Laura Crisponi

List of Publications by Year
in descending order

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Version: 2024-02-01

72 papers	18,304 citations	87401 40 h-index	104191 69 g-index
76 all docs	76 docs citations	76 times ranked	29742 citing authors

#	Article	IF	Citations
1	CRLF1 and CLCF1 in Development, Health and Disease. International Journal of Molecular Sciences, 2022, 23, 992.	1.8	11
2	Editorial: Female Infertility: Genetics of Reproductive Ageing, Menopause and Primary Ovarian Insufficiency. Frontiers in Genetics, 2022, 13, 839758.	1.1	0
3	Genetic insights into biological mechanisms governing human ovarian ageing. Nature, 2021, 596, 393-397.	13.7	183
4	Crisponi/coldâ€induced sweating syndrome: Differential diagnosis, pathogenesis and treatment concepts. Clinical Genetics, 2020, 97, 209-221.	1.0	12
5	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
6	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
7	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
8	Exome sequencing in Crisponi/coldâ€induced sweating syndromeâ€“like individuals reveals unpredicted alternative diagnoses. Clinical Genetics, 2019, 95, 607-614.	1.0	7
9	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
10	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
11	Novel NALCN biallelic truncating mutations in siblings with IHPRF1 syndrome. Clinical Genetics, 2018, 93, 1245-1247.	1.0	14
12	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
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	Investigating the genetic relationship between Alzheimerâ€™s disease and cancer using GWAS summary statistics. Human Genetics, 2017, 136, 1341-1351.	1.8	46
15	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
16	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
17	Novel action of FOXL2 as mediator of Col1a2 gene autoregulation. Developmental Biology, 2016, 416, 200-211.	0.9	9
18	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

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19	Rare coding variants and X-linked loci associated with age at menarche. Nature Communications, 2015, 6, 7756.	5.8	32
20	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
21	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
22	Genome-wide association study of susceptibility loci for breast cancer in Sardinian population. BMC Cancer, 2015, 15, 383.	1.1	12
23	Modeling Linkage Disequilibrium Increases Accuracy of Polygenic Risk Scores. American Journal of Human Genetics, 2015, 97, 576-592.	2.6	1,098
24	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
25	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
26	FOXL2 modulates cartilage, skeletal development and IGF1-dependent growth in mice. BMC Developmental Biology, 2015, 15, 27.	2.1	27
27	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
28	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. Nature, 2014, 514, 92-97.	13.7	548
29	Expanding the Mutational Spectrum ofCRLF1in Crisponi/CISS1 Syndrome. Human Mutation, 2014, 35, 424-433.	1.1	21
30	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
31	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
32	A Novel Null Homozygous Mutation Confirms CACNA2D2 as a Gene Mutated in Epileptic Encephalopathy. PLoS ONE, 2013, 8, e82154.	1.1	67
33	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
34	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
35	Reproductive aging-associated common genetic variants and the risk of breast cancer. Breast Cancer Research, 2012, 14, R54.	2.2	17
36	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

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37	A Genome-Wide Association Search for Type 2 Diabetes Genes in African Americans. PLoS ONE, 2012, 7, e29202.	1.1	197
38	Genome-wide association study identifies loci influencing concentrations of liver enzymes in plasma. Nature Genetics, 2011, 43, 1131-1138.	9.4	501
39	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
40	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
41	Neuroticism, Depressive Symptoms, and Serum BDNF. Psychosomatic Medicine, 2011, 73, 638-642.	1.3	67
42	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
43	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
44	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
45	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
46	Determination and Stability of Gonadal Sex. Journal of Andrology, 2010, 31, 16-25.	2.0	46
47	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
48	Common variants in the SLCO1B3 locus are associated with bilirubin levels and unconjugated hyperbilirubinemia. Human Molecular Genetics, 2009, 18, 2711-2718.	1.4	126
49	A role of BRCA1 and BRCA2germline mutations in breast cancer susceptibility within Sardinian population. BMC Cancer, 2009, 9, 245.	1.1	18
50	Variants in MTNR1B influence fasting glucose levels. Nature Genetics, 2009, 41, 77-81.	9.4	662
51	Common variants at ten loci modulate the QT interval duration in the QTSCD Study. Nature Genetics, 2009, 41, 407-414.	9.4	356
52	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
53	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
54	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

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55	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
56	Common variants in the GDF5-UQCC region are associated with variation in human height. Nature Genetics, 2008, 40, 198-203.	9.4	369
57	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
58	Transcriptional Control of Ovarian Development in Somatic Cells. Seminars in Reproductive Medicine, 2007, 25, 252-263.	0.5	5
59	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
60	IRAK-M Is Involved in the Pathogenesis of Early-Onset Persistent Asthma. American Journal of Human Genetics, 2007, 80, 1103-1114.	2.6	144
61	The GLUT9 Gene Is Associated with Serum Uric Acid Levels in Sardinia and Chianti Cohorts. PLoS Genetics, 2007, 3, e194.	1.5	249
62	Determination and stability of sex. BioEssays, 2007, 29, 15-25.	1.2	49
63	Foxl2 is required for commitment to ovary differentiation. Human Molecular Genetics, 2005, 14, 2053-2062.	1.4	298
64	Foxl2 disruption causes mouse ovarian failure by pervasive blockage of follicle development. Human Molecular Genetics, 2004, 13, 1171-1181.	1.4	468
65	Aging of Oocyte, Ovary, and Human Reproduction. Annals of the New York Academy of Sciences, 2004, 1034, 117-131.	1.8	77
66	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
67	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
68	Genes and translocations involved in POF. American Journal of Medical Genetics Part A, 2002, 111, 328-333.	2.4	146
69	The putative forkhead transcription factor FOXL2 is mutated in blepharophimosis/ptosis/epicanthus inversus syndrome. Nature Genetics, 2001, 27, 159-166.	9.4	886
70	Jagged-1 mutation analysis in Italian Alagille syndrome patients. , 1999, 14, 394-400.		44
71	Glypican 3 and glypican 4 are juxtaposed in Xq26.1. Gene, 1998, 225, 9-16.	1.0	14
72	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