Daniela Toniolo

List of Publications by Year
in descending order

187	24,934	10389 72	8396 147
papers	citations	h-index	g-index

203	203	203	34275
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. Communications Biology, 2022, 5, .	4.4	17
2	Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. Science Advances, 2021, 7, .	10.3	36
3	Relationship between clone metrics and clinical outcome in clonal cytopenia. Blood, 2021, 138, 965-976.	1.4	58
4	Genetic insights into biological mechanisms governing human ovarian ageing. Nature, 2021, 596, 393-397.	27.8	183
5	A birdâ€™s-eye view of Italian genomic variation through whole-genome sequencing. European Journal of Human Genetics, 2020, 28, 435-444.	2.8	29
6	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	12.8	84
7	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. Nature Genetics, 2019, 51, 1459-1474.	21.4	251
8	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.	0.9	32
9	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	21.4	549
10	Factors associated with food liking and their relationship with metabolic traits in Italian cohorts. Food Quality and Preference, 2019, 75, 64-70.	4.6	9
11	Big Data in Medicine, the Present and Hopefully the Future. Frontiers in Medicine, 2019, 6, 263.	2.6	22
12	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.	21.4	86
13	Reconstructing the genetic history of Italians: new insights from a male (Y-chromosome) perspective. Annals of Human Biology, 2018, 45, 44-56.	1.0	19
14	Genome-Wide Meta-Analysis Unravels Interactions between Magnesium Homeostasis and Metabolic Phenotypes. Journal of the American Society of Nephrology: JASN, 2018, 29, 335-348.	6.1	34
15	Genome-wide analyses identify a role for SLC17A4 and AADAT in thyroid hormone regulation. Nature Communications, 2018, 9, 4455.	12.8	181
16	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. Nature Genetics, 2018, 50, 1412-1425.	21.4	924
17	MCM8 and MCM9 Nucleotide Variants in Women with Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2017, 102, jc.2016-2565.	3.6	68
18	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. Nature Genetics, 2017, 49, 403-415.	21.4	492

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19	COL6A5variants in familial neuropathic chronic itch. Brain, 2017, 140, aww343.	7.6	25
20	Clinical significance of somatic mutation in unexplained blood cytopenia. Blood, 2017, 129, 3371-3378.	1.4	379
21	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.	21.4	426
22	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. Scientific Reports, 2017, 7, 45040.	3.3	98
23	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.	2.8	27
24	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. American Journal of Human Genetics, 2017, 100, 865-884.	6.2	131
25	SOS2 and ACP1 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.	6.1	39
26	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. Hypertension, 2017, 70, .	2.7	123
27	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. Nature Communications, 2017, 8, 15927.	12.8	64
28	Fertility Preservation in Endometriosis Patients: Anti-MÃ¼llerian Hormone Is a Reliable Marker of the Ovarian Follicle Density. Frontiers in Surgery, 2017, 4, 40.	1.4	26
29	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
30	A Genome-Wide Association Study in isolated populations reveals new genes associated to common food likings. Reviews in Endocrine and Metabolic Disorders, 2016, 17, 209-219.	5.7	22
31	Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps. Nature Genetics, 2016, 48, 1303-1312.	21.4	66
32	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	21.4	2,421
33	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. Nature Genetics, 2016, 48, 1462-1472.	21.4	284
34	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. Nature Communications, 2016, 7, 10023.	12.8	412
35	Six Novel Loci Associated with Circulating VEGF Levels Identified by a Meta-analysis of Genome-Wide Association Studies. PLoS Genetics, 2016, 12, e1005874.	3.5	56
36	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

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37	Whole-genome sequence-based analysis of thyroid function. Nature Communications, 2015, 6, 5681.	12.8	75
38	Rare coding variants and X-linked loci associated with age at menarche. Nature Communications, 2015, 6, 7756.	12.8	32
39	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. Nature Communications, 2015, 6, 5897.	12.8	173
40	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	27.8	173
41	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, 1717-1722.	2.8	12
42	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	21.4	357
43	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.	21.4	294
44	Multicohort analysis of the maternal age effect on recombination. Nature Communications, 2015, 6, 7846.	12.8	29
45	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
46	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. Nature Genetics, 2015, 47, 1272-1281.	21.4	193
47	Modulation of Genetic Associations with Serum Urate Levels by Body-Mass-Index in Humans. PLoS ONE, 2015, 10, e0119752.	2.5	64
48	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.	2.5	41
49	Identification of Novel Genetic Loci Associated with Thyroid Peroxidase Antibodies and Clinical Thyroid Disease. PLoS Genetics, 2014, 10, e1004123.	3.5	150
50	A General Approach for Haplotype Phasing across the Full Spectrum of Relatedness. PLoS Genetics, 2014, 10, e1004234.	3.5	553
51	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. Nature Communications, 2014, 5, 4926.	12.8	192
52	Common Variants in UMOD Associate with Urinary Uromodulin Levels. Journal of the American Society of Nephrology: JASN, 2014, 25, 1869-1882.	6.1	85
53	Small effective population size and genetic homogeneity in the Val Borbera isolate. European Journal of Human Genetics, 2013, 21, 89-94.	2.8	32
54	The A736V TMPRSS6 polymorphism influences hepcidin and iron metabolism in chronic hemodialysis patients: TMPRSS6 and hepcidin in hemodialysis. BMC Nephrology, 2013, 14, 48.	1.8	20

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55	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. European Journal of Human Genetics, 2013, 21, 659-665.	2.8	64
56	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. Nature Genetics, 2013, 45, 145-154.	21.4	675
57	Common Variants in Mendelian Kidney Disease Genes and Their Association with Renal Function. Journal of the American Society of Nephrology: JASN, 2013, 24, 2105-2117.	6.1	33
58	Identification of TRIM22 single nucleotide polymorphisms associated with loss of inhibition of HIV-1 transcription and advanced HIV-1 disease. Aids, 2013, 27, 2335-2344.	2.2	17
59	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.	3.5	194
60	A genome-wide association study of early menopause and the combined impact of identified variants. Human Molecular Genetics, 2013, 22, 1465-1472.	2.9	104
61	Association of Adiposity Genetic Variants With Menarche Timing in 92,105 Women of European Descent. American Journal of Epidemiology, 2013, 178, 451-460.	3.4	51
62	Age- And Sex-Related Variations in Platelet Count in Italy: A Proposal of Reference Ranges Based on 40987 Subjects' Data. PLoS ONE, 2013, 8, e54289.	2.5	190
63	Genome Wide Association Analysis of a Founder Population Identified TAF3 as a Gene for MCHC in Humans. PLoS ONE, 2013, 8, e69206.	2.5	9
64	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
65	Evidence of Inbreeding Depression on Human Height. PLoS Genetics, 2012, 8, e1002655.	3.5	79
66	Genome-Wide Association and Functional Follow-Up Reveals New Loci for Kidney Function. PLoS Genetics, 2012, 8, e1002584.	3.5	166
67	Integration of genome-wide association studies with biological knowledge identifies six novel genes related to kidney function. Human Molecular Genetics, 2012, 21, 5329-5343.	2.9	64
68	Serum levels of the hepcidin-20 isoform in a large general population: The Val Borbera study. Journal of Proteomics, 2012, 76, 28-35.	2.4	29
69	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	27.8	320
70	Genome-wide meta-analysis of common variant differences between men and women. Human Molecular Genetics, 2012, 21, 4805-4815.	2.9	33
71	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. Nature Genetics, 2012, 44, 260-268.	21.4	303
72	Increased Serum Hepcidin Levels in Subjects with the Metabolic Syndrome: A Population Study. PLoS ONE, 2012, 7, e48250.	2.5	68

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73	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, 384-387.	2.5	33
74	Forebrain Deletion of Î±GDI in Adult Mice Worsens the Pre-Synaptic Deficit at Cortico-Lateral Amygdala Synaptic Connections. PLoS ONE, 2012, 7, e29763.	2.5	18
75	New gene functions in megakaryopoiesis and platelet formation. Nature, 2011, 480, 201-208.	27.8	401
76	General population low-count CLL-like MBL persists over time without clinical progression, although carrying the same cytogenetic abnormalities of CLL. Blood, 2011, 118, 6618-6625.	1.4	131
77	TMPRSS6 rs855791 modulates hepcidin transcription in vitro and serum hepcidin levels in normal individuals. Blood, 2011, 118, 4459-4462.	1.4	97
78	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, 593-596.	2.8	13
79	Computer-based genealogy reconstruction in founder populations. Journal of Biomedical Informatics, 2011, 44, 997-1003.	4.3	9
80	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, 629-634.	3.2	84
81	ThePOF1Bcandidate gene for premature ovarian failure regulates epithelial polarity. Journal of Cell Science, 2011, 124, 3356-3368.	2.0	20
82	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	21.4	403
83	Multiple Loci Are Associated with White Blood Cell Phenotypes. PLoS Genetics, 2011, 7, e1002113.	3.5	106
84	Serum Hepcidin Levels Correlate with Phenotypes of the Metabolic Syndrome At Population Level. Blood, 2011, 118, 348-348.	1.4	10
85	Inherited genetic susceptibility to monoclonal B-cell lymphocytosis. Blood, 2010, 116, 5957-5960.	1.4	42
86	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, 185-195.	6.2	220
87	Temporal gene expression profile of the hippocampus following trace fear conditioning. Brain Research, 2010, 1308, 14-23.	2.2	13
88	Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies. Nature Genetics, 2010, 42, 1077-1085.	21.4	445
89	Normal serum concentrations of anti-MÃ¼llerian hormone in women with regular menstrual cycles. Reproductive BioMedicine Online, 2010, 21, 463-469.	2.4	91
90	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

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91	Genetic Structure of Europeans: A View from the Northâ€“East. PLoS ONE, 2009, 4, e5472.	2.5	279
92	Heritability and Demographic Analyses in the Large Isolated Population of Val Borbera Suggest Advantages in Mapping Complex Traits Genes. PLoS ONE, 2009, 4, e7554.	2.5	37
93	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, 2023-2028.	0.9	30
94	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, 105-117.	2.9	50
95	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, 585-592.	3.2	33
96	Singleâ€cell expression profiling of dopaminergic neurons combined with association analysis identifies pyridoxal kinase as Parkinson's disease gene. Annals of Neurology, 2009, 66, 792-798.	5.3	49
97	BMP15 mutations associated with primary ovarian insufficiency cause a defective production of bioactive protein. Human Mutation, 2009, 30, 804-810.	2.5	126
98	The immunoglobulin gene repertoire of low-count chronic lymphocytic leukemia (CLL)â€“like monoclonal B lymphocytosis is different from CLL: diagnostic implications for clinical monitoring. Blood, 2009, 114, 26-32.	1.4	122
99	A 12Mb deletion at 7q33â€“q35 associated with autism spectrum disorders and primary amenorrhea. European Journal of Medical Genetics, 2008, 51, 631-638.	1.3	68
100	Alterations in the expression, structure and function of progesterone receptor membrane component-1 (PGRMC1) in premature ovarian failure. Human Molecular Genetics, 2008, 17, 3776-3783.	2.9	114
101	Variation of hemoglobin levels in normal Italian populations from genetic isolates. Haematologica, 2008, 93, 1372-1375.	3.5	25
102	Highly Conserved Non-Coding Sequences and the 18q Critical Region for Short Stature: A Common Mechanism of Disease?. PLoS ONE, 2008, 3, e1460.	2.5	7
103	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
104	Sequence variation at the human FOXO3 locus: a study of premature ovarian failure and primary amenorrhea. Human Reproduction, 2007, 23, 216-221.	0.9	49
105	X Chromosome and Ovarian Failure. Seminars in Reproductive Medicine, 2007, 25, 264-271.	1.1	56
106	Spatial and temporal expression of POF1B, a gene expressed in epithelia. Gene Expression Patterns, 2007, 7, 529-534.	0.8	12
107	Epigenetic control of the critical region for premature ovarian failure on autosomal genes translocated to the X chromosome: a hypothesis. Human Genetics, 2007, 121, 441-450.	3.8	35
108	Influence of intermediate and uninterrupted FMR1 CGG expansions in premature ovarian failure manifestation. Human Reproduction, 2006, 21, 952-957.	0.9	162

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109	X-linked premature ovarian failure: a complex disease. Current Opinion in Genetics and Development, 2006, 16, 293-300.	3.3	78
110	Barth syndrome presenting with acute metabolic decompensation in the neonatal period. Journal of Inherited Metabolic Disease, 2006, 29, 684-684.	3.6	22
111	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, 140A, 1349-1351.	1.2	16
112	Chromosomal rearrangements in Xq and premature ovarian failure: mapping of 25 new cases and review of the literature. Human Reproduction, 2006, 21, 1477-1483.	0.9	105
113	Mutation analysis of two candidate genes for premature ovarian failure, DACH2 and POF1B. Human Reproduction, 2004, 19, 2759-2766.	0.9	82
114	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, 1895-1905.	2.6	50
115	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, 531-534.	3.3	9
116	DNA variants in the human RAB3A gene are not associated with autism. Genes, Brain and Behavior, 2004, 3, 123-124.	2.2	0
117	A susceptibility gene for premature ovarian failure (POF) maps to proximal Xq28. European Journal of Human Genetics, 2004, 12, 829-834.	2.8	44
118	Long-term treatment of Barth syndrome with pantothenic acid: a retrospective study. Molecular Genetics and Metabolism, 2003, 80, 408-411.	1.1	10
119	108th ENMC International Workshop, 3rd Workshop of the MYO-CLUSTER project: EUROMEN, 7th International Emery-Dreifuss Muscular Dystrophy (EDMD) Workshop, 13â€“15 September 2002, Naarden, The Netherlands. Neuromuscular Disorders, 2003, 13, 508-515.	0.6	78
120	Mutation analysis of the lamin A/C gene (LMNA) among patients with different cardiomuscular phenotypes. Journal of Medical Genetics, 2003, 40, 132e-132.	3.2	98
121	Cardiac features of Emeryâ€“Dreifuss muscular dystrophy caused by lamin A/C gene mutations. European Heart Journal, 2003, 24, 2227-2236.	2.2	103
122	Clinical Relevance of Atrial Fibrillation/Flutter, Stroke, Pacemaker Implant, and Heart Failure in Emery-Dreifuss Muscular Dystrophy. Stroke, 2003, 34, 901-908.	2.0	158
123	Deletion of the mental retardation gene Gdi1 impairs associative memory and alters social behavior in mice. Human Molecular Genetics, 2002, 11, 2567-2580.	2.9	100
124	Frequent low penetrance mutations in the Lamin A/C gene, causing Emery Dreifuss muscular dystrophy. Neuromuscular Disorders, 2002, 12, 958-963.	0.6	52
125	A mutation in the X-linked Emeryâ€“Dreifuss muscular dystrophy gene in a patient affected with conduction cardiomyopathy. Neuromuscular Disorders, 2001, 11, 411-413.	0.6	37
126	Direct Selection of cDNAs by Genomic Clones. , 2001, 175, 189-199.		0

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127	In search of the MRX genes. American Journal of Medical Genetics Part A, 2000, 97, 221-227.	2.4	17
128	Emerin presence in platelets. Acta Neuropathologica, 2000, 100, 291-298.	7.7	12
129	X Chromosome Genes and Premature Ovarian Failure. Seminars in Reproductive Medicine, 2000, 18, 051-058.	1.1	23
130	Different Mutations in the LMNA Gene Cause Autosomal Dominant and Autosomal Recessive Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2000, 66, 1407-1412.	6.2	384
131	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, 982-985.	6.2	213
132	Emerin expression at the early stages of myogenic differentiation. Differentiation, 2000, 66, 208-217.	1.9	30
133	Unusual expression of emerin in a patient with X-linked Emeryâ€“Dreifuss muscular dystrophy. Neuromuscular Disorders, 2000, 10, 567-571.	0.6	13
134	X-linked non-specific mental retardation. Current Opinion in Genetics and Development, 2000, 10, 280-285.	3.3	25
135	Neurite Extension Occurs in the Absence of Regulated Exocytosis in PC12 Subclones. Molecular Biology of the Cell, 1999, 10, 2919-2931.	2.1	43
136	Mutations in the gene encoding lamin A/C cause autosomal dominant Emery-Dreifuss muscular dystrophy. Nature Genetics, 1999, 21, 285-288.	21.4	1,245
137	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.	2.9	12
138	Mutations in GDI1 are responsible for X-linked non-specific mental retardation. Nature Genetics, 1998, 19, 134-139.	21.4	304
139	Immunocytochemical detection of emerin within the nuclear matrix. Neuromuscular Disorders, 1998, 8, 338-344.	0.6	44
140	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, 533-541.	6.2	248
141	X Chromosome Inactivation in Carriers of Barth Syndrome. American Journal of Human Genetics, 1998, 63, 1457-1463.	6.2	71
142	Ultrastructural abnormality of sarcolemmal nuclei in Emery-Dreifuss muscular dystrophy (EDMD). Journal of the Neurological Sciences, 1998, 159, 88-93.	0.6	115
143	Non-specific X-linked semidominant mental retardation by mutations in a Rab GDP-dissociation inhibitor. Human Molecular Genetics, 1998, 7, 1311-1315.	2.9	65
144	Heart-specific localization of emerin: new insights into Emery-Dreifuss muscular dystrophy. Human Molecular Genetics, 1997, 6, 2257-2264.	2.9	138

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145	Eleven X Chromosome Breakpoints Associated with Premature Ovarian Failure (POF) Map to a 15-Mb YAC Contig Spanning Xq21. Genomics, 1997, 40, 123-131.	2.9	133
146	The X-Linked Gene G4.5 Is Responsible for Different Infantile Dilated Cardiomyopathies. American Journal of Human Genetics, 1997, 61, 862-867.	6.2	236
147	Xâ€linked severe mental retardation and a progressive neurological disorder in a Belgian family: clinical and genetic studies. Clinical Genetics, 1997, 52, 155-161.	2.0	11
148	Selection and Fine Mapping of Chromosome-Specific cDNAs: Application to Human Chromosome 1. Genomics, 1996, 38, 149-154.	2.9	5
149	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, 674-678.	7.1	169
150	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, 695-699.	7.1	50
151	A novel X-linked gene, G4.5. is responsible for Barth syndrome. Nature Genetics, 1996, 12, 385-389.	21.4	718
152	FISH characterization of the Xq21 breakpoint in a translocation carrier with premature ovarian failure. Clinical Genetics, 1996, 50, 267-269.	2.0	15
153	Identification of new mutations in the Emery-Dreifuss muscular dystrophy gene and evidence for genetic heterogeneity of the disease. Human Molecular Genetics, 1995, 4, 1859-1863.	2.9	93
154	Sequence and gene content in 52 kb including and centromeric to the G6PD gene in Xq28. DNA Sequence, 1995, 6, 1-11.	0.7	11
155	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, 377-382.	2.9	7
156	G6PD Ferrara I has the same two mutations as G6PD A(-) but a distinct biochemical phenotype. Human Genetics, 1994, 93, 139-142.	3.8	15
157	Identification of a novel X-linked gene responsible for Emery-Dreifuss muscular dystrophy. Nature Genetics, 1994, 8, 323-327.	21.4	857
158	Biochemical and molecular characterization of a new sporadic glucose-6-phosphate dehydrogenase variant described in Italy: G6PD Modena. British Journal of Haematology, 1994, 87, 209-211.	2.5	8
159	The Exon-Intron Organization of the Human X-Linked Gene (FLN1) Encoding Actin-Binding Protein 280. Genomics, 1994, 21, 71-76.	2.9	29
160	Comparative Mapping of the Actin-Binding Protein 280 Genes in Human and Mouse. Genomics, 1994, 21, 428-430.	2.9	21
161	The use of recombinant human growth hormone for radioiodination and standard preparation in radioimmunoassay. Journal of Immunological Methods, 1993, 159, 269-274.	1.4	22
162	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, 761-766.	2.9	73

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163	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, 10977-10981.	7.1	67
164	Methylation and sequence analysis around Eagi sites: identification of 28 new CpG islands in XQ24-XQ28. Nucleic Acids Research, 1992, 20, 727-733.	14.5	44
165	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, 275-280.	2.9	26
166	Identification and characterization of a new gene in the human Xq28 region. Human Molecular Genetics, 1992, 1, 269-273.	2.9	22
167	The CpG island in the 5â€² region of the G6PD gene of man and mouse. Gene, 1991, 102, 197-203.	2.2	46
168	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, 551-554.	2.9	16
169	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, 8302-8306.	7.1	76
170	Is DNA methylation of X chromosome genes stable during aging?. Somatic Cell and Molecular Genetics, 1991, 17, 101-103.	0.7	2
171	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, 519-521.	3.8	35
172	Molecular cloning and analysis of the fragile X region in man. Nucleic Acids Research, 1991, 19, 2567-2572.	14.5	50
173	Isolation of sequences that span the fragile X and identification of a fragile X-related CpG island. Science, 1991, 251, 1236-1239.	12.6	181
174	Stability of DNA methylation of X-chromosome genes during aging. Somatic Cell and Molecular Genetics, 1990, 16, 79-84.	0.7	9
175	Probes for CpG islands on the distal long arm of the human X chromosome are clustered in Xq24 and Xq28. Genomics, 1990, 8, 664-670.	2.9	24
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