

Daniela Toniolo

List of Publications by Year in descending order

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187
papers

24,934
citations

10389

72
h-index

8396

147
g-index

203
all docs

203
docs citations

203
times ranked

34275
citing authors

#	ARTICLE	IF	CITATIONS
1	A reference panel of 64,976 haplotypes for genotype imputation. <i>Nature Genetics</i> , 2016, 48, 1279-1283.	21.4	2,421
2	Mutations in the gene encoding lamin A/C cause autosomal dominant Emery-Dreifuss muscular dystrophy. <i>Nature Genetics</i> , 1999, 21, 285-288.	21.4	1,245
3	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , 2015, 526, 82-90.	27.8	1,014
4	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. <i>Nature Genetics</i> , 2018, 50, 1412-1425.	21.4	924
5	Identification of a novel X-linked gene responsible for Emery-Dreifuss muscular dystrophy. <i>Nature Genetics</i> , 1994, 8, 323-327.	21.4	857
6	A novel X-linked gene, G4.5, is responsible for Barth syndrome. <i>Nature Genetics</i> , 1996, 12, 385-389.	21.4	718
7	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. <i>Nature Genetics</i> , 2013, 45, 145-154.	21.4	675
8	A General Approach for Haplotype Phasing across the Full Spectrum of Relatedness. <i>PLoS Genetics</i> , 2014, 10, e1004234.	3.5	553
9	A catalog of genetic loci associated with kidney function from analyses of a million individuals. <i>Nature Genetics</i> , 2019, 51, 957-972.	21.4	549
10	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. <i>Nature Genetics</i> , 2017, 49, 403-415.	21.4	492
11	Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies. <i>Nature Genetics</i> , 2010, 42, 1077-1085.	21.4	445
12	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. <i>Nature Genetics</i> , 2017, 49, 834-841.	21.4	426
13	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. <i>Nature Communications</i> , 2016, 7, 10023.	12.8	412
14	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. <i>Nature Genetics</i> , 2011, 43, 1005-1011.	21.4	403
15	New gene functions in megakaryopoiesis and platelet formation. <i>Nature</i> , 2011, 480, 201-208.	27.8	401
16	Different Mutations in the LMNA Gene Cause Autosomal Dominant and Autosomal Recessive Emery-Dreifuss Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2000, 66, 1407-1412.	6.2	384
17	Clinical significance of somatic mutation in unexplained blood cytopenia. <i>Blood</i> , 2017, 129, 3371-3378.	1.4	379
18	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. <i>Nature Genetics</i> , 2015, 47, 1294-1303.	21.4	357

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19	Seventy-five genetic loci influencing the human red blood cell. <i>Nature</i> , 2012, 492, 369-375.	27.8	320
20	Mutations in GDI1 are responsible for X-linked non-specific mental retardation. <i>Nature Genetics</i> , 1998, 19, 134-139.	21.4	304
21	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. <i>Nature Genetics</i> , 2012, 44, 260-268.	21.4	303
22	Trans-ancestry genome-wide association study identifies 12 genetic loci influencing blood pressure and implicates a role for DNA methylation. <i>Nature Genetics</i> , 2015, 47, 1282-1293.	21.4	294
23	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. <i>Nature Genetics</i> , 2016, 48, 1462-1472.	21.4	284
24	Genetic Structure of Europeans: A View from the North-East. <i>PLoS ONE</i> , 2009, 4, e5472.	2.5	279
25	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. <i>Nature Genetics</i> , 2019, 51, 1459-1474.	21.4	251
26	A Human Homologue of the <i>Drosophila melanogaster</i> diaphanous Gene Is Disrupted in a Patient with Premature Ovarian Failure: Evidence for Conserved Function in Oogenesis and Implications for Human Sterility. <i>American Journal of Human Genetics</i> , 1998, 62, 533-541.	6.2	248
27	Isolation of human glucose-6-phosphate dehydrogenase (G6PD) cDNA clones: primary structure of the protein and unusual 5' non-coding region. <i>Nucleic Acids Research</i> , 1986, 14, 2511-2522.	14.5	242
28	The X-Linked Gene G4.5 Is Responsible for Different Infantile Dilated Cardiomyopathies. <i>American Journal of Human Genetics</i> , 1997, 61, 862-867.	6.2	236
29	Mutations in the Small GTPase Gene RAB39B Are Responsible for X-linked Mental Retardation Associated with Autism, Epilepsy, and Macrocephaly. <i>American Journal of Human Genetics</i> , 2010, 86, 185-195.	6.2	220
30	A Mutation in the Rett Syndrome Gene, MECP2, Causes X-Linked Mental Retardation and Progressive Spasticity in Males. <i>American Journal of Human Genetics</i> , 2000, 67, 982-985.	6.2	213
31	A Meta-Analysis of Thyroid-Related Traits Reveals Novel Loci and Gender-Specific Differences in the Regulation of Thyroid Function. <i>PLoS Genetics</i> , 2013, 9, e1003266.	3.5	194
32	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. <i>Nature Genetics</i> , 2015, 47, 1272-1281.	21.4	193
33	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. <i>Nature Communications</i> , 2014, 5, 4926.	12.8	192
34	Age- And Sex-Related Variations in Platelet Count in Italy: A Proposal of Reference Ranges Based on 40987 Subjects' Data. <i>PLoS ONE</i> , 2013, 8, e54289.	2.5	190
35	Genetic insights into biological mechanisms governing human ovarian ageing. <i>Nature</i> , 2021, 596, 393-397.	27.8	183
36	Isolation of sequences that span the fragile X and identification of a fragile X-related CpG island. <i>Science</i> , 1991, 251, 1236-1239.	12.6	181

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37	Genome-wide analyses identify a role for SLC17A4 and AADAT in thyroid hormone regulation. <i>Nature Communications</i> , 2018, 9, 4455.	12.8	181
38	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. <i>Nature Communications</i> , 2015, 6, 5897.	12.8	173
39	Directional dominance on stature and cognition in diverse human populations. <i>Nature</i> , 2015, 523, 459-462.	27.8	173
40	A family of transmembrane proteins with homology to the MET-hepatocyte growth factor receptor. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1996, 93, 674-678.	7.1	169
41	Genome-Wide Association and Functional Follow-Up Reveals New Loci for Kidney Function. <i>PLoS Genetics</i> , 2012, 8, e1002584.	3.5	166
42	Influence of intermediate and uninterrupted FMR1 CGG expansions in premature ovarian failure manifestation. <i>Human Reproduction</i> , 2006, 21, 952-957.	0.9	162
43	Clinical Relevance of Atrial Fibrillation/Flutter, Stroke, Pacemaker Implant, and Heart Failure in Emery-Dreifuss Muscular Dystrophy. <i>Stroke</i> , 2003, 34, 901-908.	2.0	158
44	Identification of Novel Genetic Loci Associated with Thyroid Peroxidase Antibodies and Clinical Thyroid Disease. <i>PLoS Genetics</i> , 2014, 10, e1004123.	3.5	150
45	Heart-specific localization of emerin: new insights into Emery-Dreifuss muscular dystrophy. <i>Human Molecular Genetics</i> , 1997, 6, 2257-2264.	2.9	138
46	Eleven X Chromosome Breakpoints Associated with Premature Ovarian Failure (POF) Map to a 15-Mb YAC Contig Spanning Xq21. <i>Genomics</i> , 1997, 40, 123-131.	2.9	133
47	General population low-count CLL-like MBL persists over time without clinical progression, although carrying the same cytogenetic abnormalities of CLL. <i>Blood</i> , 2011, 118, 6618-6625.	1.4	131
48	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. <i>American Journal of Human Genetics</i> , 2017, 100, 865-884.	6.2	131
49	BMP15 mutations associated with primary ovarian insufficiency cause a defective production of bioactive protein. <i>Human Mutation</i> , 2009, 30, 804-810.	2.5	126
50	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. <i>Hypertension</i> , 2017, 70, .	2.7	123
51	The immunoglobulin gene repertoire of low-count chronic lymphocytic leukemia (CLL) "like" monoclonal B lymphocytosis is different from CLL: diagnostic implications for clinical monitoring. <i>Blood</i> , 2009, 114, 26-32.	1.4	122
52	Ultrastructural abnormality of sarcolemmal nuclei in Emery-Dreifuss muscular dystrophy (EDMD). <i>Journal of the Neurological Sciences</i> , 1998, 159, 88-93.	0.6	115
53	Alterations in the expression, structure and function of progesterone receptor membrane component-1 (PGRMC1) in premature ovarian failure. <i>Human Molecular Genetics</i> , 2008, 17, 3776-3783.	2.9	114
54	Multiple Loci Are Associated with White Blood Cell Phenotypes. <i>PLoS Genetics</i> , 2011, 7, e1002113.	3.5	106

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55	Chromosomal rearrangements in Xq and premature ovarian failure: mapping of 25 new cases and review of the literature. <i>Human Reproduction</i> , 2006, 21, 1477-1483.	0.9	105
56	A genome-wide association study of early menopause and the combined impact of identified variants. <i>Human Molecular Genetics</i> , 2013, 22, 1465-1472.	2.9	104
57	Cardiac features of Emery-Dreifuss muscular dystrophy caused by lamin A/C gene mutations. <i>European Heart Journal</i> , 2003, 24, 2227-2236.	2.2	103
58	Deletion of the mental retardation gene <i>Gdi1</i> impairs associative memory and alters social behavior in mice. <i>Human Molecular Genetics</i> , 2002, 11, 2567-2580.	2.9	100
59	Mutation analysis of the lamin A/C gene (<i>LMNA</i>) among patients with different cardiomyopathy phenotypes. <i>Journal of Medical Genetics</i> , 2003, 40, 132e-132.	3.2	98
60	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. <i>Scientific Reports</i> , 2017, 7, 45040.	3.3	98
61	<i>TMPRSS6</i> rs855791 modulates hepcidin transcription in vitro and serum hepcidin levels in normal individuals. <i>Blood</i> , 2011, 118, 4459-4462.	1.4	97
62	Complete concordance between glucose-6-phosphate dehydrogenase activity and hypomethylation of 3 CpG clusters: implications for X chromosome dosage compensation. <i>Nucleic Acids Research</i> , 1984, 12, 9333-9348.	14.5	94
63	Identification of new mutations in the Emery-Dreifuss muscular dystrophy gene and evidence for genetic heterogeneity of the disease. <i>Human Molecular Genetics</i> , 1995, 4, 1859-1863.	2.9	93
64	Normal serum concentrations of anti-Müllerian hormone in women with regular menstrual cycles. <i>Reproductive BioMedicine Online</i> , 2010, 21, 463-469.	2.4	91
65	Cytological mapping of the human glucose-6-phosphate dehydrogenase gene distal to the fragile-X site suggests a high rate of meiotic recombination across this site. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1984, 81, 7855-7859.	7.1	86
66	Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability. <i>Nature Genetics</i> , 2018, 50, 652-656.	21.4	86
67	Common Variants in <i>UMOD</i> Associate with Urinary Uromodulin Levels. <i>Journal of the American Society of Nephrology: JASN</i> , 2014, 25, 1869-1882.	6.1	85
68	Association of <i>HFE</i> and <i>TMPRSS6</i> genetic variants with iron and erythrocyte parameters is only in part dependent on serum hepcidin concentrations. <i>Journal of Medical Genetics</i> , 2011, 48, 629-634.	3.2	84
69	Associations of autozygosity with a broad range of human phenotypes. <i>Nature Communications</i> , 2019, 10, 4957.	12.8	84
70	Mutation analysis of two candidate genes for premature ovarian failure, <i>DACH2</i> and <i>POF1B</i> . <i>Human Reproduction</i> , 2004, 19, 2759-2766.	0.9	82
71	A "housekeeping" gene on the X chromosome encodes a protein similar to ubiquitin. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1988, 85, 851-855.	7.1	80
72	Evidence of Inbreeding Depression on Human Height. <i>PLoS Genetics</i> , 2012, 8, e1002655.	3.5	79

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73	108th ENMC International Workshop, 3rd Workshop of the MYO-CLUSTER project: EUROMEN, 7th International Emery-Dreifuss Muscular Dystrophy (EDMD) Workshop, 13-15 September 2002, Naarden, The Netherlands. <i>Neuromuscular Disorders</i> , 2003, 13, 508-515.	0.6	78
74	X-linked premature ovarian failure: a complex disease. <i>Current Opinion in Genetics and Development</i> , 2006, 16, 293-300.	3.3	78
75	Physical map of human Xq27-qter: localizing the region of the fragile X mutation.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1991, 88, 8302-8306.	7.1	76
76	Whole-genome sequence-based analysis of thyroid function. <i>Nature Communications</i> , 2015, 6, 5681.	12.8	75
77	Mapping of two genes encoding isoforms of the actin binding protein ABP-280, a dystrophin like protein, to Xq28 and to chromosome 7. <i>Human Molecular Genetics</i> , 1993, 2, 761-766.	2.9	73
78	Tissue-specific levels of human glucose-6-phosphate dehydrogenase correlate with methylation of specific sites at the 3' end of the gene.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1985, 82, 1465-1469.	7.1	72
79	X Chromosome Inactivation in Carriers of Barth Syndrome. <i>American Journal of Human Genetics</i> , 1998, 63, 1457-1463.	6.2	71
80	A 12Mb deletion at 7q33-q35 associated with autism spectrum disorders and primary amenorrhea. <i>European Journal of Medical Genetics</i> , 2008, 51, 631-638.	1.3	68
81	Increased Serum Hepcidin Levels in Subjects with the Metabolic Syndrome: A Population Study. <i>PLoS ONE</i> , 2012, 7, e48250.	2.5	68
82	<i>MCM8</i> and <i>MCM9</i> Nucleotide Variants in Women with Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, jc.2016-2565.	3.6	68
83	Transcriptional organization of a 450-kb region of the human X chromosome in Xq28. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1993, 90, 10977-10981.	7.1	67
84	Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps. <i>Nature Genetics</i> , 2016, 48, 1303-1312.	21.4	66
85	Non-specific X-linked semidominant mental retardation by mutations in a Rab GDP-dissociation inhibitor. <i>Human Molecular Genetics</i> , 1998, 7, 1311-1315.	2.9	65
86	Integration of genome-wide association studies with biological knowledge identifies six novel genes related to kidney function. <i>Human Molecular Genetics</i> , 2012, 21, 5329-5343.	2.9	64
87	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. <i>European Journal of Human Genetics</i> , 2013, 21, 659-665.	2.8	64
88	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. <i>Nature Communications</i> , 2017, 8, 15927.	12.8	64
89	Modulation of Genetic Associations with Serum Urate Levels by Body-Mass-Index in Humans. <i>PLoS ONE</i> , 2015, 10, e0119752.	2.5	64
90	cDNA sequences of human glucose 6-phosphate dehydrogenase cloned in pBR322. <i>Nature</i> , 1981, 294, 778-780.	27.8	60

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91	Relationship between clone metrics and clinical outcome in clonal cytopenia. <i>Blood</i> , 2021, 138, 965-976.	1.4	58
92	X Chromosome and Ovarian Failure. <i>Seminars in Reproductive Medicine</i> , 2007, 25, 264-271.	1.1	56
93	Six Novel Loci Associated with Circulating VEGF Levels Identified by a Meta-analysis of Genome-Wide Association Studies. <i>PLoS Genetics</i> , 2016, 12, e1005874.	3.5	56
94	Frequent low penetrance mutations in the Lamin A/C gene, causing Emery Dreifuss muscular dystrophy. <i>Neuromuscular Disorders</i> , 2002, 12, 958-963.	0.6	52
95	Association of Adiposity Genetic Variants With Menarche Timing in 92,105 Women of European Descent. <i>American Journal of Epidemiology</i> , 2013, 178, 451-460.	3.4	51
96	Molecular cloning and analysis of the fragile X region in man. <i>Nucleic Acids Research</i> , 1991, 19, 2567-2572.	14.5	50
97	An X chromosome-linked gene encoding a protein with characteristics of a rhoGAP predominantly expressed in hematopoietic cells.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1996, 93, 695-699.	7.1	50
98	Mice deficient for the synaptic vesicle protein Rab3a show impaired spatial reversal learning and increased explorative activity but none of the behavioral changes shown by mice deficient for the Rab3a regulator Gdi1. <i>European Journal of Neuroscience</i> , 2004, 19, 1895-1905.	2.6	50
99	Cognitive impairment in Gdi1-deficient mice is associated with altered synaptic vesicle pools and short-term synaptic plasticity, and can be corrected by appropriate learning training. <i>Human Molecular Genetics</i> , 2009, 18, 105-117.	2.9	50
100	CpG islands of the X chromosome are gene associated. <i>Nucleic Acids Research</i> , 1988, 16, 9527-9543.	14.5	49
101	Sequence variation at the human FOXO3 locus: a study of premature ovarian failure and primary amenorrhea. <i>Human Reproduction</i> , 2007, 23, 216-221.	0.9	49
102	Single-cell expression profiling of dopaminergic neurons combined with association analysis identifies pyridoxal kinase as Parkinson's disease gene. <i>Annals of Neurology</i> , 2009, 66, 792-798.	5.3	49
103	The CpG island in the 5' region of the G6PD gene of man and mouse. <i>Gene</i> , 1991, 102, 197-203.	2.2	46
104	Methylation and sequence analysis around Eagi sites: identification of 28 new CpG islands in XQ24-XQ28. <i>Nucleic Acids Research</i> , 1992, 20, 727-733.	14.5	44
105	Immunocytochemical detection of emerin within the nuclear matrix. <i>Neuromuscular Disorders</i> , 1998, 8, 338-344.	0.6	44
106	A susceptibility gene for premature ovarian failure (POF) maps to proximal Xq28. <i>European Journal of Human Genetics</i> , 2004, 12, 829-834.	2.8	44
107	Neurite Extension Occurs in the Absence of Regulated Exocytosis in PC12 Subclones. <i>Molecular Biology of the Cell</i> , 1999, 10, 2919-2931.	2.1	43
108	Inherited genetic susceptibility to monoclonal B-cell lymphocytosis. <i>Blood</i> , 2010, 116, 5957-5960.	1.4	42

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109	Association Analysis of Bitter Receptor Genes in Five Isolated Populations Identifies a Significant Correlation between TAS2R43 Variants and Coffee Liking. <i>PLoS ONE</i> , 2014, 9, e92065.	2.5	41
110	SOS2 and ACP1 Loci Identified through Large-Scale Exome Chip Analysis Regulate Kidney Development and Function. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 981-994.	6.1	39
111	A mutation in the X-linked Emeryâ€Dreifuss muscular dystrophy gene in a patient affected with conduction cardiomyopathy. <i>Neuromuscular Disorders</i> , 2001, 11, 411-413.	0.6	37
112	Heritability and Demographic Analyses in the Large Isolated Population of Val Borbera Suggest Advantages in Mapping Complex Traits Genes. <i>PLoS ONE</i> , 2009, 4, e7554.	2.5	37
113	Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. <i>Science Advances</i> , 2021, 7, .	10.3	36
114	Duplications of the X chromosome in males: evidence that most parts of the X chromosome can be active in two copies. <i>Human Genetics</i> , 1991, 86, 519-521.	3.8	35
115	Epigenetic control of the critical region for premature ovarian failure on autosomal genes translocated to the X chromosome: a hypothesis. <i>Human Genetics</i> , 2007, 121, 441-450.	3.8	35
116	Genome-Wide Meta-Analysis Unravels Interactions between Magnesium Homeostasis and Metabolic Phenotypes. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 335-348.	6.1	34
117	Epigenetic analysis of the critical region I for premature ovarian failure: demonstration of a highly heterochromatic domain on the long arm of the mammalian X chromosome. <i>Journal of Medical Genetics</i> , 2009, 46, 585-592.	3.2	33
118	Genome-wide meta-analysis of common variant differences between men and women. <i>Human Molecular Genetics</i> , 2012, 21, 4805-4815.	2.9	33
119	Influence of age, sex and ethnicity on platelet count in five Italian geographic isolates: mild thrombocytopenia may be physiological. <i>British Journal of Haematology</i> , 2012, 157, 384-387.	2.5	33
120	Common Variants in Mendelian Kidney Disease Genes and Their Association with Renal Function. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 2105-2117.	6.1	33
121	Small effective population size and genetic homogeneity in the Val Borbera isolate. <i>European Journal of Human Genetics</i> , 2013, 21, 89-94.	2.8	32
122	Rare coding variants and X-linked loci associated with age at menarche. <i>Nature Communications</i> , 2015, 6, 7756.	12.8	32
123	High-resolution array-CGH analysis on 46,XX patients affected by early onset primary ovarian insufficiency discloses new genes involved in ovarian function. <i>Human Reproduction</i> , 2019, 34, 574-583.	0.9	32
124	Emerin expression at the early stages of myogenic differentiation. <i>Differentiation</i> , 2000, 66, 208-217.	1.9	30
125	A large-scale association study to assess the impact of known variants of the human INHA gene on premature ovarian failure. <i>Human Reproduction</i> , 2009, 24, 2023-2028.	0.9	30
126	The Exon-Intron Organization of the Human X-Linked Gene (FLN1) Encoding Actin-Binding Protein 280. <i>Genomics</i> , 1994, 21, 71-76.	2.9	29

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127	Serum levels of the hepcidin-20 isoform in a large general population: The Val Borbera study. <i>Journal of Proteomics</i> , 2012, 76, 28-35.	2.4	29
128	Multicohort analysis of the maternal age effect on recombination. <i>Nature Communications</i> , 2015, 6, 7846.	12.8	29
129	A bird's-eye view of Italian genomic variation through whole-genome sequencing. <i>European Journal of Human Genetics</i> , 2020, 28, 435-444.	2.8	29
130	Common variants in CLDN14 are associated with differential excretion of magnesium over calcium in urine. <i>Pflügers Archiv European Journal of Physiology</i> , 2017, 469, 91-103.	2.8	27
131	An archipelago of CpG islands in Xq28: identification and fine mapping of 20 new CpG islands of the human X chromosome. <i>Human Molecular Genetics</i> , 1992, 1, 275-280.	2.9	26
132	Fertility Preservation in Endometriosis Patients: Anti-Müllerian Hormone Is a Reliable Marker of the Ovarian Follicle Density. <i>Frontiers in Surgery</i> , 2017, 4, 40.	1.4	26
133	X-linked non-specific mental retardation. <i>Current Opinion in Genetics and Development</i> , 2000, 10, 280-285.	3.3	25
134	Variation of hemoglobin levels in normal Italian populations from genetic isolates. <i>Haematologica</i> , 2008, 93, 1372-1375.	3.5	25
135	<i>COL6A5</i> variants in familial neuropathic chronic itch. <i>Brain</i> , 2017, 140, aww343.	7.6	25
136	Probes for CpG islands on the distal long arm of the human X chromosome are clustered in Xq24 and Xq28. <i>Genomics</i> , 1990, 8, 664-670.	2.9	24
137	Complementation of a defect in the production of ribosomal RNA in somatic cell hybrids. <i>Nature</i> , 1974, 248, 411-413.	27.8	23
138	X Chromosome Genes and Premature Ovarian Failure. <i>Seminars in Reproductive Medicine</i> , 2000, 18, 051-058.	1.1	23
139	Regulation of glucose 6-phosphate dehydrogenase expression in CHO-human fibroblast somatic cell hybrids. <i>Somatic Cell Genetics</i> , 1983, 9, 429-443.	2.7	22
140	Identification and characterization of a new gene in the human Xq28 region. <i>Human Molecular Genetics</i> , 1992, 1, 269-273.	2.9	22
141	The use of recombinant human growth hormone for radioiodination and standard preparation in radioimmunoassay. <i>Journal of Immunological Methods</i> , 1993, 159, 269-274.	1.4	22
142	Barth syndrome presenting with acute metabolic decompensation in the neonatal period. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 684-684.	3.6	22
143	A Genome-Wide Association Study in isolated populations reveals new genes associated to common food likings. <i>Reviews in Endocrine and Metabolic Disorders</i> , 2016, 17, 209-219.	5.7	22
144	Big Data in Medicine, the Present and Hopefully the Future. <i>Frontiers in Medicine</i> , 2019, 6, 263.	2.6	22

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145	SV40-transformed cells with temperature-dependent serum requirements. <i>Cell</i> , 1975, 4, 255-262.	28.9	21
146	Comparative Mapping of the Actin-Binding Protein 280 Genes in Human and Mouse. <i>Genomics</i> , 1994, 21, 428-430.	2.9	21
147	Localization of glucose-6-phosphate dehydrogenase in mouse and man by in situ hybridization: evidence for a single locus and transposition of homologous X-linked genes. <i>Cytogenetic and Genome Research</i> , 1985, 39, 87-92.	1.1	20
148	Linkage and sequence conservation of the X-linked genes DXS253E (P3) and DXS254E (GdX) in mouse and man. <i>Genomics</i> , 1990, 7, 453-457.	2.9	20
149	The <i>POF1B</i> candidate gene for premature ovarian failure regulates epithelial polarity. <i>Journal of Cell Science</i> , 2011, 124, 3356-3368.	2.0	20
150	The A736V <i>TMPRSS6</i> polymorphism influences hepcidin and iron metabolism in chronic hemodialysis patients: <i>TMPRSS6</i> and hepcidin in hemodialysis. <i>BMC Nephrology</i> , 2013, 14, 48.	1.8	20
151	Reconstructing the genetic history of Italians: new insights from a male (Y-chromosome) perspective. <i>Annals of Human Biology</i> , 2018, 45, 44-56.	1.0	19
152	Forebrain Deletion of $\hat{\pm}$ GDI in Adult Mice Worsens the Pre-Synaptic Deficit at Cortico-Lateral Amygdala Synaptic Connections. <i>PLoS ONE</i> , 2012, 7, e29763.	2.5	18
153	In search of the MRX genes. <i>American Journal of Medical Genetics Part A</i> , 2000, 97, 221-227.	2.4	17
154	Identification of <i>TRIM22</i> single nucleotide polymorphisms associated with loss of inhibition of HIV-1 transcription and advanced HIV-1 disease. <i>Aids</i> , 2013, 27, 2335-2344.	2.2	17
155	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. <i>Communications Biology</i> , 2022, 5, .	4.4	17
156	The nucleotide sequence of a CpG island demonstrates the presence of the first exon of the gene encoding the human lysosomal membrane protein lamp2 and assigns the gene to Xq24. <i>Genomics</i> , 1991, 9, 551-554.	2.9	16
157	Skewed X-chromosome inactivation is not associated with premature ovarian failure in a large cohort of Italian patients. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1349-1351.	1.2	16
158	G6PD Ferrara I has the same two mutations as G6PD A(-) but a distinct biochemical phenotype. <i>Human Genetics</i> , 1994, 93, 139-142.	3.8	15
159	FISH characterization of the Xq21 breakpoint in a translocation carrier with premature ovarian failure. <i>Clinical Genetics</i> , 1996, 50, 267-269.	2.0	15
160	Unusual expression of emerin in a patient with X-linked Emeryâ€Dreifuss muscular dystrophy. <i>Neuromuscular Disorders</i> , 2000, 10, 567-571.	0.6	13
161	Temporal gene expression profile of the hippocampus following trace fear conditioning. <i>Brain Research</i> , 2010, 1308, 14-23.	2.2	13
162	Association of a variant in the <i>CHRNA5-A3-B4</i> gene cluster region to heavy smoking in the Italian population. <i>European Journal of Human Genetics</i> , 2011, 19, 593-596.	2.8	13

#	ARTICLE	IF	CITATIONS
163	Selection and Mapping of Replication Origins from a 500-kb Region of the Human X Chromosome and Their Relationship to Gene Expression. <i>Genomics</i> , 1999, 62, 11-20.	2.9	12
164	Emerin presence in platelets. <i>Acta Neuropathologica</i> , 2000, 100, 291-298.	7.7	12
165	Spatial and temporal expression of POF1B, a gene expressed in epithelia. <i>Gene Expression Patterns</i> , 2007, 7, 529-534.	0.8	12
166	Genome-wide association analysis on five isolated populations identifies variants of the HLA-DOA gene associated with white wine liking. <i>European Journal of Human Genetics</i> , 2015, 23, 1717-1722.	2.8	12
167	Sequence and gene content in 52 kb including and centromeric to the G6PD gene in Xq28. <i>DNA Sequence</i> , 1995, 6, 1-11.	0.7	11
168	X-linked severe mental retardation and a progressive neurological disorder in a Belgian family: clinical and genetic studies. <i>Clinical Genetics</i> , 1997, 52, 155-161.	2.0	11
169	Long-term treatment of Barth syndrome with pantothenic acid: a retrospective study. <i>Molecular Genetics and Metabolism</i> , 2003, 80, 408-411.	1.1	10
170	Serum Hepcidin Levels Correlate with Phenotypes of the Metabolic Syndrome At Population Level. <i>Blood</i> , 2011, 118, 348-348.	1.4	10
171	Stability of DNA methylation of X-chromosome genes during aging. <i>Somatic Cell and Molecular Genetics</i> , 1990, 16, 79-84.	0.7	9
172	The screening for X-linked Emery-Dreifuss muscular dystrophy amongst young patients with idiopathic heart conduction system disease treated by a pacemaker implant. <i>European Journal of Neurology</i> , 2004, 11, 531-534.	3.3	9
173	Computer-based genealogy reconstruction in founder populations. <i>Journal of Biomedical Informatics</i> , 2011, 44, 997-1003.	4.3	9
174	Factors associated with food liking and their relationship with metabolic traits in Italian cohorts. <i>Food Quality and Preference</i> , 2019, 75, 64-70.	4.6	9
175	Genome Wide Association Analysis of a Founder Population Identified TAF3 as a Gene for MCHC in Humans. <i>PLoS ONE</i> , 2013, 8, e69206.	2.5	9
176	Biochemical and molecular characterization of a new sporadic glucose-6-phosphate dehydrogenase variant described in Italy: G6PD Modena. <i>British Journal of Haematology</i> , 1994, 87, 209-211.	2.5	8
177	A Comparative Transcriptional Map of a Region of 250 kb on the Human and Mouse X Chromosome between the G6PD and the FLN1 Genes. <i>Genomics</i> , 1995, 28, 377-382.	2.9	7
178	Highly Conserved Non-Coding Sequences and the 18q Critical Region for Short Stature: A Common Mechanism of Disease?. <i>PLoS ONE</i> , 2008, 3, e1460.	2.5	7
179	Selection and Fine Mapping of Chromosome-Specific cDNAs: Application to Human Chromosome 1. <i>Genomics</i> , 1996, 38, 149-154.	2.9	5
180	Is DNA methylation of X chromosome genes stable during aging?. <i>Somatic Cell and Molecular Genetics</i> , 1991, 17, 101-103.	0.7	2

#	ARTICLE	IF	CITATIONS
181	Meta-GWAS and Meta-Analysis of Exome Array Studies Do Not Reveal Genetic Determinants of Serum Hepcidin. PLoS ONE, 2016, 11, e0166628.	2.5	2
182	CLL-Like MBL In the General Population Persist Over Time, without Clinical Progression, Though Carrying the Same Cytogenetic Abnormalities of CLL. Blood, 2010, 116, 2440-2440.	1.4	1
183	Direct Selection of cDNAs by Genomic Clones. , 2001, 175, 189-199.		0
184	DNA variants in the human RAB3A gene are not associated with autism. Genes, Brain and Behavior, 2004, 3, 123-124.	2.2	0
185	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.	0.4	0
186	The Immunoglobulin Gene Repertoire of Low-Count CLL-Like MBL Is Different from CLL: Diagnostic Considerations and Implications for Clinical Monitoring. Blood, 2008, 112, 779-779.	1.4	0
187	Iron Status Independently Associates With Bone Mineral Density At Population Level. Insights From The Val Borbera Study. Blood, 2013, 122, 4672-4672.	1.4	0