Marco Fichera

List of Publications by Year
in descending order

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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	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
2	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	13.5	637
3	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
4	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	9.4	539
5	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	9.4	511
6	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	9.4	509
7	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	9.4	418
8	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. PLoS Genetics, 2011, 7, e1002334.	1.5	293
9	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	9.4	293
10	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
11	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
12	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
13	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	1.5	172
14	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
15	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. Molecular Psychiatry, 2016, 21, 126-132.	4.1	142
16	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
17	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
18	Evidence of kinesin heavy chain (KIF5A) involvement in pure hereditary spastic paraplegia. Neurology, 2004, 63, 1108-1110.	1.5	105

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19	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
20	The molecular landscape of ASPM mutations in primary microcephaly. Journal of Medical Genetics, 2009, 46, 249-253.	1.5	91
21	CDKL5 mutations in boys with severe encephalopathy and early-onset intractable epilepsy. Neurology, 2008, 71, 997-999.	1.5	84
22	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	1.4	71
23	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.9	61
24	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
25	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
26	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
27	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
28	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
29	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
30	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
31	Sox11 Is Required to Maintain Proper Levels of Hedgehog Signaling during Vertebrate Ocular Morphogenesis. PLoS Genetics, 2014, 10, e1004491.	1.5	48
32	Interhemispheric Balance in Parkinson's Disease: A Transcranial Magnetic Stimulation Study. Brain Stimulation, 2013, 6, 892-897.	0.7	46
33	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
34	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
35	Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. Clinical Genetics, 2007, 71, 599-601.	1.0	38
36	Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. Nature Methods, 2013, 10, 903-909.	9.0	31

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37	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
38	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
39	Molecular basis of Î±-thalassemia in Sicily. Human Genetics, 1997, 99, 381-386.	1.8	24
40	6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies. Molecular Cytogenetics, 2013, 6, 4.	0.4	23
41	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	0.7	22
42	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
43	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
44	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
45	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
46	Two novel mutations in the spastin gene (SPG4) found by DHPLC mutation analysis. Neuromuscular Disorders, 2004, 14, 750-753.	0.3	19
47	CDKL5 MUTATIONS IN BOYS WITH SEVERE ENCEPHALOPATHY AND EARLY-ONSET INTRACTABLE EPILEPSY. Neurology, 2009, 73, 77-78.	1.5	19
48	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
49	Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. Gene, 2014, 534, 435-439.	1.0	19
50	Genetic variations in human fetal globin gene microsatellites and their functional relevance. Human Genetics, 1999, 104, 307-314.	1.8	18
51	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
52	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
53	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
54	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

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55	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
56	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
57	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
58	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
59	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
60	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
61	Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. Clinical Genetics, 2005, 67, 446-447.	1.0	11
62	Severe encephalomyopathy in a patient with homoplasmic A5814G point mutation in mitochondrial tRNACys gene. Neuromuscular Disorders, 2007, 17, 258-261.	0.3	11
63	Posterior fossa abnormalities in hereditary spastic paraparesis with spastin mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 80, 440-443.	0.9	11
64	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
65	12q12 deletion: A new patient contributing to genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2008, 146A, 1354-1357.	0.7	10
66	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
67	Evaluation of a mutation screening strategy for sporadic cases of ATR-X syndrome. Journal of Medical Genetics, 1999, 36, 183-6.	1.5	10
68	A novel L1CAM mutation in a fetus detected by prenatal diagnosis. European Journal of Pediatrics, 2010, 169, 415-419.	1.3	9
69	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
70	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
71	Denaturing HPLC-Based Assay for Molecular Screening of Nondeletional Mutations Causing Î±-Thalassemias. Clinical Chemistry, 2004, 50, 1242-1245.	1.5	8
72	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. Clinica Chimica Acta, 2007, 384, 35-40.	0.5	8

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73	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
74	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
75	Three new patients with dup(17)(p11.2p11.2) without autism. Clinical Genetics, 2008, 73, 294-296.	1.0	7
76	Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. Neurological Sciences, 2006, 27, 425-431.	0.9	6
77	An unusual presentation ofÂBecker Nevus. European Journal of Dermatology, 2010, 20, 522-523.	0.3	6
78	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
79	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
80	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
81	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
82	Familial 1q22 microduplication associated with psychiatric disorders, intellectual disability and late-onset autoimmune inflammatory response. Molecular Cytogenetics, 2014, 7, 90.	0.4	5
83	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
84	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
85	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
86	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
87	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
88	Secondary cervical dystonic tremor after Japanese encephalitis. Neurological Sciences, 2014, 35, 491-493.	0.9	3
89	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
90	Denaturing HPLC-Based Assay for Detection of ATRX Gene Mutations. Clinical Chemistry, 2005, 51, 1314-1315.	1.5	2

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91	The Italian XLMR bank: a clinical and molecular database. Human Mutation, 2007, 28, 13-18.	1.1	2
92	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
93	Coexistence of mitochondrial and nuclear DNA mutations in a woman with mitochondrial encephalomyopathy and double cortex. Mitochondrion, 2010, 10, 548-554.	1.6	2
94	Quantitative evaluation of partial deletions of the DAZ gene cluster. International Journal of Molecular Medicine, 0, , .	1.8	2
95	A Syndrome with Coarse Face, Mental Retardation and Unusual Stereotyped Movements. Neuropediatrics, 2009, 40, 186-188.	0.3	1
96	Identification of novel mutations in L1CAM gene by a DHPLC-based assay. Genes and Genomics, 2016, 38, 1159-1164.	0.5	1
97	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