Francesca Mari

List of Publications by Year
in descending order

131 papers	6,157 citations	76196 40 h-index	85405 71 g-index
145 all docs	145 docs citations	145 times ranked	8960 citing authors

#	Article	IF	Citations
1	Dyskeratosis Congenita and Cancer in Mice Deficient in Ribosomal RNA Modification. Science, 2003, 299, 259-262.	6.0	387
2	FOXG1 Is Responsible for the Congenital Variant of Rett Syndrome. American Journal of Human Genetics, 2008, 83, 89-93.	2.6	366
3	CDKL5 belongs to the same molecular pathway of MeCP2 and it is responsible for the early-onset seizure variant of Rett syndrome. Human Molecular Genetics, 2005, 14, 1935-1946.	1.4	279
4	A Mutation in the Rett Syndrome Gene, MECP2, Causes X-Linked Mental Retardation and Progressive Spasticity in Males. American Journal of Human Genetics, 2000, 67, 982-985.	2.6	213
5	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. European Journal of Human Genetics, 2020, 28, 1602-1614.	1.4	208
6	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. Journal of Medical Genetics, 2005, 42, 103-107.	1.5	206
7	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	1.4	190
8	COL4A3/COL4A4 mutations: From familial hematuria to autosomal-dominant or recessive Alport syndrome. Kidney International, 2002, 61, 1947-1956.	2.6	187
9	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males: findings from a nested case-control study. ELife, 2021, 10, .	2.8	145
10	Evidence of digenic inheritance in Alport syndrome. Journal of Medical Genetics, 2015, 52, 163-174.	1.5	129
11	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
12	Autosomal-dominant Alport syndrome: Natural history of a disease due to COL4A3 or COL4A4 gene. Kidney International, 2004, 65, 1598-1603.	2.6	124
13	Unbiased next generation sequencing analysis confirms the existence of autosomal dominant Alport syndrome in a relevant fraction of cases. Clinical Genetics, 2014, 86, 252-257.	1.0	121
14	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
15	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. Pediatric Nephrology, 2019, 34, 1175-1189.	0.9	97
16	Real-time quantitative PCR as a routine method for screening large rearrangements in Rett syndrome: Report of one case of MECP2 deletion and one case of MECP2 duplication. Human Mutation, 2004, 24, 172-177.	1.1	96
17	Identification of sixty-two novel and twelve known FBN1 mutations in eighty-one unrelated probands with Marfan syndrome and other fibrillinopathies. Human Mutation, 2005, 26, 494-494.	1.1	83
18	Autosomal dominant Alport syndrome: molecular analysis of the COL4A4 gene and clinical outcome. Nephrology Dialysis Transplantation, 2009, 24, 1464-1471.	0.4	81

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19	Rett syndrome: the complex nature of a monogenic disease. Journal of Molecular Medicine, 2003, 81, 346-354.	1.7	80
20	iPS cells to model CDKL5-related disorders. European Journal of Human Genetics, 2011, 19, 1246-1255.	1.4	80
21	Advances in Alport syndrome diagnosis using next-generation sequencing. European Journal of Human Genetics, 2012, 20, 50-57.	1.4	76
22	Study ofMECP2 gene in Rett syndrome variants and autistic girls. American Journal of Medical Genetics Part A, 2003, 119B, 102-107.	2.4	67
23	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	1.1	67
24	GluD1 is a common altered player in neuronal differentiation from both MECP2-mutated and CDKL5-mutated iPS cells. European Journal of Human Genetics, 2015, 23, 195-201.	1.4	65
25	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. European Journal of Human Genetics, 2021, 29, 1186-1197.	1.4	61
26	Phosphatase and Tensin Homolog (PTEN) Gene Mutations and Autism: Literature Review and a Case Report of a Patient With Cowden Syndrome, Autistic Disorder, and Epilepsy. Journal of Child Neurology, 2012, 27, 392-397.	0.7	60
27	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. Orphanet Journal of Rare Diseases, 2013, 8, 63.	1.2	60
28	Private inherited microdeletion/microduplications: Implications in clinical practice. European Journal of Medical Genetics, 2008, 51, 409-416.	0.7	59
29	A 3 Mb deletion in 14q12 causes severe mental retardation, mild facial dysmorphisms and Rettâ€like features. American Journal of Medical Genetics, Part A, 2008, 146A, 1994-1998.	0.7	56
30	Revealing the Complexity of a Monogenic Disease: Rett Syndrome Exome Sequencing. PLoS ONE, 2013, 8, e56599.	1.1	54
31	Alport syndrome: impact of digenic inheritance in patients management. Clinical Genetics, 2017, 92, 34-44.	1.0	52
32	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	2.7	52
33	2q24â€“q31 Deletion: Report of a case and review of the literature. European Journal of Medical Genetics, 2007, 50, 21-32.	0.7	49
34	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
35	Guidelines for Genetic Testing and Management of Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 143-154.	2.2	49
36	Chromosome 2 deletion encompassing the MAP2 gene in a patient with autism and Rett-like features. Clinical Genetics, 2003, 64, 497-501.	1.0	48

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37	Epilepsy in Rett syndromeâ€"Lessons from the Rett networked database. Epilepsia, 2015, 56, 569-576.	2.6	47
38	MECP2 deletions and genotypeâ€"phenotype correlation in Rett syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 2775-2784.	0.7	45
39	Redox Imbalance and Morphological Changes in Skin Fibroblasts in Typical Rett Syndrome. Oxidative Medicine and Cellular Longevity, 2014, 2014, 1-10.	1.9	44
40	Xq28 duplications including MECP2 in five females: Expanding the phenotype toÂsevere mental retardation. European Journal of Medical Genetics, 2012, 55, 404-413.	0.7	42
41	Epilepsy in Mowatâ€"Wilson syndrome: Delineation of the electroclinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 273-284.	0.7	42
42	Genomic differences between retinoma and retinoblastoma. Acta OncolÃ³gica, 2008, 47, 1483-1492.	0.8	41
43	Visual impairment in FOXG1-mutated individuals and mice. Neuroscience, 2016, 324, 496-508.	1.1	41
44	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients. Genes and Immunity, 2022, 23, 51-56.	2.2	41
45	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. Nephrology Dialysis Transplantation, 2006, 21, 665-671.	0.4	40
46	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. European Journal of Medical Genetics, 2009, 52, 148-152.	0.7	40
47	Advances and unmet needs in genetic, basic and clinical science in Alport syndrome: report from the 2015 International Workshop on Alport Syndrome. Nephrology Dialysis Transplantation, 2017, 32, gfw095.	0.4	40
48	Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes, 2021, 12, 596.	1.0	39
49	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. Cancer Science, 2009, 100, 465-471.	1.7	38
50	Clinical and molecular characterization of a patient with a 2q31.2-32.3 deletion identified by array-CGH. American Journal of Medical Genetics, Part A, 2007, 143A, 858-865.	0.7	37
51	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. European Journal of Human Genetics, 2015, 23, 1523-1530.	1.4	37
52	Non-syndromic X-linked mental retardation: From a molecular to a clinical point of view. Journal of Cellular Physiology, 2005, 204, 8-20.	2.0	36
53	Dual copy number variants involving 16p11 and 6q22 in a case of childhood apraxia of speech and pervasive developmental disorder. European Journal of Human Genetics, 2013, 21, 361-365.	1.4	36
54	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated Î±-tubulin defect which improves after iHDAC6 treatment in Rett syndrome. Experimental Cell Research, 2018, 368, 225-235.	1.2	36

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55	Employing a systematic approach to biobanking and analyzing clinical and genetic data for advancing COVID-19 research. European Journal of Human Genetics, 2021, 29, 745-759.	1.4	35
56	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. Brain, 2020, 143, 2380-2387.	3.7	34
57	Refinement of the 12q14 microdeletion syndrome: primordial dwarfism and developmental delay with or without osteopoikilosis. European Journal of Human Genetics, 2009, 17, 1141-1147.	1.4	33
58	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. Clinical Genetics, 2005, 67, 258-260.	1.0	32
59	Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes are a common well recognizable cause of intellectual disability. Brain and Development, 2015, 37, 527-536.	0.6	32
60	High frequency of COH1 intragenic deletions and duplications detected by MLPA in patients with Cohen syndrome. European Journal of Human Genetics, 2010, 18, 1133-1140.	1.4	31
61	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. Lung Cancer, 2014, 85, 168-174.	0.9	30
62	Mutational screening of the RB1 gene in Italian patients with retinoblastoma reveals 11 novel mutations. Journal of Human Genetics, 2006, 51, 209-216.	1.1	29
63	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
64	Italian Rett database and biobank. Human Mutation, 2007, 28, 329-335.	1.1	27
65	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. Pediatric Nephrology, 2011, 26, 717-724.	0.9	27
66	Thin glomerular basement membrane disease: clinical significance of a morphological diagnosis--a collaborative study of the Italian Renal Immunopathology Group. Nephrology Dialysis Transplantation, 2005, 20, 545-551.	0.4	26
67	A spectrum of LMX1B mutations in Nail-Patella syndrome: New point mutations, deletion, and evidence of mosaicism in unaffected parents. Genetics in Medicine, 2010, 12, 431-439.	1.1	26
68	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. Journal of Human Genetics, 2007, 52, 1011-1017.	1.1	25
69	Investigation of modifier genes within copy number variations in Rett syndrome. Journal of Human Genetics, 2011, 56, 508-515.	1.1	25
70	Clinical and molecular characterization of COVID-19 hospitalized patients. PLoS ONE, 2020, 15, e0242534.	1.1	25
71	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males. Autophagy, 2022, 18, 1662-1672.	4.3	25
72	Syndromic mental retardation with thrombocytopenia due to 21q22.11q22.12 deletion: Report of three patients. American Journal of Medical Genetics, Part A, 2010, 152A, 1711-1717.	0.7	23

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73	Five patients with novel overlapping interstitial deletions in 8q22.2q22.3. American Journal of Medical Genetics, Part A, 2011, 155, 1857-1864.	0.7	23
74	Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9.	0.8	23
75	MEIS2 gene is responsible for intellectual disability, cardiac defects and a distinct facial phenotype. European Journal of Medical Genetics, 2020, 63, 103627.	0.7	23
76	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. Brain, 2020, 143, 3564-3573.	3.7	23
77	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing. International Journal of Molecular Sciences, 2021, 22, 13439.	1.8	23
78	Delineation of the phenotype associated with 7q36.1q36.2 deletion: Long QT syndrome, renal hypoplasia and mental retardation. American Journal of Medical Genetics, Part A, 2008, 146A, 1195-1199.	0.7	22
79	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	1.8	22
80	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. Molecular Genetics and Metabolism, 2016, 119, 214-222.	0.5	21
81	Spectrum ofPTCH mutations in Italian nevoid basal cell-carcinoma syndrome patients: Identification of thirteen novel alleles. Human Mutation, 2004, 24, 441-441.	1.1	20
82	CHARGEâ€like presentation, craniosynostosis and mild Mowatâ€“Wilson Syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases. American Journal of Medical Genetics, Part A, 2014, 164, 2557-2566.	0.7	20
83	Retinoblastoma and mental retardation microdeletion syndrome: clinical characterization and molecular dissection using array CGH. Journal of Human Genetics, 2007, 52, 535-542.	1.1	19
84	Alâ€Awadiâ€“Raasâ€Rothschild (limb/pelvis/uterusâ€“hypoplasia/aplasia) syndrome and WNT7A mutations: Genetic homogeneity and nosological delineation. American Journal of Medical Genetics, Part A, 2011, 155, 332-336.	0.7	19
85	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
86	Rett networked database: An integrated clinical and genetic network of rett syndrome databases. Human Mutation, 2012, 33, 1031-1036.	1.1	19
87	A 9.3Mb microdeletion of 3q27.3q29 associated with psychomotor and growth delay, tricuspid valve dysplasia and bifid thumb. European Journal of Medical Genetics, 2009, 52, 131-133.	0.7	18
88	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. Clinical Dysmorphology, 2008, 17, 13-17.	0.1	17
89	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. Brain Sciences, 2021, 11, 936.	1.1	17
90	3.2 Mb microdeletion in chromosome 7 bands q22.2â€“q22.3 associated with overgrowth and delayed bone age. European Journal of Medical Genetics, 2010, 53, 168-170.	0.7	16

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91	Ambiguous external genitalia due to defect of 5-Î±-reductase in seven Iraqi patients: Prevalence of a novel mutation. Gene, 2013, 526, 490-493.	1.0	16
92	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. Human Mutation, 2018, 39, 302-314.	1.1	16
93	Evidence of predisposing epimutation in retinoblastoma. Human Mutation, 2019, 40, 201-206.	1.1	16
94	Severe COVID-19 in Hospitalized Carriers of Single CFTR Pathogenic Variants. Journal of Personalized Medicine, 2021, 11, 558.	1.1	16
95	A 2.6Mb deletion of 6q24.3â€“25.1 in a patient with growth failure, cardiac septal defect, thin upperlip and asymmetric dysmorphic ears. European Journal of Medical Genetics, 2007, 50, 315-321.	0.7	15
96	Leukoencephalopathy in 21-Î² hydroxylase deficiency: Report of a family. Brain and Development, 2010, 32, 421-424.	0.6	13
97	EEG features and epilepsy in MECP2-mutated patients with the Zappella variant of Rett syndrome. Clinical Neurophysiology, 2010, 121, 652-657.	0.7	13
98	Optineurin gene is not involved in the common high-tension form of primary open-angle glaucoma. Graefe's Archive for Clinical and Experimental Ophthalmology, 2006, 244, 1077-1082.	1.0	12
99	Frequency of the LRRK2 G2019S mutation in Italian patients affected by Parkinson's disease. Journal of Human Genetics, 2007, 52, 201-204.	1.1	12
100	Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome: Clinical and Molecular Analysis of a Case. Journal of AAPOS, 2006, 10, 279-280.	0.2	11
101	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. European Journal of Medical Genetics, 2014, 57, 163-168.	0.7	11
102	Exome sequencing analysis in a pair of monozygotic twins re-evaluates the genetics behind their intellectual disability and reveals a CHD2 mutation. Brain and Development, 2016, 38, 590-596.	0.6	11
103	<scp>IQSEC2</scp> disorder: A new disease entity or a Rett spectrum continuum?. Clinical Genetics, 2021, 99, 462-474.	1.0	11
104	SELP Asp603Asn and severe thrombosis in COVID-19 males. Journal of Hematology and Oncology, 2021, 14, 123.	6.9	11
105	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. European Journal of Human Genetics, 2016, 24, 252-257.	1.4	10
106	9q31.1q31.3 deletion in two patients with similar clinical features: A newly recognized microdeletion syndrome?. American Journal of Medical Genetics, Part A, 2014, 164, 685-690.	0.7	9
107	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. Clinica Chimica Acta, 2007, 384, 35-40.	0.5	8
108	Disruption of the IQSEC2 transcript in a female with X;autosome translocation t(X;20)(p11.2;q11.2) and a phenotype resembling X-linked infantile spasms (ISSX) syndrome. Molecular Medicine Reports, 2008, , .	1.1	8

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109	A Unique Patient Presenting With Concomitant Klinefelter Syndrome, Alport Syndrome, and Craniopharyngioma. Journal of Andrology, 2012, 33, 1155-1159.	2.0	7
110	Personalized therapy in a GRIN1 mutated girl with intellectual disability and epilepsy. Clinical Dysmorphology, 2018, 27, 18-20.	0.1	7
111	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
112	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	1.1	6
113	Otosclerosis: exclusion of linkage to the OTSC1 and OTSC2 loci in four Italian families: Otoesclerosis: exclusiÃ³n de enlaces entre los loci OTSC1 y OTSC2 en cuatro familias italianas. International Journal of Audiology, 2003, 42, 475-480.	0.9	5
114	Potentially Treatable Disorder Diagnosed Post Mortem by Exome Analysis in a Boy with Respiratory Distress. International Journal of Molecular Sciences, 2016, 17, 306.	1.8	5
115	Should a syndrome be called by its correct name? The example of the preserved speech variant of Rett syndrome. European Journal of Pediatrics, 2005, 164, 710-710.	1.3	4
116	13q Deletion syndrome and retinoblastoma in identical dichorionic diamniotic monozygotic twins. European Journal of Ophthalmology, 2012, 22, 857-860.	0.7	4
117	Nicolaidesâ€“Baraitser syndrome: defining a phenotype. Journal of Neurology, 2016, 263, 1659-1660.	1.8	4
118	Genetics and mechanisms of disease in Rett syndrome. Drug Discovery Today Disease Mechanisms, 2005, 2, 419-425.	0.8	3
119	Sporadic hereditary motor and sensory neuropathies: Advances in the diagnosis using next generation sequencing technology. Journal of the Neurological Sciences, 2015, 359, 409-417.	0.3	3
120	Combined ultrasound and exome sequencing approach recognizes Opitz G/BBB syndrome in two malformed fetuses. Clinical Dysmorphology, 2017, 26, 18-25.	0.1	3
121	Understanding the new <scp>BRD4</scp>â€related syndrome: Clinical and genomic delineation with an international cohort study. Clinical Genetics, 2022, 102, 117-122.	1.0	3
122	The Italian XLMR bank: a clinical and molecular database. Human Mutation, 2007, 28, 13-18.	1.1	2
123	Response to Phelan K. et al.: Letter to the Editor Regarding Disciglio et al: Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1681-1681.	0.7	2
124	Epilepsy in Nicolaidesâ€“Baraitser Syndrome: Review of Literature and Report of 25 Patients Focusing on Treatment Aspects. Neuropediatrics, 2021, 52, 109-122.	0.3	2
125	13q Deletion Syndrome Involving RB1: Characterization of a New Minimal Critical Region for Psychomotor Delay. Genes, 2021, 12, 1318.	1.0	2
126	Shorter Androgen Receptor PolyQ Alleles Protect Against Life-Threatening COVID-19 Disease in Males. SSRN Electronic Journal, 0, , .	0.4	2

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127	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. Frontiers in Oncology, 2021, 11, 649435.	1.3	2
128	Huntington's disease gene expansion associates with early onset nonprogressive chorea. Movement Disorders, 2013, 28, 684-684.	2.2	1
129	Triorchidism: Genetic and imaging evaluation in an adult male. Archivio Italiano Di Urologia Andrologia, 2014, 86, 156.	0.4	1
130	Autism Spectrum Disorders: Analysis of Mobile Elements at 7q11.23 Williamsâ€“Beuren Region by Comparative Genomics. Genes, 2021, 12, 1605.	1.0	1
131	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis. Neurological Sciences, 2022, 43, 2849-2852.	0.9	0