

Francesca Mari

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

132
papers

4,666
citations

36
h-index

64
g-index

145
ext. papers

5,498
ext. citations

4.7
avg, IF

4.4
L-index

#	Paper	IF	Citations
132	Dyskeratosis congenita and cancer in mice deficient in ribosomal RNA modification. <i>Science</i> , 2003 , 299, 259-62	33.3	340
131	FOXP1 is responsible for the congenital variant of Rett syndrome. <i>American Journal of Human Genetics</i> , 2008 , 83, 89-93	11	312
130	CDKL5 belongs to the same molecular pathway of MeCP2 and it is responsible for the early-onset seizure variant of Rett syndrome. <i>Human Molecular Genetics</i> , 2005 , 14, 1935-46	5.6	248
129	A mutation in the rett syndrome gene, MECP2, causes X-linked mental retardation and progressive spasticity in males. <i>American Journal of Human Genetics</i> , 2000 , 67, 982-5	11	196
128	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. <i>Journal of Medical Genetics</i> , 2005 , 42, 103-7	5.8	180
127	COL4A3/COL4A4 mutations: from familial hematuria to autosomal-dominant or recessive Alport syndrome. <i>Kidney International</i> , 2002 , 61, 1947-56	9.9	143
126	A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. <i>Human Molecular Genetics</i> , 2013 , 22, 5121-35	5.6	138
125	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. <i>European Journal of Human Genetics</i> , 2020 , 28, 1602-1614	5.3	132
124	Evidence of digenic inheritance in Alport syndrome. <i>Journal of Medical Genetics</i> , 2015 , 52, 163-74	5.8	95
123	Unbiased next generation sequencing analysis confirms the existence of autosomal dominant Alport syndrome in a relevant fraction of cases. <i>Clinical Genetics</i> , 2014 , 86, 252-7	4	92
122	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. <i>Human Mutation</i> , 2004 , 24, 172-7	4.7	92
121	Autosomal-dominant Alport syndrome: natural history of a disease due to COL4A3 or COL4A4 gene. <i>Kidney International</i> , 2004 , 65, 1598-603	9.9	90
120	Mowat-Wilson syndrome: facial phenotype changing with age: study of 19 Italian patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 417-26	2.5	83
119	Identification of sixty-two novel and twelve known FBN1 mutations in eighty-one unrelated probands with Marfan syndrome and other fibrillinopathies. <i>Human Mutation</i> , 2005 , 26, 494	4.7	74
118	Autosomal dominant Alport syndrome: molecular analysis of the COL4A4 gene and clinical outcome. <i>Nephrology Dialysis Transplantation</i> , 2009 , 24, 1464-71	4.3	71
117	iPS cells to model CDKL5-related disorders. <i>European Journal of Human Genetics</i> , 2011 , 19, 1246-55	5.3	71
116	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. <i>Genetics in Medicine</i> , 2019 , 21, 816-825	8.1	71

115	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. <i>Pediatric Nephrology</i> , 2019 , 34, 1175-1189	3.2	70
114	Rett syndrome: the complex nature of a monogenic disease. <i>Journal of Molecular Medicine</i> , 2003 , 81, 346-54	5.5	67
113	Advances in Alport syndrome diagnosis using next-generation sequencing. <i>European Journal of Human Genetics</i> , 2012 , 20, 50-7	5.3	66
112	Study of MECP2 gene in Rett syndrome variants and autistic girls. <i>American Journal of Medical Genetics Part A</i> , 2003 , 119B, 102-7		61
111	GluD1 is a common altered player in neuronal differentiation from both MECP2-mutated and CDKL5-mutated iPS cells. <i>European Journal of Human Genetics</i> , 2015 , 23, 195-201	5.3	56
110	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. <i>Journal of Child Neurology</i> , 2012 , 27, 392-7	2.5	52
109	Private inherited microdeletion/microduplications: implications in clinical practice. <i>European Journal of Medical Genetics</i> , 2008 , 51, 409-16	2.6	51
108	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males: findings from a nested case-control study. <i>ELife</i> , 2021 , 10,	8.9	51
107	A 3 Mb deletion in 14q12 causes severe mental retardation, mild facial dysmorphisms and Rett-like features. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 1994-8	2.5	50
106	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. <i>Orphanet Journal of Rare Diseases</i> , 2013 , 8, 63	4.2	47
105	2q24-q31 deletion: report of a case and review of the literature. <i>European Journal of Medical Genetics</i> , 2007 , 50, 21-32	2.6	46
104	Revealing the complexity of a monogenic disease: rett syndrome exome sequencing. <i>PLoS ONE</i> , 2013 , 8, e56599	3.7	45
103	MECP2 deletions and genotype-phenotype correlation in Rett syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 2775-84	2.5	41
102	Chromosome 2 deletion encompassing the MAP2 gene in a patient with autism and Rett-like features. <i>Clinical Genetics</i> , 2003 , 64, 497-501	4	41
101	Xq28 duplications including MECP2 in five females: Expanding the phenotype to severe mental retardation. <i>European Journal of Medical Genetics</i> , 2012 , 55, 404-13	2.6	38
100	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. <i>Nephrology Dialysis Transplantation</i> , 2006 , 21, 665-71	4.3	38
99	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. <i>Genetics in Medicine</i> , 2018 , 20, 965-975	8.1	37
98	Interstitial 22q13 deletions not involving SHANK3 gene: a new contiguous gene syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1666-76	2.5	36

97	Epilepsy in Mowat-Wilson syndrome: delineation of the electroclinical phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 273-84	2.5	36
96	Redox imbalance and morphological changes in skin fibroblasts in typical Rett syndrome. <i>Oxidative Medicine and Cellular Longevity</i> , 2014 , 2014, 195935	6.7	36
95	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. <i>European Journal of Medical Genetics</i> , 2009 , 52, 148-52	2.6	36
94	Genomic differences between retinoma and retinoblastoma. <i>Acta Oncologica</i> , 2008 , 47, 1483-92	3.2	34
93	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. <i>European Journal of Human Genetics</i> , 2015 , 23, 1523-30	5.3	33
92	Alport syndrome: impact of digenic inheritance in patients management. <i>Clinical Genetics</i> , 2017 , 92, 34-44		33
91	Clinical and molecular characterization of a patient with a 2q31.2-32.3 deletion identified by array-CGH. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 858-65	2.5	32
90	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated β -tubulin defect which improves after iHDAC6 treatment in Rett syndrome. <i>Experimental Cell Research</i> , 2018 , 368, 225-235	4.2	31
89	Dual copy number variants involving 16p11 and 6q22 in a case of childhood apraxia of speech and pervasive developmental disorder. <i>European Journal of Human Genetics</i> , 2013 , 21, 361-5	5.3	31
88	Advances and unmet needs in genetic, basic and clinical science in Alport syndrome: report from the 2015 International Workshop on Alport Syndrome. <i>Nephrology Dialysis Transplantation</i> , 2017 , 32, 916-924	4.3	31
87	Epilepsy in Rett syndrome--lessons from the Rett networked database. <i>Epilepsia</i> , 2015 , 56, 569-76	6.4	30
86	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. <i>Cancer Science</i> , 2009 , 100, 465-71	6.9	30
85	Refinement of the 12q14 microdeletion syndrome: primordial dwarfism and developmental delay with or without osteopoikilosis. <i>European Journal of Human Genetics</i> , 2009 , 17, 1141-7	5.3	30
84	Non-syndromic X-linked mental retardation: from a molecular to a clinical point of view. <i>Journal of Cellular Physiology</i> , 2005 , 204, 8-20	7	30
83	High frequency of COH1 intragenic deletions and duplications detected by MLPA in patients with Cohen syndrome. <i>European Journal of Human Genetics</i> , 2010 , 18, 1133-40	5.3	28
82	Visual impairment in FOXP1-mutated individuals and mice. <i>Neuroscience</i> , 2016 , 324, 496-508	3.9	27
81	Coffin-Siris and Nicolaides-Baraitser syndromes are a common well recognizable cause of intellectual disability. <i>Brain and Development</i> , 2015 , 37, 527-36	2.2	26
80	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. <i>Clinical Genetics</i> , 2005 , 67, 258-60		25

79	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. <i>EBioMedicine</i> , 2021 , 65, 103246	8.8	25
78	Investigation of modifier genes within copy number variations in Rett syndrome. <i>Journal of Human Genetics</i> , 2011 , 56, 508-15	4.3	23
77	Syndromic mental retardation with thrombocytopenia due to 21q22.11q22.12 deletion: Report of three patients. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 1711-7	2.5	23
76	Italian Rett database and biobank. <i>Human Mutation</i> , 2007 , 28, 329-35	4.7	23
75	Mutational screening of the RB1 gene in Italian patients with retinoblastoma reveals 11 novel mutations. <i>Journal of Human Genetics</i> , 2006 , 51, 209-216	4.3	23
74	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. <i>Pediatric Nephrology</i> , 2011 , 26, 717-24	3.2	22
73	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. <i>Lung Cancer</i> , 2014 , 85, 168-74	5.9	21
72	A spectrum of LMX1B mutations in Nail-Patella syndrome: new point mutations, deletion, and evidence of mosaicism in unaffected parents. <i>Genetics in Medicine</i> , 2010 , 12, 431-9	8.1	21
71	Thin glomerular basement membrane disease: clinical significance of a morphological diagnosis--a collaborative study of the Italian Renal Immunopathology Group. <i>Nephrology Dialysis Transplantation</i> , 2005 , 20, 545-51	4.3	21
70	Five patients with novel overlapping interstitial deletions in 8q22.2q22.3. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 1857-64	2.5	20
69	Employing a systematic approach to biobanking and analyzing clinical and genetic data for advancing COVID-19 research. <i>European Journal of Human Genetics</i> , 2021 , 29, 745-759	5.3	20
68	Delineation of the phenotype associated with 7q36.1q36.2 deletion: long QT syndrome, renal hypoplasia and mental retardation. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 1195-9	2.5	19
67	Spectrum of PTCH mutations in Italian nevoid basal cell-carcinoma syndrome patients: identification of thirteen novel alleles. <i>Human Mutation</i> , 2004 , 24, 441	4.7	19
66	Exome sequencing coupled with mRNA analysis identifies NDUFA6 as a Leigh gene. <i>Molecular Genetics and Metabolism</i> , 2016 , 119, 214-222	3.7	19
65	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. <i>Journal of Human Genetics</i> , 2016 , 61, 95-101	4.3	18
64	CHARGE-like presentation, craniosynostosis and mild Mowat-Wilson Syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2557-66	2.5	18
63	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. <i>Journal of Human Genetics</i> , 2007 , 52, 1011-1017	4.3	18
62	Al-Awadi-Raas-Rothschild (limb/pelvis/uterus-hypoplasia/aplasia) syndrome and WNT7A mutations: genetic homogeneity and nosological delineation. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 332-6	2.5	16

61	A 9.3 Mb microdeletion of 3q27.3q29 associated with psychomotor and growth delay, tricuspid valve dysplasia and bifid thumb. <i>European Journal of Medical Genetics</i> , 2009 , 52, 131-3	2.6	16
60	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. <i>Clinical Dysmorphology</i> , 2008 , 17, 13-17	0.9	16
59	Creatine transporter defect diagnosed by proton NMR spectroscopy in males with intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 2446-52	2.5	15
58	Retinoblastoma and mental retardation microdeletion syndrome: clinical characterization and molecular dissection using array CGH. <i>Journal of Human Genetics</i> , 2007 , 52, 535-542	4.3	15
57	A 2.6 Mb deletion of 6q24.3-25.1 in a patient with growth failure, cardiac septal defect, thin upperlip and asymmetric dysmorphic ears. <i>European Journal of Medical Genetics</i> , 2007 , 50, 315-21	2.6	15
56	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. <i>Brain</i> , 2020 , 143, 2380-2387	11.2	15
55	Analysis of the Phenotypes in the Rett Networked Database. <i>International Journal of Genomics</i> , 2019 , 2019, 6956934	2.5	14
54	Rett networked database: an integrated clinical and genetic network of Rett syndrome databases. <i>Human Mutation</i> , 2012 , 33, 1031-6	4.7	14
53	Clinical and molecular characterization of COVID-19 hospitalized patients. <i>PLoS ONE</i> , 2020 , 15, e0242534	3.7	14
52	Protective Role of a Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. <i>Genes</i> , 2021 , 12,	4.2	14
51	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. <i>European Journal of Human Genetics</i> , 2021 , 29, 1186-1197	5.3	14
50	Evidence of predisposing epimutation in retinoblastoma. <i>Human Mutation</i> , 2019 , 40, 201-206	4.7	14
49	3.2 Mb microdeletion in chromosome 7 bands q22.2-q22.3 associated with overgrowth and delayed bone age. <i>European Journal of Medical Genetics</i> , 2010 , 53, 168-70	2.6	12
48	Ambiguous external genitalia due to defect of 5- β -reductase in seven Iraqi patients: prevalence of a novel mutation. <i>Gene</i> , 2013 , 526, 490-3	3.8	11
47	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. <i>Human Mutation</i> , 2018 , 39, 302-314	4.7	11
46	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. <i>European Journal of Medical Genetics</i> , 2014 , 57, 163-8	2.6	10
45	EEG features and epilepsy in MECP2-mutated patients with the Zappella variant of Rett syndrome. <i>Clinical Neurophysiology</i> , 2010 , 121, 652-7	4.3	10
44	Exome sequencing analysis in a pair of monozygotic twins re-evaluates the genetics behind their intellectual disability and reveals a CHD2 mutation. <i>Brain and Development</i> , 2016 , 38, 590-6	2.2	10

43	MEIS2 gene is responsible for intellectual disability, cardiac defects and a distinct facial phenotype. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103627	2.6	10
42	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. <i>European Journal of Human Genetics</i> , 2016 , 24, 252-7	5.3	9
41	Frequency of the LRRK2 G2019S mutation in Italian patients affected by Parkinson's disease. <i>Journal of Human Genetics</i> , 2007 , 52, 201-204	4.3	9
40	Blepharophimosis, ptosis, and epicanthus inversus syndrome: clinical and molecular analysis of a case. <i>Journal of AAPOS</i> , 2006 , 10, 279-80	1.3	9
39	Clinical and molecular characterization of COVID-19 hospitalized patients		9
38	Optineurin gene is not involved in the common high-tension form of primary open-angle glaucoma. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2006 , 244, 1077-82	3.8	8
37	A unique patient presenting with concomitant Klinefelter syndrome, Alport syndrome, and craniopharyngioma. <i>Journal of Andrology</i> , 2012 , 33, 1155-9		7
36	Leukoencephalopathy in 21-beta hydroxylase deficiency: report of a family. <i>Brain and Development</i> , 2010 , 32, 421-4	2.2	7
35	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. <i>Brain</i> , 2020 , 143, 3564-3573	11.2	7
34	9q31.1q31.3 deletion in two patients with similar clinical features: a newly recognized microdeletion syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 685-90	2.5	6
33	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. <i>Clinica Chimica Acta</i> , 2007 , 384, 35-40	6.2	5
32	Otosclerosis: exclusion of linkage to the OTSC1 and OTSC2 loci in four Italian families. <i>International Journal of Audiology</i> , 2003 , 42, 475-80	2.6	5
31	The polymorphism L412F in inhibits autophagy and is a marker of severe COVID-19 in males.. <i>Autophagy</i> , 2021 , 1-11	10.2	5
30	Severe COVID-19 in Hospitalized Carriers of Single Pathogenic Variants. <i>Journal of Personalized Medicine</i> , 2021 , 11,	3.6	5
29	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. <i>Brain Sciences</i> , 2021 , 11,	3.4	5
28	Potentially Treatable Disorder Diagnosed Post Mortem by Exome Analysis in a Boy with Respiratory Distress. <i>International Journal of Molecular Sciences</i> , 2016 , 17, 306	6.3	5
27	Guidelines for Genetic Testing and Management of Alport Syndrome.. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2021 ,	6.9	5
26	Employing a Systematic Approach to Biobanking and Analyzing Clinical and Genetic Data for Advancing COVID-19 Research		4

25	Nicolaides-Baraitser syndrome: defining a phenotype. <i>Journal of Neurology</i> , 2016 , 263, 1659-60	5.5	4
24	Personalized therapy in a GRIN1 mutated girl with intellectual disability and epilepsy. <i>Clinical Dysmorphology</i> , 2018 , 27, 18-20	0.9	4
23	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients.. <i>Genes and Immunity</i> , 2021 ,	4.4	4
22	Sporadic hereditary motor and sensory neuropathies: Advances in the diagnosis using next generation sequencing technology. <i>Journal of the Neurological Sciences</i> , 2015 , 359, 409-17	3.2	3
21	Combined ultrasound and exome sequencing approach recognizes Opitz G/BBB syndrome in two malformed fetuses. <i>Clinical Dysmorphology</i> , 2017 , 26, 18-25	0.9	3
20	13q deletion syndrome and retinoblastoma in identical dichorionic diamniotic monozygotic twins. <i>European Journal of Ophthalmology</i> , 2012 , 22, 857-60	1.9	3
19	Should a syndrome be called by its correct name? The example of the preserved speech variant of Rett syndrome. <i>European Journal of Pediatrics</i> , 2005 , 164, 710; author reply 711-2	4.1	3
18	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. <i>Human Genetics</i> , 2021 , 141, 147	6.3	3
17	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males		3
16	IQSEC2 disorder: A new disease entity or a Rett spectrum continuum?. <i>Clinical Genetics</i> , 2021 , 99, 462-474		3
15	SELP Asp603Asn and severe thrombosis in COVID-19 males. <i>Journal of Hematology and Oncology</i> , 2021 , 14, 123	22.4	3
14	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing.. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	3
13	The Italian XLMR bank: a clinical and molecular database. <i>Human Mutation</i> , 2007 , 28, 13-8	4.7	2
12	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. <i>Frontiers in Oncology</i> , 2021 , 11, 649435	5.3	2
11	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males		2
10	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. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167, 1681	2.5	1
9	Huntington's disease gene expansion associates with early onset nonprogressive chorea. <i>Movement Disorders</i> , 2013 , 28, 684	7	1
8	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. <i>Molecular Medicine Reports</i> , 2008 ,	2.9	1

7	Genetics and mechanisms of disease in Rett syndrome. <i>Drug Discovery Today Disease Mechanisms</i> , 2005 , 2, 419-425		1
6	Shorter Androgen Receptor PolyQ Alleles Protect Against Life-Threatening COVID-19 Disease in Males. <i>SSRN Electronic Journal</i> ,	1	1
5	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in males		1
4	Post-Mendelian genetic model in COVID-19		1
3	Epilepsy in Nicolaides-Baraitser Syndrome: Review of Literature and Report of 25 Patients Focusing on Treatment Aspects. <i>Neuropediatrics</i> , 2021 , 52, 109-122	1.6	1
2	Triorchidism: genetic and imaging evaluation in an adult male. <i>Archivio Italiano Di Urologia Andrologia</i> , 2014 , 86, 156-7	1.6	0
1	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis.. <i>Neurological Sciences</i> , 2022 , 43, 2849	3.5	