Ilaria Meloni

List of Publications by Citations

78 papers	3,107 citations	31 h-index	54 g-index
82 ext. papers	3,620 ext. citations	5.8 avg, IF	3.97 L-index

#	Paper	IF	Citations
78	FOXG1 is responsible for the congenital variant of Rett syndrome. *American Journal of Human Genetics*, **2008**, 83, 89-93	11	312
77	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
76	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. *Journal of Medical Genetics*, **2005**, 42, 103-7	5.8	180
75	A natural history of cleidocranial dysplasia. *American Journal of Medical Genetics Part A*, **2001**, 104, 1-6		147
74	COL4A3/COL4A4 mutations: from familial hematuria to autosomal-dominant or recessive Alport syndrome. *Kidney International*, **2002**, 61, 1947-56	9.9	143
73	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
72	FACL4, encoding fatty acid-CoA ligase 4, is mutated in nonspecific X-linked mental retardation. *Nature Genetics*, **2002**, 30, 436-40	36.3	123
71	Preserved speech variant is allelic of classic Rett syndrome. *European Journal of Human Genetics*, **2000**, 8, 325-30	5.3	105
70	Preserved speech variants of the Rett syndrome: molecular and clinical analysis. *American Journal of Medical Genetics Part A*, **2001**, 104, 14-22		100
69	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
68	iPS cells to model CDKL5-related disorders. *European Journal of Human Genetics*, **2011**, 19, 1246-55	5.3	71
67	Diagnostic criteria for the Zappella variant of Rett syndrome (the preserved speech variant). *Brain and Development*, **2009**, 31, 208-16	2.2	68
66	Rett syndrome: the complex nature of a monogenic disease. *Journal of Molecular Medicine*, **2003**, 81, 346-54	5.5	67
65	Study of MECP2 gene in Rett syndrome variants and autistic girls. *American Journal of Medical Genetics Part A*, **2003**, 119B, 102-7		61
64	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
63	Private inherited microdeletion/microduplications: implications in clinical practice. *European Journal of Medical Genetics*, **2008**, 51, 409-16	2.6	51
62	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

61	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
60	Mutation analysis of the MECP2 gene in British and Italian Rett syndrome females. *Journal of Molecular Medicine*, **2001**, 78, 648-55	5.5	47
59	Revealing the complexity of a monogenic disease: rett syndrome exome sequencing. *PLoS ONE*, **2013**, 8, e56599	3.7	45
58	Epstein syndrome: another renal disorder with mutations in the nonmuscle myosin heavy chain 9 gene. *Human Genetics*, **2002**, 110, 182-6	6.3	43
57	Chromosome 2 deletion encompassing the MAP2 gene in a patient with autism and Rett-like features. *Clinical Genetics*, **2003**, 64, 497-501	4	41
56	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
55	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. *Nephrology Dialysis Transplantation*, **2006**, 21, 665-71	4.3	38
54	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
53	Redox imbalance and morphological changes in skin fibroblasts in typical Rett syndrome. *Oxidative Medicine and Cellular Longevity*, **2014**, 2014, 195935	6.7	36
52	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. *European Journal of Medical Genetics*, **2009**, 52, 148-52	2.6	36
51	Genomic differences between retinoma and retinoblastoma. *Acta Oncologica*, **2008**, 47, 1483-92	3.2	34
50	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. *European Journal of Human Genetics*, **2015**, 23, 1523-30	5.3	33
49	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	11.5	31
48	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated α-tubulin defect which improves after iHDAC6 treatment in Rett syndrome. *Experimental Cell Research*, **2018**, 368, 225-235	4.2	31
47	Epilepsy in Rett syndrome--lessons from the Rett networked database. *Epilepsia*, **2015**, 56, 569-76	6.4	30
46	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. *Cancer Science*, **2009**, 100, 465-71	6.9	30
45	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
44	The XLMR gene ACSL4 plays a role in dendritic spine architecture. *Neuroscience*, **2009**, 159, 657-69	3.9	28

43	Pseudoxanthoma elasticum: Point mutations in the ABCC6 gene and a large deletion including also ABCC1 and MYH11. *Human Mutation*, **2001**, 18, 85	4.7	28
42	Visual impairment in FOXG1-mutated individuals and mice. *Neuroscience*, **2016**, 324, 496-508	3.9	27
41	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. *Clinical Genetics*, **2003**, 63, 510-5	4	26
40	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. *Clinical Genetics*, **2005**, 67, 258-60	4	25
39	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. *EBioMedicine*, **2021**, 65, 103246	8.8	25
38	Italian Rett database and biobank. *Human Mutation*, **2007**, 28, 329-35	4.7	23
37	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. *Pediatric Nephrology*, **2011**, 26, 717-24	3.2	22
36	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 Suppl 1, S246-50	2.2	21
35	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
34	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
33	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
32	Expanding the phenotype associated with FOXG1 mutations and in vivo FoxG1 chromatin-binding dynamics. *Clinical Genetics*, **2012**, 82, 395-403	4	18
31	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. *Journal of Human Genetics*, **2007**, 52, 1011-1017	4.3	18
30	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. *Clinical Dysmorphology*, **2008**, 17, 13-17	0.9	16
29	Alteration of serum lipid profile, SRB1 loss, and impaired Nrf2 activation in CDKL5 disorder. *Free Radical Biology and Medicine*, **2015**, 86, 156-65	7.8	15
28	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, 717-20	5.3	15
27	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
26	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	4.2	14

25	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, 713-7	2.5	12
24	Gene replacement ameliorates deficits in mouse and human models of cyclin-dependent kinase-like 5 disorder. *Brain*, **2020**, 143, 811-832	11.2	11
23	Three Rett patients with both MECP2 mutation and 15q11-13 rearrangements. *European Journal of Human Genetics*, **2004**, 12, 682-5	5.3	10
22	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. *European Journal of Human Genetics*, **2016**, 24, 252-7	5.3	9
21	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
20	Identification and characterization of mouse orthologs of the AMMECR1 and FACL4 genes deleted in AMME syndrome: orthology of Xq22.3 and MmuXF1-F3. *Cytogenetic and Genome Research*, **2000**, 88, 259-63	1.9	8
19	Intermediate Repeats Confer Genetic Risk for Severe COVID-19 Pneumonia Independently of Age. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	7
18	AAV-mediated FOXG1 gene editing in human Rett primary cells. *European Journal of Human Genetics*, **2020**, 28, 1446-1458	5.3	6
17	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. *Clinica Chimica Acta*, **2007**, 384, 35-40	6.2	5
16	Optic disc drusen, angioid streaks, and mottled fundus in various combinations in a Sicilian family. *Graefers Archive for Clinical and Experimental Ophthalmology*, **2002**, 240, 771-6	3.8	5
15	The polymorphism L412F in inhibits autophagy and is a marker of severe COVID-19 in males.. *Autophagy*, **2021**, 1-11	10.2	5
14	Lymphoblastoid cell lines of Rett syndrome patients exposed to oxidative-stress-induced apoptosis. *Brain and Development*, **2004**, 26, 384-8	2.2	4
13	Employing a Systematic Approach to Biobanking and Analyzing Clinical and Genetic Data for Advancing COVID-19 Research		4
12	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
11	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. *Human Genetics*, **2021**, 141, 147	6.3	3
10	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males		3
9	JNK signaling provides a novel therapeutic target for Rett syndrome.. *BMC Biology*, **2021**, 19, 256	7.3	3
8	High rate of HDR in gene editing of p.(Thr158Met) MECP2 mutational hotspot. *European Journal of Human Genetics*, **2020**, 28, 1231-1242	5.3	2

7	The Italian XLMR bank: a clinical and molecular database. *Human Mutation*, **2007**, 28, 13-8	4.7	2
6	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,		2
5	Low-level TP53 mutational load antecedes clonal expansion in chronic lymphocytic leukaemia. *British Journal of Haematology*, **2019**, 184, 657-659	4.5	2
4	Huntingtons disease gene expansion associates with early onset nonprogressive chorea. *Movement Disorders*, **2013**, 28, 684	7	1
3	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in males		1
2	The phenomenon of multidrug resistance in glioblastomas. *Hematology/ Oncology and Stem Cell Therapy*, **2021**,	2.7	1
1	Post-Mendelian genetic model in COVID-19		1