Ilaria Meloni

List of Publications by Year
in descending order

76 papers	3,996 citations	109264 35 h-index	123376 61 g-index
82 all docs	82 docs citations	82 times ranked	5601 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	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, 982-985.	2.6	213
3	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.	1.4	208
4	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. Journal of Medical Genetics, 2005, 42, 103-107.	1.5	206
5	COL4A3/COL4A4 mutations: From familial hematuria to autosomal-dominant or recessive Alport syndrome. Kidney International, 2002, 61, 1947-1956.	2.6	187
6	A natural history of cleidocranial dysplasia. American Journal of Medical Genetics Part A, 2001, 104, 1-6.	2.4	179
7	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, .	2.8	145
8	FACL4, encoding fatty acid-CoA ligase 4, is mutated in nonspecific X-linked mental retardation. Nature Genetics, 2002, 30, 436-440.	9.4	135
9	Preserved speech variants of the Rett syndrome: Molecular and clinical analysis. American Journal of Medical Genetics Part A, 2001, 104, 14-22.	2.4	117
10	Preserved speech variant is allelic of classic Rett syndrome. European Journal of Human Genetics, 2000, 8, 325-330.	1.4	116
11	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
12	Diagnostic criteria for the Zappella variant of Rett syndrome (the preserved speech variant). Brain and Development, 2009, 31, 208-216.	0.6	83
13	Rett syndrome: the complex nature of a monogenic disease. Journal of Molecular Medicine, 2003, 81, 346-354.	1.7	80
14	iPS cells to model CDKL5-related disorders. European Journal of Human Genetics, 2011, 19, 1246-1255.	1.4	80
15	Study ofMECP2 gene in Rett syndrome variants and autistic girls. American Journal of Medical Genetics Part A, 2003, 119B, 102-107.	2.4	67
16	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
17	Private inherited microdeletion/microduplications: Implications in clinical practice. European Journal of Medical Genetics, 2008, 51, 409-416.	0.7	59
18	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

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19	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
20	Revealing the Complexity of a Monogenic Disease: Rett Syndrome Exome Sequencing. PLoS ONE, 2013, 8, e56599.	1.1	54
21	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	2.7	52
22	Mutation analysis of the MECP2 gene in British and Italian Rett syndrome females. Journal of Molecular Medicine, 2001, 78, 648-655.	1.7	51
23	Inherited human IRAK-1 deficiency selectively impairs TLR signaling in fibroblasts. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E514-E523.	3.3	49
24	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
25	Epilepsy in Rett syndromeâ€"Lessons from the Rett networked database. Epilepsia, 2015, 56, 569-576.	2.6	47
26	Epstein syndrome: another renal disorder with mutations in the nonmuscle myosin heavy chainÂ9 gene. Human Genetics, 2002, 110, 182-186.	1.8	45
27	Redox Imbalance and Morphological Changes in Skin Fibroblasts in Typical Rett Syndrome. Oxidative Medicine and Cellular Longevity, 2014, 2014, 1-10.	1.9	44
28	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
29	Genomic differences between retinoma and retinoblastoma. Acta OncolÃ³gica, 2008, 47, 1483-1492.	0.8	41
30	Visual impairment in FOXG1-mutated individuals and mice. Neuroscience, 2016, 324, 496-508.	1.1	41
31	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients. Genes and Immunity, 2022, 23, 51-56.	2.2	41
32	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. Nephrology Dialysis Transplantation, 2006, 21, 665-671.	0.4	40
33	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. European Journal of Medical Genetics, 2009, 52, 148-152.	0.7	40
34	The alliance between genetic biobanks and patient organisations: the experience of the telethon network of genetic biobanks. Orphanet Journal of Rare Diseases, 2016, 11, 142.	1.2	40
35	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. Cancer Science, 2009, 100, 465-471.	1.7	38
36	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. European Journal of Human Genetics, 2015, 23, 1523-1530.	1.4	37

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37	Pseudoxanthoma elasticum: Point mutations in the ABCC6 gene and a large deletion including also ABCC1 and MYH11. Human Mutation, 2001, 18, 85-85.	1.1	36
38	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
39	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
40	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.	1.4	35
41	The XLMR gene ACSL4 plays a role in dendritic spine architecture. Neuroscience, 2009, 159, 657-669.	1.1	34
42	Gene replacement ameliorates deficits in mouse and human models of cyclin-dependent kinase-like 5 disorder. Brain, 2020, 143, 811-832.	3.7	34
43	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. Clinical Genetics, 2005, 67, 258-260.	1.0	32
44	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
45	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. Clinical Genetics, 2003, 63, 510-515.	1.0	27
46	Italian Rett database and biobank. Human Mutation, 2007, 28, 329-335.	1.1	27
47	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. Pediatric Nephrology, 2011, 26, 717-724.	0.9	27
48	MECP2 gene mutation analysis in the British and Italian Rett Syndrome patients: hot spot map of the most recurrent mutations and bioinformatic analysis of a new MECP2 conserved region. Brain and Development, 2001, 23, S246-S250.	0.6	25
49	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. Journal of Human Genetics, 2007, 52, 1011-1017.	1.1	25
50	Expanding the phenotype associated with FOXG1 mutations and in vivo FoxG1 chromatinâ€binding dynamics. Clinical Genetics, 2012, 82, 395-403.	1.0	25
51	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males. Autophagy, 2022, 18, 1662-1672.	4.3	25
52	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
53	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	1.8	22
54	Hybridisation-based resequencing of 17 X-linked intellectual disability genes in 135 patients reveals novel mutations in ATRX, SLC6A8 and PQBP1. European Journal of Human Genetics, 2011, 19, 717-720.	1.4	21

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55	Intellectual disability, midface hypoplasia, facial hypotonia, and alport syndrome are associated with a deletion in Xq22.3. American Journal of Medical Genetics, Part A, 2010, 152A, 713-717.	0.7	19
56	Alteration of serum lipid profile, SRB1 loss, and impaired Nrf2 activation in CDKL5 disorder. Free Radical Biology and Medicine, 2015, 86, 156-165.	1.3	19
57	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. Clinical Dysmorphology, 2008, 17, 13-17.	0.1	17
58	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
59	Is Rett syndrome a loss-of-imprinting disorder?. Nature Genetics, 2005, 37, 10-11.	9.4	14
60	AAV-mediated FOXG1 gene editing in human Rett primary cells. European Journal of Human Genetics, 2020, 28, 1446-1458.	1.4	12
61	C9orf72 Intermediate Repeats Confer Genetic Risk for Severe COVID-19 Pneumonia Independently of Age. International Journal of Molecular Sciences, 2021, 22, 6991.	1.8	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	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. European Journal of Human Genetics, 2016, 24, 252-257.	1.4	10
64	High rate of HDR in gene editing of p.(Thr158Met) MECP2 mutational hotspot. European Journal of Human Genetics, 2020, 28, 1231-1242.	1.4	10
65	The phenomenon of multidrug resistance in glioblastomas. Hematology/ Oncology and Stem Cell Therapy, 2021, , .	0.6	10
66	Identification and characterization of mouse orthologs of the AMMECR1 and FACL4 genes deleted in AMME syndrome: orthology of Xq22.3 and MmuXF1â€“F3. Cytogenetic and Genome Research, 2000, 88, 259-263.	0.6	9
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	Optic disc drusen, angioid streaks, and mottled fundus in various combinations in a Sicilian family. Graefe's Archive for Clinical and Experimental Ophthalmology, 2002, 240, 771-776.	1.0	6
70	JNK signaling provides a novel therapeutic target for Rett syndrome. BMC Biology, 2021, 19, 256.	1.7	6
71	Lymphoblastoid cell lines of Rett syndrome patients exposed to oxidative-stress-induced apoptosis. Brain and Development, 2004, 26, 384-388.	0.6	4
72	A Genome Wide Copy Number Variations Analysis in Autism Spectrum Disorder (Asd) and Intellectual Disability (Id) in Italian Families. Journal of Genetic Syndromes & Gene Therapy, 2016, 7, .	0.2	3

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73	The Italian XLMR bank: a clinical and molecular database. Human Mutation, 2007, 28, 13-18.	1.1	2
74	Lowâ€level <scp>TP</scp>53 mutational load antecedes clonal expansion in chronic lymphocytic leukaemia. British Journal of Haematology, 2019, 184, 657-659.	1.2	2
75	Huntington's disease gene expansion associates with early onset nonprogressive chorea. Movement Disorders, 2013, 28, 684-684.	2.2	1
76	Corrigendum to â€œMECP2 gene mutation analysis in the British and Italian Rett Syndrome patients: hot spot map of the most recurrent mutations and bioinformatic analysis of a new MECP2 conserved regionâ€•[Brain Dev 2001; 23: S246â€“50]. Brain and Development, 2012, 34, 891.	0.6	0