

Francesca Ariani

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.

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	FOXG1 is responsible for the congenital variant of Rett syndrome. <i>American Journal of Human Genetics</i> , 2008 , 83, 89-93	11	312
78	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
77	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. <i>Journal of Medical Genetics</i> , 2005 , 42, 103-7	5.8	180
76	Evidence of digenic inheritance in Alport syndrome. <i>Journal of Medical Genetics</i> , 2015 , 52, 163-74	5.8	95
75	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
74	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
73	Novel FOXG1 mutations associated with the congenital variant of Rett syndrome. <i>Journal of Medical Genetics</i> , 2010 , 47, 49-53	5.8	91
72	iPS cells to model CDKL5-related disorders. <i>European Journal of Human Genetics</i> , 2011 , 19, 1246-55	5.3	71
71	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. <i>Pediatric Nephrology</i> , 2019 , 34, 1175-1189	3.2	70
70	Diagnostic criteria for the Zappella variant of Rett syndrome (the preserved speech variant). <i>Brain and Development</i> , 2009 , 31, 208-16	2.2	68
69	Rett syndrome: the complex nature of a monogenic disease. <i>Journal of Molecular Medicine</i> , 2003 , 81, 346-54	5.5	67
68	Advances in Alport syndrome diagnosis using next-generation sequencing. <i>European Journal of Human Genetics</i> , 2012 , 20, 50-7	5.3	66
67	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
66	Early-onset seizure variant of Rett syndrome: definition of the clinical diagnostic criteria. <i>Brain and Development</i> , 2010 , 32, 17-24	2.2	51
65	Private inherited microdeletion/microduplications: implications in clinical practice. <i>European Journal of Medical Genetics</i> , 2008 , 51, 409-16	2.6	51
64	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
63	Revealing the complexity of a monogenic disease: rett syndrome exome sequencing. <i>PLoS ONE</i> , 2013 , 8, e56599	3.7	45

62	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
61	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
60	Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXG1(+/-) patients and in foxg1(+/-) mice. <i>European Journal of Human Genetics</i> , 2016 , 24, 871-80	5.3	39
59	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
58	Epigenetic and copy number variation analysis in retinoblastoma by MS-MLPA. <i>Pathology and Oncology Research</i> , 2012 , 18, 703-12	2.6	37
57	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. <i>Journal of Medical Genetics</i> , 2003 , 40, 11-7	5.8	37
56	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
55	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. <i>European Journal of Medical Genetics</i> , 2009 , 52, 148-52	2.6	36
54	Genomic differences between retinoma and retinoblastoma. <i>Acta Oncologica</i> , 2008 , 47, 1483-92	3.2	34
53	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. <i>European Journal of Human Genetics</i> , 2015 , 23, 1523-30	5.3	33
52	Alport syndrome: impact of digenic inheritance in patients management. <i>Clinical Genetics</i> , 2017 , 92, 34-44	4.4	33
51	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated tubulin defect which improves after iHDAC6 treatment in Rett syndrome. <i>Experimental Cell Research</i> , 2018 , 368, 225-235	4.2	31
50	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
49	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. <i>Cancer Science</i> , 2009 , 100, 465-71	6.9	30
48	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
47	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
46	The XLMR gene ACSL4 plays a role in dendritic spine architecture. <i>Neuroscience</i> , 2009 , 159, 657-69	3.9	28
45	Visual impairment in FOXG1-mutated individuals and mice. <i>Neuroscience</i> , 2016 , 324, 496-508	3.9	27

44	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. <i>Clinical Genetics</i> , 2003 , 63, 510-5	4	26
43	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. <i>Clinical Genetics</i> , 2005 , 67, 258-60	4	25
42	Investigation of modifier genes within copy number variations in Rett syndrome. <i>Journal of Human Genetics</i> , 2011 , 56, 508-15	4-3	23
41	Italian Rett database and biobank. <i>Human Mutation</i> , 2007 , 28, 329-35	4-7	23
40	Variant of Rett syndrome and CDKL5 gene: clinical and autonomic description of 10 cases. <i>Neuropediatrics</i> , 2012 , 43, 37-43	1.6	22
39	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. <i>Lung Cancer</i> , 2014 , 85, 168-74	5-9	21
38	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
37	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
36	Expanding the phenotype associated with FOXP1 mutations and in vivo FoxG1 chromatin-binding dynamics. <i>Clinical Genetics</i> , 2012 , 82, 395-403	4	18
35	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. <i>Journal of Human Genetics</i> , 2007 , 52, 1011-1017	4-3	18
34	Usefulness and Limitations of Comprehensive Characterization of mRNA Splicing Profiles in the Definition of the Clinical Relevance of Variants of Uncertain Significance. <i>Cancers</i> , 2019 , 11,	6.6	16
33	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. <i>Clinical Dysmorphology</i> , 2008 , 17, 13-17	0.9	16
32	Vav1 haploinsufficiency in a common variable immunodeficiency patient with defective T-cell function. <i>International Journal of Immunopathology and Pharmacology</i> , 2012 , 25, 811-7	3	15
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. <i>European Journal of Medical Genetics</i> , 2007 , 50, 315-21	2.6	15
30	Evidence of predisposing epimutation in retinoblastoma. <i>Human Mutation</i> , 2019 , 40, 201-206	4-7	14
29	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
28	Omic Approach in Non-smoker Female with Lung Squamous Cell Carcinoma Pinpoints to Germline Susceptibility and Personalized Medicine. <i>Cancer Research and Treatment</i> , 2018 , 50, 356-365	5-2	12
27	Parent-of-origin effect of hypomorphic pathogenic variants and somatic mosaicism impact on phenotypic expression of retinoblastoma. <i>European Journal of Human Genetics</i> , 2018 , 26, 1026-1037	5-3	11

26	p53 Arg72Pro and MDM2 309 SNPs in hereditary retinoblastoma. <i>Journal of Human Genetics</i> , 2011 , 56, 685-6	4.3	11
25	Efficacy and safety of moxifloxacin in acute exacerbations of chronic bronchitis and COPD: a systematic review and meta-analysis. <i>Journal of Thoracic Disease</i> , 2014 , 6, 221-9	2.6	11
24	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
23	Three Rett patients with both MECP2 mutation and 15q11-13 rearrangements. <i>European Journal of Human Genetics</i> , 2004 , 12, 682-5	5.3	10
22	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
21	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
20	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. <i>European Journal of Human Genetics</i> , 2016 , 24, 252-7	5.3	9
19	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
18	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
17	Exome sequencing overrides formal genetics: ASPM mutations in a case study of apparent X-linked microcephalic intellectual deficit. <i>Clinical Genetics</i> , 2013 , 83, 288-90	4	8
16	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
15	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
14	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. <i>Brain Sciences</i> , 2021 , 11,	3.4	5
13	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
12	Exploiting the potential of next-generation sequencing in genomic medicine. <i>Expert Review of Molecular Diagnostics</i> , 2016 , 16, 1037-47	3.8	4
11	IQSEC2 disorder: A new disease entity or a Rett spectrum continuum?. <i>Clinical Genetics</i> , 2021 , 99, 462-474		3
10	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	3
9	The Italian XLMR bank: a clinical and molecular database. <i>Human Mutation</i> , 2007 , 28, 13-8	4.7	2

8	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
7	Variant of Rett Syndrome and CDKL5 Gene: Clinical and Autonomic Description of 10 Cases. <i>Neuropediatrics</i> , 2013 , 44, 237-238	1.6	1
6	Variation in novel exons (RACEfrags) of the MECP2 gene in Rett syndrome patients and controls. <i>Human Mutation</i> , 2009 , 30, E866-79	4.7	1
5	Germline Variant Predisposing to a Rare Ovarian Germ Cell Tumor: A Case Report. <i>Frontiers in Oncology</i> , 2020 , 10, 1467	5.3	1
4	Spondyloocular Syndrome: A Novel Variant with Description of the Neonatal Phenotype.. <i>Frontiers in Genetics</i> , 2021 , 12, 761264	4.5	1
3	Identification of a Novel Pathogenic Variant in a Patient with a Neurodevelopmental Disorder.. <i>Genes</i> , 2022 , 13,	4.2	1
2	Identification of a Novel Pathogenic Variant in the NAGLU Gene in a Child with Neurodevelopmental Delay. <i>Journal of Autism and Developmental Disorders</i> , 2021 , 1	4.6	
1	Retinoma and Retinoblastoma: Genomic Hybridisation 2012 , 93-102		