

Jian-Min Chen

List of Publications by Year in descending order

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166
papers

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47006

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8173
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#	ARTICLE	IF	CITATIONS
1	NGS mismapping confounds the clinical interpretation of the <i>PRSS1</i> p.Ala16Val (c.47C>T) variant in chronic pancreatitis. <i>Gut</i> , 2022, 71, 841-842.	12.1	8
2	In situ delivery of apoptotic bodies derived from mesenchymal stem cells via a hyaluronic acid hydrogel: A therapy for intrauterine adhesions. <i>Bioactive Materials</i> , 2022, 12, 107-119.	15.6	36
3	Trypsinogen (<i>PRSS1</i> and <i>PRSS2</i>) gene dosage correlates with pancreatitis risk across genetic and transgenic studies: a systematic review and re-analysis. <i>Human Genetics</i> , 2022, 141, 1327-1338.	3.8	8
4	The <i>CEL-HYB1</i> Hybrid Allele Promotes Digestive Enzyme Misfolding and Pancreatitis in Mice. <i>Cellular and Molecular Gastroenterology and Hepatology</i> , 2022, 14, 55-74.	4.5	8
5	Functionally deficient <i>TRPV6</i> variants contribute to hereditary and familial chronic pancreatitis. <i>Human Mutation</i> , 2022, 43, 228-239.	2.5	7
6	Variants in the pancreatic CUB and zona pellucida-like domains 1 (<i>CUZD1</i>) gene in early-onset chronic pancreatitis - A possible new susceptibility gene. <i>Pancreatology</i> , 2022, 22, 564-571.	1.1	4
7	Splicing analysis of <i>SLC40A1</i> missense variations and contribution to hemochromatosis type 4 phenotypes. <i>Blood Cells, Molecules, and Diseases</i> , 2021, 87, 102527.	1.4	5
8	The reversion variant (p.Arg90Leu) at the evolutionarily adaptive p.Arg90 site in <i>CELA3B</i> predisposes to chronic pancreatitis. <i>Human Mutation</i> , 2021, 42, 385-391.	2.5	6
9	The effect of laparoscopic excisional and ablative surgery on ovarian reserve in patients with endometriomas. <i>Medicine (United States)</i> , 2021, 100, e24362.	1.0	6
10	The p.E152K- <i>STIM1</i> mutation deregulates Ca ²⁺ signaling contributing to chronic pancreatitis. <i>Journal of Cell Science</i> , 2021, 134, .	2.0	4
11	Scale and Scope of Gene-Alcohol Interactions in Chronic Pancreatitis: A Systematic Review. <i>Genes</i> , 2021, 12, 471.	2.4	9
12	Digenic Inheritance and Gene-Environment Interaction in a Patient With Hypertriglyceridemia and Acute Pancreatitis. <i>Frontiers in Genetics</i> , 2021, 12, 640859.	2.3	7
13	Missense RHD single nucleotide variants induce weakened D antigen expression by altering splicing and/or protein expression. <i>Transfusion</i> , 2021, 61, 2468-2476.	1.6	1
14	The three common polymorphisms p.A986S, p.R990G and p.Q1011E in the calcium sensing receptor (<i>CASR</i>) are not associated with chronic pancreatitis. <i>Pancreatology</i> , 2021, 21, 1299-1304.	1.1	3
15	Splicing Outcomes of 5' Splice Site GT>GC Variants That Generate Wild-Type Transcripts Differ Significantly Between Full-Length and Minigene Splicing Assays. <i>Frontiers in Genetics</i> , 2021, 12, 701652.	2.3	9
16	Effect of Low Dietary Vitamin D Fed Prior to and During Pregnancy and Lactation on Maternal Bone Mineral Density, Structure, and Strength in C57BL/6 Mice. <i>Current Developments in Nutrition</i> , 2021, 5, nzab114.	0.3	1
17	Asian Population Is More Prone to Develop High-Risk Myelodysplastic Syndrome, Concordantly with Their Propensity to Exhibit High-Risk Cytogenetic Aberrations. <i>Cancers</i> , 2021, 13, 481.	3.7	22
18	No Convincing Evidence to Support a Bimodal Age of Onset in Idiopathic Chronic Pancreatitis. <i>Clinical Gastroenterology and Hepatology</i> , 2021, , .	4.4	1

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19	Chronic Pancreatitis: The True Pathogenic Culprit within the SPINK1 N34S-Containing Haplotype Is No Longer at Large. <i>Genes</i> , 2021, 12, 1683.	2.4	5
20	The corrected breakpoint sequence of the homozygous SPINK1 deletion causing severe infantile isolated exocrine pancreatic insufficiency. <i>Human Mutation</i> , 2021, 42, 216-217.	2.5	0
21	Most unambiguous loss-of-function <i>CPA1</i> mutations are unlikely to predispose to chronic pancreatitis. <i>Gut</i> , 2020, 69, 785-786.	12.1	6
22	Variants That Affect Function of Calcium Channel TRPV6 Are Associated With Early-Onset Chronic Pancreatitis. <i>Gastroenterology</i> , 2020, 158, 1626-1641.e8.	1.3	77
23	Analysis of GPRC6A variants in different pancreatitis etiologies. <i>Pancreatology</i> , 2020, 20, 1262-1267.	1.1	1
24	Role of the Common PRSS1-PRSS2 Haplotype in Alcoholic and Non-Alcoholic Chronic Pancreatitis: Meta- and Re-Analyses. <i>Genes</i> , 2020, 11, 1349.	2.4	14
25	Pathogenic and likely pathogenic variants in at least five genes account for approximately 3% of mild isolated nonsyndromic thrombocytopenia. <i>Transfusion</i> , 2020, 60, 2419-2431.	1.6	6
26	EXT1 and EXT2 Variants in 22 Chinese Families With Multiple Osteochondromas: Seven New Variants and Potentiation of Preimplantation Genetic Testing and Prenatal Diagnosis. <i>Frontiers in Genetics</i> , 2020, 11, 607838.	2.3	4
27	5' splice site GC and GT variants differ markedly in terms of their functionality and pathogenicity. <i>Human Mutation</i> , 2020, 41, 1358-1364.	2.5	7
28	Identification and functional characterization of a novel heterozygous missense variant in the <i>LPL</i> associated with recurrent hypertriglyceridemia-induced acute pancreatitis in pregnancy. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1048.	1.2	11
29	Characterization of CEL-DUP2: Complete duplication of the carboxyl ester lipase gene is unlikely to influence risk of chronic pancreatitis. <i>Pancreatology</i> , 2020, 20, 377-384.	1.1	5
30	The Experimentally Obtained Functional Impact Assessments of 5' Splice Site GT and GC Variants Differ Markedly from Those Predicted. <i>Current Genomics</i> , 2020, 21, 56-66.	1.6	16
31	<i>Citrobacter rodentium</i> alters the mouse colonic miRNome. <i>Genes and Immunity</i> , 2019, 20, 207-213.	4.1	2
32	Common variants in glyoxalase I do not increase chronic pancreatitis risk. <i>PLoS ONE</i> , 2019, 14, e0222927.	2.5	0
33	First estimate of the scale of canonical 5' splice site GT and GC variants capable of generating wild-type transcripts. <i>Human Mutation</i> , 2019, 40, 1856-1873.	2.5	25
34	Identification of a novel and heterozygous LMF1 nonsense mutation in an acute pancreatitis patient with severe hypertriglyceridemia, severe obesity and heavy smoking. <i>Lipids in Health and Disease</i> , 2019, 18, 68.	3.0	17
35	Toward a clinical diagnostic pipeline for SPINK1 intronic variants. <i>Human Genomics</i> , 2019, 13, 8.	2.9	8
36	Protease-Sensitive Pancreatic Lipase Variants Are Associated With Early Onset Chronic Pancreatitis. <i>American Journal of Gastroenterology</i> , 2019, 114, 974-983.	0.4	48

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37	Cornual Suture at the Time of Laparoscopic Salpingectomy Reduces the Incidence of Interstitial Pregnancy after In Vitro Fertilization. <i>Journal of Minimally Invasive Gynecology</i> , 2018, 25, 1080-1087.	0.6	9
38	<i>PRSS1</i> copy number variants and promoter polymorphisms in pancreatitis: common pathogenetic mechanism, different genetic effects. <i>Gut</i> , 2018, 67, 592-593.	12.1	12
39	Genome-wide association study identifies inversion in the <i>CTRB1-CTRB2</i> locus to modify risk for alcoholic and non-alcoholic chronic pancreatitis. <i>Gut</i> , 2018, 67, 1855-1863.	12.1	97
40	The <i>CTRB1-CTRB2</i> risk allele for chronic pancreatitis discovered in European populations does not contribute to disease risk variation in the Chinese population due to near allele fixation. <i>Gut</i> , 2018, 67, 1368-1369.	12.1	12
41	<i>SPINK1</i> , <i>PRSS1</i> , <i>CTRC</i> , and <i>CFTR</i> Genotypes Influence Disease Onset and Clinical Outcomes in Chronic Pancreatitis. <i>Clinical and Translational Gastroenterology</i> , 2018, 9, e204.	2.5	76
42	<i>TPX2</i> silencing mediated by joint action of microvesicles and ultrasonic radiation inhibits the migration and invasion of SKOV3 cells. <i>Molecular Medicine Reports</i> , 2018, 17, 7627-7635.	2.4	4
43	Identification of compound heterozygous variants in the noncoding <i>RNU4ATAC</i> gene in a Chinese family with two successive foetuses with severe microcephaly. <i>Human Genomics</i> , 2018, 12, 3.	2.9	12
44	No significant enrichment of rare functionally defective <i>CPA1</i> variants in a large Chinese idiopathic chronic pancreatitis cohort. <i>Human Mutation</i> , 2017, 38, 959-963.	2.5	19
45	Identification of a novel <i>SPINK1</i> deletion in a teenager with idiopathic chronic pancreatitis. <i>Digestive and Liver Disease</i> , 2017, 49, 941-943.	0.9	1
46	Identification of a functional enhancer variant within the chronic pancreatitis-associated <i>SPINK1</i> c.101A>G (p.Asn34Ser)-containing haplotype. <i>Human Mutation</i> , 2017, 38, 1014-1024.	2.5	18
47	<i>PKD2</i> -Related Autosomal Dominant Polycystic Kidney Disease: Prevalence, Clinical Presentation, Mutation Spectrum, and Prognosis. <i>American Journal of Kidney Diseases</i> , 2017, 70, 476-485.	1.9	50
48	In vitro and in silico evidence against a significant effect of the <i>SPINK1</i> c.194G>A variant on pre-mRNA splicing. <i>Gut</i> , 2017, 66, 2195-2196.	12.1	12
49	Severe infantile isolated exocrine pancreatic insufficiency caused by the complete functional loss of the <i>SPINK1</i> gene. <i>Human Mutation</i> , 2017, 38, 1660-1665.	2.5	24
50	In silico prioritization and further functional characterization of <i>SPINK1</i> intronic variants. <i>Human Genomics</i> , 2017, 11, 7.	2.9	10
51	Deletions Overlapping <i>VCAN</i> Exon 8 Are New Molecular Defects for Wagner Disease. <i>Human Mutation</i> , 2017, 38, 43-47.	2.5	16
52	Analysis of the Impact of Known <i>SPINK1</i> Missense Variants on Pre-mRNA Splicing and/or mRNA Stability in a Full-Length Gene Assay. <i>Genes</i> , 2017, 8, 263.	2.4	10
53	Pathogenetics of Chronic Pancreatitis. , 2017, , 63-77.		0
54	A Short History of Research into Chronic Pancreatitis. , 2017, , 1-5.		0

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55	No Association Between CELâ€“HYB Hybrid Allele and Chronic Pancreatitis in Asian Populations. <i>Gastroenterology</i> , 2016, 150, 1558-1560.e5.	1.3	59
56	Discovery and Functional Annotation of<i>PRSS1</i> Promoter Variants in Chronic Pancreatitis. <i>Human Mutation</i> , 2016, 37, 1149-1152.	2.5	5
57	Incidence of Interstitial Pregnancy After InÂ“vitro Fertilization/Embryo Transfer and the Outcome of a ConsecutiveÂ“Series of 38 Cases Managed by Laparoscopic Cornuostomy or Cornual Repair. <i>Journal of Minimally Invasive Gynecology</i> , 2016, 23, 739-747.	0.6	32
58	Digging deeper into the intronic sequences of the<i>SPINK1</i> gene: TableÂ“1. <i>Gut</i> , 2016, 65, 1055-1056.	12.1	10
59	Clarifying the clinical relevance of<i>SPINK1</i> intronic variants in chronic pancreatitis. <i>Gut</i> , 2016, 65, 884-886.	12.1	32
60	Overrepresentation of Rare CASR Coding Variants in a Sample of Young French Patients With Idiopathic Chronic Pancreatitis. <i>Pancreas</i> , 2015, 44, 996-998.	1.1	15
61	Disclosing the Hidden Structure and Underlying Mutational Mechanism of a Novel Type of Duplication CNV Responsible for Hereditary Multiple Osteochondromas. <i>Human Mutation</i> , 2015, 36, 758-763.	2.5	6
62	Complex Multiple-Nucleotide Substitution Mutations Causing Human Inherited Disease Reveal Novel Insights into the Action of Translesion Synthesis DNA Polymerases. <i>Human Mutation</i> , 2015, 36, 1034-1038.	2.5	12
63	Report of 2 CTRC Intronic Mutations Associated With Acute or Chronic Pancreatitis and Delineation of Their Pathogenic Molecular Mechanisms. <i>Pancreas</i> , 2015, 44, 999-1001.	1.1	2
64	Concurrent Nucleotide Substitution Mutations in the Human Genome Are Characterized by a Significantly Decreased Transition/Transversion Ratio. <i>Human Mutation</i> , 2015, 36, 333-341.	2.5	9
65	A recombined allele of the lipase gene CEL and its pseudogene CELP confers susceptibility to chronic pancreatitis. <i>Nature Genetics</i> , 2015, 47, 518-522.	21.4	157
66	Characterization of 26 deletion CNVs reveals the frequent occurrence of micro-mutations within the breakpoint-flanking regions and frequent repair of double-strand breaks by templated insertions derived from remote genomic regions. <i>Human Genetics</i> , 2015, 134, 589-603.	3.8	25
67	Clinical Features and Endoscopic Treatment of Chinese Patients With Hereditary Pancreatitis. <i>Pancreas</i> , 2015, 44, 59-63.	1.1	16
68	Characterization of the second <scp>HFE</scp> gross deletion highlights the potential importance of Aluâ€“mediated recombination in haemochromatosis. <i>British Journal of Haematology</i> , 2015, 168, 759-762.	2.5	0
69	Identification of a functional<i>PRSS1</i> promoter variant in linkage disequilibrium with the chronic pancreatitis-protecting rs10273639. <i>Gut</i> , 2015, 64, 1837-1838.	12.1	35
70	Flaxseed oil enhances the effectiveness of trastuzumab in reducing the growth of HER2-overexpressing human breast tumors (BT-474). <i>Journal of Nutritional Biochemistry</i> , 2015, 26, 16-23.	4.2	27
71	Polymorphisms at<i>PRSS1</i>â€“PRSS2</i> and<i>CLDN2</i>â€“MORC4</i> loci associate with alcoholic and non-alcoholic chronic pancreatitis in a European replication study. <i>Gut</i> , 2015, 64, 1426-1433.	12.1	105
72	Long-Term Vitamin D3 Supplementation Does Not Prevent Colonic Inflammation or Modulate Bone Health in IL-10 Knockout Mice at Young Adulthood. <i>Nutrients</i> , 2014, 6, 3847-3862.	4.1	14

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73	Genetics and Pathogenesis of Autosomal Dominant Polycystic Kidney Disease: 20 Years On. <i>Human Mutation</i> , 2014, 35, 1393-1406.	2.5	74
74	A New and More Accurate Estimate of the Rate of Concurrent Tandem-Base Substitution Mutations in the Human Germline: $\hat{\approx}$ 1/40.4% of the Single-Nucleotide Substitution Mutation Rate. <i>Human Mutation</i> , 2014, 35, 392-394.	2.5	15
75	Small deletions within the <i>RHD</i> coding sequence: a report of two novel mutational events and a survey of the underlying pathophysiologic mechanisms. <i>Transfusion</i> , 2013, 53, 206-210.	1.6	9
76	Dissecting the Structure and Mechanism of a Complex Duplication-Triplication Rearrangement in the <i>DMD</i> Gene. <i>Human Mutation</i> , 2013, 34, 1080-1084.	2.5	31
77	Human Trypsins. , 2013, , 2600-2609.		7
78	Variants in <i>CPA1</i> are strongly associated with early onset chronic pancreatitis. <i>Nature Genetics</i> , 2013, 45, 1216-1220.	21.4	255
79	Establishment of a medium-throughput approach for the genotyping of <i>RHD</i> variants and report of nine novel rare alleles. <i>Transfusion</i> , 2013, 53, 1821-1828.	1.6	39
80	Characterization of two deletions of the <i>CTRC</i> locus. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 296-300.	1.1	12
81	Type of <i>PKD1</i> Mutation Influences Renal Outcome in ADPKD. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 1006-1013.	6.1	403
82	Patterns and Mutational Signatures of Tandem Base Substitutions Causing Human Inherited Disease. <i>Human Mutation</i> , 2013, 34, 1119-1130.	2.5	34
83	Cationic Trypsin (Human). , 2013, , 2609-2614.		1
84	Anionic Trypsin (Human). , 2013, , 2614-2616.		1
85	A Missense Mutation in the Alpha-Actinin 1 Gene (<i>ACTN1</i>) Is the Cause of Autosomal Dominant Macrothrombocytopenia in a Large French Family. <i>PLoS ONE</i> , 2013, 8, e74728.	2.5	49
86	A Conservative Assessment of the Major Genetic Causes of Idiopathic Chronic Pancreatitis: Data from a Comprehensive Analysis of <i>PRSS1</i> , <i>SPINK1</i> , <i>CTRC</i> and <i>CFTR</i> Genes in 253 Young French Patients. <i>PLoS ONE</i> , 2013, 8, e73522.	2.5	89
87	First description of <i>ABCB4</i> gene deletions in familial low phospholipid-associated cholelithiasis and oral contraceptives-induced cholestasis. <i>European Journal of Human Genetics</i> , 2012, 20, 277-282.	2.8	42
88	Functional Analysis of Eight Missense Mutations in the <i>SPINK1</i> Gene. <i>Pancreas</i> , 2012, 41, 329-330.	1.1	17
89	Local sequence determinants of two in-frame triplet deletion/duplication hotspots in the <i>RHD/RHCE</i> genes. <i>Human Genomics</i> , 2012, 6, 8.	2.9	2
90	Genetics and pathogenesis of chronic pancreatitis: The 2012 update. <i>Clinics and Research in Hepatology and Gastroenterology</i> , 2012, 36, 334-340.	1.5	49

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91	Autosomal dominant polycystic kidney disease: Comprehensive mutation analysis of PKD1 and PKD2 in 700 unrelated patients. <i>Human Mutation</i> , 2012, 33, 1239-1250.	2.5	144
92	Variant screening of the <i>RHD</i> gene in a large cohort of subjects with D phenotype ambiguity: report of 17 novel rare alleles. <i>Transfusion</i> , 2012, 52, 759-764.	1.6	24
93	Weak D caused by a founder deletion in the <i>RHD</i> gene. <i>Transfusion</i> , 2012, 52, 2348-2355.	1.6	22
94	Transient hypermutability, chromothripsis and replication-based mechanisms in the generation of concurrent clustered mutations. <i>Mutation Research - Reviews in Mutation Research</i> , 2012, 750, 52-59.	5.5	25
95	Effects of Flaxseed Lignan and Oil on Bone Health of Breast-Tumor-Bearing Mice Treated With or Without Tamoxifen. <i>Journal of Toxicology and Environmental Health - Part A: Current Issues</i> , 2011, 74, 757-768.	2.3	13
96	Local DNA sequence determinants of <i>FUT2</i> copy number variation. <i>Transfusion</i> , 2011, 51, 1359-1361.	1.6	3
97	Assessing the pathological relevance of SPINK1 promoter variants. <i>European Journal of Human Genetics</i> , 2011, 19, 1066-1073.	2.8	18
98	On the sequence-directed nature of human gene mutation: The role of genomic architecture and the local DNA sequence environment in mediating gene mutations underlying human inherited disease. <i>Human Mutation</i> , 2011, 32, 1075-1099.	2.5	99
99	Detection and characterisation of large SERPINC1 deletions in type I inherited antithrombin deficiency. <i>Human Genetics</i> , 2010, 127, 45-53.	3.8	27
100	Genetics of osteoporosis: accelerating pace in gene identification and validation. <i>Human Genetics</i> , 2010, 127, 249-285.	3.8	85
101	Genomic rearrangements in inherited disease and cancer. <i>Seminars in Cancer Biology</i> , 2010, 20, 222-233.	9.6	140
102	Complete ascertainment of intragenic copy number mutations (CNMs) in the CFTR gene and its implications for CNM formation at other autosomal loci. <i>Human Mutation</i> , 2010, 31, 421-428.	2.5	31
103	Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. <i>Human Mutation</i> , 2010, 31, 631-655.	2.5	161
104	Gene Conversion in Human Genetic Disease. <i>Genes</i> , 2010, 1, 550-563.	2.4	16
105	Genetic Analysis of the Glycoprotein 2 Gene in Patients With Chronic Pancreatitis. <i>Pancreas</i> , 2010, 39, 353-358.	1.1	10
106	The c.1275A>G putative chronic pancreatitis-associated synonymous polymorphism in the glycoprotein 2 (GP2) gene decreases exon 9 inclusion. <i>Molecular Genetics and Metabolism</i> , 2010, 99, 319-324.	1.1	10
107	Genetics of osteoporosis: perspectives for personalized medicine. <i>Personalized Medicine</i> , 2010, 7, 655-668.	1.5	13
108	The true culprit within the SPINK1 p.N34S-containing haplotype is still at large. <i>Gut</i> , 2009, 58, 478-480.	12.1	13

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109	Elucidation of the complex structure and origin of the human trypsinogen locus triplication. <i>Human Molecular Genetics</i> , 2009, 18, 3605-3614.	2.9	22
110	Gene conversion causing human inherited disease: Evidence for involvement of non-B-DNA-forming sequences and recombination-promoting motifs in DNA breakage and repair. <i>Human Mutation</i> , 2009, 30, 1189-1198.	2.5	63
111	Closely spaced multiple mutations as potential signatures of transient hypermutability in human genes. <i>Human Mutation</i> , 2009, 30, 1435-1448.	2.5	51
112	Chronic Pancreatitis: Genetics and Pathogenesis. <i>Annual Review of Genomics and Human Genetics</i> , 2009, 10, 63-87.	6.2	104
113	Association of rare chymotrypsinogen C (CTRC) gene variations in patients with idiopathic chronic pancreatitis. <i>Human Genetics</i> , 2008, 123, 83-91.	3.8	159
114	Hereditary pancreatitis caused by a double gain-of-function trypsinogen mutation. <i>Human Genetics</i> , 2008, 123, 521-529.	3.8	42
115	The 10â€Mb paracentric inversion of chromosome arm 2p in activating <i>MSH2</i> and causing hereditary nonpolyposis colorectal cancer: Reâ€annotation and mutational mechanisms. <i>Genes Chromosomes and Cancer</i> , 2008, 47, 543-545.	2.8	18
116	Role of nonâ€B DNA conformations in initiating the nonallelic homologous recombinationâ€derived <i>Se^{sup}fus</i> allele and the interlocus gene conversionâ€derived <i>Sec1â€FUT2â€Sec1</i> hybrid allele. <i>Transfusion</i> , 2008, 48, 1522-1523.	1.6	5
117	Detection of two Alu insertions in the CFTR gene. <i>Journal of Cystic Fibrosis</i> , 2008, 7, 37-43.	0.7	38
118	Trypsinogen Copy Number Mutations in Patients With Idiopathic Chronic Pancreatitis. <i>Clinical Gastroenterology and Hepatology</i> , 2008, 6, 82-88.	4.4	75
119	Homozygous deletion of HFE produces a phenotype similar to the HFE p.C282Y/p.C282Y genotype. <i>Blood</i> , 2008, 112, 5238-5240.	1.4	16
120	Absence of Mesotrypsinogen Gene (PRSS3) Copy Number Variations in Patients With Chronic Pancreatitis. <i>Pancreas</i> , 2008, 37, 227-228.	1.1	10
121	Co-inheritance of a novel deletion of the entire SPINK1 gene with a CFTR missense mutation (L997F) in a family with chronic pancreatitis. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 168-175.	1.1	25
122	Dietary Flaxseed Interaction With Tamoxifen Induced Tumor Regression in Athymic Mice With MCF-7 Xenografts by Downregulating the Expression of Estrogen Related Gene Products and Signal Transduction Pathways. <i>Nutrition and Cancer</i> , 2007, 58, 162-170.	2.0	31
123	Flaxseed Alone or in Combination with Tamoxifen Inhibits MCF-7 Breast Tumor Growth in Ovariectomized Athymic Mice with High Circulating Levels of Estrogen. <i>Experimental Biology and Medicine</i> , 2007, 232, 1071-1080.	2.4	55
124	A large genomic deletion in the PDHX gene caused by the retrotranspositional insertion of a full-length LINE-1 element. <i>Human Mutation</i> , 2007, 28, 137-142.	2.5	62
125	Signal peptide variants that impair secretion of pancreatic secretory trypsin inhibitor (SPINK1) cause autosomal dominant hereditary pancreatitis. <i>Human Mutation</i> , 2007, 28, 469-476.	2.5	68
126	Double complex mutations involving F8 and FUNDC2 caused by distinct break-induced replication. <i>Human Mutation</i> , 2007, 28, 1198-1206.	2.5	57

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127	Gene conversion: mechanisms, evolution and human disease. <i>Nature Reviews Genetics</i> , 2007, 8, 762-775.	16.3	576
128	Functional analysis of pancreatitis-associated missense mutations in the pancreatic secretory trypsin inhibitor (SPINK1) gene. <i>European Journal of Human Genetics</i> , 2007, 15, 936-942.	2.8	64
129	Searching for potential microRNA-binding site mutations amongst known disease-associated 3' UTR variants. <i>Genomic Medicine</i> , 2007, 1, 29-33.	0.3	7
130	Mechanism of Alu integration into the human genome. <i>Genomic Medicine</i> , 2007, 1, 9-17.	0.3	16
131	Flaxseed and its components reduce metastasis after surgical excision of solid human breast tumor in nude mice. <i>Cancer Letters</i> , 2006, 234, 168-175.	7.2	67
132	A degradation-sensitive anionic trypsinogen (PRSS2) variant protects against chronic pancreatitis. <i>Nature Genetics</i> , 2006, 38, 668-673.	21.4	220
133	Hereditary pancreatitis caused by triplication of the trypsinogen locus. <i>Nature Genetics</i> , 2006, 38, 1372-1374.	21.4	182
134	Gross genomic rearrangements involving deletions in the CFTR gene: characterization of six new events from a large cohort of hitherto unidentified cystic fibrosis chromosomes and meta-analysis of the underlying mechanisms. <i>European Journal of Human Genetics</i> , 2006, 14, 567-576.	2.8	77
135	Detection of a large genomic deletion in the pancreatic secretory trypsin inhibitor (SPINK1) gene. <i>European Journal of Human Genetics</i> , 2006, 14, 1204-1208.	2.8	28
136	A systematic analysis of disease-associated variants in the 3' regulatory regions of human protein-coding genes I: general principles and overview. <i>Human Genetics</i> , 2006, 120, 1-21.	3.8	135
137	A systematic analysis of disease-associated variants in the 3' regulatory regions of human protein-coding genes II: the importance of mRNA secondary structure in assessing the functionality of 3' UTR variants. <i>Human Genetics</i> , 2006, 120, 301-333.	3.8	125
138	LINE-1 Endonuclease-Dependent Retrotranspositional Events Causing Human Genetic Disease: Mutation Detection Bias and Multiple Mechanisms of Target Gene Disruption. <i>Journal of Biomedicine and Biotechnology</i> , 2006, 2006, 1-9.	3.0	51
139	Interactive effects of sesame seed and tamoxifen on estrogen dependent breast cancer in athymic nude mice. <i>FASEB Journal</i> , 2006, 20, A993.	0.5	0
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