Daniela Toniolo

List of Publications by Year in Descending Order

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	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. *Communications Biology*, **2022**, 5,	6.7	1
189	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
188	Relationship between clone metrics and clinical outcome in clonal cytopenia. *Blood*, **2021**, 138, 965-976	2.2	5
187	Genetic insights into biological mechanisms governing human ovarian ageing. *Nature*, **2021**, 596, 393-397	50.4	28
186	A birds-eye view of Italian genomic variation through whole-genome sequencing. *European Journal of Human Genetics*, **2020**, 28, 435-444	5.3	16
185	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics*, **2019**, 51, 1459-1474	36.3	122
184	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
183	A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics*, **2019**, 51, 957-972	36.3	217
182	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
181	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
180	Big Data in Medicine, the Present and Hopefully the Future. *Frontiers in Medicine*, **2019**, 6, 263	4.9	11
179	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
178	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
177	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
176	Genome-wide analyses identify a role for SLC17A4 and AADAT in thyroid hormone regulation. *Nature Communications*, **2018**, 9, 4455	17.4	75
175	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
174	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

173	COL6A5 variants in familial neuropathic chronic itch. *Brain*, **2017**, 140, 555-567	11.2	17
172	Clinical significance of somatic mutation in unexplained blood cytopenia. *Blood*, **2017**, 129, 3371-3378	2.2	229
171	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
170	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Scientific Reports*, **2017**, 7, 45040	4.9	70
169	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
168	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. *American Journal of Human Genetics*, **2017**, 100, 865-884	11	74
167	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
166	MCM8 and MCM9 Nucleotide Variants in Women With Primary Ovarian Insufficiency. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 576-582	5.6	52
165	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
164	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. *Nature Communications*, **2017**, 8, 15927	17.4	37
163	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
162	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
161	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. *Nature Genetics*, **2016**, 48, 1462-1472	36.3	198
160	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. *Nature Communications*, **2016**, 7, 10023	17.4	295
159	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
158	Meta-GWAS and Meta-Analysis of Exome Array Studies Do Not Reveal Genetic Determinants of Serum Hepcidin. *PLoS ONE*, **2016**, 11, e0166628	3.7	1
157	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
156	Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps. *Nature Genetics*, **2016**, 48, 1303-1312	36.3	51

155	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	50.4	119
154	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
153	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
152	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
151	Multicohort analysis of the maternal age effect on recombination. *Nature Communications*, **2015**, 6, 7846	17.4	21
150	The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90	50.4	776
149	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
148	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	2.4	
147	Whole-genome sequence-based analysis of thyroid function. *Nature Communications*, **2015**, 6, 5681	17.4	56
146	Rare coding variants and X-linked loci associated with age at menarche. *Nature Communications*, **2015**, 6, 7756	17.4	23
145	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. *Nature Communications*, **2015**, 6, 5897	17.4	147
144	Modulation of genetic associations with serum urate levels by body-mass-index in humans. *PLoS ONE*, **2015**, 10, e0119752	3.7	31
143	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. *Nature Communications*, **2014**, 5, 4926	17.4	121
142	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
141	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
140	Identification of novel genetic Loci associated with thyroid peroxidase antibodies and clinical thyroid disease. *PLoS Genetics*, **2014**, 10, e1004123	6	122
139	A general approach for haplotype phasing across the full spectrum of relatedness. *PLoS Genetics*, **2014**, 10, e1004234	6	377
138	Small effective population size and genetic homogeneity in the Val Borbera isolate. *European Journal of Human Genetics*, **2013**, 21, 89-94	5.3	20

137	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
136	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
135	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. *Nature Genetics*, **2013**, 45, 145-54	36.3	505
134	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
133	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
132	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
131	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
130	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
129	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
128	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
127	Iron Status Independently Associates With Bone Mineral Density At Population Level. Insights From The Val Borbera Study. *Blood*, **2013**, 122, 4672-4672	2.2	
126	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
125	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
124	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257
123	Genome-wide meta-analysis of common variant differences between men and women. *Human Molecular Genetics*, **2012**, 21, 4805-15	5.6	24
122	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
121	Increased serum hepcidin levels in subjects with the metabolic syndrome: a population study. *PLoS ONE*, **2012**, 7, e48250	3.7	53
120	Evidence of inbreeding depression on human height. *PLoS Genetics*, **2012**, 8, e1002655	6	62

119	Genome-wide association and functional follow-up reveals new loci for kidney function. *PLoS Genetics*, **2012**, 8, e1002584	6	143
118	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
117	Forebrain deletion of GDI in adult mice worsens the pre-synaptic deficit at cortico-lateral amygdala synaptic connections. *PLoS ONE*, **2012**, 7, e29763	3.7	12
116	New gene functions in megakaryopoiesis and platelet formation. *Nature*, **2011**, 480, 201-8	50.4	330
115	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
114	TMPRSS6 rs855791 modulates hepcidin transcription in vitro and serum hepcidin levels in normal individuals. *Blood*, **2011**, 118, 4459-62	2.2	80
113	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
112	Computer-based genealogy reconstruction in founder populations. *Journal of Biomedical Informatics*, **2011**, 44, 997-1003	10.2	5
111	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
110	The POF1B candidate gene for premature ovarian failure regulates epithelial polarity. *Journal of Cell Science*, **2011**, 124, 3356-68	5.3	15
109	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
108	Multiple loci are associated with white blood cell phenotypes. *PLoS Genetics*, **2011**, 7, e1002113	6	92
107	Serum Hepcidin Levels Correlate with Phenotypes of the Metabolic Syndrome At Population Level. *Blood*, **2011**, 118, 348-348	2.2	1
106	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
105	Normal serum concentrations of anti-Müllerian hormone in women with regular menstrual cycles. *Reproductive BioMedicine Online*, **2010**, 21, 463-9	4	81
104	Inherited genetic susceptibility to monoclonal B-cell lymphocytosis. *Blood*, **2010**, 116, 5957-60	2.2	39
103	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
102	Temporal gene expression profile of the hippocampus following trace fear conditioning. *Brain Research*, **2010**, 1308, 14-23	3.7	13

101	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	2.2	1
100	Genetic structure of Europeans: a view from the North-East. *PLoS ONE*, **2009**, 4, e5472	3.7	237
99	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
98	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
97	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
96	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
95	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
94	BMP15 mutations associated with primary ovarian insufficiency cause a defective production of bioactive protein. *Human Mutation*, **2009**, 30, 804-10	4.7	101
93	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
92	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
91	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
90	Variation of hemoglobin levels in normal Italian populations from genetic isolates. *Haematologica*, **2008**, 93, 1372-5	6.6	23
89	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
88	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
87	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	2.2	
86	Spatial and temporal expression of POF1B, a gene expressed in epithelia. *Gene Expression Patterns*, **2007**, 7, 529-34	1.5	10
85	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
84	X chromosome and ovarian failure. *Seminars in Reproductive Medicine*, **2007**, 25, 264-71	1.4	45

83	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
82	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
81	Influence of intermediate and uninterrupted FMR1 CGG expansions in premature ovarian failure manifestation. *Human Reproduction*, **2006**, 21, 952-7	5.7	152
80	X-linked premature ovarian failure: a complex disease. *Current Opinion in Genetics and Development*, **2006**, 16, 293-300	4.9	70
79	Barth syndrome presenting with acute metabolic decompensation in the neonatal period. *Journal of Inherited Metabolic Disease*, **2006**, 29, 684	5.4	17
78	Mutation analysis of two candidate genes for premature ovarian failure, DACH2 and POF1B. *Human Reproduction*, **2004**, 19, 2759-66	5.7	65
77	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
76	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
75	DNA variants in the human RAB3A gene are not associated with autism. *Genes, Brain and Behavior*, **2004**, 3, 123-4	3.6	
74	A susceptibility gene for premature ovarian failure (POF) maps to proximal Xq28. *European Journal of Human Genetics*, **2004**, 12, 829-34	5.3	32
73	Cardiac features of Emery-Dreifuss muscular dystrophy caused by lamin A/C gene mutations. *European Heart Journal*, **2003**, 24, 2227-36	9.5	78
72	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
71	Long-term treatment of Barth syndrome with pantothenic acid: a retrospective study. *Molecular Genetics and Metabolism*, **2003**, 80, 408-11	3.7	10
70	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
69	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
68	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
67	Frequent low penetrance mutations in the Lamin A/C gene, causing Emery Dreifuss muscular dystrophy. *Neuromuscular Disorders*, **2002**, 12, 958-63	2.9	37
66	Direct selection of cDNAs by genomic clones. *Methods in Molecular Biology*, **2001**, 175, 189-99	1.4	

65	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
64	In search of the MRX genes. *American Journal of Medical Genetics Part A*, **2000**, 97, 221-7		16
63	Emerin presence in platelets. *Acta Neuropathologica*, **2000**, 100, 291-8	14.3	11
62	X chromosome genes and premature ovarian failure. *Seminars in Reproductive Medicine*, **2000**, 18, 51-7	1.4	18
61	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
60	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
59	Emerin expression at the early stages of myogenic differentiation. *Differentiation*, **2000**, 66, 208-17	3.5	29
58	Unusual expression of emerin in a patient with X-linked Emery-Dreifuss muscular dystrophy. *Neuromuscular Disorders*, **2000**, 10, 567-71	2.9	10
57	X-linked non-specific mental retardation. *Current Opinion in Genetics and Development*, **2000**, 10, 280-5	4.9	21
56	Neurite extension occurs in the absence of regulated exocytosis in PC12 subclones. *Molecular Biology of the Cell*, **1999**, 10, 2919-31	3.5	42
55	Mutations in the gene encoding lamin A/C cause autosomal dominant Emery-Dreifuss muscular dystrophy. *Nature Genetics*, **1999**, 21, 285-8	36.3	1076
54	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
53	Mutations in GDI1 are responsible for X-linked non-specific mental retardation. *Nature Genetics*, **1998**, 19, 134-9	36.3	277
52	Immunocytochemical detection of emerin within the nuclear matrix. *Neuromuscular Disorders*, **1998**, 8, 338-44	2.9	37
51	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
50	X chromosome inactivation in carriers of Barth syndrome. *American Journal of Human Genetics*, **1998**, 63, 1457-63	11	64
49	Ultrastructural abnormality of sarcolemmal nuclei in Emery-Dreifuss muscular dystrophy (EDMD). *Journal of the Neurological Sciences*, **1998**, 159, 88-93	3.2	102
48	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

47	Heart-specific localization of emerin: new insights into Emery-Dreifuss muscular dystrophy. *Human Molecular Genetics*, **1997**, 6, 2257-64	5.6	111
46	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
45	The X-linked gene G4.5 is responsible for different infantile dilated cardiomyopathies. *American Journal of Human Genetics*, **1997**, 61, 862-7	11	212
44	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
43	FISH characterization of the Xq21 breakpoint in a translocation carrier with premature ovarian failure. *Clinical Genetics*, **1996**, 50, 267-9	4	13
42	Selection and fine mapping of chromosome-specific cDNAs: application to human chromosome 1. *Genomics*, **1996**, 38, 149-54	4.3	4
41	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
40	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
39	A novel X-linked gene, G4.5. is responsible for Barth syndrome. *Nature Genetics*, **1996**, 12, 385-9	36.3	620
38	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
37	Sequence and gene content in 52 kb including and centromeric to the G6PD gene in Xq28. *DNA Sequence*, **1995**, 6, 1-11		11
36	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
35	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
34	Identification of a novel X-linked gene responsible for Emery-Dreifuss muscular dystrophy. *Nature Genetics*, **1994**, 8, 323-7	36.3	762
33	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
32	The exon-intron organization of the human X-linked gene (FLN1) encoding actin-binding protein 280. *Genomics*, **1994**, 21, 71-6	4.3	25
31	Comparative mapping of the actin-binding protein 280 genes in human and mouse. *Genomics*, **1994**, 21, 428-30	4.3	17
30	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

29	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
28	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
27	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
26	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
25	Identification and characterization of a new gene in the human Xq28 region. *Human Molecular Genetics*, **1992**, 1, 269-73	5.6	22
24	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
23	Is DNA methylation of X chromosome genes stable during aging?. *Somatic Cell and Molecular Genetics*, **1991**, 17, 101-3		2
22	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
21	Molecular cloning and analysis of the fragile X region in man. *Nucleic Acids Research*, **1991**, 19, 2567-72	20.1	47
20	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
19	The CpG island in the 5Sregion of the G6PD gene of man and mouse. *Gene*, **1991**, 102, 197-203	3.8	39
18	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
17	Stability of DNA methylation of X-chromosome genes during aging. *Somatic Cell and Molecular Genetics*, **1990**, 16, 79-84		9
16	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
15	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
14	CpG islands of the X chromosome are gene associated. *Nucleic Acids Research*, **1988**, 16, 9527-43	20.1	48
13	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
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3	A reference panel of 64,976 haplotypes for genotype imputation		15
2	Novel blood pressure locus and gene discovery using GWAS and expression datasets from blood and the kidney		1
1	Genetic analysis of over one million people identifies 535 novel loci for blood pressure		4