

# Elisabetta Tabolacci

## List of Publications by Year in descending order

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43  
papers

1,600  
citations

304743

22  
h-index

315739

38  
g-index

43  
all docs

43  
docs citations

43  
times ranked

2087  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mechanisms of the FMR1 Repeat Instability: How Does the CGG Sequence Expand?. International Journal of Molecular Sciences, 2022, 23, 5425.	4.1	7
2	Mother and Daughter Carrying of the Same Pathogenic Variant in FGFR2 with Discordant Phenotype. Genes, 2022, 13, 1161.	2.4	0
3	Infantile Liver Failure Syndrome 1 associated with a novel variant of the <sc><i>LARS1</i></sc> gene: Clinical, genetic, and functional characterization. Clinical Genetics, 2021, 99, 601-603.	2.0	0
4	DNA Methylation, Mechanisms of FMR1 Inactivation and Therapeutic Perspectives for Fragile X Syndrome. Biomolecules, 2021, 11, 296.	4.0	19
5	Co-Occurrence of Fragile X Syndrome with a Second Genetic Condition: Three Independent Cases of Double Diagnosis. Genes, 2021, 12, 1909.	2.4	4
6	Methylated premutation of the FMR1 gene in three sisters: correlating CGG expansion and epigenetic inactivation. European Journal of Human Genetics, 2020, 28, 567-575.	2.8	6
7	The emerging role of the BDNF-TrkB signaling pathway in the modulation of pain perception. Journal of Neuroimmunology, 2020, 349, 577406.	2.3	66
8	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
9	Reversion to Normal of FMR1 Expanded Alleles: A Rare Event in Two Independent Fragile X Syndrome Families. Genes, 2020, 11, 248.	2.4	7
10	Altered mitochondrial function in cells carrying a premutation or unmethylated full mutation of the FMR1 gene. Human Genetics, 2020, 139, 227-245.	3.8	16
11	The mTOR kinase inhibitor rapamycin enhances the expression and release of pro-inflammatory cytokine interleukin 6 modulating the activation of human microglial cells. EXCLI Journal, 2019, 18, 779-798.	0.7	12
12	Simpsonâ€“Golabiâ€“Behmel syndrome in a female: A case report and an unsolved issue. American Journal of Medical Genetics, Part A, 2017, 173, 285-288.	1.2	14
13	Reactivation of the FMR1 Gene. , 2017, , 341-360.		0
14	Epigenetic causes of intellectual disabilityâ€“the fragile X syndrome paradigm. , 2017, , 107-127.		0
15	Transcriptional Reactivation of the FMR1 Gene. A Possible Approach to the Treatment of the Fragile X Syndrome. Genes, 2016, 7, 49.	2.4	28
16	Genome-wide methylation analysis demonstrates that 5-aza-2-deoxycytidine treatment does not cause random DNA demethylation in fragile X syndrome cells. Epigenetics and Chromatin, 2016, 9, 12.	3.9	28
17	CGG Repeat-Induced FMR1 Silencing Depends on the Expansion Size in Human iPSCs and Neurons Carrying Unmethylated Full Mutations. Stem Cell Reports, 2016, 7, 1059-1071.	4.8	25
18	Defining the role of the CGGBP1 protein in FMR1 gene expression. European Journal of Human Genetics, 2016, 24, 697-703.	2.8	6

#	ARTICLE	IF	CITATIONS
19	What Mechanisms Induce Methylation of FMR1 Gene Full Mutation? A Still Unanswered Question. <i>Epigenetics and Human Health</i> , 2016, , 145-173.	0.2	0
20	Epigenetics, fragile X syndrome and transcriptional therapy. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2797-2808.	1.2	31
21	Role of CTCF Protein in Regulating FMR1 Locus Transcription. <i>PLoS Genetics</i> , 2013, 9, e1003601.	3.5	38
22	Epigenetic Modifications of the FMR1 Gene. <i>Methods in Molecular Biology</i> , 2013, 1010, 141-153.	0.9	4
23	The mGluR5 antagonist AFQ056 does not affect methylation and transcription of the mutant FMR1 gene in vitro. <i>BMC Medical Genetics</i> , 2012, 13, 13.	2.1	15
24	The FRAXopathies: Definition, overview, and update. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1803-1816.	1.2	42
25	Insertion of 16 amino acids in the BAR domain of the oligophrenin 1 protein causes mental retardation and cerebellar hypoplasia in an Italian family. <i>Human Mutation</i> , 2011, 32, E2294-E2307.	2.5	20
26	Treatment with valproic acid ameliorates ADHD symptoms in fragile X syndrome boys. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1420-1427.	1.2	48
27	Tumorigenic Potential of Olfactory Bulb-Derived Human Adult Neural Stem Cells Associates with Activation of TERT and NOTCH1. <i>PLoS ONE</i> , 2009, 4, e4434.	2.5	41
28	A double-blind, parallel, multicenter comparison of L-carnitine with placebo on the attention deficit hyperactivity disorder in fragile X syndrome boys. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 803-812.	1.2	91
29	Epigenetic analysis reveals a euchromatic configuration in the FMR1 unmethylated full mutations. <i>European Journal of Human Genetics</i> , 2008, 16, 1487-1498.	2.8	87
30	A unique case of reversion to normal size of a maternal premutation FMR1 allele in a normal boy. <i>European Journal of Human Genetics</i> , 2008, 16, 209-214.	2.8	24
31	Modest reactivation of the mutant FMR1 gene by valproic acid is accompanied by histone modifications but not DNA demethylation. <i>Pharmacogenetics and Genomics</i> , 2008, 18, 738-741.	1.5	39
32	MID1 mutation screening in a large cohort of Opitz G/BBB syndrome patients: twenty-nine novel mutations identified. <i>Human Mutation</i> , 2007, 28, 206-207.	2.5	55
33	Prevalence of spinocerebellar ataxia type 2 mutation among Italian Parkinsonian patients. <i>Movement Disorders</i> , 2007, 22, 324-327.	3.9	42
34	A new function for the fragile X mental retardation protein in regulation of PSD-95 mRNA stability. <i>Nature Neuroscience</i> , 2007, 10, 578-587.	14.8	318
35	A truncating mutation in the L1RAPL1 gene is responsible for X-linked mental retardation in the MRX21 family. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 482-487.	1.2	51
36	Two brothers with 22q13 deletion syndrome and features suggestive of the Clark-Baraitser syndrome. <i>Clinical Dysmorphology</i> , 2005, 14, 127-132.	0.3	24

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37	Differential epigenetic modifications in the FMR1 gene of the fragile X syndrome after reactivating pharmacological treatments. <i>European Journal of Human Genetics</i> , 2005, 13, 641-648.	2.8	83
38	Molecular dissection of the events leading to inactivation of the FMR1 gene. <i>Human Molecular Genetics</i> , 2005, 14, 267-277.	2.9	120
39	Two brothers with 22q13 deletion syndrome and features suggestive of the Clark-Baraitser syndrome. <i>Clinical Dysmorphology</i> , 2005, 14, 127-132.	0.3	10
40	X-Linked Mental Retardation (XLMR): From Clinical Conditions to Cloned Genes. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 2004, 41, 117-158.	6.1	27
41	Assisted reproductive technology and congenital overgrowth: Some speculations on a case of Pallister-Killian syndrome. <i>American Journal of Medical Genetics Part A</i> , 2004, 130A, 315-316.	2.4	14
42	Quantitative analysis of DNA demethylation and transcriptional reactivation of the FMR1 gene in fragile X cells treated with 5-azadeoxycytidine. <i>Nucleic Acids Research</i> , 2002, 30, 3278-3285.	14.5	103
43	Telomeric associations and chromosome instability in ataxia telangiectasia T cells characterized by TCL1 expression. <i>Cancer Genetics and Cytogenetics</i> , 2001, 125, 46-51.	1.0	7