Laura Crisponi

List of Publications by Year
in descending order

72	18,304	71061 41	88593 70
papers	citations	h-index	g-index

76	76	76	27027
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
2	New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. Nature Genetics, 2010, 42, 105-116.	9.4	1,982
3	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
4	Modeling Linkage Disequilibrium Increases Accuracy of Polygenic Risk Scores. American Journal of Human Genetics, 2015, 97, 576-592.	2.6	1,098
5	The putative forkhead transcription factor FOXL2 is mutated in blepharophimosis/ptosis/epicanthus inversus syndrome. Nature Genetics, 2001, 27, 159-166.	9.4	886
6	Genome-Wide Association Analysis Identifies Variants Associated with Nonalcoholic Fatty Liver Disease That Have Distinct Effects on Metabolic Traits. PLoS Genetics, 2011, 7, e1001324.	1.5	796
7	Variants in MTNR1B influence fasting glucose levels. Nature Genetics, 2009, 41, 77-81.	9.4	662
8	Genome-wide association study shows BCL11A associated with persistent fetal hemoglobin and amelioration of the phenotype of Î²-thalassemia. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 1620-1625.	3.3	561
9	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. Nature, 2014, 514, 92-97.	13.7	548
10	Genome-wide association study identifies loci influencing concentrations of liver enzymes in plasma. Nature Genetics, 2011, 43, 1131-1138.	9.4	501
11	Foxl2 disruption causes mouse ovarian failure by pervasive blockage of follicle development. Human Molecular Genetics, 2004, 13, 1171-1181.	1.4	468
12	Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies. Nature Genetics, 2010, 42, 1077-1085.	9.4	445
13	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. Nature Genetics, 2017, 49, 834-841.	9.4	426
14	Novel Loci for Adiponectin Levels and Their Influence on Type 2 Diabetes and Metabolic Traits: A Multi-Ethnic Meta-Analysis of 45,891 Individuals. PLoS Genetics, 2012, 8, e1002607.	1.5	419
15	Common variants in the GDF5-UQCC region are associated with variation in human height. Nature Genetics, 2008, 40, 198-203.	9.4	369
16	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	9.4	357
17	Common variants at ten loci modulate the QT interval duration in the QTSCD Study. Nature Genetics, 2009, 41, 407-414.	9.4	356
18	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. Nature Genetics, 2012, 44, 260-268.	9.4	303

#	Article	IF	Citations
19	Foxl2 is required for commitment to ovary differentiation. Human Molecular Genetics, 2005, 14, 2053-2062.	1.4	298
20	The GLUT9 Gene Is Associated with Serum Uric Acid Levels in Sardinia and Chianti Cohorts. PLoS Genetics, 2007, 3, e194.	1.5	249
21	A Genome-Wide Association Search for Type 2 Diabetes Genes in African Americans. PLoS ONE, 2012, 7, e29202.	1.1	197
22	Genetic insights into biological mechanisms governing human ovarian ageing. Nature, 2021, 596, 393-397.	13.7	183
23	Genes and translocations involved in POF. American Journal of Medical Genetics Part A, 2002, 111, 328-333.	2.4	146
24	IRAK-M Is Involved in the Pathogenesis of Early-Onset Persistent Asthma. American Journal of Human Genetics, 2007, 80, 1103-1114.	2.6	144
25	A Genome-Wide Association Scan on the Levels of Markers of Inflammation in Sardinians Reveals Associations That Underpin Its Complex Regulation. PLoS Genetics, 2012, 8, e1002480.	1.5	141
26	Common variants in the SLCO1B3 locus are associated with bilirubin levels and unconjugated hyperbilirubinemia. Human Molecular Genetics, 2009, 18, 2711-2718.	1.4	126
27	Phosphodiesterase 8B Gene Variants Are Associated with Serum TSH Levels and Thyroid Function. American Journal of Human Genetics, 2008, 82, 1270-1280.	2.6	124
28	Overgrowth of a Mouse Model of the Simpsonâ€“Golabiâ€“Behmel Syndrome Is Independent of IGF Signaling. Developmental Biology, 2002, 243, 185-206.	0.9	89
29	Genome-wide association study of sexual maturation in males and females highlights a role for body mass and menarche loci in male puberty. Human Molecular Genetics, 2014, 23, 4452-4464.	1.4	82
30	Aging of Oocyte, Ovary, and Human Reproduction. Annals of the New York Academy of Sciences, 2004, 1034, 117-131.	1.8	77
31	Crisponi Syndrome Is Caused by Mutations in the CRLF1 Gene and Is Allelic to Cold-Induced Sweating Syndrome Type 1. American Journal of Human Genetics, 2007, 80, 971-981.	2.6	76
32	Neuroticism, Depressive Symptoms, and Serum BDNF. Psychosomatic Medicine, 2011, 73, 638-642.	1.3	67
33	A Novel Null Homozygous Mutation Confirms CACNA2D2 as a Gene Mutated in Epileptic Encephalopathy. PLoS ONE, 2013, 8, e82154.	1.1	67
34	Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index. Molecular Psychiatry, 2017, 22, 192-201.	4.1	63
35	Genetics of serum BDNF: Meta-analysis of the Val66Met and genome-wide association study. World Journal of Biological Psychiatry, 2013, 14, 583-589.	1.3	57
36	Association of Adiposity Genetic Variants With Menarche Timing in 92,105 Women of European Descent. American Journal of Epidemiology, 2013, 178, 451-460.	1.6	51

#	Article	IF	Citations
37	FOXL2 inactivation by a translocation 171 kb away: analysis of 500 kb of chromosome 3 for candidate long-range regulatory sequences. Genomics, 2004, 83, 757-764.	1.3	50
38	Determination and stability of sex. BioEssays, 2007, 29, 15-25.	1.2	49
39	Determination and Stability of Gonadal Sex. Journal of Andrology, 2010, 31, 16-25.	2.0	46
40	Investigating the genetic relationship between Alzheimerâ€™s disease and cancer using GWAS summary statistics. Human Genetics, 2017, 136, 1341-1351.	1.8	46
41	Jagged-1 mutation analysis in Italian Alagille syndrome patients. Human Mutation, 1999, 14, 394-400.	1.1	44
42	Two patients with balanced translocations and autistic disorder: CSMD3 as a candidate gene for autism found in their common 8q23 breakpoint area. European Journal of Human Genetics, 2008, 16, 696-704.	1.4	37
43	Differential secretion of the mutated protein is a major component affecting phenotypic severity in CRLF1-associated disorders. European Journal of Human Genetics, 2011, 19, 525-533.	1.4	34
44	Rare coding variants and X-linked loci associated with age at menarche. Nature Communications, 2015, 6, 7756.	5.8	32
45	Analysis of Exon/Intron Structure and 400 kb of Genomic Sequence Surrounding the 5â€²-Promoter and 3â€²-Terminal Ends of the Human Glypican 3 (GPC3) Gene. Genomics, 1997, 45, 48-58.	1.3	29
46	Bi-allelic Mutations in KLHL7 Cause a Crisponi/CISS1-like Phenotype Associated with Early-Onset Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 236-245.	2.6	28
47	FOXL2 modulates cartilage, skeletal development and IGF1-dependent growth in mice. BMC Developmental Biology, 2015, 15, 27.	2.1	27
48	SUMOylation of the Forkhead Transcription Factor FOXL2 Promotes Its Stabilization/Activation through Transient Recruitment to PML Bodies. PLoS ONE, 2011, 6, e25463.	1.1	24
49	Mandibular hypoplasia, deafness, progeroid features and lipodystrophy (MDPL) syndrome in the context of inherited lipodystrophies. Metabolism: Clinical and Experimental, 2015, 64, 1530-1540.	1.5	24
50	The Forkhead Transcription Factor Foxl2 Is Sumoylated in Both Human and Mouse: Sumoylation Affects Its Stability, Localization, and Activity. PLoS ONE, 2010, 5, e9477.	1.1	21
51	Expanding the Mutational Spectrum ofCRLF1in Crisponi/CISS1 Syndrome. Human Mutation, 2014, 35, 424-433.	1.1	21
52	A role of BRCA1 and BRCA2germline mutations in breast cancer susceptibility within Sardinian population. BMC Cancer, 2009, 9, 245.	1.1	18
53	Reproductive aging-associated common genetic variants and the risk of breast cancer. Breast Cancer Research, 2012, 14, R54.	2.2	17
54	Crisponi syndrome: A new case with additional features and new mutation in CRLF1. American Journal of Medical Genetics, Part A, 2008, 146A, 3237-3239.	0.7	16

#	Article	IF	Citations
55	Successful treatment of coldâ€induced sweating in Crisponi syndrome and its possible mechanism of action. Developmental Medicine and Child Neurology, 2010, 52, 494-497.	1.1	16
56	Glypican 3 and glypican 4 are juxtaposed in Xq26.1. Gene, 1998, 225, 9-16.	1.0	14
57	Novel NALCN biallelic truncating mutations in siblings with IHPRF1 syndrome. Clinical Genetics, 2018, 93, 1245-1247.	1.0	14
58	Novel ANKRD11 gene mutation in an individual with a mild phenotype of KBG syndrome associated to a GEFS+ phenotypic spectrum: a case report. BMC Medical Genetics, 2019, 20, 16.	2.1	14
59	Confirmation of a new phenotype in an individual with a variant in the last part of exon 30 of CREBBP. American Journal of Medical Genetics, Part A, 2019, 179, 634-638.	0.7	13
60	Crisponi syndrome in an Indian patient: A rare differential diagnosis for neonatal tetanus. American Journal of Medical Genetics, Part A, 2008, 146A, 2831-2834.	0.7	12
61	Genome-wide association study of susceptibility loci for breast cancer in Sardinian population. BMC Cancer, 2015, 15, 383.	1.1	12
62	Crisponi/coldâ€induced sweating syndrome: Differential diagnosis, pathogenesis and treatment concepts. Clinical Genetics, 2020, 97, 209-221.	1.0	12
63	CRLF1 and CLCF1 in Development, Health and Disease. International Journal of Molecular Sciences, 2022, 23, 992.	1.8	11
64	Novel action of FOXL2 as mediator of Col1a2 gene autoregulation. Developmental Biology, 2016, 416, 200-211.	0.9	9
65	Exome sequencing in Crisponi/coldâ€induced sweating syndromeâ€"like individuals reveals unpredicted alternative diagnoses. Clinical Genetics, 2019, 95, 607-614.	1.0	7
66	Transcriptional Control of Ovarian Development in Somatic Cells. Seminars in Reproductive Medicine, 2007, 25, 252-263.	0.5	5
67	A new case series of Crisponi syndrome in a Turkish family and review of the literature. Clinical Dysmorphology, 2017, 26, 66-72.	0.1	5
68	Large-Scale Genomic Analyses Link Reproductive Aging to Hypothalamic Signaling, Breast Cancer Susceptibility, and BRCA1-Mediated DNA Repair. Obstetrical and Gynecological Survey, 2015, 70, 758-762.	0.2	0
69	Crisponi/Cold Induced Sweating Syndrome Type 1 With a Private Cytokine Receptor Like Factor 1 (CRLF1) Mutation in an Indian Family. Indian Pediatrics, 2020, 57, 1075-1077.	0.2	0
70	Crisponi syndrome/cold-induced sweating syndrome type 2: Reprogramming of CS/CISS2 individual derived fibroblasts into three clones of one iPSC line. Stem Cell Research, 2020, 46, 101855.	0.3	0
71	Generation of induced pluripotent stem cell lines from a Crisponi/Cold induced sweating syndrome type 1 individual. Stem Cell Research, 2020, 46, 101820.	0.3	0
72	Editorial: Female Infertility: Genetics of Reproductive Ageing, Menopause and Primary Ovarian Insufficiency. Frontiers in Genetics, 2022, 13, 839758.	1.1	0