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	Mutations in GPC3, a glypican gene, cause the Simpson-Golabi-Behmel overgrowth syndrome. Nature Genetics, 1996, 12, 241-247.	21.4	732
2	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
3	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
4	Fragile X syndrome: causes, diagnosis, mechanisms, and therapeutics. Journal of Clinical Investigation, 2012, 122, 4314-4322.	8.2	269
5	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
6	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	21.4	196
7	Clinical and molecular aspects of the Simpson-Golabi-Behmel syndrome. American Journal of Medical Genetics Part A, 1998, 79, 279-283.	2.4	156
8	XLMR genes: update 2007. European Journal of Human Genetics, 2008, 16, 422-434.	2.8	155
9	Cardio-Facio-Cutaneous Syndrome: Clinical Features, Diagnosis, and Management Guidelines. Pediatrics, 2014, 134, e1149-e1162.	2.1	148
10	Molecular dissection of the events leading to inactivation of the FMR1 gene. Human Molecular Genetics, 2005, 14, 267-277.	2.9	120
11	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
12	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
13	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
14	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
15	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
16	Xâ€linked intellectual disability update 2017. American Journal of Medical Genetics, Part A, 2018, 176, 1375-1388.	1.2	88
17	Epigenetic analysis reveals a euchromatic configuration in the FMR1 unmethylated full mutations. European Journal of Human Genetics, 2008, 16, 1487-1498.	2.8	87
18	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

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19	Fragile X syndrome: a preclinical review on metabotropic glutamate receptor 5 (mGluR5) antagonists and drug development. Psychopharmacology, 2014, 231, 1217-1226.	3.1	79
20	XLMR genes: update 2000. European Journal of Human Genetics, 2001, 9, 71-81.	2.8	74
21	Characterization ofMSH2 andMLH1 mutations in Italian families with hereditary nonpolyposis colorectal cancer. , 1997, 18, 8-18.		67
22	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
23	Cardiac anomalies in the Simpson-Golabi-Behmel syndrome. American Journal of Medical Genetics Part A, 1999, 83, 378-381.	2.4	61
24	Fragile X syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 137C, 32-37.	1.6	60
25	The ring 14 syndrome: Clinical and molecular definition. American Journal of Medical Genetics, Part A, 2009, 149A, 1116-1124.	1.2	54
26	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
27	MLH1 and MSH2 constitutinal mutations in colorectal cancer families not meeting the standard criteria for hereditary nonpolyposis colorectal cancer. , 1998, 75, 835-839.		50
28	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
29	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
30	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
31	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
32	Involvement of MBD4 inactivation in mismatch repair-deficient tumorigenesis. Oncotarget, 2015, 6, 42892-42904.	1.8	43
33	The FRAXopathies: Definition, overview, and update. American Journal of Medical Genetics, Part A, 2011, 155, 1803-1816.	1.2	42
34	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
35	XLMR genes: Update 1990. American Journal of Medical Genetics Part A, 1991, 38, 186-189.	2.4	38
36	Role of CTCF Protein in Regulating FMR1 Locus Transcription. PLoS Genetics, 2013, 9, e1003601.	3.5	38

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37	Congenital generalized hypertrichosis: the skin as a clue to complex malformation syndromes. Italian Journal of Pediatrics, 2015, 41, 55.	2.6	38
38	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
39	The ring 14 syndrome. European Journal of Medical Genetics, 2012, 55, 374-380.	1.3	34
40	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
41	Further delineation of the Simpson-Golabi-Behmel (SGB) syndrome. American Journal of Medical Genetics Part A, 1992, 44, 136-137.	2.4	31
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	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
44	Transcriptional Reactivation of the FMR1 Gene. A Possible Approach to the Treatment of the Fragile X Syndrome. Genes, 2016, 7, 49.	2.4	28
45	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
46	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
47	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
48	X-Linked Mental Retardation (XLMR): From Clinical Conditions to Cloned Genes. Critical Reviews in Clinical Laboratory Sciences, 2004, 41, 117-158.	6.1	27
49	Understanding the biological underpinnings of fragile X syndrome. Current Opinion in Pediatrics, 2003, 15, 559-566.	2.0	26
50	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
51	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
52	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
53	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
54	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

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55	More on the Noonan-CFC controversy. , 1996, 65, 100-100.		19
56	DNA Methylation, Mechanisms of FMR1 Inactivation and Therapeutic Perspectives for Fragile X Syndrome. Biomolecules, 2021, 11, 296.	4.0	19
57	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
58	Epilepsy in ring 14 syndrome: A clinical and <scp>EEG</scp> study of 22 patients. Epilepsia, 2013, 54, 2204-2213.	5.1	17
59	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
60	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
61	X-Linked Mental Retardation. Advances in Genetics, 1999, 41, 55-94.	1.8	16
62	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
63	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
64	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
65	Pharmacological reactivation of inactive genes: the fragile X experience. Brain Research Bulletin, 2001, 56, 383-387.	3.0	13
66	Overgrowth Syndromes: A Classification. Endocrine Development, 2009, 14, 53-60.	1.3	13
67	Biomarkers in Rare Disorders: The Experience with Spinal Muscular Atrophy. International Journal of Molecular Sciences, 2011, 12, 24-38.	4.1	13
68	Syndromal (and nonsyndromal) forms of male pseudohermaphroditism. American Journal of Medical Genetics Part A, 1999, 89, 201-209.	2.4	12
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	Xâ€linked intellectual disability: Phenotypic expression in carrier females. Clinical Genetics, 2020, 97, 418-425.	2.0	12
71	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
72	Linguistic and psychomotor development in children with chromosome 14 deletions. Clinical Linguistics and Phonetics, 2012, 26, 962-974.	0.9	10

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73	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
74	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
75	CFC syndrome. American Journal of Medical Genetics Part A, 2003, 116A, 410-410.	2.4	7
76	The Helena syndromes. American Journal of Medical Genetics, Part A, 2006, 140A, 2007-2012.	1.2	7
77	Reversion to Normal of FMR1 Expanded Alleles: A Rare Event in Two Independent Fragile X Syndrome Families. Genes, 2020, 11, 248.	2.4	7
78	Defining the role of the CGGBP1 protein in FMR1 gene expression. European Journal of Human Genetics, 2016, 24, 697-703.	2.8	6
79	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
80	Heterogeneity of cardio-facio-cutaneous syndrome. American Journal of Medical Genetics Part A, 2000, 95, 144-144.	2.4	5
81	ATP1A3 mutant patient with alternating hemiplegia of childhood and brain spectroscopic abnormalities. Journal of the Neurological Sciences, 2017, 379, 36-38.	0.6	4
82	Chromosome 14 deletions, rings, and epilepsy genes: A riddle wrapped in a mystery inside an enigma. Epilepsia, 2021, 62, 25-40.	5.1	4
83	Co-Occurrence of Fragile X Syndrome with a Second Genetic Condition: Three Independent Cases of Double Diagnosis. Genes, 2021, 12, 1909.	2.4	4
84	Phenotypic map in ring 14 syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 237-237.	1.2	3
85	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
86	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
87	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
88	â€œMinimalâ€•holoprosencephaly in a 14q deletion syndrome patient. American Journal of Medical Genetics, Part A, 2017, 173, 3216-3220.	1.2	2
89	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
90	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

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91	The Clinical Phenotype of the Fragile X Syndrome and Related Disorders. , 2017, , 1-16.		1
92	Reply of the Authors: 45,X infertile males: not so rare. Fertility and Sterility, 2009, 92, e50.	1.0	0
93	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
94	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
95	What Mechanisms Induce Methylation of FMR1 Gene Full Mutation? A Still Unanswered Question. Epigenetics and Human Health, 2016, , 145-173.	0.2	0