Fiorella Gurrieri

List of Publications by Year
in descending order

68 papers	3,555 citations	185998 28 h-index	143772 57 g-index
70 all docs	70 docs citations	70 times ranked	5091 citing authors

#	Article	IF	Citations
1	De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. Nature Genetics, 2012, 44, 1030-1034.	9.4	345
2	p63 Gene Mutations in EEC Syndrome, Limb-Mammary Syndrome, and Isolated Split Handâ€“Split Foot Malformation Suggest a Genotype-Phenotype Correlation. American Journal of Human Genetics, 2001, 69, 481-492.	2.6	331
3	Prevalence of SHANK3 variants in patients with different subtypes of autism spectrum disorders. European Journal of Human Genetics, 2013, 21, 310-316.	1.4	210
4	Distinct neurological disorders with ATP1A3 mutations. Lancet Neurology, The, 2014, 13, 503-514.	4.9	206
5	Coding exons function as tissue-specific enhancers of nearby genes. Genome Research, 2012, 22, 1059-1068.	2.4	202
6	Clinical and molecular aspects of the Simpson-Golabi-Behmel syndrome. , 1998, 79, 279-283.		156
7	Oralâ€“facialâ€“digital syndromes: Review and diagnostic guidelines. American Journal of Medical Genetics, Part A, 2007, 143A, 3314-3323.	0.7	134
8	Clinical profile of patients with ATP1A3 mutations in Alternating Hemiplegia of Childhoodâ€”a study of 155 patients. Orphanet Journal of Rare Diseases, 2015, 10, 123.	1.2	117
9	Linkage of a human brain malformation, familial holoprosencephaly, to chromosome 7 and evidence for genetic heterogeneity.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 8102-8106.	3.3	111
10	Mowatâ€“Wilson syndrome: Facial phenotype changing with age: Study of 19 Italian patients and review of the literature. American Journal of Medical Genetics, Part A, 2009, 149A, 417-426.	0.7	97
11	Physical mapping of the holoprosencephaly critical region on chromosome 7q36. Nature Genetics, 1993, 3, 247-251.	9.4	95
12	Hypo-Phosphorylation of Salivary Peptidome as a Clue to the Molecular Pathogenesis of Autism Spectrum Disorders. Journal of Proteome Research, 2008, 7, 5327-5332.	1.8	90
13	Elements of morphology: Standard terminology for the hands and feet. American Journal of Medical Genetics, Part A, 2009, 149A, 93-127.	0.7	89
14	A genomic rearrangement resulting in a tandem duplication is associated with split hand-split foot malformation 3 (SHFM3) at 10q24. Human Molecular Genetics, 2003, 12, 1959-1971.	1.4	88
15	Advances in the genetics of progressive myoclonus epilepsy. American Journal of Medical Genetics Part A, 2001, 106, 129-138.	2.4	83
16	Mutational spectrum of the oral-facial-digital type I syndrome: a study on a large collection of patients. Human Mutation, 2008, 29, 1237-1246.	1.1	82
17	A New Susceptibility Locus for Migraine with Aura in the 15q11-q13 Genomic Region Containing Three GABA-A Receptor Genes. American Journal of Human Genetics, 2005, 76, 327-333.	2.6	72
18	Further evidence that the rs1858830 C variant in the promoter region of the MET gene is associated with autistic disorder. Autism Research, 2009, 2, 232-236.	2.1	67

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19	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	1.1	64
20	A split hand-split foot (SHFM3) gene is located at 10Q24â†'25. American Journal of Medical Genetics Part A, 1996, 62, 427-436.	2.4	57
21	Functional characterization of tissue-specific enhancers in the DLX5/6 locus. Human Molecular Genetics, 2012, 21, 4930-4938.	1.4	54
22	Clinical, genetic, and molecular aspects of splitâ€hand/foot malformation: An update. American Journal of Medical Genetics, Part A, 2013, 161, 2860-2872.	0.7	53
23	Split hand/split foot malformation with hearing loss: first report of families linked to the SHFM1 locus in 7q21. Clinical Genetics, 2001, 59, 28-36.	1.0	44
24	XLMR genes: Update 1990. American Journal of Medical Genetics Part A, 1991, 38, 186-189.	2.4	38
25	Oral-facial-skeletal syndromes. American Journal of Medical Genetics Part A, 1995, 59, 365-368.	2.4	37
26	Genetic Imprinting: The Paradigm of Prader-Willi and Angelman Syndromes. Endocrine Development, 2009, 14, 20-28.	1.3	37
27	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.	1.4	35
28	Split-hand/split-foot malformation 3 (SHFM3) at 10q24, development of rapid diagnostic methods and gene expression from the region. American Journal of Medical Genetics, Part A, 2006, 140A, 1384-1395.	0.7	33
29	Further delineation of the Simpson-Golabi-Behmel (SGB) syndrome. American Journal of Medical Genetics Part A, 1992, 44, 136-137.	2.4	31
30	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	1.0	28
31	Limb anomalies: Developmental and evolutionary aspects. American Journal of Medical Genetics Part A, 2002, 115, 231-244.	2.4	26
32	Integrated analysis of clinical signs and literature data for the diagnosis and therapy of a previously undescribed 6p21.3 deletion syndrome. European Journal of Human Genetics, 2011, 19, 239-242.	1.4	25
33	Two brothers with 22q13 deletion syndrome and features suggestive of the Clark???Baraitser syndrome. Clinical Dysmorphology, 2005, 14, 127-132.	0.1	24
34	Defective oxytocin function: a clue to understanding the cause of autism?. BMC Medicine, 2009, 7, 63.	2.3	24
35	Working up autism: The practical role of medical genetics. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 104-110.	0.7	24
36	NFIX mutations affecting the DNA-binding domain cause a peculiar overgrowth syndrome (Malan) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.7	23

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37	X-linked mental retardation with marfanoid habitus: First report of four Italian patients. American Journal of Medical Genetics Part A, 1991, 38, 228-232.	2.4	22
38	Ulnar ray defect in an infant with a 6q21;7q31.2 translocation: Further evidence for the existence of a limb defect gene in 6q21. American Journal of Medical Genetics Part A, 1995, 55, 315-318.	2.4	22
39	Clinical and molecular studies on two further families with Simpson-Golabi-Behmel syndrome. American Journal of Medical Genetics, Part A, 2005, 138A, 272-277.	0.7	22
40	Frequency of genomic rearrangements involving the SHFM3 locus at chromosome 10q24 in syndromic and non-syndromic split-hand/foot malformation. American Journal of Medical Genetics, Part A, 2006, 140A, 1375-1383.	0.7	22
41	Megalencephaly and Perisylvian Polymicrogyria with Postaxial Polydactyly and Hydrocephalus (MPPH): Report of a New Case. Neuropediatrics, 2007, 38, 200-203.	0.3	22
42	Rare missense variants in the ALPK1 gene may predispose to periodic fever, aphthous stomatitis, pharyngitis and adenitis (PFAPA) syndrome. European Journal of Human Genetics, 2019, 27, 1361-1368.	1.4	21
43	PCR and serology find no association between xenotropic murine leukemia virus-related virus (XMRV) and autism. Molecular Autism, 2010, 1, 14.	2.6	19
44	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
45	Rearrangements of chromosome 15 in epilepsy. American Journal of Medical Genetics Part A, 2001, 106, 125-128.	2.4	16
46	A SPRY2 mutation leading to MAPK/ERK pathway inhibition is associated with an autosomal dominant form of IgA nephropathy. European Journal of Human Genetics, 2015, 23, 1673-1678.	1.4	15
47	ATP1A3 spectrum disorders: A video-documented history of 7 genetically confirmed early onset cases. European Journal of Paediatric Neurology, 2018, 22, 264-271.	0.7	15
48	Genes for split hand/split foot and laterality defects on 7q22.1 and xq24-q27.1. American Journal of Medical Genetics Part A, 1994, 50, 101-101.	2.4	12
49	Genomic organization and embryonic expression ofSuppressor of Fused, a candidate gene for the split-hand/split-foot malformation type 3. FEBS Letters, 2001, 505, 13-17.	1.3	12
50	A novel microdeletion syndrome with loss of the MSH2 locus and hereditary non-polyposis colorectal cancer. Clinical Genetics, 2004, 67, 178-182.	1.0	11
51	Three Rett patients with both MECP2 mutation and 15q11â€“13 rearrangements. European Journal of Human Genetics, 2004, 12, 682-685.	1.4	11
52	New autosomal recessive syndrome of mental retardation, epilepsy, short stature, and skeletal dysplasia. American Journal of Medical Genetics Part A, 1992, 44, 315-320.	2.4	9
53	Recognizable facial features in patients with alternating hemiplegia of childhood. American Journal of Medical Genetics, Part A, 2016, 170, 2698-2705.	0.7	9
54	Identification of new candidate genes for spina bifida through exome sequencing. Child's Nervous System, 2021, 37, 2589-2596.	0.6	9

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55	The Simpsonâ€“Golabiâ€“Behmel syndrome: A clinical case and a detective story. American Journal of Medical Genetics, Part A, 2011, 155, 145-148.	0.7	8
56	Defective activation of the MAPK/ERK pathway, leading to PARP1 and DNMT1 dysregulation, is a common defect in IgA nephropathy and Henoch-SchÃ¶nlein purpura. Journal of Nephrology, 2018, 31, 731-741.	0.9	7
57	New syndrome of mental retardation, Robin sequence, and brachydactyly. American Journal of Medical Genetics Part A, 2001, 100, 49-51.	2.4	4
58	Variable expressivity of a familial 1.9ÂMb microdeletion in 3q28 leading to haploinsufficiency of TP63: Refinement of the critical region for a new microdeletion phenotype. European Journal of Medical Genetics, 2015, 58, 400-405.	0.7	4
59	ATP1A3 mutant patient with alternating hemiplegia of childhood and brain spectroscopic abnormalities. Journal of the Neurological Sciences, 2017, 379, 36-38.	0.3	4
60	The genetics of epilepsy. American Journal of Medical Genetics Part A, 2001, 106, 117-118.	2.4	2
61	Mental retardation, Robin sequence, and brachydactyly: Further confirmation of a new syndrome. American Journal of Medical Genetics Part A, 2004, 126A, 204-207.	2.4	2
62	Colchicine trial in PFAPA Syndrome and MEFV-negative patients. Pediatric Rheumatology, 2015, 13, .	0.9	2
63	In Silico Design of E3 Ubiquitin-Protein Ligase NEDD4-1 Inhibitors: An Alternative Approach for Targeting the MAPK Pathway in Cancer Therapy. Proceedings (mdpi), 2019, 22, .	0.2	1
64	A split hand-split foot (SHFM3) gene is located at 10Q24â†'25. , 1996, 62, 427.		1
65	Reply to Dr. Lin: Dilated cardiomyopathy in the SGB syndrome. American Journal of Medical Genetics Part A, 1993, 46, 607-607.	2.4	0
66	Encomium: Giovanni Neriâ€"Polyhedral and downâ€toâ€earth mentor. American Journal of Medical Genetics, Part A, 2013, 161, 2687-2690.	0.7	0
67	PFAPA syndrome as an hereditary autoinflamatory disorder. Pediatric Rheumatology, 2015, 13, .	0.9	0
68	Hypo-Phosphorylation of Salivary Peptidome as Indicator of Molecular Pathogenesis of Autism Spectrum Disorders. , 2014, , 1543-1563.		0