Francesca Mari

List of Publications by Year in Descending Order

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	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis.. *Neurological Sciences*, **2022**, 43, 2849	3.5	
131	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. *Human Genetics*, **2021**, 141, 147	6.3	3
130	The polymorphism L412F in inhibits autophagy and is a marker of severe COVID-19 in males.. *Autophagy*, **2021**, 1-11	10.2	5
129	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. *Frontiers in Oncology*, **2021**, 11, 649435	5.3	2
128	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. *EBioMedicine*, **2021**, 65, 103246	8.8	25
127	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,	8.9	51
126	Protective Role of a Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. *Genes*, **2021**, 12,	4.2	14
125	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	5.3	14
124	Severe COVID-19 in Hospitalized Carriers of Single Pathogenic Variants. *Journal of Personalized Medicine*, **2021**, 11,	3.6	5
123	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. *Brain Sciences*, **2021**, 11,	3.4	5
122	IQSEC2 disorder: A new disease entity or a Rett spectrum continuum?. *Clinical Genetics*, **2021**, 99, 462-474	4	3
121	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	5.3	20
120	Epilepsy in Nicolaides-Baraitser Syndrome: Review of Literature and Report of 25 Patients Focusing on Treatment Aspects. *Neuropediatrics*, **2021**, 52, 109-122	1.6	1
119	SELP Asp603Asn and severe thrombosis in COVID-19 males. *Journal of Hematology and Oncology*, **2021**, 14, 123	22.4	3
118	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients.. *Genes and Immunity*, **2021**,	4.4	4
117	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing.. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
116	Guidelines for Genetic Testing and Management of Alport Syndrome.. *Clinical Journal of the American Society of Nephrology: CJASN*, **2021**,	6.9	5

115	Clinical and molecular characterization of COVID-19 hospitalized patients. *PLoS ONE*, **2020**, 15, e0242534	3.7	14
114	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. *Brain*, **2020**, 143, 2380-2387	11.2	15
113	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	5.3	132
112	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. *Brain*, **2020**, 143, 3564-3573	11.2	7
111	MEIS2 gene is responsible for intellectual disability, cardiac defects and a distinct facial phenotype. *European Journal of Medical Genetics*, **2020**, 63, 103627	2.6	10
110	Analysis of the Phenotypes in the Rett Networked Database. *International Journal of Genomics*, **2019**, 2019, 6956934	2.5	14
109	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. *Pediatric Nephrology*, **2019**, 34, 1175-1189	3.2	70
108	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. *Genetics in Medicine*, **2019**, 21, 816-825	8.1	71
107	Evidence of predisposing epimutation in retinoblastoma. *Human Mutation*, **2019**, 40, 201-206	4.7	14
106	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. *Genetics in Medicine*, **2018**, 20, 965-975	8.1	37
105	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated Ⅱtubulin defect which improves after iHDAC6 treatment in Rett syndrome. *Experimental Cell Research*, **2018**, 368, 225-235	4.2	31
104	Personalized therapy in a GRIN1 mutated girl with intellectual disability and epilepsy. *Clinical Dysmorphology*, **2018**, 27, 18-20	0.9	4
103	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. *Human Mutation*, **2018**, 39, 302-314	4.7	11
102	Combined ultrasound and exome sequencing approach recognizes Opitz G/BBB syndrome in two malformed fetuses. *Clinical Dysmorphology*, **2017**, 26, 18-25	0.9	3
101	Alport syndrome: impact of digenic inheritance in patients management. *Clinical Genetics*, **2017**, 92, 34-44	4	33
100	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, 916-924	4.3	31
99	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. *European Journal of Human Genetics*, **2016**, 24, 252-7	5.3	9
98	Visual impairment in FOXG1-mutated individuals and mice. *Neuroscience*, **2016**, 324, 496-508	3.9	27

97	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. *Journal of Human Genetics*, **2016**, 61, 95-101	4.3	18
96	Potentially Treatable Disorder Diagnosed Post Mortem by Exome Analysis in a Boy with Respiratory Distress. *International Journal of Molecular Sciences*, **2016**, 17, 306	6.3	5
95	Nicolaides-Baraitser syndrome: defining a phenotype. *Journal of Neurology*, **2016**, 263, 1659-60	5.5	4
94	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, 590-6	2.2	10
93	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. *Molecular Genetics and Metabolism*, **2016**, 119, 214-222	3.7	19
92	Evidence of digenic inheritance in Alport syndrome. *Journal of Medical Genetics*, **2015**, 52, 163-74	5.8	95
91	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. *European Journal of Human Genetics*, **2015**, 23, 1523-30	5.3	33
90	Sporadic hereditary motor and sensory neuropathies: Advances in the diagnosis using next generation sequencing technology. *Journal of the Neurological Sciences*, **2015**, 359, 409-17	3.2	3
89	Epilepsy in Rett syndrome--lessons from the Rett networked database. *Epilepsia*, **2015**, 56, 569-76	6.4	30
88	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	5.3	56
87	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	2.5	1
86	Coffin-Siris and Nicolaides-Baraitser syndromes are a common well recognizable cause of intellectual disability. *Brain and Development*, **2015**, 37, 527-36	2.2	26
85	9q31.1q31.3 deletion in two patients with similar clinical features: a newly recognized microdeletion syndrome?. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 685-90	2.5	6
84	CHARGE-like presentation, craniosynostosis and mild Mowat-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**, 164A, 2557-66	2.5	18
83	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. *European Journal of Medical Genetics*, **2014**, 57, 163-8	2.6	10
82	Interstitial 22q13 deletions not involving SHANK3 gene: a new contiguous gene syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1666-76	2.5	36
81	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. *Lung Cancer*, **2014**, 85, 168-74	5.9	21
80	Redox imbalance and morphological changes in skin fibroblasts in typical Rett syndrome. *Oxidative Medicine and Cellular Longevity*, **2014**, 2014, 195935	6.7	36

79	Triorchidism: genetic and imaging evaluation in an adult male. *Archivio Italiano Di Urologia Andrologia*, **2014**, 86, 156-7	1.6	0
78	Unbiased next generation sequencing analysis confirms the existence of autosomal dominant Alport syndrome in a relevant fraction of cases. *Clinical Genetics*, **2014**, 86, 252-7	4	92
77	Epilepsy in Mowat-Wilson syndrome: delineation of the electroclinical phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 273-84	2.5	36
76	Ambiguous external genitalia due to defect of 5-□reductase in seven Iraqi patients: prevalence of a novel mutation. *Gene*, **2013**, 526, 490-3	3.8	11
75	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	4.2	47
74	A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. *Human Molecular Genetics*, **2013**, 22, 5121-35	5.6	138
73	Huntington□s disease gene expansion associates with early onset nonprogressive chorea. *Movement Disorders*, **2013**, 28, 684	7	1
72	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, 361-5	5.3	31
71	Revealing the complexity of a monogenic disease: rett syndrome exome sequencing. *PLoS ONE*, **2013**, 8, e56599	3.7	45
70	Xq28 duplications including MECP2 in five females: Expanding the phenotype to□severe mental retardation. *European Journal of Medical Genetics*, **2012**, 55, 404-13	2.6	38
69	A unique patient presenting with concomitant Klinefelter syndrome, Alport syndrome, and craniopharyngioma. *Journal of Andrology*, **2012**, 33, 1155-9		7
68	13q deletion syndrome and retinoblastoma in identical dichorionic diamniotic monozygotic twins. *European Journal of Ophthalmology*, **2012**, 22, 857-60	1.9	3
67	Rett networked database: an integrated clinical and genetic network of Rett syndrome databases. *Human Mutation*, **2012**, 33, 1031-6	4.7	14
66	Advances in Alport syndrome diagnosis using next-generation sequencing. *European Journal of Human Genetics*, **2012**, 20, 50-7	5.3	66
65	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, 392-7	2.5	52
64	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. *Pediatric Nephrology*, **2011**, 26, 717-24	3.2	22
63	Al-Awadi-Raas-Rothschild (limb/pelvis/uterus-hypoplasia/aplasia) syndrome and WNT7A mutations: genetic homogeneity and nosological delineation. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 332-6	2.5	16
62	Five patients with novel overlapping interstitial deletions in 8q22.2q22.3. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1857-64	2.5	20

61	Creatine transporter defect diagnosed by proton NMR spectroscopy in males with intellectual disability. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2446-52	2.5	15
60	iPS cells to model CDKL5-related disorders. *European Journal of Human Genetics*, **2011**, 19, 1246-55	5.3	71
59	Investigation of modifier genes within copy number variations in Rett syndrome. *Journal of Human Genetics*, **2011**, 56, 508-15	4.3	23
58	High frequency of COH1 intragenic deletions and duplications detected by MLPA in patients with Cohen syndrome. *European Journal of Human Genetics*, **2010**, 18, 1133-40	5.3	28
57	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, 431-9	8.1	21
56	3.2 Mb microdeletion in chromosome 7 bands q22.2-q22.3 associated with overgrowth and delayed bone age. *European Journal of Medical Genetics*, **2010**, 53, 168-70	2.6	12
55	EEG features and epilepsy in MECP2-mutated patients with the Zappella variant of Rett syndrome. *Clinical Neurophysiology*, **2010**, 121, 652-7	4.3	10
54	Leukoencephalopathy in 21-beta hydroxylase deficiency: report of a family. *Brain and Development*, **2010**, 32, 421-4	2.2	7
53	Syndromic mental retardation with thrombocytopenia due to 21q22.11q22.12 deletion: Report of three patients. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1711-7	2.5	23
52	Autosomal dominant Alport syndrome: molecular analysis of the COL4A4 gene and clinical outcome. *Nephrology Dialysis Transplantation*, **2009**, 24, 1464-71	4.3	71
51	Mowat-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, 417-26	2.5	83
50	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. *Cancer Science*, **2009**, 100, 465-71	6.9	30
49	Refinement of the 12q14 microdeletion syndrome: primordial dwarfism and developmental delay with or without osteopoikilosis. *European Journal of Human Genetics*, **2009**, 17, 1141-7	5.3	30
48	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. *European Journal of Medical Genetics*, **2009**, 52, 148-52	2.6	36
47	A 9.3 Mb microdeletion of 3q27.3q29 associated with psychomotor and growth delay, tricuspid valve dysplasia and bifid thumb. *European Journal of Medical Genetics*, **2009**, 52, 131-3	2.6	16
46	Private inherited microdeletion/microduplications: implications in clinical practice. *European Journal of Medical Genetics*, **2008**, 51, 409-16	2.6	51
45	Genomic differences between retinoma and retinoblastoma. *Acta Oncologica*, **2008**, 47, 1483-92	3.2	34
44	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**,	2.9	1

43	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. *Clinical Dysmorphology*, **2008**, 17, 13-17	0.9	16
42	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, 1195-9	2.5	19
41	A 3 Mb deletion in 14q12 causes severe mental retardation, mild facial dysmorphisms and Rett-like features. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1994-8	2.5	50
40	FOXG1 is responsible for the congenital variant of Rett syndrome. *American Journal of Human Genetics*, **2008**, 83, 89-93	11	312
39	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, 858-65	2.5	32
38	MECP2 deletions and genotype-phenotype correlation in Rett syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2775-84	2.5	41
37	The Italian XLMR bank: a clinical and molecular database. *Human Mutation*, **2007**, 28, 13-8	4.7	2
36	Italian Rett database and biobank. *Human Mutation*, **2007**, 28, 329-35	4.7	23
35	Frequency of the LRRK2 G2019S mutation in Italian patients affected by Parkinson's disease. *Journal of Human Genetics*, **2007**, 52, 201-204	4.3	9
34	Retinoblastoma and mental retardation microdeletion syndrome: clinical characterization and molecular dissection using array CGH. *Journal of Human Genetics*, **2007**, 52, 535-542	4.3	15
33	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. *Journal of Human Genetics*, **2007**, 52, 1011-1017	4.3	18
32	2q24-q31 deletion: report of a case and review of the literature. *European Journal of Medical Genetics*, **2007**, 50, 21-32	2.6	46
31	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. *European Journal of Medical Genetics*, **2007**, 50, 315-21	2.6	15
30	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. *Clinica Chimica Acta*, **2007**, 384, 35-40	6.2	5
29	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. *Nephrology Dialysis Transplantation*, **2006**, 21, 665-71	4.3	38
28	Blepharophimosis, ptosis, and epicanthus inversus syndrome: clinical and molecular analysis of a case. *Journal of AAPOS*, **2006**, 10, 279-80	1.3	9
27	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, 1077-82	3.8	8
26	Mutational screening of the RB1 gene in Italian patients with retinoblastoma reveals 11 novel mutations. *Journal of Human Genetics*, **2006**, 51, 209-216	4.3	23

25	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, 545-51	4.3	21
24	Genetics and mechanisms of disease in Rett syndrome. *Drug Discovery Today Disease Mechanisms*, **2005**, 2, 419-425		1
23	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. *Clinical Genetics*, **2005**, 67, 258-60	4	25
22	Non-syndromic X-linked mental retardation: from a molecular to a clinical point of view. *Journal of Cellular Physiology*, **2005**, 204, 8-20	7	30
21	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	4.7	74
20	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; author reply 711-2	4.1	3
19	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. *Journal of Medical Genetics*, **2005**, 42, 103-7	5.8	180
18	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, 1935-46	5.6	248
17	Autosomal-dominant Alport syndrome: natural history of a disease due to COL4A3 or COL4A4 gene. *Kidney International*, **2004**, 65, 1598-603	9.9	90
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, 172-7	4.7	92
15	Spectrum of PTCH mutations in Italian nevoid basal cell-carcinoma syndrome patients: identification of thirteen novel alleles. *Human Mutation*, **2004**, 24, 441	4.7	19
14	Rett syndrome: the complex nature of a monogenic disease. *Journal of Molecular Medicine*, **2003**, 81, 346-54	5.5	67
13	Study of MECP2 gene in Rett syndrome variants and autistic girls. *American Journal of Medical Genetics Part A*, **2003**, 119B, 102-7		61
12	Chromosome 2 deletion encompassing the MAP2 gene in a patient with autism and Rett-like features. *Clinical Genetics*, **2003**, 64, 497-501	4	41
11	Dyskeratosis congenita and cancer in mice deficient in ribosomal RNA modification. *Science*, **2003**, 299, 259-62	33.3	340
10	Otosclerosis: exclusion of linkage to the OTSC1 and OTSC2 loci in four Italian families. *International Journal of Audiology*, **2003**, 42, 475-80	2.6	5
9	COL4A3/COL4A4 mutations: from familial hematuria to autosomal-dominant or recessive Alport syndrome. *Kidney International*, **2002**, 61, 1947-56	9.9	143
8	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, 982-5	11	196

List of Publications

7	Shorter Androgen Receptor PolyQ Alleles Protect Against Life-Threatening COVID-19 Disease in Males. *SSRN Electronic Journal*,	1	1
6	Clinical and molecular characterization of COVID-19 hospitalized patients		9
5	Employing a Systematic Approach to Biobanking and Analyzing Clinical and Genetic Data for Advancing COVID-19 Research		4
4	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in males		1
3	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males		2
2	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males		3
1	Post-Mendelian genetic model in COVID-19		1