Elisabetta Tabolacci

List of Publications by Year
in descending order

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	A new function for the fragile X mental retardation protein in regulation of PSD-95 mRNA stability. Nature Neuroscience, 2007, 10, 578-587.	14.8	318
2	Molecular dissection of the events leading to inactivation of the FMR1 gene. Human Molecular Genetics, 2005, 14, 267-277.	2.9	120
3	Quantitative analysis of DNA demethylation and transcriptional reactivation of the FMR1 gene in fragile X cells treated with 5-azadeoxycytidine. Nucleic Acids Research, 2002, 30, 3278-3285.	14.5	103
4	A doubleâ€blind, parallel, multicenter comparison of <scp>L</scp>â€acetylcarnitine with placebo on the attention deficit hyperactivity disorder in fragile X syndrome boys. American Journal of Medical Genetics, Part A, 2008, 146A, 803-812.	1.2	91
5	Epigenetic analysis reveals a euchromatic configuration in the FMR1 unmethylated full mutations. European Journal of Human Genetics, 2008, 16, 1487-1498.	2.8	87
6	Differential epigenetic modifications in the FMR1 gene of the fragile X syndrome after reactivating pharmacological treatments. European Journal of Human Genetics, 2005, 13, 641-648.	2.8	83
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	MID1 mutation screening in a large cohort of Opitz G/BBB syndrome patients: twenty-nine novel mutations identified. Human Mutation, 2007, 28, 206-207.	2.5	55
9	A truncating mutation in theIL1RAPL1 gene is responsible for X-linked mental retardation in the MRX21 family. American Journal of Medical Genetics, Part A, 2006, 140A, 482-487.	1.2	51
10	Treatment with valproic acid ameliorates ADHD symptoms in fragile X syndrome boys. American Journal of Medical Genetics, Part A, 2010, 152A, 1420-1427.	1.2	48
11	Prevalence of spinocerebellar ataxia type 2 mutation among Italian Parkinsonian patients. Movement Disorders, 2007, 22, 324-327.	3.9	42
12	The FRAXopathies: Definition, overview, and update. American Journal of Medical Genetics, Part A, 2011, 155, 1803-1816.	1.2	42
13	Tumorigenic Potential of Olfactory Bulb-Derived Human Adult Neural Stem Cells Associates with Activation of TERT and NOTCH1. PLoS ONE, 2009, 4, e4434.	2.5	41
14	Modest reactivation of the mutant FMR1 gene by valproic acid is accompanied by histone modifications but not DNA demethylation. Pharmacogenetics and Genomics, 2008, 18, 738-741.	1.5	39
15	Role of CTCF Protein in Regulating FMR1 Locus Transcription. PLoS Genetics, 2013, 9, e1003601.	3.5	38
16	Epigenetics, fragile X syndrome and transcriptional therapy. American Journal of Medical Genetics, Part A, 2013, 161, 2797-2808.	1.2	31
17	Transcriptional Reactivation of the FMR1 Gene. A Possible Approach to the Treatment of the Fragile X Syndrome. Genes, 2016, 7, 49.	2.4	28
18	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

#	Article	IF	Citations
19	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
20	X-Linked Mental Retardation (XLMR): From Clinical Conditions to Cloned Genes. Critical Reviews in Clinical Laboratory Sciences, 2004, 41, 117-158.	6.1	27
21	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
22	Two brothers with 22q13 deletion syndrome and features suggestive of the Clark???Baraitser syndrome. Clinical Dysmorphology, 2005, 14, 127-132.	0.3	24
23	A unique case of reversion to normal size of a maternal premutation FMR1 allele in a normal boy. European Journal of Human Genetics, 2008, 16, 209-214.	2.8	24
24	Insertion of 16 amino acids in the BAR domain of the oligophrenin 1 protein causes mental retardation and cerebellar hypoplasia in an Italian family. Human Mutation, 2011, 32, E2294-E2307.	2.5	20
25	DNA Methylation, Mechanisms of FMR1 Inactivation and Therapeutic Perspectives for Fragile X Syndrome. Biomolecules, 2021, 11, 296.	4.0	19
26	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
27	The mGluR5 antagonist AFQ056 does not affect methylation and transcription of the mutant FMR1 gene in vitro. BMC Medical Genetics, 2012, 13, 13.	2.1	15
28	Assisted reproductive technology and congenital overgrowth: Some speculations on a case of Pallister-Killian syndrome. American Journal of Medical Genetics Part A, 2004, 130A, 315-316.	2.4	14
29	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
30	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
31	Two brothers with 22q13 deletion syndrome and features suggestive of the Clark-Baraitser syndrome. Clinical Dysmorphology, 2005, 14, 127-132.	0.3	10
32	Telomeric associations and chromosome instability in ataxia telangiectasia T cells characterized by TCL1 expression. Cancer Genetics and Cytogenetics, 2001, 125, 46-51.	1.0	7
33	Reversion to Normal of FMR1 Expanded Alleles: A Rare Event in Two Independent Fragile X Syndrome Families. Genes, 2020, 11, 248.	2.4	7
34	Mechanisms of the FMR1 Repeat Instability: How Does the CGG Sequence Expand?. International Journal of Molecular Sciences, 2022, 23, 5425.	4.1	7
35	Defining the role of the CGGBP1 protein in FMR1 gene expression. European Journal of Human Genetics, 2016, 24, 697-703.	2.8	6
36	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

#	Article	IF	Citations
37	Epigenetic Modifications of the FMR1 Gene. Methods in Molecular Biology, 2013, 1010, 141-153.	0.9	4
38	Co-Occurrence of Fragile X Syndrome with a Second Genetic Condition: Three Independent Cases of Double Diagnosis. Genes, 2021, 12, 1909.	2.4	4
39	Reactivation of the FMR1 Gene. , 2017, , 341-360.		0
40	Infantile Liver Failure Syndrome 1 associated with a novel variant of the <scp>LARS1</scp> gene: Clinical, genetic, and functional characterization. Clinical Genetics, 2021, 99, 601-603.	2.0	0
41	What Mechanisms Induce Methylation of FMR1 Gene Full Mutation? A Still Unanswered Question. Epigenetics and Human Health, 2016, , 145-173.	0.2	0
42	Epigenetic causes of intellectual disabilityâ€"the fragile X syndrome paradigm. , 2017, , 107-127.		0
43	Mother and Daughter Carrying of the Same Pathogenic Variant in FGFR2 with Discordant Phenotype. Genes, 2022, 13, 1161.	2.4	0