition, overview, and update. American Journal of Medical Genetics, Part A, 2011, 155, 1803-1816.	0.7	42
59	Conflicts regarding genetic counseling for fragile X syndrome screening: A survey of clinical geneticists and genetic counselors in Israel. American Journal of Medical Genetics, Part A, 2011, 155, 2154-2160.	0.7	3
60	Fibromyalgia in fragile X mental retardation 1 gene premutation carriers. Rheumatology, 2011, 50, 2233-2236.	0.9	51
61	A Quantitative Assessment of Tremor and Ataxia in Female <i>FMR1</i> Premutation Carriers Using CATSYS. Current Gerontology and Geriatrics Research, 2011, 2011, 1-7.	1.6	26
62	Cortisol response to behavior problems in FMR1 premutation mothers of adolescents and adults with fragile X syndrome. International Journal of Behavioral Development, 2012, 36, 53-61.	1.3	52
63	Genetic polymorphism studies in humans. Middle East Journal of Medical Genetics, 2012, 1, 57-63.	0.0	32
64	Differential sensitivity to life stress in FMR1 premutation carrier mothers of children with Fragile X Syndrome.. Health Psychology, 2012, 31, 612-622.	1.3	94
65	Capturing the fragile X premutation phenotypes: A collaborative effort across multiple cohorts.. Neuropsychology, 2012, 26, 156-164.	1.0	36
66	Fragile X-associated tremor/ataxia syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 103, 373-386.	1.0	38
67	Daily Health Symptoms of Mothers of Adolescents and Adults with Fragile X Syndrome and Mothers of Adolescents and Adults with Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2012, 42, 1836-1846.	1.7	55
68	Diagnosis of Fragile X Syndrome: A Qualitative Study of African American Families. Journal of Genetic Counseling, 2012, 21, 845-853.	0.9	11
69	Genetic Counseling and Testing for <i>FMR1</i> Gene Mutations: Practice Guidelines of the National Society of Genetic Counselors. Journal of Genetic Counseling, 2012, 21, 752-760.	0.9	61
70	Identification of Expanded Alleles of the <i>FMR1</i> Gene Among High-Risk Population in Indonesia by Using Blood Spot Screening. Genetic Testing and Molecular Biomarkers, 2012, 16, 162-166.	0.3	9
71	Investigation of C9orf72 in 4 Neurodegenerative Disorders. Archives of Neurology, 2012, 69, 1583.	4.9	89
72	Screening for the presence of FMR1 premutation alleles in a Spanish population with fibromyalgia. Clinical Rheumatology, 2012, 31, 1611-1615.	1.0	8
73	The Fragile X-Associated Tremor Ataxia Syndrome. Results and Problems in Cell Differentiation, 2012, 54, 337-357.	0.2	26
74	Immune-mediated disorders among women carriers of fragile X premutation alleles. American Journal of Medical Genetics, Part A, 2012, 158A, 2473-2481.	0.7	86

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75	Carriers of the fragile X mental retardation 1 (FMR1) premutation allele present with increased levels of cytokine IL-10. <i>Journal of Neuroinflammation</i> , 2012, 9, 238.	3.1	20
76	Fragile X syndrome: a pilot proton magnetic resonance spectroscopy study in premutation carriers. <i>Journal of Neurodevelopmental Disorders</i> , 2012, 4, 23.	1.5	3
77	Newborn, Carrier, and Early Childhood Screening Recommendations for Fragile X. <i>Pediatrics</i> , 2012, 130, 1126-1135.	1.0	39
78	Distribution and frequency of intranuclear inclusions in female CGG KI mice modeling the fragile X premutation. <i>Brain Research</i> , 2012, 1472, 124-137.	1.1	13
79	Behavioral Phenotype of Fragile X Syndrome in Adolescence and Adulthood. <i>American Journal on Intellectual and Developmental Disabilities</i> , 2012, 117, 1-17.	0.8	96
80	Unstable Mutations in the FMR1 Gene and the Phenotypes. <i>Advances in Experimental Medicine and Biology</i> , 2012, 769, 78-114.	0.8	52
81	FMR1 CGG allele size and prevalence ascertained through newborn screening in the United States. <i>Genome Medicine</i> , 2012, 4, 100.	3.6	258
82	Hypertension in <i>FMR1</i> premutation males with and without fragile X-associated tremor/ataxia syndrome (FXTAS). <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1304-1309.	0.7	41
83	Depression and anxiety symptoms among women who carry the <i>FMR1</i> premutation: Impact of raising a child with fragile X syndrome is moderated by <i>CRHR1</i> polymorphisms. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 549-559.	1.1	32
84	Prevalence of CGG expansions of the <i>FMR1</i> gene in a US population-based sample. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 589-597.	1.1	162
85	The FMR1 Premutation and Attention-Deficit Hyperactivity Disorder (ADHD): Evidence for a Complex Inheritance. <i>Behavior Genetics</i> , 2012, 42, 415-422.	1.4	41
86	Self-injurious behaviour in intellectual disability syndromes: evidence for aberrant pain signalling as a contributing factor. <i>Journal of Intellectual Disability Research</i> , 2012, 56, 441-452.	1.2	41
87	Neuropathological, clinical and molecular pathology in female fragile X premutation carriers with and without FXTAS. <i>Genes, Brain and Behavior</i> , 2012, 11, 577-585.	1.1	117
88	Fragile X-associated disorders: a clinical overview. <i>Journal of Neurology</i> , 2012, 259, 401-413.	1.8	91
89	Advances in clinical and molecular understanding of the FMR1 premutation and fragile X-associated tremor/ataxia syndrome. <i>Lancet Neurology</i> , The, 2013, 12, 786-798.	4.9	288
90	Fragile X syndrome: An aging perspective. <i>Developmental Disabilities Research Reviews</i> , 2013, 18, 68-74.	2.9	14
91	Fragile X Syndrome and X-linked Intellectual Disability. , 2013, , 1-27.		1
92	Phenotypes of hypofrontality in older female fragile x premutation carriers. <i>Annals of Neurology</i> , 2013, 74, n/a-n/a.	2.8	25

#	ARTICLE	IF	CITATIONS
93	Abnormal GABA-mediated and cerebellar inhibition in women with the fragile X premutation. <i>Journal of Neurophysiology</i> , 2013, 109, 1315-1322.	0.9	26
94	œœlt's about having the choiceœœ Stakeholder perceptions of population-based genetic carrier screening for fragile X syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 48-58.	0.7	25
95	Newborn screening and cascade testing for <i>FMR1</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 59-69.	0.7	24
96	Current controversies in prenatal diagnosis 1: screening for fragile X syndrome. <i>Prenatal Diagnosis</i> , 2013, 33, 6-8.	1.1	2
97	Cognitive-motor interference during postural control indicates at-risk cerebellar profiles in females with the FMR1 premutation. <i>Behavioural Brain Research</i> , 2013, 253, 329-336.	1.2	27
98	Neurobehavioural evidence for the involvement of the FMR1 gene in female carriers of fragile X syndrome. <i>Neuroscience and Biobehavioral Reviews</i> , 2013, 37, 522-547.	2.9	45
99	A Dual-Mode Single-Molecule Fluorescence Assay for the Detection of Expanded CGG Repeats in Fragile X Syndrome. <i>Molecular Biotechnology</i> , 2013, 53, 19-28.	1.3	10
100	The fragile X-associated tremor ataxia syndrome (<sc>FXTAS</sc>) in Indonesia. <i>Clinical Genetics</i> , 2013, 83, 263-268.	1.0	2
101	Fragile X-associated tremor/ataxia syndrome (FXTAS): pathology and mechanisms. <i>Acta Neuropathologica</i> , 2013, 126, 1-19.	3.9	142
102	Screening for the presence of FMR1 premutation alleles in women with fibromyalgia. <i>Gene</i> , 2013, 512, 305-308.	1.0	13
103	Caregiver Burden in Fragile X Families. <i>Current Psychiatry Reviews</i> , 2013, 9, 85-91.	0.9	12
104	Postpartum Depression in Women with the FMR1 Premutation. <i>Current Psychiatry Reviews</i> , 2013, 9, 72-77.	0.9	4
105	Fragile X Clinical Features and Neurobiology. , 2013, , 631-650.		0
106	Neural Substrates of Executive Dysfunction in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS): a Brain Potential Study. <i>Cerebral Cortex</i> , 2013, 23, 2657-2666.	1.6	28
107	Prevalence and risk of migraine headaches in adult fragile X premutation carriers. <i>Clinical Genetics</i> , 2013, 84, 546-551.	1.0	45
108	Ages of Onset of Mood and Anxiety Disorders in Fragile X Premutation Carriers. <i>Current Psychiatry Reviews</i> , 2013, 9, 65-71.	0.9	21
109	Cognitive Dysfunction in FMR1 Premutation Carriers. <i>Current Psychiatry Reviews</i> , 2013, 9, 78-84.	0.9	14
110	Psychiatric Disorders Associated with FXTAS. <i>Current Psychiatry Reviews</i> , 2013, 9, 59-64.	0.9	18

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111	Prenatal Screening Using Maternal Markers. <i>Journal of Clinical Medicine</i> , 2014, 3, 504-520.	1.0	13
112	Immune Dysregulation as a Cause of Autoinflammation in Fragile X Premutation Carriers: Link between FMRI CGG Repeat Number and Decreased Cytokine Responses. <i>PLoS ONE</i> , 2014, 9, e94475.	1.1	26
113	Current research, diagnosis, and treatment of fragile X-associated tremor/ataxia syndrome. <i>Intractable and Rare Diseases Research</i> , 2014, 3, 101-109.	0.3	18
114	Addictive substances may induce a rapid neurological deterioration in fragile X-associated tremor ataxia syndrome: A report of two cases. <i>Intractable and Rare Diseases Research</i> , 2014, 3, 162-165.	0.3	34
115	Fragile X-Associated Disorders. , 0, , 221-253.		7
116	Neurodevelopmental Disabilities in Children With Intermediate and Premutation Range Fragile X Cytosine-Guanine-Guanine Expansions. <i>Journal of Child Neurology</i> , 2014, 29, 326-330.	0.7	16
117	Health and reproductive experiences of women with an FMR1 premutation with and without fragile X premature ovarian insufficiency. <i>Frontiers in Genetics</i> , 2014, 5, 300.	1.1	26
118	Repeat-mediated genetic and epigenetic changes at the FMR1 locus in the Fragile X-related disorders. <i>Frontiers in Genetics</i> , 2014, 5, 226.	1.1	33
119	New observations in the fragile X-associated tremor/ataxia syndrome (FXTAS) phenotype. <i>Frontiers in Genetics</i> , 2014, 5, 365.	1.1	29
120	Towards an understanding of neuropsychiatric manifestations in fragile X premutation carriers. <i>Future Neurology</i> , 2014, 9, 227-239.	0.9	29
121	Fragile X protein in newborn dried blood spots. <i>BMC Medical Genetics</i> , 2014, 15, 119.	2.1	18
122	Fragile X spectrum disorders. <i>Intractable and Rare Diseases Research</i> , 2014, 3, 134-146.	0.3	150
123	Genomic studies in fragile X premutation carriers. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 27.	1.5	24
124	Climbing the Branches of a Family Tree: Diagnosis of Fragile X Syndrome. <i>Journal of Pediatrics</i> , 2014, 164, 1292-1295.	0.9	13
125	Advances in the Genetics of Parkinson's Disease: A Guide for the Clinician. <i>Movement Disorders Clinical Practice</i> , 2014, 1, 3-13.	0.8	17
126	Association between fragile X premutation and premature ovarian failure: a case-control study and meta-analysis. <i>Archives of Gynecology and Obstetrics</i> , 2014, 289, 1255-1262.	0.8	11
127	Blood expression profiles of fragile X premutation carriers identify candidate genes involved in neurodegenerative and infertility phenotypes. <i>Neurobiology of Disease</i> , 2014, 65, 43-54.	2.1	23
128	Abnormal semantic processing in females with fragile X-associated tremor/ataxia syndrome. <i>Genes, Brain and Behavior</i> , 2014, 13, 152-162.	1.1	10



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129	The cognitive neuropsychological phenotype of carriers of the FMR1 premutation. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 28.	1.5	74
130	Associated features in females with an FMR1 premutation. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 30.	1.5	116
131	Emerging topics in FXTAS. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 31.	1.5	76
132	Molecular Advances Leading to Treatment Implications for Fragile X Premutation Carriers. <i>Brain Disorders &amp; Therapy</i> , 2014, 03, .	0.1	40
134	Eye movements reveal impaired inhibitory control in adult male fragile X premutation carriers asymptomatic for FXTAS.. <i>Neuropsychology</i> , 2014, 28, 571-584.	1.0	14
135	Methadone use in a male with the <i>FMR1</i> premutation and FXTAS. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1354-1359.	0.7	24
136	Tremorâ€Predominant Fragile Xâ€Associated Tremor/Ataxia Syndrome in a Female. <i>Movement Disorders Clinical Practice</i> , 2015, 2, 45-46.	0.8	0
137	Axonal neuropathy in female carriers of the fragile X premutation with fragile xâ€associated tremor ataxia syndrome. <i>Muscle and Nerve</i> , 2015, 52, 234-239.	1.0	5
138	Simplified strategy for rapid first-line screening of fragile X syndrome: closed-tube triplet-primed PCR and amplicon melt peak analysis. <i>Expert Reviews in Molecular Medicine</i> , 2015, 17, e7.	1.6	23
139	Anti-Neuronal Antibodies in Patients with Fragile X Syndrome: Is there a Role of Autoimmunity in Its Pathogenesis?. <i>Neurodegenerative Diseases</i> , 2015, 15, 45-49.	0.8	3
140	Dysregulated iron metabolism in the choroid plexus in fragile X-associated tremor/ataxia syndrome. <i>Brain Research</i> , 2015, 1598, 88-96.	1.1	41
141	Fragile Xâ€associated tremor/ataxia syndrome. <i>Annals of the New York Academy of Sciences</i> , 2015, 1338, 58-70.	1.8	139
142	Fragile X premutation carriers: A systematic review of neuroimaging findings. <i>Journal of the Neurological Sciences</i> , 2015, 352, 19-28.	0.3	38
143	Mouse Models of the Fragile X Tremor/Ataxia Syndrome (FXTAS) and the Fragile X Premutation. , 2015, , 641-652.		0
144	Characterization and Early Detection of Balance Deficits in Fragile X Premutation Carriers With and Without Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). <i>Cerebellum</i> , 2015, 14, 650-662.	1.4	31
145	Metabolic and Genetic Causes of Autism. , 2015, , 209-217.		0
146	A genetic study of the FMR1 gene in a Sardinian multiple sclerosis population. <i>Neurological Sciences</i> , 2015, 36, 2213-2220.	0.9	1
147	Immune mediated disorders in women with a fragile X expansion and FXTAS. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 190-197.	0.7	25

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148	Associated Clinical Disorders Diagnosed by Medical Specialists in 188 FMR1 Premutation Carriers Found in the Last 25 Years in the Spanish Basque Country: A Retrospective Study. <i>Genes</i> , 2016, 7, 90.	1.0	6
150	Molecular Correlates and Recent Advancements in the Diagnosis and Screening of FMR1-Related Disorders. <i>Genes</i> , 2016, 7, 87.	1.0	17
151	Endocrine Dysfunction in Female FMR1 Premutation Carriers: Characteristics and Association with Ill Health. <i>Genes</i> , 2016, 7, 101.	1.0	6
153	FXTAS: Neuropsychological and Neuropsychiatric Phenotypes. , 2016, , 39-69.		0
154	Mouse Models for FXTAS and the Fragile X Premutation. , 2016, , 161-179.		0
155	Risk Factors for Cognitive Impairment in Fragile X-Associated Tremor/Ataxia Syndrome. <i>Journal of Geriatric Psychiatry and Neurology</i> , 2016, 29, 328-337.	1.2	15
156	Premature Ovarian Insufficiency: New Perspectives on Genetic Cause and Phenotypic Spectrum. <i>Endocrine Reviews</i> , 2016, 37, 609-635.	8.9	170
157	Fragile X-associated tremor/ataxia syndrome: phenotypic comparisons with other movement disorders. <i>Clinical Neuropsychologist</i> , 2016, 30, 849-900.	1.5	21
158	Small Molecule Recognition and Tools to Study Modulation of r(CGG) <sup>exp</sup> in Fragile X-Associated Tremor Ataxia Syndrome. <i>ACS Chemical Biology</i> , 2016, 11, 2456-2465.	1.6	44
159	Neurological and endocrine phenotypes of fragile X carrier women. <i>Clinical Genetics</i> , 2016, 89, 60-67.	1.0	17
160	Fragile X-associated tremor/ataxia syndrome: another phenotype of the fragile X gene. <i>Clinical Neuropsychologist</i> , 2016, 30, 810-814.	1.5	7
161	The Molecular Biology of Premutation Expanded Alleles. , 2016, , 101-127.		0
162	Psychiatric disorders among women with the fragile X premutation without children affected by fragile X syndrome. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2016, 171, 1139-1147.	1.1	21
163	Reproductive and gynecologic care of women with fragile X primary ovarian insufficiency (FXPOI). <i>Menopause</i> , 2016, 23, 993-999.	0.8	45
164	Carriage of One or Two FMR1 Premutation Alleles Seems to Have No Effect on Illness Severity in a FXTAS Female with an Autozygous FMR1 Premutation Allele. <i>Cerebellum</i> , 2016, 15, 570-577.	1.4	2
165	Fragile X-associated tremor/ataxia syndrome " features, mechanisms and management. <i>Nature Reviews Neurology</i> , 2016, 12, 403-412.	4.9	221
166	Three Faces of Fragile X. <i>Physical Therapy</i> , 2016, 96, 1782-1790.	1.1	11
167	Aging in Fragile X Premutation Carriers. <i>Cerebellum</i> , 2016, 15, 587-594.	1.4	14

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168	The fragile X mental retardation 1 gene ( <i>FMR1</i> ): historical perspective, phenotypes, mechanism, pathology, and epidemiology. <i>Clinical Neuropsychologist</i> , 2016, 30, 815-833.	1.5	22
169	Granulosa cell and oocyte mitochondrial abnormalities in a mouse model of fragile X primary ovarian insufficiency. <i>Molecular Human Reproduction</i> , 2016, 22, 384-396.	1.3	58
170	Etiologies of Primary Ovarian Insufficiency. , 2016, , 19-35.		1
171	X-inactivation in the clinical phenotype of fragile X premutation carrier sisters. <i>Neurology: Genetics</i> , 2016, 2, e45.	0.9	25
172	Development of prenatal screening—A historical overview. <i>Seminars in Perinatology</i> , 2016, 40, 12-22.	1.1	55
173	Improving Health Education for Women Who Carry an <i>FMR1</i> Premutation. <i>Journal of Genetic Counseling</i> , 2016, 25, 228-238.	0.9	6
174	Iron accumulation and dysregulation in the putamen in fragile X-associated tremor/ataxia syndrome. <i>Movement Disorders</i> , 2017, 32, 585-591.	2.2	32
175	<i>FMR1</i> genotype interacts with parenting stress to shape health and functional abilities in older age. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2017, 174, 399-412.	1.1	10
176	Fragile X premutation in women: recognizing the health challenges beyond primary ovarian insufficiency. <i>Journal of Assisted Reproduction and Genetics</i> , 2017, 34, 315-323.	1.2	30
177	Implications of the <i>FMR1</i> Premutation for Children, Adolescents, Adults, and Their Families. <i>Pediatrics</i> , 2017, 139, S172-S182.	1.0	38
178	Adult <i>Fmr1</i> knockout mice present with deficiencies in hippocampal interleukin-6 and tumor necrosis factor- $\alpha$ expression. <i>NeuroReport</i> , 2017, 28, 1246-1249.	0.6	12
179	Diagnostic et prise en charge d'un syndrome tremblement-ataxie lié à une pr-mutation de l'X fragile: le syndrome FXTAS. <i>Pratique Neurologique - FMC</i> , 2017, 8, 204-210.	0.1	0
180	Reduced vagal tone in women with the <i>FMR1</i> premutation is associated with <i>FMR1</i> mRNA but not depression or anxiety. <i>Journal of Neurodevelopmental Disorders</i> , 2017, 9, 16.	1.5	12
181	Prognostic dilemmas and genetic counseling for prenatally detected fragile X gene expansions. <i>Prenatal Diagnosis</i> , 2017, 37, 37-42.	1.1	4
182	Global transcriptome dysregulation in second trimester fetuses with <i>FMR1</i> expansions. <i>Prenatal Diagnosis</i> , 2017, 37, 43-52.	1.1	5
183	Screening for intermediate CGG alleles of <i>FMR1</i> gene in male Iranian patients with Parkinsonism. <i>Neurological Sciences</i> , 2017, 38, 123-128.	0.9	4
184	Molecular Diagnostics and Genetic Counseling in Fragile X Syndrome and <i>FMR1</i> -Associated Disorders. , 2017, , 41-55.		1
185	Fragile X-Associated Tremor/Ataxia Syndrome: From Molecular Pathogenesis to Development of Therapeutics. <i>Frontiers in Cellular Neuroscience</i> , 2017, 11, 128.	1.8	49

#	ARTICLE	IF	CITATIONS
186	Fragile X syndrome and fragile X-associated disorders. F1000Research, 2017, 6, 2112.	0.8	38
187	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
188	The Clinical Phenotype of the Fragile X Syndrome and Related Disorders. , 2017, , 1-16.		1
189	Fragile X syndrome and fragile X-associated tremor ataxia syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 377-391.	1.0	26
190	Mindfulness and Acceptance as Potential Protective Factors for Mothers of Children With Fragile X Syndrome. Frontiers in Public Health, 2018, 6, 316.	1.3	8
191	Microglial cell activation and senescence are characteristic of the pathology FXTAS. Movement Disorders, 2018, 33, 1887-1894.	2.2	19
192	Fragile X-Associated Neuropsychiatric Disorders (FXAND). Frontiers in Psychiatry, 2018, 9, 564.	1.3	132
193	Cognitive function impacts gait, functional mobility and falls in fragile X-associated tremor/ataxia syndrome. Gait and Posture, 2018, 66, 288-293.	0.6	24
194	Health Profiles of Mosaic Versus Non-mosaic FMR1 Premutation Carrier Mothers of Children With Fragile X Syndrome. Frontiers in Genetics, 2018, 9, 173.	1.1	18
195	Deregulation of RNA Metabolism in Microsatellite Expansion Diseases. Advances in Neurobiology, 2018, 20, 213-238.	1.3	5
196	Noncoding CGG repeat expansions in neuronal intranuclear inclusion disease, oculopharyngodistal myopathy and an overlapping disease. Nature Genetics, 2019, 51, 1222-1232.	9.4	265
197	Fragile X-associated neuropsychiatric disorders: a case report. Future Neurology, 2019, 14, FNL14.	0.9	5
198	Current Genetic Service Delivery Models for the Provision of Genetic Testing in Europe: A Systematic Review of the Literature. Frontiers in Genetics, 2019, 10, 552.	1.1	36
199	Decreased functional brain response to emotional arousal and increased psychiatric symptomology in FMR1 premutation carriers. Psychiatry Research - Neuroimaging, 2019, 285, 9-17.	0.9	4
200	Piperine Modulates Protein Mediated Toxicity in Fragile X-Associated Tremor/Ataxia Syndrome through Interacting Expanded CGG Repeat (r(CGG)exp) RNA. ACS Chemical Neuroscience, 2019, 10, 3778-3788.	1.7	20
201	The spectrum of tremor among carriers of the FMR1 premutation with or without the fragile X-associated tremor/ataxia syndrome (FXTAS). Parkinsonism and Related Disorders, 2019, 65, 32-38.	1.1	8
202	Pathophysiology Mechanisms in Fragile-X Primary Ovarian Insufficiency. Methods in Molecular Biology, 2019, 1942, 165-171.	0.4	5
203	Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS). Methods in Molecular Biology, 2019, 1942, 173-189.	0.4	6

#	ARTICLE	IF	CITATIONS
204	The FMRpolyGlycine Protein Mediates Aggregate Formation and Toxicity Independent of the CGG mRNA Hairpin in a Cellular Model for FXTAS. <i>Frontiers in Genetics</i> , 2019, 10, 249.	1.1	18
205	Exploring the Potential of Small Molecule-Based Therapeutic Approaches for Targeting Trinucleotide Repeat Disorders. <i>Molecular Neurobiology</i> , 2020, 57, 566-584.	1.9	17
206	Clustering of comorbid conditions among women who carry an FMR1 premutation. <i>Genetics in Medicine</i> , 2020, 22, 758-766.	1.1	31
207	Cerebellar-cortical function and connectivity during sensorimotor behavior in aging FMR1 gene premutation carriers. <i>NeuroImage: Clinical</i> , 2020, 27, 102332.	1.4	10
208	Cardiovascular Problems in the Fragile X Premutation. <i>Frontiers in Genetics</i> , 2020, 11, 586910.	1.1	11
209	The RNA-binding fragile-X mental retardation protein and its role beyond the brain. <i>Biophysical Reviews</i> , 2020, 12, 903-916.	1.5	11
210	Women with Fragile X-associated Tremor/Ataxia Syndrome. <i>Movement Disorders Clinical Practice</i> , 2020, 7, 910-919.	0.8	13
211	Fragile X clinical features and neurobiology. , 2020, , 351-375.		0
212	Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS): Pathophysiology and Clinical Implications. <i>International Journal of Molecular Sciences</i> , 2020, 21, 4391.	1.8	52
213	Curcumin Regulates the r(CGG)exp RNA Hairpin Structure and Ameliorate Defects in Fragile X-Associated Tremor Ataxia Syndrome. <i>Frontiers in Neuroscience</i> , 2020, 14, 295.	1.4	13
214	The Effects of Dual Task Cognitive Interference and Fast-Paced Walking on Gait, Turns, and Falls in Men and Women with FXTAS. <i>Cerebellum</i> , 2021, 20, 212-221.	1.4	8
215	Fragile X premutation and associated health conditions: A review. <i>Clinical Genetics</i> , 2021, 99, 751-760.	1.0	12
216	Premutations in the FMR1 gene in Serbian patients with undetermined tremor, ataxia and parkinsonism. <i>Neurological Research</i> , 2021, 43, 321-326.	0.6	1
217	EpiMOGA: An Epistasis Detection Method Based on a Multi-Objective Genetic Algorithm. <i>Genes</i> , 2021, 12, 191.	1.0	12
218	Men with an FMR1 premutation and their health education needs. <i>Journal of Genetic Counseling</i> , 2021, 30, 1156-1167.	0.9	0
220	Relationships between motor scores and cognitive functioning in FMR1 female premutation X carriers indicate early involvement of cerebello-cerebral pathways. <i>Cerebellum and Ataxias</i> , 2021, 8, 15.	1.9	12
221	Dysregulation of anti-Mullerian hormone expression levels in mural granulosa cells of FMR1 premutation carriers. <i>Scientific Reports</i> , 2021, 11, 14139.	1.6	1
223	Prevalence of Fragile X-Associated Tremor/Ataxia Syndrome in Patients with Cerebellar Ataxia in Japan. <i>Cerebellum</i> , 2022, 21, 851-860.	1.4	11

#	ARTICLE	IF	CITATIONS
224	Predictors of Comorbid Conditions in Women Who Carry an FMR1 Premutation. <i>Frontiers in Psychiatry</i> , 2021, 12, 715922.	1.3	4
225	Differential Progression of Motor Dysfunction Between Male and Female Fragile X Premutation Carriers Reveals Novel Aspects of Sex-Specific Neural Involvement. <i>Frontiers in Molecular Biosciences</i> , 2020, 7, 577246.	1.6	13
226	Health knowledge of women with a fragile X premutation: Improving understanding with targeted educational material. <i>Journal of Genetic Counseling</i> , 2020, 29, 983-991.	0.9	7
228	Clinical Neurological Phenotype of FXTAS. , 2010, , 1-16.		2
229	The Epidemiology of FXTAS. , 2010, , 17-30.		6
230	FXTAS: Neuropsychological/Neuropsychiatric Phenotypes. , 2010, , 31-53.		2
231	Genotype/Phenotype Relationships in FXTAS. , 2010, , 95-122.		1
232	Treatment and Management of FXTAS. , 2010, , 137-154.		4
233	Prevalence Studies on Fragile X Alleles in Autism. , 2014, , 2755-2771.		1
234	Clinical Neurological Phenotype of FXTAS. , 2016, , 1-24.		3
235	Clinical Manifestation and Management of FXPOI. , 2016, , 199-224.		2
236	Genotype/Phenotype Relationships in FXTAS. , 2016, , 129-160.		1
237	Treatment and Management of FXTAS. , 2016, , 181-197.		4
238	Fragile X Mental Retardation Protein (FMRP) and the Spinal Sensory System. <i>Results and Problems in Cell Differentiation</i> , 2012, 54, 41-59.	0.2	15
239	Health problems in females carriers of premutation in the FMR1 gene. <i>Psychiatria Polska</i> , 2017, 51, 899-907.	0.2	6
241	Advances in the Understanding of the Gabaergic Neurobiology of FMR1 Expanded Alleles Leading to Targeted Treatments for Fragile X Spectrum Disorder. <i>Current Pharmaceutical Design</i> , 2015, 21, 4972-4979.	0.9	20
243	A Review of Fragile X Premutation Disorders. <i>Journal of Clinical Psychiatry</i> , 2009, 70, e1-e11.	1.1	119
244	Memantine for Fragile Xâ€‘Associated Tremor/Ataxia Syndrome. <i>Journal of Clinical Psychiatry</i> , 2014, 75, 264-271.	1.1	44

#	ARTICLE	IF	CITATIONS
245	Case Series: Deep Brain Stimulation in Patients with FXTAS. <i>Brain Disorders &amp; Therapy</i> , 2012, 01, .	0.1	10
246	Current Gaps in Understanding the Molecular Basis of FXTAS. <i>Tremor and Other Hyperkinetic Movements</i> , 2012, 2, .	1.1	21
247	Fragile x-associated tremor ataxia syndrome: the expanding clinical picture, pathophysiology, epidemiology, and update on treatment. <i>Tremor and Other Hyperkinetic Movements</i> , 2012, 2, .	1.1	29
248	Primary Ovarian Insufficiency. , 2010, , 55-82.		0
249	Genetic Counseling for FXTAS and FMR1-Associated Disorders. , 2010, , 155-176.		1
250	The Molecular Biology of FXTAS. , 2010, , 77-93.		0
253	Fragile Xâ€™A Family of Disorders: Changing Phenotype and Molecular Genetics. , 2013, , 453-471.		2
255	Genetic Counseling for FXTAS and Fragile X-Associated Disorders. , 2016, , 263-285.		0
256	Preutation-Associated Disorders in Childhood and Adulthood. , 2016, , 241-262.		3
257	Reduction in the number of CGG repeats on the FMR1 gene in carriers of genetic disorders versus noncarriers. <i>Jornal Brasileiro De Reproducao Assistida</i> , 2017, 21, 327-329.	0.3	0
258	Matrix Metalloproteinases in Fragile X Syndrome. , 2017, , 301-322.		1
261	3 SekundÃre Parkinson-Syndrome. , 2020, , .		0
262	Mystery of Expansion: DNA Metabolism and Unstable Repeats. <i>Advances in Experimental Medicine and Biology</i> , 2020, 1241, 101-124.	0.8	3
263	Fragile X clinical features and neurobiology. , 2020, , 311-332.		0
267	Maternal Mental Health and Parenting Stress and Their Relationships to Characteristics of the Child With Fragile X Syndrome. <i>Frontiers in Psychiatry</i> , 2021, 12, 716585.	1.3	3
268	Allopregnanolone Improves Locomotor Activity and Arousal in the Aged CGG Knock-in Mouse Model of Fragile X-Associated Tremor/Ataxia Syndrome. <i>Frontiers in Neuroscience</i> , 2021, 15, 752973.	1.4	1
269	Increased Pain Symptomatology Among Females vs. Males With Fragile X-Associated Tremor/Ataxia Syndrome. <i>Frontiers in Psychiatry</i> , 2021, 12, 762915.	1.3	6
270	Family history of FXTAS is associated with age-related cognitive-linguistic decline among mothers with the FMR1 preutation. <i>Journal of Neurodevelopmental Disorders</i> , 2022, 14, 7.	1.5	3

#	ARTICLE	IF	CITATIONS
271	Expression of FMRpolyG in Peripheral Blood Mononuclear Cells of Women with Fragile X Mental Retardation 1 Gene Premutation. <i>Genes</i> , 2022, 13, 451.	1.0	1
272	Female fragile X premutation carriers are at increased risk for metabolic syndrome from early adulthood. <i>Nutrition, Metabolism and Cardiovascular Diseases</i> , 2022, 32, 1010-1018.	1.1	1
280	Connective Tissue Disorders and Fragile X Molecular Status in Females: A Case Series and Review. <i>International Journal of Molecular Sciences</i> , 2022, 23, 9090.	1.8	0
281	The effect of college degree attainment on neurodegenerative symptoms in genetically at-risk women. <i>SSM - Population Health</i> , 2022, 19, 101262.	1.3	3
282	The diagnostic experience of women with fragile X-associated primary ovarian insufficiency (FXPOI). <i>Journal of Assisted Reproduction and Genetics</i> , 0, , .	1.2	2
283	Fragile-X-associated Tremor/Ataxia Syndrome (FXTAS) in a Female with FMR1 Premutation: Case Report. <i>The Journal of Tepecik Education and Research Hospital</i> , 2022, 32, 488-491.	0.2	0
284	Evaluation of AQP4 functional variants and its association with fragile X-associated tremor/ataxia syndrome. <i>Frontiers in Aging Neuroscience</i> , 0, 14, .	1.7	0
285	Autism Spectrum Disorder May Be Highly Prevalent in People with Functional Neurological Disorders. <i>Journal of Clinical Medicine</i> , 2023, 12, 299.	1.0	4