Daniela Toniolo

List of Publications by Citations

190 papers	18,484 citations	69 h-index	133 g-index
203 ext. papers	22,306 ext. citations	11.6 avg, IF	5.22 L-index

#	Paper	IF	Citations
190	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
189	Mutations in the gene encoding lamin A/C cause autosomal dominant Emery-Dreifuss muscular dystrophy. *Nature Genetics*, **1999**, 21, 285-8	36.3	1076
188	The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90	50.4	776
187	Identification of a novel X-linked gene responsible for Emery-Dreifuss muscular dystrophy. *Nature Genetics*, **1994**, 8, 323-7	36.3	762
186	A novel X-linked gene, G4.5. is responsible for Barth syndrome. *Nature Genetics*, **1996**, 12, 385-9	36.3	620
185	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. *Nature Genetics*, **2013**, 45, 145-54	36.3	505
184	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. *Nature Genetics*, **2018**, 50, 1412-1425	36.3	386
183	A general approach for haplotype phasing across the full spectrum of relatedness. *PLoS Genetics*, **2014**, 10, e1004234	6	377
182	Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies. *Nature Genetics*, **2010**, 42, 1077-85	36.3	372
181	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
180	New gene functions in megakaryopoiesis and platelet formation. *Nature*, **2011**, 480, 201-8	50.4	330
179	Different mutations in the LMNA gene cause autosomal dominant and autosomal recessive Emery-Dreifuss muscular dystrophy. *American Journal of Human Genetics*, **2000**, 66, 1407-12	11	330
178	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. *Nature Genetics*, **2017**, 49, 403-415	36.3	313
177	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. *Nature Communications*, **2016**, 7, 10023	17.4	295
176	Mutations in GDI1 are responsible for X-linked non-specific mental retardation. *Nature Genetics*, **1998**, 19, 134-9	36.3	277
175	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	36.3	257
174	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257

173	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. *Nature Genetics*, **2012**, 44, 260-8	36.3	243
172	Genetic structure of Europeans: a view from the North-East. *PLoS ONE*, **2009**, 4, e5472	3.7	237
171	Clinical significance of somatic mutation in unexplained blood cytopenia. *Blood*, **2017**, 129, 3371-3378	2.2	229
170	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
169	Trans-ancestry genome-wide association study identifies 12 genetic loci influencing blood pressure and implicates a role for DNA methylation. *Nature Genetics*, **2015**, 47, 1282-1293	36.3	223
168	A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics*, **2019**, 51, 957-972	36.3	217
167	Isolation of human glucose-6-phosphate dehydrogenase (G6PD) cDNA clones: primary structure of the protein and unusual 5Snon-coding region. *Nucleic Acids Research*, **1986**, 14, 2511-22	20.1	213
166	The X-linked gene G4.5 is responsible for different infantile dilated cardiomyopathies. *American Journal of Human Genetics*, **1997**, 61, 862-7	11	212
165	A human homologue of the Drosophila melanogaster diaphanous gene is disrupted in a patient with premature ovarian failure: evidence for conserved function in oogenesis and implications for human sterility. *American Journal of Human Genetics*, **1998**, 62, 533-41	11	209
164	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. *Nature Genetics*, **2016**, 48, 1462-1472	36.3	198
163	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
162	Mutations in the small GTPase gene RAB39B are responsible for X-linked mental retardation associated with autism, epilepsy, and macrocephaly. *American Journal of Human Genetics*, **2010**, 86, 185-95	11	176
161	Isolation of sequences that span the fragile X and identification of a fragile X-related CpG island. *Science*, **1991**, 251, 1236-9	33.3	161
160	A family of transmembrane proteins with homology to the MET-hepatocyte growth factor receptor. *Proceedings of the National Academy of Sciences of the United States of America*, **1996**, 93, 674-8	11.5	153
159	Influence of intermediate and uninterrupted FMR1 CGG expansions in premature ovarian failure manifestation. *Human Reproduction*, **2006**, 21, 952-7	5.7	152
158	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. *Nature Communications*, **2015**, 6, 5897	17.4	147
157	A meta-analysis of thyroid-related traits reveals novel loci and gender-specific differences in the regulation of thyroid function. *PLoS Genetics*, **2013**, 9, e1003266	6	146
156	Age- and sex-related variations in platelet count in Italy: a proposal of reference ranges based on 40987 subjectsSdata. *PLoS ONE*, **2013**, 8, e54289	3.7	146

155	Genome-wide association and functional follow-up reveals new loci for kidney function. *PLoS Genetics*, **2012**, 8, e1002584	6	143
154	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. *Nature Genetics*, **2015**, 47, 1272-1281	36.3	129
153	Clinical relevance of atrial fibrillation/flutter, stroke, pacemaker implant, and heart failure in Emery-Dreifuss muscular dystrophy: a long-term longitudinal study. *Stroke*, **2003**, 34, 901-8	6.7	126
152	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics*, **2019**, 51, 1459-1474	36.3	122
151	Identification of novel genetic Loci associated with thyroid peroxidase antibodies and clinical thyroid disease. *PLoS Genetics*, **2014**, 10, e1004123	6	122
150	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. *Nature Communications*, **2014**, 5, 4926	17.4	121
149	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	50.4	119
148	Eleven X chromosome breakpoints associated with premature ovarian failure (POF) map to a 15-Mb YAC contig spanning Xq21. *Genomics*, **1997**, 40, 123-31	4.3	114
147	General population low-count CLL-like MBL persists over time without clinical progression, although carrying the same cytogenetic abnormalities of CLL. *Blood*, **2011**, 118, 6618-25	2.2	112
146	Heart-specific localization of emerin: new insights into Emery-Dreifuss muscular dystrophy. *Human Molecular Genetics*, **1997**, 6, 2257-64	5.6	111
145	The immunoglobulin gene repertoire of low-count chronic lymphocytic leukemia (CLL)-like monoclonal B lymphocytosis is different from CLL: diagnostic implications for clinical monitoring. *Blood*, **2009**, 114, 26-32	2.2	107
144	Alterations in the expression, structure and function of progesterone receptor membrane component-1 (PGRMC1) in premature ovarian failure. *Human Molecular Genetics*, **2008**, 17, 3776-83	5.6	102
143	Ultrastructural abnormality of sarcolemmal nuclei in Emery-Dreifuss muscular dystrophy (EDMD). *Journal of the Neurological Sciences*, **1998**, 159, 88-93	3.2	102
142	BMP15 mutations associated with primary ovarian insufficiency cause a defective production of bioactive protein. *Human Mutation*, **2009**, 30, 804-10	4.7	101
141	Multiple loci are associated with white blood cell phenotypes. *PLoS Genetics*, **2011**, 7, e1002113	6	92
140	Complete concordance between glucose-6-phosphate dehydrogenase activity and hypomethylation of 3SCpG clusters: implications for X chromosome dosage compensation. *Nucleic Acids Research*, **1984**, 12, 9333-48	20.1	88
139	Chromosomal rearrangements in Xq and premature ovarian failure: mapping of 25 new cases and review of the literature. *Human Reproduction*, **2006**, 21, 1477-83	5.7	87
138	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. *Hypertension*, **2017**,	8.5	85

137	Deletion of the mental retardation gene Gdi1 impairs associative memory and alters social behavior in mice. *Human Molecular Genetics*, **2002**, 11, 2567-80	5.6	85
136	A genome-wide association study of early menopause and the combined impact of identified variants. *Human Molecular Genetics*, **2013**, 22, 1465-72	5.6	82
135	Normal serum concentrations of anti-Müllerian hormone in women with regular menstrual cycles. *Reproductive BioMedicine Online*, **2010**, 21, 463-9	4	81
134	TMPRSS6 rs855791 modulates hepcidin transcription in vitro and serum hepcidin levels in normal individuals. *Blood*, **2011**, 118, 4459-62	2.2	80
133	Identification of new mutations in the Emery-Dreifuss muscular dystrophy gene and evidence for genetic heterogeneity of the disease. *Human Molecular Genetics*, **1995**, 4, 1859-63	5.6	79
132	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. *Proceedings of the National Academy of Sciences of the United States of America*, **1984**, 81, 7855-9	11.5	79
131	Cardiac features of Emery-Dreifuss muscular dystrophy caused by lamin A/C gene mutations. *European Heart Journal*, **2003**, 24, 2227-36	9.5	78
130	Mutation analysis of the lamin A/C gene (LMNA) among patients with different cardiomuscular phenotypes. *Journal of Medical Genetics*, **2003**, 40, e132	5.8	77
129	Genome-wide analyses identify a role for SLC17A4 and AADAT in thyroid hormone regulation. *Nature Communications*, **2018**, 9, 4455	17.4	75
128	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. *American Journal of Human Genetics*, **2017**, 100, 865-884	11	74
127	A "housekeeping" gene on the X chromosome encodes a protein similar to ubiquitin. *Proceedings of the National Academy of Sciences of the United States of America*, **1988**, 85, 851-5	11.5	72
126	Common variants in UMOD associate with urinary uromodulin levels: a meta-analysis. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 1869-82	12.7	71
125	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Scientific Reports*, **2017**, 7, 45040	4.9	70
124	X-linked premature ovarian failure: a complex disease. *Current Opinion in Genetics and Development*, **2006**, 16, 293-300	4.9	70
123	Association of HFE and TMPRSS6 genetic variants with iron and erythrocyte parameters is only in part dependent on serum hepcidin concentrations. *Journal of Medical Genetics*, **2011**, 48, 629-34	5.8	69
122	Physical map of human Xq27-qter: localizing the region of the fragile X mutation. *Proceedings of the National Academy of Sciences of the United States of America*, **1991**, 88, 8302-6	11.5	69
121	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. *Neuromuscular Disorders*, **2003**, 13, 508-15	2.9	68
120	Mapping of two genes encoding isoforms of the actin binding protein ABP-280, a dystrophin like protein, to Xq28 and to chromosome 7. *Human Molecular Genetics*, **1993**, 2, 761-6	5.6	66

119	Mutation analysis of two candidate genes for premature ovarian failure, DACH2 and POF1B. *Human Reproduction*, **2004**, 19, 2759-66	5.7	65
118	X chromosome inactivation in carriers of Barth syndrome. *American Journal of Human Genetics*, **1998**, 63, 1457-63	11	64
117	Evidence of inbreeding depression on human height. *PLoS Genetics*, **2012**, 8, e1002655	6	62
116	A 12Mb deletion at 7q33-q35 associated with autism spectrum disorders and primary amenorrhea. *European Journal of Medical Genetics*, **2008**, 51, 631-8	2.6	61
115	Tissue-specific levels of human glucose-6-phosphate dehydrogenase correlate with methylation of specific sites at the 3Send of the gene. *Proceedings of the National Academy of Sciences of the United States of America*, **1985**, 82, 1465-9	11.5	61
114	Transcriptional organization of a 450-kb region of the human X chromosome in Xq28. *Proceedings of the National Academy of Sciences of the United States of America*, **1993**, 90, 10977-81	11.5	60
113	Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability. *Nature Genetics*, **2018**, 50, 652-656	36.3	59
112	cDNA sequences of human glucose 6-phosphate dehydrogenase cloned in pBR322. *Nature*, **1981**, 294, 778-80	50.4	57
111	Whole-genome sequence-based analysis of thyroid function. *Nature Communications*, **2015**, 6, 5681	17.4	56
110	Integration of genome-wide association studies with biological knowledge identifies six novel genes related to kidney function. *Human Molecular Genetics*, **2012**, 21, 5329-43	5.6	54
109	Increased serum hepcidin levels in subjects with the metabolic syndrome: a population study. *PLoS ONE*, **2012**, 7, e48250	3.7	53
108	MCM8 and MCM9 Nucleotide Variants in Women With Primary Ovarian Insufficiency. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 576-582	5.6	52
107	Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps. *Nature Genetics*, **2016**, 48, 1303-1312	36.3	51
106	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. *European Journal of Human Genetics*, **2013**, 21, 659-65	5.3	50
105	Association of adiposity genetic variants with menarche timing in 92,105 women of European descent. *American Journal of Epidemiology*, **2013**, 178, 451-60	3.8	48
104	Non-specific X-linked semidominant mental retardation by mutations in a Rab GDP-dissociation inhibitor. *Human Molecular Genetics*, **1998**, 7, 1311-5	5.6	48
103	CpG islands of the X chromosome are gene associated. *Nucleic Acids Research*, **1988**, 16, 9527-43	20.1	48
102	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. *European Journal of Neuroscience*, **2004**, 19, 1895-905	3.5	47

101	An X chromosome-linked gene encoding a protein with characteristics of a rhoGAP predominantly expressed in hematopoietic cells. *Proceedings of the National Academy of Sciences of the United States of America*, **1996**, 93, 695-9	11.5	47
100	Molecular cloning and analysis of the fragile X region in man. *Nucleic Acids Research*, **1991**, 19, 2567-72	20.1	47
99	X chromosome and ovarian failure. *Seminars in Reproductive Medicine*, **2007**, 25, 264-71	1.4	45
98	Six Novel Loci Associated with Circulating VEGF Levels Identified by a Meta-analysis of Genome-Wide Association Studies. *PLoS Genetics*, **2016**, 12, e1005874	6	43
97	Neurite extension occurs in the absence of regulated exocytosis in PC12 subclones. *Molecular Biology of the Cell*, **1999**, 10, 2919-31	3.5	42
96	Methylation and sequence analysis around EagI sites: identification of 28 new CpG islands in XQ24-XQ28. *Nucleic Acids Research*, **1992**, 20, 727-33	20.1	42
95	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
94	Single-cell expression profiling of dopaminergic neurons combined with association analysis identifies pyridoxal kinase as Parkinson's disease gene. *Annals of Neurology*, **2009**, 66, 792-8	9.4	40
93	Sequence variation at the human FOXO3 locus: a study of premature ovarian failure and primary amenorrhea. *Human Reproduction*, **2008**, 23, 216-21	5.7	40
92	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. *Human Molecular Genetics*, **2009**, 18, 105-17	5.6	39
91	Inherited genetic susceptibility to monoclonal B-cell lymphocytosis. *Blood*, **2010**, 116, 5957-60	2.2	39
90	The CpG island in the 5Sregion of the G6PD gene of man and mouse. *Gene*, **1991**, 102, 197-203	3.8	39
89	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. *Nature Communications*, **2017**, 8, 15927	17.4	37
88	Immunocytochemical detection of emerin within the nuclear matrix. *Neuromuscular Disorders*, **1998**, 8, 338-44	2.9	37
87	Frequent low penetrance mutations in the Lamin A/C gene, causing Emery Dreifuss muscular dystrophy. *Neuromuscular Disorders*, **2002**, 12, 958-63	2.9	37
86	Duplications of the X chromosome in males: evidence that most parts of the X chromosome can be active in two copies. *Human Genetics*, **1991**, 86, 519-21	6.3	33
85	Association analysis of bitter receptor genes in five isolated populations identifies a significant correlation between TAS2R43 variants and coffee liking. *PLoS ONE*, **2014**, 9, e92065	3.7	32
84	Heritability and demographic analyses in the large isolated population of Val Borbera suggest advantages in mapping complex traits genes. *PLoS ONE*, **2009**, 4, e7554	3.7	32

83	A susceptibility gene for premature ovarian failure (POF) maps to proximal Xq28. *European Journal of Human Genetics*, **2004**, 12, 829-34	5.3	32
82	Modulation of genetic associations with serum urate levels by body-mass-index in humans. *PLoS ONE*, **2015**, 10, e0119752	3.7	31
81	and Loci Identified through Large-Scale Exome Chip Analysis Regulate Kidney Development and Function. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 981-994	12.7	30
80	A mutation in the X-linked Emery-Dreifuss muscular dystrophy gene in a patient affected with conduction cardiomyopathy. *Neuromuscular Disorders*, **2001**, 11, 411-3	2.9	30
79	Epigenetic control of the critical region for premature ovarian failure on autosomal genes translocated to the X chromosome: a hypothesis. *Human Genetics*, **2007**, 121, 441-50	6.3	29
78	Emerin expression at the early stages of myogenic differentiation. *Differentiation*, **2000**, 66, 208-17	3.5	29
77	Influence of age, sex and ethnicity on platelet count in five Italian geographic isolates: mild thrombocytopenia may be physiological. *British Journal of Haematology*, **2012**, 157, 384-7	4.5	28
76	Genetic insights into biological mechanisms governing human ovarian ageing. *Nature*, **2021**, 596, 393-397	50.4	28
75	Common variants in Mendelian kidney disease genes and their association with renal function. *Journal of the American Society of Nephrology: JASN*, **2013**, 24, 2105-17	12.7	27
74	Serum levels of the hepcidin-20 isoform in a large general population: the Val Borbera study. *Journal of Proteomics*, **2012**, 76 Spec No., 28-35	3.9	27
73	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. *Journal of Medical Genetics*, **2009**, 46, 585-92	5.8	27
72	A large-scale association study to assess the impact of known variants of the human INHA gene on premature ovarian failure. *Human Reproduction*, **2009**, 24, 2023-8	5.7	26
71	The exon-intron organization of the human X-linked gene (FLN1) encoding actin-binding protein 280. *Genomics*, **1994**, 21, 71-6	4.3	25
70	An archipelago of CpG islands in Xq28: identification and fine mapping of 20 new CpG islands of the human X chromosome. *Human Molecular Genetics*, **1992**, 1, 275-80	5.6	25
69	Genome-wide meta-analysis of common variant differences between men and women. *Human Molecular Genetics*, **2012**, 21, 4805-15	5.6	24
68	High-resolution array-CGH analysis on 46,XX patients affected by early onset primary ovarian insufficiency discloses new genes involved in ovarian function. *Human Reproduction*, **2019**, 34, 574-583	5.7	23
67	Rare coding variants and X-linked loci associated with age at menarche. *Nature Communications*, **2015**, 6, 7756	17.4	23
66	Variation of hemoglobin levels in normal Italian populations from genetic isolates. *Haematologica*, **2008**, 93, 1372-5	6.6	23

65	Probes for CpG islands on the distal long arm of the human X chromosome are clustered in Xq24 and Xq28. *Genomics*, **1990**, 8, 664-70	4.3	23
64	Complementation of a defect in the production of ribosomal RNA in somatic cell hybrids. *Nature*, **1974**, 248, 411-3	50.4	23
63	Identification and characterization of a new gene in the human Xq28 region. *Human Molecular Genetics*, **1992**, 1, 269-73	5.6	22
62	Common variants in CLDN14 are associated with differential excretion of magnesium over calcium in urine. *Pflugers Archiv European Journal of Physiology*, **2017**, 469, 91-103	4.6	21
61	Multicohort analysis of the maternal age effect on recombination. *Nature Communications*, **2015**, 6, 7846	17.4	21
60	X-linked non-specific mental retardation. *Current Opinion in Genetics and Development*, **2000**, 10, 280-5	4.9	21
59	Small effective population size and genetic homogeneity in the Val Borbera isolate. *European Journal of Human Genetics*, **2013**, 21, 89-94	5.3	20
58	Regulation of glucose 6-phosphate dehydrogenase expression in CHO-human fibroblast somatic cell hybrids. *Somatic Cell Genetics*, **1983**, 9, 429-43		20
57	Genome-Wide Meta-Analysis Unravels Interactions between Magnesium Homeostasis and Metabolic Phenotypes. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 335-348	12.7	19
56	SV40-transformed cells with temperature-dependent serum requirements. *Cell*, **1975**, 4, 255-62	56.2	19
55	X chromosome genes and premature ovarian failure. *Seminars in Reproductive Medicine*, **2000**, 18, 51-7	1.4	18
54	Linkage and sequence conservation of the X-linked genes DXS253E (P3) and DXS254E (GdX) in mouse and man. *Genomics*, **1990**, 7, 453-7	4.3	18
53	COL6A5 variants in familial neuropathic chronic itch. *Brain*, **2017**, 140, 555-567	11.2	17
52	The A736V TMPRSS6 polymorphism influences hepcidin and iron metabolism in chronic hemodialysis patients: TMPRSS6 and hepcidin in hemodialysis. *BMC Nephrology*, **2013**, 14, 48	2.7	17
51	Barth syndrome presenting with acute metabolic decompensation in the neonatal period. *Journal of Inherited Metabolic Disease*, **2006**, 29, 684	5.4	17
50	Comparative mapping of the actin-binding protein 280 genes in human and mouse. *Genomics*, **1994**, 21, 428-30	4.3	17
49	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. *Cytogenetic and Genome Research*, **1985**, 39, 87-92	1.9	17
48	A Genome-Wide Association Study in isolated populations reveals new genes associated to common food likings. *Reviews in Endocrine and Metabolic Disorders*, **2016**, 17, 209-19	10.5	17

47	Fertility Preservation in Endometriosis Patients: Anti-Müllerian Hormone Is a Reliable Marker of the Ovarian Follicle Density. *Frontiers in Surgery*, **2017**, 4, 40	2.3	16
46	In search of the MRX genes. *American Journal of Medical Genetics Part A*, **2000**, 97, 221-7		16
45	A birds-eye view of Italian genomic variation through whole-genome sequencing. *European Journal of Human Genetics*, **2020**, 28, 435-444	5.3	16
44	The POF1B candidate gene for premature ovarian failure regulates epithelial polarity. *Journal of Cell Science*, **2011**, 124, 3356-68	5.3	15
43	The use of recombinant human growth hormone for radioiodination and standard preparation in radioimmunoassay. *Journal of Immunological Methods*, **1993**, 159, 269-74	2.5	15
42	G6PD Ferrara I has the same two mutations as G6PD A(-) but a distinct biochemical phenotype. *Human Genetics*, **1994**, 93, 139-42	6.3	15
41	A reference panel of 64,976 haplotypes for genotype imputation		15
40	Identification of TRIM22 single nucleotide polymorphisms associated with loss of inhibition of HIV-1 transcription and advanced HIV-1 disease. *Aids*, **2013**, 27, 2335-44	3.5	14
39	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. *Genomics*, **1991**, 9, 551-4	4.3	14
38	Association of a variant in the CHRNA5-A3-B4 gene cluster region to heavy smoking in the Italian population. *European Journal of Human Genetics*, **2011**, 19, 593-6	5.3	13
37	Temporal gene expression profile of the hippocampus following trace fear conditioning. *Brain Research*, **2010**, 1308, 14-23	3.7	13
36	FISH characterization of the Xq21 breakpoint in a translocation carrier with premature ovarian failure. *Clinical Genetics*, **1996**, 50, 267-9	4	13
35	Skewed X-chromosome inactivation is not associated with premature ovarian failure in a large cohort of Italian patients. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1349-51	2.5	13
34	Selection and mapping of replication origins from a 500-kb region of the human X chromosome and their relationship to gene expression. *Genomics*, **1999**, 62, 11-20	4.3	12
33	Forebrain deletion of CDI in adult mice worsens the pre-synaptic deficit at cortico-lateral amygdala synaptic connections. *PLoS ONE*, **2012**, 7, e29763	3.7	12
32	Emerin presence in platelets. *Acta Neuropathologica*, **2000**, 100, 291-8	14.3	11
31	Sequence and gene content in 52 kb including and centromeric to the G6PD gene in Xq28. *DNA Sequence*, **1995**, 6, 1-11		11
30	Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. *Science Advances*, **2021**, 7,	14.3	11

29	Big Data in Medicine, the Present and Hopefully the Future. *Frontiers in Medicine*, **2019**, 6, 263	4.9	11
28	Genome-wide association analysis on five isolated populations identifies variants of the HLA-DOA gene associated with white wine liking. *European Journal of Human Genetics*, **2015**, 23, 1717-22	5.3	10
27	X-linked severe mental retardation and a progressive neurological disorder in a Belgian family: clinical and genetic studies. *Clinical Genetics*, **1997**, 52, 155-61	4	10
26	Spatial and temporal expression of POF1B, a gene expressed in epithelia. *Gene Expression Patterns*, **2007**, 7, 529-34	1.5	10
25	Long-term treatment of Barth syndrome with pantothenic acid: a retrospective study. *Molecular Genetics and Metabolism*, **2003**, 80, 408-11	3.7	10
24	Unusual expression of emerin in a patient with X-linked Emery-Dreifuss muscular dystrophy. *Neuromuscular Disorders*, **2000**, 10, 567-71	2.9	10
23	Reconstructing the genetic history of Italians: new insights from a male (Y-chromosome) perspective. *Annals of Human Biology*, **2018**, 45, 44-56	1.7	9
22	Stability of DNA methylation of X-chromosome genes during aging. *Somatic Cell and Molecular Genetics*, **1990**, 16, 79-84		9
21	Genome wide association analysis of a founder population identified TAF3 as a gene for MCHC in humans. *PLoS ONE*, **2013**, 8, e69206	3.7	9
20	A comparative transcriptional map of a region of 250 kb on the human and mouse X chromosome between the G6PD and the FLN1 genes. *Genomics*, **1995**, 28, 377-82	4.3	7
19	Highly conserved non-coding sequences and the 18q critical region for short stature: a common mechanism of disease?. *PLoS ONE*, **2008**, 3, e1460	3.7	7
18	Factors associated with food liking and their relationship with metabolic traits in Italian cohorts. *Food Quality and Preference*, **2019**, 75, 64-70	5.8	7
17	The screening for X-linked Emery-Dreifuss muscular dystrophy amongst young patients with idiopathic heart conduction system disease treated by a pacemaker implant. *European Journal of Neurology*, **2004**, 11, 531-4	6	6
16	Computer-based genealogy reconstruction in founder populations. *Journal of Biomedical Informatics*, **2011**, 44, 997-1003	10.2	5
15	Biochemical and molecular characterization of a new sporadic glucose-6-phosphate dehydrogenase variant described in Italy: G6PD Modena. *British Journal of Haematology*, **1994**, 87, 209-11	4.5	5
14	Relationship between clone metrics and clinical outcome in clonal cytopenia. *Blood*, **2021**, 138, 965-976	2.2	5
13	Selection and fine mapping of chromosome-specific cDNAs: application to human chromosome 1. *Genomics*, **1996**, 38, 149-54	4.3	4
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