Giovanni Neri

List of Publications by Year
in descending order

95 papers	5,289 citations	109321 35 h-index	88630 70 g-index
97 all docs	97 docs citations	97 times ranked	6015 citing authors

#	Article	IF	Citations
1	Chromosome 14 deletions, rings, and epilepsy genes: A riddle wrapped in a mystery inside an enigma. Epilepsia, 2021, 62, 25-40.	5.1	4
2	DNA Methylation, Mechanisms of FMR1 Inactivation and Therapeutic Perspectives for Fragile X Syndrome. Biomolecules, 2021, 11, 296.	4.0	19
3	Co-Occurrence of Fragile X Syndrome with a Second Genetic Condition: Three Independent Cases of Double Diagnosis. Genes, 2021, 12, 1909.	2.4	4
4	Xâ€linked intellectual disability: Phenotypic expression in carrier females. Clinical Genetics, 2020, 97, 418-425.	2.0	12
5	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
6	Reversion to Normal of FMR1 Expanded Alleles: A Rare Event in Two Independent Fragile X Syndrome Families. Genes, 2020, 11, 248.	2.4	7
7	Xâ€linked intellectual disability update 2017. American Journal of Medical Genetics, Part A, 2018, 176, 1375-1388.	1.2	88
8	Guideline recommendations for diagnosis and clinical management of Ring14 syndromeâ€"first report of an ad hoc task force. Orphanet Journal of Rare Diseases, 2017, 12, 69.	2.7	18
9	ATP1A3 mutant patient with alternating hemiplegia of childhood and brain spectroscopic abnormalities. Journal of the Neurological Sciences, 2017, 379, 36-38.	0.6	4
10	Variations on the theme of how to write a scientific article. American Journal of Medical Genetics, Part A, 2017, 173, 1149-1151.	1.2	1
11	â€œMinimalâ€holoprosencephaly in a 14q deletion syndrome patient. American Journal of Medical Genetics, Part A, 2017, 173, 3216-3220.	1.2	2
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	The Clinical Phenotype of the Fragile X Syndrome and Related Disorders. , 2017, , 1-16.		1
14	Transcriptional Reactivation of the FMR1 Gene. A Possible Approach to the Treatment of the Fragile X Syndrome. Genes, 2016, 7, 49.	2.4	28
15	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
16	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
17	Defining the role of the CGGBP1 protein in FMR1 gene expression. European Journal of Human Genetics, 2016, 24, 697-703.	2.8	6
18	What Mechanisms Induce Methylation of FMR1 Gene Full Mutation? A Still Unanswered Question. Epigenetics and Human Health, 2016, , 145-173.	0.2	0

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19	Congenital generalized hypertrichosis: the skin as a clue to complex malformation syndromes. Italian Journal of Pediatrics, 2015, 41, 55.	2.6	38
20	ZC4H2, an XLID gene, is required for the generation of a specific subset of CNS interneurons. Human Molecular Genetics, 2015, 24, 4848-4861.	2.9	48
21	Involvement of MBD4 inactivation in mismatch repair-deficient tumorigenesis. Oncotarget, 2015, 6, 42892-42904.	1.8	43
22	Fragile X syndrome: a preclinical review on metabotropic glutamate receptor 5 (mGluR5) antagonists and drug development. Psychopharmacology, 2014, 231, 1217-1226.	3.1	79
23	Cardio-Facio-Cutaneous Syndrome: Clinical Features, Diagnosis, and Management Guidelines. Pediatrics, 2014, 134, e1149-e1162.	2.1	148
24	Mild Beckwith-Wiedemann and severe long-QT syndrome due to deletion of the imprinting center 2 on chromosome 11p. European Journal of Human Genetics, 2013, 21, 965-969.	2.8	35
25	Role of CTCF Protein in Regulating FMR1 Locus Transcription. PLoS Genetics, 2013, 9, e1003601.	3.5	38
26	The Fragile X Protein binds m <scp>RNA</scp> s involved in cancer progression and modulates metastasis formation. EMBO Molecular Medicine, 2013, 5, 1523-1536.	6.9	106
27	Epilepsy in ring 14 syndrome: A clinical and <scp>EEG</scp> study of 22 patients. Epilepsia, 2013, 54, 2204-2213.	5.1	17
28	The ring 14 syndrome. European Journal of Medical Genetics, 2012, 55, 374-380.	1.3	34
29	Three unrelated patients with congenital anterior pituitary aplasia and a characteristic physical and neuropsychological phenotype: A new syndrome?. American Journal of Medical Genetics, Part A, 2012, 158A, 2750-2755.	1.2	2
30	Linguistic and psychomotor development in children with chromosome 14 deletions. Clinical Linguistics and Phonetics, 2012, 26, 962-974.	0.9	10
31	Fragile X syndrome: causes, diagnosis, mechanisms, and therapeutics. Journal of Clinical Investigation, 2012, 122, 4314-4322.	8.2	269
32	Expanding the spectrum of rearrangements involving chromosome 19: A mild phenotype associated with a 19p13.12â€“p13.13 deletion. American Journal of Medical Genetics, Part A, 2012, 158A, 888-893.	1.2	8
33	Chromosome 9p deletion syndrome and sex reversal: Novel findings and redefinition of the critically deleted regions. American Journal of Medical Genetics, Part A, 2012, 158A, 2266-2271.	1.2	33
34	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
35	Epigenetic Modification of the FMR1 Gene in Fragile X Syndrome Is Associated with Differential Response to the mGluR5 Antagonist AFQ056. Science Translational Medicine, 2011, 3, 64ra1.	12.4	344
36	The FRAXopathies: Definition, overview, and update. American Journal of Medical Genetics, Part A, 2011, 155, 1803-1816.	1.2	42

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37	Biomarkers in Rare Disorders: The Experience with Spinal Muscular Atrophy. International Journal of Molecular Sciences, 2011, 12, 24-38.	4.1	13
38	Phenotypic map in ring 14 syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 237-237.	1.2	3
39	Unexpected finding of a paternal premutation of the fragile X FMR1 gene in a female fetus of a premutation carrier mother. American Journal of Medical Genetics, Part A, 2010, 152A, 409-412.	1.2	0
40	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
41	Response to â€œValproic Acid and ADHD Symptoms in Fragile X Syndrome: More Evidence Is Neededâ€. American Journal of Medical Genetics, Part A, 2010, 152A, 2682-2682.	1.2	0
42	A case of Beckwith-Wiedemann syndrome caused by a cryptic 11p15 deletion encompassing the centromeric imprinted domain of the BWS locus. Journal of Medical Genetics, 2010, 47, 429-432.	3.2	30
43	The ring 14 syndrome: Clinical and molecular definition. American Journal of Medical Genetics, Part A, 2009, 149A, 1116-1124.	1.2	54
44	Down syndrome: Comments and reflections on the 50th anniversary of Lejeune's discovery. American Journal of Medical Genetics, Part A, 2009, 149A, 2647-2654.	1.2	28
45	Reply of the Authors: 45,X infertile males: not so rare. Fertility and Sterility, 2009, 92, e50.	1.0	0
46	Overgrowth Syndromes: A Classification. Endocrine Development, 2009, 14, 53-60.	1.3	13
47	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
48	Epigenetic analysis reveals a euchromatic configuration in the FMR1 unmethylated full mutations. European Journal of Human Genetics, 2008, 16, 1487-1498.	2.8	87
49	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
50	XLMR genes: update 2007. European Journal of Human Genetics, 2008, 16, 422-434.	2.8	155
51	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
52	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
53	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
54	The Helena syndromes. American Journal of Medical Genetics, Part A, 2006, 140A, 2007-2012.	1.2	7

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55	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
56	Fragile X syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 137C, 32-37.	1.6	60
57	Molecular dissection of the events leading to inactivation of the FMR1 gene. Human Molecular Genetics, 2005, 14, 267-277.	2.9	120
58	X-Linked Mental Retardation (XLMR): From Clinical Conditions to Cloned Genes. Critical Reviews in Clinical Laboratory Sciences, 2004, 41, 117-158.	6.1	27
59	CFC syndrome. American Journal of Medical Genetics Part A, 2003, 116A, 410-410.	2.4	7
60	Cryptic t(1;12)(q44;p13.3) translocation in a previously described syndrome with polymicrogyria, segregating as an apparently X-linked trait. American Journal of Medical Genetics Part A, 2003, 117A, 65-71.	2.4	25
61	Understanding the biological underpinnings of fragile X syndrome. Current Opinion in Pediatrics, 2003, 15, 559-566.	2.0	26
62	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
63	Pharmacological reactivation of inactive genes: the fragile X experience. Brain Research Bulletin, 2001, 56, 383-387.	3.0	13
64	Mutations of the 'minor' mismatch repair gene MSH6 in typical and atypical hereditary nonpolyposis colorectal cancer. Familial Cancer, 2001, 1, 95-101.	1.9	24
65	Premature termination mutations in exon 3 of the SMN1 gene are associated with exon skipping and a relatively mild SMA phenotype. European Journal of Human Genetics, 2001, 9, 113-120.	2.8	48
66	XLMR genes: update 2000. European Journal of Human Genetics, 2001, 9, 71-81.	2.8	74
67	Heterogeneity of cardio-facio-cutaneous syndrome. American Journal of Medical Genetics Part A, 2000, 95, 144-144.	2.4	5
68	Partial deletion of chromosome 12q is not usually associated with CFC syndrome. American Journal of Medical Genetics Part A, 2000, 95, 296-296.	2.4	10
69	Sixty tears of X-linked mental retardation: A historical footnote. American Journal of Medical Genetics Part A, 2000, 97, 228-233.	2.4	12
70	SMN protein analysis in fibroblast, amniocyte and CVS cultures from spinal muscular atrophy patients and its relevance for diagnosis. European Journal of Human Genetics, 1999, 7, 301-309.	2.8	43
71	The drastic reduction of SMN protein in SMA I spinal cord motor neurons is not due to inefficient transcription. Neurogenetics, 1999, 2, 97-100.	1.4	2
72	Cardiac anomalies in the Simpson-Golabi-Behmel syndrome. American Journal of Medical Genetics Part A, 1999, 83, 378-381.	2.4	61

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73	Syndromal (and nonsyndromal) forms of male pseudohermaphroditism. American Journal of Medical Genetics Part A, 1999, 89, 201-209.	2.4	12
74	X-Linked Mental Retardation. Advances in Genetics, 1999, 41, 55-94.	1.8	16
75	MLH1 and MSH2 constitutinal mutations in colorectal cancer families not meeting the standard criteria for hereditary nonpolyposis colorectal cancer. , 1998, 75, 835-839.		50
76	Clinical and molecular aspects of the Simpson-Golabi-Behmel syndrome. American Journal of Medical Genetics Part A, 1998, 79, 279-283.	2.4	156
77	Cytogenetic evidence for a less malignant leukemic cell population in the central nervous system in a critical case of acute myeloblastic leukemia. , 1998, 30, 91-94.		1
78	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	21.4	196
79	Characterization ofMSH2 andMLH1 mutations in Italian families with hereditary nonpolyposis colorectal cancer. , 1997, 18, 8-18.		67
80	Deletions in the SMN gene in infantile and adult spinal muscular atrophy patients from the same family. Human Genetics, 1996, 97, 315-318.	3.8	8
81	More on the Noonan-CFC controversy. , 1996, 65, 100-100.		19
82	Mutations in GPC3, a glypican gene, cause the Simpson-Golabi-Behmel overgrowth syndrome. Nature Genetics, 1996, 12, 241-247.	21.4	732
83	Gene for Simpson-Golabi-Behmel syndrome is linked to HPRT in Xq26 in two European families. American Journal of Medical Genetics Part A, 1994, 50, 388-390.	2.4	16
84	No apparent involvement of the FMR1 gene in five patients with phenotypic manifestations of the fragile X syndrome. American Journal of Medical Genetics Part A, 1994, 51, 309-314.	2.4	13
85	Butyrate and acetyl-carnitine inhibit the cytogenetic expression of the fragile X in vitro. American Journal of Medical Genetics Part A, 1994, 51, 447-450.	2.4	23
86	Unstable triplets and their mutational mechanism: Size reduction of the CGG repeat vs. germline mosaicism in the fragile X syndrome. American Journal of Medical Genetics Part A, 1994, 51, 517-521.	2.4	27
87	Costello syndrome: Further clinical delineation, natural history, genetic definition, and nosology. American Journal of Medical Genetics Part A, 1993, 47, 176-183.	2.4	96
88	Further delineation of the Simpson-Golabi-Behmel (SGB) syndrome. American Journal of Medical Genetics Part A, 1992, 44, 136-137.	2.4	31
89	XLMR genes: Update 1990. American Journal of Medical Genetics Part A, 1991, 38, 186-189.	2.4	38
90	Conference report: Third International Workshop on the fragile X and X-linked mental retardation. American Journal of Medical Genetics Part A, 1988, 30, 1-29.	2.4	16

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91	Role of chromosome aberrations in recurrent abortion: A study of 269 balanced translocations. American Journal of Medical Genetics Part A, 1986, 24, 341-356.	2.4	99
92	New multiple congenital anomalies/mental retardation syndrome with cardioâ€facioâ€cutaneous involvementâ€"the CFC syndrome. American Journal of Medical Genetics Part A, 1986, 25, 413-427.	2.4	208
93	The Perlman syndrome: Familial renal dysplasia with Wilms tumor, fetal gigantism and multiple congenital anomalies. American Journal of Medical Genetics Part A, 1984, 19, 195-207.	2.4	64
94	Sensorineural deafness in the FG syndrome: Report on four new cases. American Journal of Medical Genetics Part A, 1984, 19, 369-377.	2.4	27
95	Behavior of A Cell Line Derived from A Mouse Submaxillary Adenocarcinoma during the Initial 480 Days in Vitro. Tumori, 1978, 64, 1-14.	1.1	2