Xiaoming Liu

List of Publications by Year
in descending order

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		18482	5120
168	45,567	62	166
papers	citations	h-index	g-index

192	192	192	67300
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Extensive identification of genes involved in congenital and structural heart disorders and cardiomyopathy. , 2022, 1, 157-173.		22
2	SARS-COV-2 as potential microRNA sponge in COVID-19 patients. BMC Medical Genomics, 2022, 15, 94.	1.5	17
3	Whole genome sequence analyses of eGFR in 23,732 people representing multiple ancestries in the NHLBI trans-omics for precision medicine (TOPMed) consortium. EBioMedicine, 2021, 63, 103157.	6.1	14
4	Genome annotation of disease-causing microorganisms. Briefings in Bioinformatics, 2021, 22, 845-854.	6.5	13
5	Personalized genealogical history of UK individuals inferred from biobank-scale IBD segments. BMC Biology, 2021, 19, 32.	3.8	12
6	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. Nature, 2021, 590, 290-299.	27.8	1,069
7	A resource of targeted mutant mouse lines for 5,061 genes. Nature Genetics, 2021, 53, 416-419.	21.4	60
8	A Deep Learning Model for Ancestry Estimation with Craniometric Measurements. , 2021, , .		0
9	Identifying Putative Causal Links between MicroRNAs and Severe COVID-19 Using Mendelian Randomization. Cells, 2021, 10, 3504.	4.1	7
10	Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	2.5	27
11	Stairway Plot 2: demographic history inference with folded SNP frequency spectra. Genome Biology, 2020, 21, 280.	8.8	125
12	Reduced meiotic recombination in rhesus macaques and the origin of the human recombination landscape. PLoS ONE, 2020, 15, e0236285.	2.5	7
13	Identification of MicroRNA-Related Tumorigenesis Variants and Genes in the Cancer Genome Atlas (TCGA) Data. Genes, 2020, 11, 953.	2.4	3
14	dbNSFP v4: a comprehensive database of transcript-specific functional predictions and annotations for human nonsynonymous and splice-site SNVs. Genome Medicine, 2020, 12, 103.	8.2	300
15	False Alarms in Consumer Genomics Add to Public Fear and Potential Health Care Burden. Journal of Personalized Medicine, 2020, 10, 187.	2.5	1
16	Human Prehistoric Demography Revealed by the Polymorphic Pattern of CpG Transitions. Molecular Biology and Evolution, 2020, 37, 2691-2698.	8.9	8
17	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	2.4	36
18	MMiRNA-Viewer2, a bioinformatics tool for visualizing functional annotation for MiRNA and MRNA pairs in a network. BMC Bioinformatics, 2020, 21, 247.	2.6	2

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19	The Deep Genome Project. Genome Biology, 2020, 21, 18.	8.8	30
20	dbMTS: A comprehensive database of putative human microRNA target site SNVs and their functional predictions. Human Mutation, 2020, 41, 1123-1130.	2.5	11
21	Dynamics of Plasmodium vivax populations in border areas of the Greater Mekong sub-region during malaria elimination. Malaria Journal, 2020, 19, 145.	2.3	7
22	Human and mouse essentiality screens as a resource for disease gene discovery. Nature Communications, 2020, 11, 655.	12.8	64
23	Title is missing!. , 2020, 15, e0236285.		0
24	Title is missing!. , 2020, 15, e0236285.		0
25	Title is missing!. , 2020, 15, e0236285.		0
26	Title is missing!. , 2020, 15, e0236285.		0
27	RaPID: ultra-fast, powerful, and accurate detection of segments identical by descent (IBD) in biobank-scale cohorts. Genome Biology, 2019, 20, 143.	8.8	48
28	Pharmacogenomics of statin-related myopathy: Meta-analysis of rare variants from whole-exome sequencing. PLoS ONE, 2019, 14, e0218115.	2.5	18
29	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
30	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	8.2	42
31	Side Effects and Behavioral Outcomes Following High-Dose Carnitine Supplementation Among Young Males With Autism Spectrum Disorder: A Pilot Study. Global Pediatric Health, 2019, 6, 2333794X1983069.	0.7	12
32	Comparison of different functional prediction scores using a gene-based permutation model for identifying cancer driver genes. BMC Medical Genomics, 2019, 12, 22.	1.5	12
33	Diagnostic Utility of Genome-wide DNA Methylation Testing in Genetically Unsolved Individuals with Suspected Hereditary Conditions. American Journal of Human Genetics, 2019, 104, 685-700.	6.2	125
34	Investigation of multi-trait associations using pathway-based analysis of GWAS summary statistics. BMC Genomics, 2019, 20, 79.	2.8	22
35	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
36	Validation Studies for Single Circulating Trophoblast Genetic Testing as a Form of Noninvasive Prenatal Diagnosis. American Journal of Human Genetics, 2019, 105, 1262-1273.	6.2	47

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37	Efficient Variant Set Mixed Model Association Tests for Continuous and Binary Traits in Large-Scale Whole-Genome Sequencing Studies. American Journal of Human Genetics, 2019, 104, 260-274.	6.2	103
38	Phenotypic association of 15q11.2 CNVs of the region of breakpoints 1â€“2 (BP1â€“BP2) in a large cohort of samples referred for genetic diagnosis. Journal of Human Genetics, 2019, 64, 253-255.	2.3	9
39	Iron Hack - A symposium/hackathon focused on porphyrias, Friedreichâ€™s ataxia, and other rare iron-related diseases. F1000Research, 2019, 8, 1135.	1.6	11
40	Climate-driven range shifts of the king penguin in a fragmented ecosystem. Nature Climate Change, 2018, 8, 245-251.	18.8	95
41	Sequence-Based Analysis of Lipid-Related Metabolites in a Multiethnic Study. Genetics, 2018, 209, 607-616.	2.9	8
42	Colorectal Cancer-Associated Genes Are Associated with Tooth Agenesis and May Have a Role in Tooth Development. Scientific Reports, 2018, 8, 2979.	3.3	18
43	Genetic variants in microRNA genes and targets associated with cardiovascular disease risk factors in the African-American population. Human Genetics, 2018, 137, 85-94.	3.8	9
44	FOXP3 mutations causing earlyâ€onset insulinâ€requiring diabetes but without other features of immune dysregulation, polyendocrinopathy, enteropathy, Xâ€linked syndrome. Pediatric Diabetes, 2018, 19, 388-392.	2.9	25
45	The International Conference on Intelligent Biology and Medicine (ICIBM) 2018: bioinformatics towards translational applications. BMC Bioinformatics, 2018, 19, 492.	2.6	1
46	Next-Generation Sequencing in Human Genetic Studies: Genome Technologies and Applications to Human Genetic Studies. Human Heredity, 2018, 83, 105-106.	0.8	0
47	Identification of genes required for eye development by high-throughput screening of mouse knockouts. Communications Biology, 2018, 1, 236.	4.4	37
48	Rapid and Integrative Discovery of Retina Regulatory Molecules. Cell Reports, 2018, 24, 2506-2519.	6.4	28
49	Comparative analysis of single-stranded DNA donors to generate conditional null mouse alleles. BMC Biology, 2018, 16, 69.	3.8	64
50	2017 Victor A. McKusick Leadership Award. American Journal of Human Genetics, 2018, 102, 361-363.	6.2	0
51	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	14.5	98
52	Practical Approaches for Whole-Genome Sequence Analysis of Heart- and Blood-Related Traits. American Journal of Human Genetics, 2017, 100, 205-215.	6.2	50
53	The next generation of population-based spinal muscular atrophy carrier screening: comprehensive pan-ethnic SMN1 copy-number and sequence variant analysis by massively parallel sequencing. Genetics in Medicine, 2017, 19, 936-944.	2.4	70
54	The performance of deleteriousness prediction scores for rare non-protein-changing single nucleotide variants in human genes. Journal of Medical Genetics, 2017, 54, 134-144.	3.2	16

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55	Loss-of-Function Variants in MYLK Cause Recessive Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. American Journal of Human Genetics, 2017, 101, 123-129.	6.2	67
56	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
57	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	27.0	565
58	Analysis commons, a team approach to discovery in a big-data environment for genetic epidemiology. Nature Genetics, 2017, 49, 1560-1563.	21.4	93
59	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
60	A large scale hearing loss screen reveals an extensive unexplored genetic landscape for auditory dysfunction. Nature Communications, 2017, 8, 886.	12.8	116
61	Brain carnitine deficiency causes nonsyndromic autism with an extreme male bias: A hypothesis. BioEssays, 2017, 39, 1700012.	2.5	35
62	Polygenic Scores for Major Depressive Disorder and Risk of Alcohol Dependence. JAMA Psychiatry, 2017, 74, 1153.	11.0	73
63	Role of <scp>WNT</scp>10A in failure of tooth development in humans and zebrafish. Molecular Genetics & Genomic Medicine, 2017, 5, 730-741.	1.2	27
64	Prevalence of sexual dimorphism in mammalian phenotypic traits. Nature Communications, 2017, 8, 15475.	12.8	200
65	Disease model discovery from 3,328 gene knockouts by The International Mouse Phenotyping Consortium. Nature Genetics, 2017, 49, 1231-1238.	21.4	216
66	Whole-genome sequencing study of serum peptide levels: the Atherosclerosis Risk in Communities study. Human Molecular Genetics, 2017, 26, 3442-3450.	2.9	25
67	In Silico Prediction of Deleteriousness for Nonsynonymous and Splice-Altering Single Nucleotide Variants in the Human Genome. Methods in Molecular Biology, 2017, 1498, 191-197.	0.9	17
68	Universal Prenatal Chromosomal Microarray Analysis: Additive Value and Clinical Dilemmas in Fetuses with a Normal Karyotype. American Journal of Perinatology, 2017, 34, 340-348.	1.4	21
69	Darwin Comes to Clinic. Trends in Genetics, 2017, 33, 1-2.	6.7	3
70	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
71	Identification of Rare Variants in Metabolites of the Carnitine Pathway by Whole Genome Sequencing Analysis. Genetic Epidemiology, 2016, 40, 486-491.	1.3	10
72	dbNSFP v3.0: A One-Stop Database of Functional Predictions and Annotations for Human Nonsynonymous and Splice-Site SNVs. Human Mutation, 2016, 37, 235-241.	2.5	845

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73	Whole genome sequence analysis of serum amino acid levels. Genome Biology, 2016, 17, 237.	8.8	17
74	iCAGES: integrated CAncer GEnome Score for comprehensively prioritizing driver genes in personal cancer genomes. Genome Medicine, 2016, 8, 135.	8.2	45
75	Three-dimensional microCT imaging of mouse development from early post-implantation to early postnatal stages. Developmental Biology, 2016, 419, 229-236.	2.0	43
76	High-throughput discovery of novel developmental phenotypes. Nature, 2016, 537, 508-514.	27.8	1,001
77	Evidence for feasibility of fetal trophoblastic cellâ€based noninvasive prenatal testing. Prenatal Diagnosis, 2016, 36, 1009-1019.	2.3	78
78	The population genomics of rhesus macaques (Macaca mulatta) based on whole-genome sequences. Genome Research, 2016, 26, 1651-1662.	5.5	101
79	Genome-wide copy number analysis on DNA from fetal cells isolated from the blood of pregnant women. Prenatal Diagnosis, 2016, 36, 1127-1134.	2.3	68
80	Chromosomal microarray analysis, or comparative genomic hybridization: A high throughput approach. MethodsX, 2016, 3, 8-18.	1.6	6
81	FLAGS: A Flexible and Adaptive Association Test for Gene Sets Using Summary Statistics. Genetics, 2016, 202, 919-929.	2.9	11
82	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	2.4	186
83	Epigenetics and Human Disease. Cold Spring Harbor Perspectives in Biology, 2016, 8, a019497.	5.5	177
84	WGSA: an annotation pipeline for human genome sequencing studies. Journal of Medical Genetics, 2016, 53, 111-112.	3.2	96
85	Global genetic carrier testing: a vision for the future. Genome Medicine, 2015, 7, 79.	8.2	4
86	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	6.2	65
87	Comparison and integration of deleteriousness prediction methods for nonsynonymous SNVs in whole exome sequencing studies. Human Molecular Genetics, 2015, 24, 2125-2137.	2.9	892
88	Analysis of loss-of-function variants and 20 risk factor phenotypes in 8,554 individuals identifies loci influencing chronic disease. Nature Genetics, 2015, 47, 640-642.	21.4	49
89	Untargeted metabolomic analysis for the clinical screening of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2015, 38, 1029-1039.	3.6	169
90	Exploring population size changes using SNP frequency spectra. Nature Genetics, 2015, 47, 555-559.	21.4	332

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91	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	8.1	1,219
92	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
93	Towards a therapy for Angelman syndrome by targeting a long non-coding RNA. Nature, 2015, 518, 409-412.	27.8	423
94	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. Biological Psychiatry, 2015, 77, 775-784.	1.3	133
95	Population Genomic Analysis of 962 Whole Genome Sequences of Humans Reveals Natural Selection in Non-Coding Regions. PLoS ONE, 2015, 10, e0121644.	2.5	13
96	In silico prediction of splice-altering single nucleotide variants in the human genome. Nucleic Acids Research, 2014, 42, 13534-13544.	14.5	396
97	Sequencing of 2 Subclinical Atherosclerosis Candidate Regions in 3669 Individuals. Circulation: Cardiovascular Genetics, 2014, 7, 359-364.	5.1	18
98	Neonatal diabetes, gallbladder agenesis, duodenal atresia, and intestinal malrotation caused by a novel homozygous mutation inRFX6. Pediatric Diabetes, 2014, 15, 67-72.	2.9	57
99	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	7.4	1,171
100	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	2.8	112
101	Whole-Exome Sequencing Identifies Rare and Low-Frequency Coding Variants Associated with LDL Cholesterol. American Journal of Human Genetics, 2014, 94, 233-245.	6.2	193
102	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	6.2	92
103	Strategies to Design and Analyze Targeted Sequencing Data. Circulation: Cardiovascular Genetics, 2014, 7, 335-343.	5.1	18
104	In silico tools for splicing defect prediction: a survey from the viewpoint of end users. Genetics in Medicine, 2014, 16, 497-503.	2.4	124
105	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	6.2	57
106	Associations of NINJ2 Sequence Variants with Incident Ischemic Stroke in the Cohorts for Heart and Aging in Genomic Epidemiology (CHARGE) Consortium. PLoS ONE, 2014, 9, e99798.	2.5	11
107	Gene-Specific Function Prediction for Non-Synonymous Mutations in Monogenic Diabetes Genes. PLoS ONE, 2014, 9, e104452.	2.5	23
108	Significantly fewer protein functional changing variants for lipid metabolism in Africans than in Europeans. Journal of Translational Medicine, 2013, 11, 67.	4.4	4

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109	Translational signatures and mRNA levels are highly correlated in human stably expressed genes. BMC Genomics, 2013, 14, 268.	2.8	6
110	dbNSFP v2.0: A Database of Human Non-synonymous SNVs and Their Functional Predictions and Annotations. Human Mutation, 2013, 34, E2393-E2402.	2.5	546
111	Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine, 2013, 369, 1502-1511.	27.0	1,717
112	Integrating GWASs and Human Protein Interaction Networks Identifies a Gene Subnetwork Underlying Alcohol Dependence. American Journal of Human Genetics, 2013, 93, 1027-1034.	6.2	72
113	The Utility of Chromosomal Microarray Analysis in Developmental and Behavioral Pediatrics. Child Development, 2013, 84, 121-132.	3.0	67
114	Whole-genome sequenceâ€"based analysis of high-density lipoprotein cholesterol. Nature Genetics, 2013, 45, 899-901.	21.4	132
115	Truncation of Ube3a-ATS Unsilences Paternal Ube3a and Ameliorates Behavioral Defects in the Angelman Syndrome Mouse Model. PLoS Genetics, 2013, 9, e1004039.	3.5	124
116	Preventable Forms of Autism?. Science, 2012, 338, 342-343.	12.6	20
117	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	27.8	7,199
118	Identification of Common Prognostic Gene Expression Signatures with Biological Meanings from Microarray Gene Expression Datasets. PLoS ONE, 2012, 7, e45894.	2.5	18
119	Evolution and Functional Impact of Rare Coding Variation from Deep Sequencing of Human Exomes. Science, 2012, 337, 64-69.	12.6	1,535
120	jPopGen Suite: population genetic analysis of DNA polymorphism from nucleotide sequences with errors. Methods in Ecology and Evolution, 2012, 3, 624-627.	5.2	3
121	Progress toward Noninvasive Prenatal Diagnosis. Clinical Chemistry, 2011, 57, 802-804.	3.2	2
122	Incorporating predicted functions of nonsynonymous variants into gene-based analysis of exome sequencing data: a comparative study. BMC Proceedings, 2011, 5, S20.	1.6	18
123	dbNSFP: A lightweight database of human nonsynonymous SNPs and their functional predictions. Human Mutation, 2011, 32, 894-899.	2.5	706
124	Smaller Genetic Risk in Catabolic Process Explains Lower Energy Expenditure, More Athletic Capability and Higher Prevalence of Obesity in Africans. PLoS ONE, 2011, 6, e26027.	2.5	3
125	Estimating population genetic parameters and comparing model goodness-of-fit using DNA sequences with error. Genome Research, 2010, 20, 101-109.	5.5	16
126	Simulating Sequences of the Human Genome with Rare Variants. Human Heredity, 2010, 70, 287-291.	0.8	15

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127	Deep resequencing reveals excess rare recent variants consistent with explosive population growth. Nature Communications, 2010, 1, 131.	12.8	213
128	Ethical issues raised by common copy number variants and single nucleotide polymorphisms of certain and uncertain significance in general medical practice. Genome Medicine, 2010, 2, 42.	8.2	20
129	Inferring Population Mutation Rate and Sequencing Error Rate Using the SNP Frequency Spectrum in a Sample of DNA Sequences. Molecular Biology and Evolution, 2009, 26, 1479-1490.	8.9	10
130	Array-Based DNA Diagnostics: Let the Revolution Begin. Annual Review of Medicine, 2008, 59, 113-129.	12.2	131
131	Algorithms to estimate the lower bounds of recombination with or without recurrent mutations. BMC Genomics, 2008, 9, S24.	2.8	4
132	Summary statistics of neutral mutations in longitudinal DNA samples. Theoretical Population Biology, 2008, 74, 56-67.	1.1	2
133	Glycogen storage disease: long-term follow-up of nocturnal intragastric feeding. Clinical Genetics, 2008, 21, 136-140.	2.0	9
134	Allan Award Lecture: Rare Patients Leading to Epigenetics and Back to Genetics. American Journal of Human Genetics, 2008, 82, 1034-1038.	6.2	5
135	Epigenetics and Complex Human Disease: Is There a Role in IBD?. Journal of Pediatric Gastroenterology and Nutrition, 2008, 46, E2.	1.8	8
136	Test of Genetical Isochronism for Longitudinal Samples of DNA Sequences. Genetics, 2007, 176, 327-342.	2.9	4
137	Evidence for Recombination in Mycobacterium tuberculosis. Journal of Bacteriology, 2006, 188, 8169-8177.	2.2	66
138	Age-of-onset of hypertension vs. a single measurement of systolic blood pressure in a combined linkage and segregation analysis. BMC Genetics, 2003, 4, S80.	2.7	3
139	Is medical genetics neglecting epigenetics?. Genetics in Medicine, 2002, 4, 399-402.	2.4	24
140	A Rheostat Model for a Rapid and Reversible Form of Imprinting-Dependent Evolution. American Journal of Human Genetics, 2002, 70, 1389-1397.	6.2	58
141	Genome-wide scan for familial nasopharyngeal carcinoma reveals evidence of linkage to chromosome 4. Nature Genetics, 2002, 31, 395-399.	21.4	217
142	A population genetics model of linkage disequilibrium in admixed populations. Science Bulletin, 2001, 46, 193-197.	1.7	7
143	Rate of decay in admixture linkage disequilibrium and its implication in gene mapping. Science Bulletin, 2001, 46, 358-363.	1.7	3
144	Allelic loss and gain, but not genomic instability, as the major somatic mutation in primary hepatocellular carcinoma. Genes Chromosomes and Cancer, 2001, 31, 221-227.	2.8	64

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145	The SNRPN promoter is not required for genomic imprinting of the Prader-Willi/Angelman domain in mice. Nature Genetics, 2001, 28, 232-240.	21.4	95
146	Absence of P-Selectin, but Not Intercellular Adhesion Molecule-1, Attenuates Neointimal Growth After Arterial Injury in Apolipoprotein Eâ€“Deficient Mice. Circulation, 2001, 103, 1000-1005.	1.6	108
147	Necdin-deficient mice do not show lethality or the obesity and infertility of Prader-Willi syndrome. Nature Genetics, 1999, 22, 15-16.	21.4	81
148	Structure of the murine E-selectin ligand 1 (ESL-1) gene and assignment to Chromosome 8. Mammalian Genome, 1999, 10, 1085-1088.	2.2	5
149	Genomic DNA transfer with a high-capacity adenovirus vector results in improved in vivo gene expression and decreased toxicity. Nature Genetics, 1998, 18, 180-183.	21.4	641
150	Combinatorial requirements for adhesion molecules in mediating neutrophil emigration during bacterial peritonitis in mice. Journal of Leukocyte Biology, 1998, 64, 291-297.	3.3	31
151	De novo truncating mutations in E6-AP ubiquitin-protein ligase gene (UBE3A) in Angelman syndrome. Nature Genetics, 1997, 15, 74-77.	21.4	801
152	Imprinted expression of the murine Angelman syndrome gene, Ube3a, in hippocampal and Purkinje neurons. Nature Genetics, 1997, 17, 75-78.	21.4	466
153	Validation studies of SNRPN methylation as a diagnostic test for Prader-Willi syndrome. , 1996, 66, 77-80.		87
154	Advantages of RT-PCR and denaturing gradient gel electrophoresis for analysis of genomic imprinting: Detection of new mouse and human expressed polymorphisms. , 1996, 7, 144-148.		1
155	Simultaneous analysis of mutant and normal alleles for multiple cystic fibrosis mutations by the ligase chain reaction. Human Mutation, 1995, 6, 144-151.	2.5	13
156	Deletions of a differentially methylated CpG island at the SNRPN gene define a putative imprinting control region. Nature Genetics, 1994, 8, 52-58.	21.4	418
157	A suggested nomenclature for designating mutations. Human Mutation, 1993, 2, 245-248.	2.5	354
158	Expansion of an unstable trinucleotide CAG repeat in spinocerebellar ataxia type 1. Nature Genetics, 1993, 4, 221-226.	21.4	1,673
159	Prenatal diagnosis of citrullinaemia: Review of a 10-year experience including recent use of DNA analysis. Prenatal Diagnosis, 1990, 10, 771-779.	2.3	27
160	Spinocerebellar ataxia: Variable age of onset and linkage to human leukocyte antigen in a large kindred. Annals of Neurology, 1988, 23, 580-584.	5.3	126
161	The Perlman familial nephroblastomatosis syndrome. American Journal of Medical Genetics Part A, 1986, 24, 101-110.	2.4	59
162	Atypical presentation and neuropathological studies in 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. Annals of Neurology, 1986, 20, 367-369.	5.3	22

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163	Abnormal mRNA for argininosuccinate synthetase in citrullinaemia. Nature, 1983, 301, 533-534.	27.8	42
164	Ornithine transcarbamylase deficiency: longâ€term survival. Clinical Genetics, 1982, 22, 211-214.	2.0	2
165	Repair of bleomycin-damaged DNA by human fibroblasts. Journal of Supramolecular Structure and Cellular Biochemistry, 1981, 16, 303-309.	1.4	7
166	Cholesteryl Lignocerate Hydrolysis in Adrenoleukodystrophy. Pediatric Research, 1980, 14, 21-23.	2.3	12
167	Detection of Fabry's disease heterozygotes by hair root analysis. Clinical Genetics, 1978, 13, 251-258.	2.0	20
168	Arginase deficiency in multiple tissues in argininemia. Clinical Genetics, 1978, 13, 61-67.	2.0	53