Marco Fichera

List of Publications by Year
in descending order

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Version: 2024-02-01

97 papers	7,649 citations	117571 34 h-index	54882 84 g-index
100 all docs	100 docs citations	100 times ranked	11002 citing authors

#	Article	IF	Citations
1	Discriminatory Weight of SNPs in Spike SARS-CoV-2 Variants: A Technically Rapid, Unambiguous, and Bioinformatically Validated Laboratory Approach. Viruses, 2022, 14, 123.	1.5	0
2	Mutational Analysis of BRCA1 and BRCA2 Genes in Breast Cancer Patients from Eastern Sicily. Cancer Management and Research, 2022, Volume 14, 1341-1352.	0.9	5
3	The embryo battle against adverse genomes: Are de novo terminal deletions the rescue of unfavorable zygotic imbalances?. European Journal of Medical Genetics, 2022, 65, 104532.	0.7	4
4	Targeted next-generation sequencing identifies the disruption of the SHANK3 and RYR2 genes in a patient carrying a de novo t(1;22)(q43;q13.3) associated with signs of Phelan-McDermid syndrome. Molecular Cytogenetics, 2020, 13, 22.	0.4	4
5	Clinical correlates in children with autism spectrum disorder and CNVs: Systematic investigation in a clinical setting. International Journal of Developmental Neuroscience, 2020, 80, 276-286.	0.7	6
6	Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. European Journal of Human Genetics, 2019, 27, 909-918.	1.4	21
7	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	1.1	127
8	Mutations in ACTL6B, coding for a subunit of the neuron-specific chromatin remodeling complex nBAF, cause early onset severe developmental and epileptic encephalopathy with brain hypomyelination and cerebellar atrophy. Human Genetics, 2019, 138, 187-198.	1.8	12
9	Biallelic intragenic duplication in ADGRB3 (BAI3) gene associated with intellectual disability, cerebellar atrophy, and behavioral disorder. European Journal of Human Genetics, 2019, 27, 594-602.	1.4	15
10	Novel c.C2254T (p.Q752*) mutation in ZFYVE26 (SPG15) gene in a patient with hereditary spastic paraparesis. Journal of Genetics, 2018, 97, 1469-1472.	0.4	3
11	Familial 18q12.2 deletion supports the role of RNAâ€binding protein CELF4 in autism spectrum disorders. American Journal of Medical Genetics, Part A, 2017, 173, 1649-1655.	0.7	18
12	Identification of novel mutations in L1CAM gene by a DHPLC-based assay. Genes and Genomics, 2016, 38, 1159-1164.	0.5	1
13	Antitumoural activity of a cytotoxic peptide of Lactobacillus casei peptidoglycan and its interaction with mitochondrial-bound hexokinase. Anti-Cancer Drugs, 2016, 27, 609-619.	0.7	20
14	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. Molecular Psychiatry, 2016, 21, 126-132.	4.1	142
15	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. Journal of Human Genetics, 2016, 61, 95-101.	1.1	29
16	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	0.7	22
17	Familial 1q22 microduplication associated with psychiatric disorders, intellectual disability and late-onset autoimmune inflammatory response. Molecular Cytogenetics, 2014, 7, 90.	0.4	5
18	Sox11 Is Required to Maintain Proper Levels of Hedgehog Signaling during Vertebrate Ocular Morphogenesis. PLoS Genetics, 2014, 10, e1004491.	1.5	48

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19	TBR1 is the candidate gene for intellectual disability in patients with a 2q24.2 interstitial deletion. American Journal of Medical Genetics, Part A, 2014, 164, 828-833.	0.7	52
20	Definition of 5q11.2 microdeletion syndrome reveals overlap with CHARGE syndrome and 22q11 deletion syndrome phenotypes. American Journal of Medical Genetics, Part A, 2014, 164, 2843-2848.	0.7	8
21	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	9.4	293
22	Definition of minimal duplicated region encompassing the XIAP and STAG2 genes in the Xq25 microduplication syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1923-1930.	0.7	15
23	Klippelâ€Trenaunay syndrome in a boy with concomitant ipsilateral overgrowth and undergrowth. American Journal of Medical Genetics, Part A, 2014, 164, 1262-1267.	0.7	12
24	Secondary cervical dystonic tremor after Japanese encephalitis. Neurological Sciences, 2014, 35, 491-493.	0.9	3
25	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Human Molecular Genetics, 2014, 23, 2752-2768.	1.4	140
26	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
27	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	13.5	637
28	Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1666-1676.	0.7	49
29	Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. Gene, 2014, 534, 435-439.	1.0	19
30	6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies. Molecular Cytogenetics, 2013, 6, 4.	0.4	23
31	Interhemispheric Balance in Parkinson's Disease: A Transcranial Magnetic Stimulation Study. Brain Stimulation, 2013, 6, 892-897.	0.7	46
32	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. Journal of Medical Genetics, 2013, 50, 802-811.	1.5	93
33	The duplication 17p13.3 phenotype: Analysis of 21 families delineates developmental, behavioral and brain abnormalities, and rare variant phenotypes. American Journal of Medical Genetics, Part A, 2013, 161, 1833-1852.	0.7	53
34	Intragenic ILRAPL1 deletion in a male patient with intellectual disability, mild dysmorphic signs, deafness, and behavioral problems. American Journal of Medical Genetics, Part A, 2013, 161, 1381-1385.	0.7	14
35	Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. Nature Methods, 2013, 10, 903-909.	9.0	31
36	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.9	61

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37	Brief Report: Peculiar Evolution of Autistic Behaviors in Two Unrelated Children with Brachidactyly-Mental Retardation Syndrome. Journal of Autism and Developmental Disorders, 2012, 42, 2202-2207.	1.7	9
38	Nevus vascularis mixtus (cutaneous vascular twin nevi) associated with intracranial vascular malformation of the Dykeâ€“Davidoffâ€“Masson type in two patients. American Journal of Medical Genetics, Part A, 2012, 158A, 2870-2880.	0.7	54
39	Apneic crises: A clue for MECP2 testing in severe neonatal hypotonia-respiratory failure. European Journal of Paediatric Neurology, 2012, 16, 744-748.	0.7	9
40	Effects of deletion and duplication in a patient with a 46,XX,der(7)t(7;17)(q36;p13)mat karyotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2239-2244.	0.7	3
41	Molecular and clinical characterization of a small duplication Xp in a human female with psychiatric disorders. Journal of Genetics, 2011, 90, 473-477.	0.4	5
42	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. American Journal of Human Genetics, 2011, 89, 551-563.	2.6	195
43	Creatine transporter defect diagnosed by proton NMR spectroscopy in males with intellectual disability. American Journal of Medical Genetics, Part A, 2011, 155, 2446-2452.	0.7	19
44	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	1.5	172
45	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. PLoS Genetics, 2011, 7, e1002334.	1.5	293
46	An unusual presentation ofÂBecker Nevus. European Journal of Dermatology, 2010, 20, 522-523.	0.3	6
47	A novel L1CAM mutation in a fetus detected by prenatal diagnosis. European Journal of Pediatrics, 2010, 169, 415-419.	1.3	9
48	Decreased expression of GRAF1/OPHN-1-L in the X-linked alpha thalassemia mental retardation syndrome. BMC Medical Genomics, 2010, 3, 28.	0.7	12
49	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	1.4	71
50	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	9.4	539
51	Novel deletion of the E3A ubiquitin protein ligase gene detected by multiplex ligation-dependent probe amplification in a patient with Angelman syndrome. Experimental and Molecular Medicine, 2010, 42, 842.	3.2	5
52	Coexistence of mitochondrial and nuclear DNA mutations in a woman with mitochondrial encephalomyopathy and double cortex. Mitochondrion, 2010, 10, 548-554.	1.6	2
53	Familial 1.1ÂMb deletion in chromosome Xq22.1 associated with mental retardation and behavioural disorders in female patients. European Journal of Medical Genetics, 2010, 53, 113-116.	0.7	20
54	A Syndrome with Coarse Face, Mental Retardation and Unusual Stereotyped Movements. Neuropediatrics, 2009, 40, 186-188.	0.3	1

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55	The molecular landscape of ASPM mutations in primary microcephaly. Journal of Medical Genetics, 2009, 46, 249-253.	1.5	91
56	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	9.4	511
57	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 394-400.	2.6	60
58	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 419.	2.6	2
59	Deletion 2p25.2: A cryptic chromosome abnormality in a patient with autism and mental retardation detected using aCGH. European Journal of Medical Genetics, 2009, 52, 67-70.	0.7	16
60	Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. European Journal of Medical Genetics, 2009, 52, 94-100.	0.7	157
61	CDKL5 MUTATIONS IN BOYS WITH SEVERE ENCEPHALOPATHY AND EARLY-ONSET INTRACTABLE EPILEPSY. Neurology, 2009, 73, 77-78.	1.5	19
62	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. Journal of Medical Genetics, 2009, 46, 511-523.	1.5	250
63	12q12 deletion: A new patient contributing to genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2008, 146A, 1354-1357.	0.7	10
64	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	9.4	509
65	Identification of non-recurrent submicroscopic genome imbalances: the advantage of genome-wide microarrays over targeted approaches. European Journal of Human Genetics, 2008, 16, 395-400.	1.4	14
66	Bilateral periventricular nodular heterotopia and lissencephaly in an infant with unbalanced t(12;17)(q24.31; p13.3) translocation. Developmental Medicine and Child Neurology, 2008, 50, 473-476.	1.1	7
67	Three new patients with dup(17)(p11.2p11.2) without autism. Clinical Genetics, 2008, 73, 294-296.	1.0	7
68	Partial monosomy Xq(Xq23â†'qter) and trisomy 4p(4p15.33â†'pter) in a woman with intractable focal epilepsy, borderline intellectual functioning, and dysmorphic features. Brain and Development, 2008, 30, 425-429.	0.6	10
69	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
70	Posterior fossa abnormalities in hereditary spastic paraparesis with spastin mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 80, 440-443.	0.9	11
71	CDKL5 mutations in boys with severe encephalopathy and early-onset intractable epilepsy. Neurology, 2008, 71, 997-999.	1.5	84
72	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. Journal of Medical Genetics, 2007, 44, 750-762.	1.5	244

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73	Severe encephalomyopathy in a patient with homoplasmic A5814G point mutation in mitochondrial tRNACys gene. Neuromuscular Disorders, 2007, 17, 258-261.	0.3	11
74	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. Clinica Chimica Acta, 2007, 384, 35-40.	0.5	8
75	The Italian XLMR bank: a clinical and molecular database. Human Mutation, 2007, 28, 13-18.	1.1	2
76	1.5 Mb de novo 22q11.21 microduplication in a patient with cognitive deficits and dysmorphic facial features. Clinical Genetics, 2007, 71, 177-182.	1.0	52
77	Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. Clinical Genetics, 2007, 71, 599-601.	1.0	38
78	6q Terminal Deletion Syndrome Associated with a Distinctive EEG and Clinical Pattern: A Report of Five Cases. Epilepsia, 2006, 47, 830-838.	2.6	44
79	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	9.4	418
80	Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. Neurological Sciences, 2006, 27, 425-431.	0.9	6
81	A Missense Mutation in the Coiled-Coil Domain of the KIF5A Gene and Late-Onset Hereditary Spastic Paraplegia. Archives of Neurology, 2006, 63, 284.	4.9	49
82	Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. Clinical Genetics, 2005, 67, 446-447.	1.0	11
83	Identification of Novel Mutations in Patients with Coffinâ€“Lowry Syndrome by a Denaturing HPLC-Based Assay. Clinical Chemistry, 2005, 51, 2356-2358.	1.5	5
84	Denaturing HPLC-Based Assay for Detection of ATRX Gene Mutations. Clinical Chemistry, 2005, 51, 1314-1315.	1.5	2
85	Narrowing the Candidate Region for Congenital Diaphragmatic Hernia in Chromosome 15q26: Contradictory Results. American Journal of Human Genetics, 2005, 77, 892-894.	2.6	20
86	Denaturing HPLC-Based Assay for Molecular Screening of Nondeletional Mutations Causing Î±-Thalassemias. Clinical Chemistry, 2004, 50, 1242-1245.	1.5	8
87	Evidence of kinesin heavy chain (KIF5A) involvement in pure hereditary spastic paraplegia. Neurology, 2004, 63, 1108-1110.	1.5	105
88	Two novel mutations in the spastin gene (SPG4) found by DHPLC mutation analysis. Neuromuscular Disorders, 2004, 14, 750-753.	0.3	19
89	Mutational analysis of the ATRX gene by DGGE: A powerful diagnostic approach for the ATRX syndrome. Human Mutation, 2003, 21, 529-534.	1.1	10
90	A new MRXS locus maps to the X chromosome pericentromeric region: a new syndrome or narrow definition of Sutherland-Haan genetic locus?. Journal of Medical Genetics, 2002, 39, 276-280.	1.5	3

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91	Spontaneous transmission from a father to his son of a Y chromosome microdeletion involving the deleted in azoospermia (DAZ) gene. Journal of Endocrinological Investigation, 2002, 25, 631-634.	1.8	38
92	Prenatal diagnosis of ATR-X syndrome in a fetus with a new G>T splicing mutation in the XNP/ATR-X gene. Prenatal Diagnosis, 2001, 21, 747-751.	1.1	17
93	Genetic variations in human fetal globin gene microsatellites and their functional relevance. Human Genetics, 1999, 104, 307-314.	1.8	18
94	Evaluation of a mutation screening strategy for sporadic cases of ATR-X syndrome. Journal of Medical Genetics, 1999, 36, 183-6.	1.5	10
95	Physical map of the D6S149-D6S193 region on chromosome 6Q27 and its involement in benign surface epithelial ovarian tumours. Oncogene, 1998, 16, 1639-1642.	2.6	26
96	Molecular basis of Î±-thalassemia in Sicily. Human Genetics, 1997, 99, 381-386.	1.8	24
97	Quantitative evaluation of partial deletions of the DAZ gene cluster. International Journal of Molecular Medicine, 0, , .	1.8	2