Francesca Ariani

List of Publications by Year in Descending Order

79 papers	2,780 citations	30 h-index	51 g-index
84 ext. papers	3,118 ext. citations	4.1 avg, IF	3.93 L-index

#	Paper	IF	Citations
79	Identification of a Novel Pathogenic Variant in a Patient with a Neurodevelopmental Disorder.. *Genes*, **2022**, 13,	4.2	1
78	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. *Frontiers in Oncology*, **2021**, 11, 649435	5.3	2
77	Identification of a Novel Pathogenic Variant in the NAGLU Gene in a Child with Neurodevelopmental Delay. *Journal of Autism and Developmental Disorders*, **2021**, 1	4.6	
76	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. *Brain Sciences*, **2021**, 11,	3.4	5
75	IQSEC2 disorder: A new disease entity or a Rett spectrum continuum?. *Clinical Genetics*, **2021**, 99, 462-474	4	3
74	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing.. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
73	Spondyloocular Syndrome: A Novel Variant with Description of the Neonatal Phenotype.. *Frontiers in Genetics*, **2021**, 12, 761264	4.5	1
72	Germline Variant Predisposing to a Rare Ovarian Germ Cell Tumor: A Case Report. *Frontiers in Oncology*, **2020**, 10, 1467	5.3	1
71	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
70	Usefulness and Limitations of Comprehensive Characterization of mRNA Splicing Profiles in the Definition of the Clinical Relevance of Variants of Uncertain Significance. *Cancers*, **2019**, 11,	6.6	16
69	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. *Pediatric Nephrology*, **2019**, 34, 1175-1189	3.2	70
68	Evidence of predisposing epimutation in retinoblastoma. *Human Mutation*, **2019**, 40, 201-206	4.7	14
67	Parent-of-origin effect of hypomorphic pathogenic variants and somatic mosaicism impact on phenotypic expression of retinoblastoma. *European Journal of Human Genetics*, **2018**, 26, 1026-1037	5.3	11
66	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated tubulin defect which improves after iHDAC6 treatment in Rett syndrome. *Experimental Cell Research*, **2018**, 368, 225-235	4.2	31
65	Omic Approach in Non-smoker Female with Lung Squamous Cell Carcinoma Pinpoints to Germline Susceptibility and Personalized Medicine. *Cancer Research and Treatment*, **2018**, 50, 356-365	5.2	12
64	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. *Human Mutation*, **2018**, 39, 302-314	4.7	11
63	Alport syndrome: impact of digenic inheritance in patients management. *Clinical Genetics*, **2017**, 92, 34-44	4	33

62	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
61	Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXG1(+/-) patients and in foxg1(+/-) mice. *European Journal of Human Genetics*, **2016**, 24, 871-80	5.3	39
60	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. *European Journal of Human Genetics*, **2016**, 24, 252-7	5.3	9
59	Exploiting the potential of next-generation sequencing in genomic medicine. *Expert Review of Molecular Diagnostics*, **2016**, 16, 1037-47	3.8	4
58	Visual impairment in FOXG1-mutated individuals and mice. *Neuroscience*, **2016**, 324, 496-508	3.9	27
57	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
56	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
55	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
54	Evidence of digenic inheritance in Alport syndrome. *Journal of Medical Genetics*, **2015**, 52, 163-74	5.8	95
53	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. *European Journal of Human Genetics*, **2015**, 23, 1523-30	5.3	33
52	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
51	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. *Lung Cancer*, **2014**, 85, 168-74	5.9	21
50	Redox imbalance and morphological changes in skin fibroblasts in typical Rett syndrome. *Oxidative Medicine and Cellular Longevity*, **2014**, 2014, 195935	6.7	36
49	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
48	Efficacy and safety of moxifloxacin in acute exacerbations of chronic bronchitis and COPD: a systematic review and meta-analysis. *Journal of Thoracic Disease*, **2014**, 6, 221-9	2.6	11
47	Exome sequencing overrides formal genetics: ASPM mutations in a case study of apparent X-linked microcephalic intellectual deficit. *Clinical Genetics*, **2013**, 83, 288-90	4	8
46	Variant of Rett Syndrome and CDKL5 Gene: Clinical and Autonomic Description of 10 Cases. *Neuropediatrics*, **2013**, 44, 237-238	1.6	1
45	Revealing the complexity of a monogenic disease: rett syndrome exome sequencing. *PLoS ONE*, **2013**, 8, e56599	3.7	45

44	Expanding the phenotype associated with FOXG1 mutations and in vivo FoxG1 chromatin-binding dynamics. *Clinical Genetics*, **2012**, 82, 395-403	4	18
43	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
42	Epigenetic and copy number variation analysis in retinoblastoma by MS-MLPA. *Pathology and Oncology Research*, **2012**, 18, 703-12	2.6	37
41	Variant of Rett syndrome and CDKL5 gene: clinical and autonomic description of 10 cases. *Neuropediatrics*, **2012**, 43, 37-43	1.6	22
40	Advances in Alport syndrome diagnosis using next-generation sequencing. *European Journal of Human Genetics*, **2012**, 20, 50-7	5.3	66
39	Vav1 haploinsufficiency in a common variable immunodeficiency patient with defective T-cell function. *International Journal of Immunopathology and Pharmacology*, **2012**, 25, 811-7	3	15
38	Retinoma and Retinoblastoma: Genomic Hybridisation **2012**, 93-102		
37	iPS cells to model CDKL5-related disorders. *European Journal of Human Genetics*, **2011**, 19, 1246-55	5.3	71
36	p53 Arg72Pro and MDM2 309 SNPs in hereditary retinoblastoma. *Journal of Human Genetics*, **2011**, 56, 685-6	4.3	11
35	Investigation of modifier genes within copy number variations in Rett syndrome. *Journal of Human Genetics*, **2011**, 56, 508-15	4.3	23
34	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
33	Novel FOXG1 mutations associated with the congenital variant of Rett syndrome. *Journal of Medical Genetics*, **2010**, 47, 49-53	5.8	91
32	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
31	Early-onset seizure variant of Rett syndrome: definition of the clinical diagnostic criteria. *Brain and Development*, **2010**, 32, 17-24	2.2	51
30	Diagnostic criteria for the Zappella variant of Rett syndrome (the preserved speech variant). *Brain and Development*, **2009**, 31, 208-16	2.2	68
29	Variation in novel exons (RACEfrags) of the MECP2 gene in Rett syndrome patients and controls. *Human Mutation*, **2009**, 30, E866-79	4.7	1
28	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. *Cancer Science*, **2009**, 100, 465-71	6.9	30
27	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. *European Journal of Medical Genetics*, **2009**, 52, 148-52	2.6	36

26	The XLMR gene ACSL4 plays a role in dendritic spine architecture. *Neuroscience*, **2009**, 159, 657-69	3.9	28
25	Private inherited microdeletion/microduplications: implications in clinical practice. *European Journal of Medical Genetics*, **2008**, 51, 409-16	2.6	51
24	Genomic differences between retinoma and retinoblastoma. *Acta Oncologica*, **2008**, 47, 1483-92	3.2	34
23	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. *Clinical Dysmorphology*, **2008**, 17, 13-17	0.9	16
22	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
21	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
20	FOXG1 is responsible for the congenital variant of Rett syndrome. *American Journal of Human Genetics*, **2008**, 83, 89-93	11	312
19	MECP2 deletions and genotype-phenotype correlation in Rett syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2775-84	2.5	41
18	The Italian XLMR bank: a clinical and molecular database. *Human Mutation*, **2007**, 28, 13-8	4.7	2
17	Italian Rett database and biobank. *Human Mutation*, **2007**, 28, 329-35	4.7	23
16	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
15	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. *Journal of Human Genetics*, **2007**, 52, 1011-1017	4.3	18
14	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
13	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. *Clinica Chimica Acta*, **2007**, 384, 35-40	6.2	5
12	Blepharophimosis, ptosis, and epicanthus inversus syndrome: clinical and molecular analysis of a case. *Journal of AAPOS*, **2006**, 10, 279-80	1.3	9
11	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
10	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. *Clinical Genetics*, **2005**, 67, 258-60	4	25
9	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

8	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. *Journal of Medical Genetics*, **2005**, 42, 103-7	5.8	180
7	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
6	Three Rett patients with both MECP2 mutation and 15q11-13 rearrangements. *European Journal of Human Genetics*, **2004**, 12, 682-5	5.3	10
5	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
4	Rett syndrome: the complex nature of a monogenic disease. *Journal of Molecular Medicine*, **2003**, 81, 346-54	5.5	67
3	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. *Clinical Genetics*, **2003**, 63, 510-5	4	26
2	Chromosome 2 deletion encompassing the MAP2 gene in a patient with autism and Rett-like features. *Clinical Genetics*, **2003**, 64, 497-501	4	41
1	A third MRX family (MRX68) is the result of mutation in the long chain fatty acid-CoA ligase 4 (FACL4) gene: proposal of a rapid enzymatic assay for screening mentally retarded patients. *Journal of Medical Genetics*, **2003**, 40, 11-7	5.8	37