Francesca Ariani

List of Publications by Year
in descending order

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81 papers	3,412 citations	136740 32 h-index	155451 55 g-index
84 all docs	84 docs citations	84 times ranked	4611 citing authors

#	Article	IF	Citations
1	FOXG1 Is Responsible for the Congenital Variant of Rett Syndrome. American Journal of Human Genetics, 2008, 83, 89-93.	2.6	366
2	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
3	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. Journal of Medical Genetics, 2005, 42, 103-107.	1.5	206
4	Evidence of digenic inheritance in Alport syndrome. Journal of Medical Genetics, 2015, 52, 163-174.	1.5	129
5	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
6	Novel FOXG1 mutations associated with the congenital variant of Rett syndrome. Journal of Medical Genetics, 2010, 47, 49-53.	1.5	106
7	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. Pediatric Nephrology, 2019, 34, 1175-1189.	0.9	97
8	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
9	Diagnostic criteria for the Zappella variant of Rett syndrome (the preserved speech variant). Brain and Development, 2009, 31, 208-216.	0.6	83
10	Rett syndrome: the complex nature of a monogenic disease. Journal of Molecular Medicine, 2003, 81, 346-354.	1.7	80
11	iPS cells to model CDKL5-related disorders. European Journal of Human Genetics, 2011, 19, 1246-1255.	1.4	80
12	Advances in Alport syndrome diagnosis using next-generation sequencing. European Journal of Human Genetics, 2012, 20, 50-57.	1.4	76
13	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
14	Early-onset seizure variant of Rett syndrome: Definition of the clinical diagnostic criteria. Brain and Development, 2010, 32, 17-24.	0.6	62
15	Private inherited microdeletion/microduplications: Implications in clinical practice. European Journal of Medical Genetics, 2008, 51, 409-416.	0.7	59
16	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
17	Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXG1+/â^' patients and in foxg1+/â^' mice. European Journal of Human Genetics, 2016, 24, 871-880.	1.4	54
18	Revealing the Complexity of a Monogenic Disease: Rett Syndrome Exome Sequencing. PLoS ONE, 2013, 8, e56599.	1.1	54

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19	Alport syndrome: impact of digenic inheritance in patients management. Clinical Genetics, 2017, 92, 34-44.	1.0	52
20	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
21	MECP2 deletions and genotypeâ€“phenotype correlation in Rett syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 2775-2784.	0.7	45
22	Redox Imbalance and Morphological Changes in Skin Fibroblasts in Typical Rett Syndrome. Oxidative Medicine and Cellular Longevity, 2014, 2014, 1-10.	1.9	44
23	Epigenetic and Copy Number Variation Analysis in Retinoblastoma by MS-MLPA. Pathology and Oncology Research, 2012, 18, 703-712.	0.9	43
24	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, 11-17.	1.5	42
25	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
26	Genomic differences between retinoma and retinoblastoma. Acta OncolÃ³gica, 2008, 47, 1483-1492.	0.8	41
27	Visual impairment in FOXG1-mutated individuals and mice. Neuroscience, 2016, 324, 496-508.	1.1	41
28	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. European Journal of Medical Genetics, 2009, 52, 148-152.	0.7	40
29	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
30	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. Cancer Science, 2009, 100, 465-471.	1.7	38
31	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. European Journal of Human Genetics, 2015, 23, 1523-1530.	1.4	37
32	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
33	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
34	The XLMR gene ACSL4 plays a role in dendritic spine architecture. Neuroscience, 2009, 159, 657-669.	1.1	34
35	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. Clinical Genetics, 2005, 67, 258-260.	1.0	32
36	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

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37	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. Lung Cancer, 2014, 85, 168-174.	0.9	30
38	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
39	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. Clinical Genetics, 2003, 63, 510-515.	1.0	27
40	Italian Rett database and biobank. Human Mutation, 2007, 28, 329-335.	1.1	27
41	Variant of Rett Syndrome and CDKL5 Gene: Clinical and Autonomic Description of 10 Cases. Neuropediatrics, 2012, 43, 037-043.	0.3	27
42	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. Journal of Human Genetics, 2007, 52, 1011-1017.	1.1	25
43	Investigation of modifier genes within copy number variations in Rett syndrome. Journal of Human Genetics, 2011, 56, 508-515.	1.1	25
44	Expanding the phenotype associated with FOXG1 mutations and in vivo FoxG1 chromatinâ€binding dynamics. Clinical Genetics, 2012, 82, 395-403.	1.0	25
45	Usefulness and Limitations of Comprehensive Characterization of mRNA Splicing Profiles in the Definition of the Clinical Relevance of BRCA1/2 Variants of Uncertain Significance. Cancers, 2019, 11, 295.	1.7	24
46	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
47	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing. International Journal of Molecular Sciences, 2021, 22, 13439.	1.8	23
48	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
49	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.	1.3	20
50	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.	1.4	19
51	Vav1Haploinsufficiency in a Common Variable Immunodeficiency Patient with Defective T-Cell Function. International Journal of Immunopathology and Pharmacology, 2012, 25, 811-817.	1.0	18
52	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. Clinical Dysmorphology, 2008, 17, 13-17.	0.1	17
53	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. Brain Sciences, 2021, 11, 936.	1.1	17
54	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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55	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. Human Mutation, 2018, 39, 302-314.	1.1	16
56	Evidence of predisposing epimutation in retinoblastoma. Human Mutation, 2019, 40, 201-206.	1.1	16
57	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
58	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.	0.6	15
59	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
60	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
61	p53 Arg72Pro and MDM2 309 SNPs in hereditary retinoblastoma. Journal of Human Genetics, 2011, 56, 685-686.	1.1	12
62	Three Rett patients with both MECP2 mutation and 15q11â€“13 rearrangements. European Journal of Human Genetics, 2004, 12, 682-685.	1.4	11
63	Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome: Clinical and Molecular Analysis of a Case. Journal of AAPOS, 2006, 10, 279-280.	0.2	11
64	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
65	<scp>IQSEC2</scp> disorder: A new disease entity or a Rett spectrum continuum?. Clinical Genetics, 2021, 99, 462-474.	1.0	11
66	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. European Journal of Human Genetics, 2016, 24, 252-257.	1.4	10
67	Exome sequencing overrides formal genetics: <scp>ASPM</scp> mutations in a case study of apparent Xâ€linked microcephalic intellectual deficit. Clinical Genetics, 2013, 83, 288-290.	1.0	9
68	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. Clinica Chimica Acta, 2007, 384, 35-40.	0.5	8
69	Identification of a Novel SHANK2 Pathogenic Variant in a Patient with a Neurodevelopmental Disorder. Genes, 2022, 13, 688.	1.0	7
70	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
71	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
72	Exploiting the potential of next-generation sequencing in genomic medicine. Expert Review of Molecular Diagnostics, 2016, 16, 1037-1047.	1.5	5

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73	Spondyloocular Syndrome: A Novel XYLT2 Variant with Description of the Neonatal Phenotype. Frontiers in Genetics, 2021, 12, 761264.	1.1	4
74	The Italian XLMR bank: a clinical and molecular database. Human Mutation, 2007, 28, 13-18.	1.1	2
75	13q Deletion Syndrome Involving RB1: Characterization of a New Minimal Critical Region for Psychomotor Delay. Genes, 2021, 12, 1318.	1.0	2
76	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. Frontiers in Oncology, 2021, 11, 649435.	1.3	2
77	Variation in novel exons (RACEfrags) of theMECP2gene in Rett syndrome patients and controls. Human Mutation, 2009, 30, E866-E879.	1.1	1
78	Variant of Rett Syndrome and CDKL5 Gene: Clinical and Autonomic Description of 10 Cases. Neuropediatrics, 2013, 44, 237-238.	0.3	1
79	RB1 Germline Variant Predisposing to a Rare Ovarian Germ Cell Tumor: A Case Report. Frontiers in Oncology, 2020, 10, 1467.	1.3	1
80	Retinoma and Retinoblastoma: Genomic Hybridisation. , 2012, , 93-102.		0
81	Identification of a Novel Pathogenic Variant in the NAGLU Gene in a Child with Neurodevelopmental Delay. Journal of Autism and Developmental Disorders, 2021, , 1.	1.7	0