

# 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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183  
docs citations

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times ranked

8173  
citing authors

#	ARTICLE	IF	CITATIONS
1	Gene conversion: mechanisms, evolution and human disease. <i>Nature Reviews Genetics</i> , 2007, 8, 762-775.	16.8	576
2	Type of PKD1 Mutation Influences Renal Outcome in ADPKD. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 1006-1013.	6.1	403
3	Variants in CPA1 are strongly associated with early onset chronic pancreatitis. <i>Nature Genetics</i> , 2013, 45, 1216-1220.	21.4	255
4	A degradation-sensitive anionic trypsinogen (PRSS2) variant protects against chronic pancreatitis. <i>Nature Genetics</i> , 2006, 38, 668-673.	21.4	220
5	A systematic analysis of LINE-1 endonuclease-dependent retrotranspositional events causing human genetic disease. <i>Human Genetics</i> , 2005, 117, 411-427.	3.8	206
6	Hereditary pancreatitis caused by triplication of the trypsinogen locus. <i>Nature Genetics</i> , 2006, 38, 1372-1374.	21.4	182
7	Dietary Flaxseed Inhibits Human Breast Cancer Growth and Metastasis and Downregulates Expression of Insulin-Like Growth Factor and Epidermal Growth Factor Receptor. <i>Nutrition and Cancer</i> , 2002, 43, 187-192.	2.0	172
8	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
9	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
10	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
11	Meta-analysis of gross insertions causing human genetic disease: Novel mutational mechanisms and the role of replication slippage. <i>Human Mutation</i> , 2005, 25, 207-221.	2.5	148
12	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
13	Genomic rearrangements in inherited disease and cancer. <i>Seminars in Cancer Biology</i> , 2010, 20, 222-233.	9.6	140
14	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
15	Determination of the relative contribution of three genes—the cystic fibrosis transmembrane conductance regulator gene, the cationic trypsinogen gene, and the pancreatic secretory trypsin inhibitor gene—to the etiology of idiopathic chronic pancreatitis. <i>European Journal of Human Genetics</i> , 2002, 10, 100-106.	2.8	131
16	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
17	Genomic rearrangements in the CFTR gene: Extensive allelic heterogeneity and diverse mutational mechanisms. <i>Human Mutation</i> , 2004, 23, 343-357.	2.5	115
18	Polymorphisms at PRSS1, PRSS2 and CLDN2/MORC4 loci associate with alcoholic and non-alcoholic chronic pancreatitis in a European replication study. <i>Gut</i> , 2015, 64, 1426-1433.	12.1	105

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19	Chronic Pancreatitis: Genetics and Pathogenesis. Annual Review of Genomics and Human Genetics, 2009, 10, 63-87.	6.2	104
20	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. Human Mutation, 2011, 32, 1075-1099.	2.5	99
21	Evolution of Trypsinogen Activation Peptides. Molecular Biology and Evolution, 2003, 20, 1767-1777.	8.9	97
22	Genome-wide association study identifies inversion in the <i>CTRB1-CTRB2</i> locus to modify risk for alcoholic and non-alcoholic chronic pancreatitis. Gut, 2018, 67, 1855-1863.	12.1	97
23	A Conservative Assessment of the Major Genetic Causes of Idiopathic Chronic Pancreatitis: Data from a Comprehensive Analysis of PRSS1, SPINK1, CTFC and CFTR Genes in 253 Young French Patients. PLoS ONE, 2013, 8, e73522.	2.5	89
24	Complex gene rearrangements caused by serial replication slippage. Human Mutation, 2005, 26, 125-134.	2.5	88
25	Genetics of osteoporosis: accelerating pace in gene identification and validation. Human Genetics, 2010, 127, 249-285.	3.8	85
26	Dietary Flaxseed Enhances the Inhibitory Effect of Tamoxifen on the Growth of Estrogen-Dependent Human Breast Cancer (MCF-7) in Nude Mice. Clinical Cancer Research, 2004, 10, 7703-7711.	7.0	81
27	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. European Journal of Human Genetics, 2006, 14, 567-576.	2.8	77
28	Variants That Affect Function of Calcium Channel TRPV6 Are Associated With Early-Onset Chronic Pancreatitis. Gastroenterology, 2020, 158, 1626-1641.e8.	1.3	77
29	SPINK1, PRSS1, CTFC, and CFTR Genotypes Influence Disease Onset and Clinical Outcomes in Chronic Pancreatitis. Clinical and Translational Gastroenterology, 2018, 9, e204.	2.5	76
30	Trypsinogen Copy Number Mutations in Patients With Idiopathic Chronic Pancreatitis. Clinical Gastroenterology and Hepatology, 2008, 6, 82-88.	4.4	75
31	Lignans and Tamoxifen, Alone or in Combination, Reduce Human Breast Cancer Cell Adhesion, Invasion and Migration in vitro. Breast Cancer Research and Treatment, 2003, 80, 163-170.	2.5	74
32	Genetics and Pathogenesis of Autosomal Dominant Polycystic Kidney Disease: 20 Years On. Human Mutation, 2014, 35, 1393-1406.	2.5	74
33	Exposure to Flaxseed or Its Purified Lignan during Suckling Inhibits Chemically Induced Rat Mammary Tumorigenesis. Experimental Biology and Medicine, 2003, 228, 951-958.	2.4	72
34	A Combined Analysis of the Cystic Fibrosis Transmembrane Conductance Regulator: Implications for Structure and Disease Models. Molecular Biology and Evolution, 2001, 18, 1771-1788.	8.9	68
35	Signal peptide variants that impair secretion of pancreatic secretory trypsin inhibitor (SPINK1) cause autosomal dominant hereditary pancreatitis. Human Mutation, 2007, 28, 469-476.	2.5	68
36	Flaxseed and its components reduce metastasis after surgical excision of solid human breast tumor in nude mice. Cancer Letters, 2006, 234, 168-175.	7.2	67

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37	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
38	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
39	Molecular pathology and evolutionary and physiological implications of pancreatitis-associated cationic trypsinogen mutations. <i>Human Genetics</i> , 2001, 109, 245-252.	3.8	62
40	Intrachromosomal serial replication slippage in <i>trans</i> gives rise to diverse genomic rearrangements involving inversions. <i>Human Mutation</i> , 2005, 26, 362-373.	2.5	62
41	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
42	No Association Between CELâ€œHYB Hybrid Allele and Chronic Pancreatitis in Asian Populations. <i>Gastroenterology</i> , 2016, 150, 1558-1560.e5.	1.3	59
43	Double complex mutations involving F8 and FUNDC2 caused by distinct break-induced replication. <i>Human Mutation</i> , 2007, 28, 1198-1206.	2.5	57
44	Interaction between trypsinogen isoforms in genetically determined pancreatitis: Mutation E79K in cationic trypsin (PRSS1) causes increased transactivation of anionic trypsinogen (PRSS2). <i>Human Mutation</i> , 2004, 23, 22-31.	2.5	55
45	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
46	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
47	Closely spaced multiple mutations as potential signatures of transient hypermutability in human genes. <i>Human Mutation</i> , 2009, 30, 1435-1448.	2.5	51
48	PKD2 -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
49	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
50	A Missense Mutation in the Alpha-Actinin 1 Gene (ACTN1) Is the Cause of Autosomal Dominant Macrothrombocytopenia in a Large French Family. <i>PLoS ONE</i> , 2013, 8, e74728.	2.5	49
51	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
52	â€œLoss of functionâ€œ mutations in the cationic trypsinogen gene (PRSS1) may act as a protective factor against pancreatitis. <i>Molecular Genetics and Metabolism</i> , 2003, 79, 67-70.	1.1	45
53	Discrimination of three mutational events that result in a disruption of the R122 primary autolysis site of the human cationic trypsinogen (PRSS1) by denaturing high performance liquid chromatography. <i>BMC Genetics</i> , 2001, 2, 19.	2.7	42
54	Hereditary pancreatitis caused by a double gain-of-function trypsinogen mutation. <i>Human Genetics</i> , 2008, 123, 521-529.	3.8	42

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55	First description of ABCB4 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
56	Genes, Cloned cDNAs, and Proteins of Human Trypsinogens and Pancreatitis-Associated Cationic Trypsinogen Mutations. <i>Pancreas</i> , 2000, 21, 57-62.	1.1	40
57	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
58	The A16V signal peptide cleavage site mutation in the cationic trypsinogen gene and chronic pancreatitis. <i>Gastroenterology</i> , 1999, 117, 1508-1509.	1.3	38
59	Detection of two Alu insertions in the CFTR gene. <i>Journal of Cystic Fibrosis</i> , 2008, 7, 37-43.	0.7	38
60	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
61	Identification of a Novel Pancreatitis-Associated Missense Mutation, R116C, in the Human Cationic Trypsinogen Gene (PRSS1). <i>Molecular Genetics and Metabolism</i> , 2001, 74, 342-344.	1.1	35
62	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
63	Patterns and Mutational Signatures of Tandem Base Substitutions Causing Human Inherited Disease. <i>Human Mutation</i> , 2013, 34, 1119-1130.	2.5	34
64	Incidence of Interstitial Pregnancy After In Vitro Fertilization/Embryo Transfer and the Outcome of a Consecutive 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
65	Clarifying the clinical relevance of <i>SPINK1</i> intronic variants in chronic pancreatitis. <i>Gut</i> , 2016, 65, 884-886.	12.1	32
66	Molecular basis of hereditary pancreatitis. <i>European Journal of Human Genetics</i> , 2000, 8, 473-479.	2.8	31
67	Gene Conversion-like Missense Mutations in the Human Cationic Trypsinogen Gene and Insights into the Molecular Evolution of the Human Trypsinogen Family. <i>Molecular Genetics and Metabolism</i> , 2000, 71, 463-469.	1.1	31
68	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
69	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
70	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
71	Detection of a large genomic deletion in the pancreatic secretory trypsin inhibitor ( <i>SPINK1</i> ) gene. <i>European Journal of Human Genetics</i> , 2006, 14, 1204-1208.	2.8	28
72	Detection and characterisation of large <i>SERPINC1</i> deletions in type I inherited antithrombin deficiency. <i>Human Genetics</i> , 2010, 127, 45-53.	3.8	27

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73	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
74	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
75	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
76	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
77	First estimate of the scale of canonical 5' splice site GT&gt;GC variants capable of generating wild&type transcripts. <i>Human Mutation</i> , 2019, 40, 1856-1873.	2.5	25
78	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
79	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
80	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
81	Weak D caused by a founder deletion in the <i>RHD</i> gene. <i>Transfusion</i> , 2012, 52, 2348-2355.	1.6	22
82	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
83	No significant enrichment of rare functionally defective CPA1 variants in a large Chinese idiopathic chronic pancreatitis cohort. <i>Human Mutation</i> , 2017, 38, 959-963.	2.5	19
84	Depletion of O6-methylguanine&quot;DNA methyltransferase and potentiation of 1,3-bis(2-chloroethyl)-1-nitrosourea antitumor activity by O6-benzylguanine in vitro. <i>Carcinogenesis</i> , 1993, 14, 1057-1060.	2.8	18
85	The 10&Mb paracentric inversion of chromosome arm 2p in activating <i>MSH2</i> and causing hereditary nonpolyposis colorectal cancer: Re&annotation and mutational mechanisms. <i>Genes Chromosomes and Cancer</i> , 2008, 47, 543-545.	2.8	18
86	Assessing the pathological relevance of SPINK1 promoter variants. <i>European Journal of Human Genetics</i> , 2011, 19, 1066-1073.	2.8	18
87	Identification of a functional enhancer variant within the chronic pancreatitis&associated <i>SPINK1</i> c.101A&gt;G (p.As<sup>n</sup>34Ser)&containing haplotype. <i>Human Mutation</i> , 2017, 38, 1014-1024.	2.5	18
88	Functional Analysis of Eight Missense Mutations in the SPINK1 Gene. <i>Pancreas</i> , 2012, 41, 329-330.	1.1	17
89	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
90	Mechanism of Alu integration into the human genome. <i>Genomic Medicine</i> , 2007, 1, 9-17.	0.3	16

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91	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
92	Gene Conversion in Human Genetic Disease. <i>Genes</i> , 2010, 1, 550-563.	2.4	16
93	Clinical Features and Endoscopic Treatment of Chinese Patients With Hereditary Pancreatitis. <i>Pancreas</i> , 2015, 44, 59-63.	1.1	16
94	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
95	The Experimentally Obtained Functional Impact Assessments of 5' Splice Site GT&gt;GC Variants Differ Markedly from Those Predicted. <i>Current Genomics</i> , 2020, 21, 56-66.	1.6	16
96	A New and More Accurate Estimate of the Rate of Concurrent Tandem-Base Substitution Mutations in the Human Germline: ~1/40.4% of the Single-Nucleotide Substitution Mutation Rate. <i>Human Mutation</i> , 2014, 35, 392-394.	2.5	15
97	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
98	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
99	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
100	The true culprit within the SPINK1 p.N34S-containing haplotype is still at large. <i>Gut</i> , 2009, 58, 478-480.	12.1	13
101	Genetics of osteoporosis: perspectives for personalized medicine. <i>Personalized Medicine</i> , 2010, 7, 655-668.	1.5	13
102	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
103	Characterization of two deletions of the CTSC locus. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 296-300.	1.1	12
104	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
105	In vitro and in silico evidence against a significant effect of the <i>SPINK1</i> c.194G&gt;A variant on pre-mRNA splicing. <i>Gut</i> , 2017, 66, 2195-2196.	12.1	12
106	<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
107	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
108	Identification of compound heterozygous variants in the noncoding RNU4ATAC gene in a Chinese family with two successive fetuses with severe microcephaly. <i>Human Genomics</i> , 2018, 12, 3.	2.9	12

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109	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 &amp; Genomic Medicine</i> , 2020, 8, e1048.	1.2	11
110	Absence of Mesotrypsinogen Gene (PRSS3) Copy Number Variations in Patients With Chronic Pancreatitis. <i>Pancreas</i> , 2008, 37, 227-228.	1.1	10
111	Genetic Analysis of the Glycoprotein 2 Gene in Patients With Chronic Pancreatitis. <i>Pancreas</i> , 2010, 39, 353-358.	1.1	10
112	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
113	Digging deeper into the intronic sequences of the <i>SPINK1</i> gene: Table 1. <i>Gut</i> , 2016, 65, 1055-1056.	12.1	10
114	In silico prioritization and further functional characterization of <i>SPINK1</i> intronic variants. <i>Human Genomics</i> , 2017, 11, 7.	2.9	10
115	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
116	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
117	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
118	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
119	Scale and Scope of Gene-Alcohol Interactions in Chronic Pancreatitis: A Systematic Review. <i>Genes</i> , 2021, 12, 471.	2.4	9
120	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
121	Definition of a "Functional R Domain" of the Cystic Fibrosis Transmembrane Conductance Regulator. <i>Molecular Genetics and Metabolism</i> , 2000, 71, 245-249.	1.1	8
122	Toward a clinical diagnostic pipeline for <i>SPINK1</i> intronic variants. <i>Human Genomics</i> , 2019, 13, 8.	2.9	8
123	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
124	Trypsinogen (PRSS1 and PRSS2) 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
125	The CEL-HYB1 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
126	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



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127	Human Trypsins. , 2013, , 2600-2609.		7
128	5â€² splice site GC&gt;GT and GT&gt;GC variants differ markedly in terms of their functionality and pathogenicity. Human Mutation, 2020, 41, 1358-1364.	2.5	7
129	Digenic Inheritance and Gene-Environment Interaction in a Patient With Hypertriglyceridemia and Acute Pancreatitis. Frontiers in Genetics, 2021, 12, 640859.	2.3	7
130	Functionally deficient <i>TRPV6</i> variants contribute to hereditary and familial chronic pancreatitis. Human Mutation, 2022, 43, 228-239.	2.5	7
131	Disclosing the Hidden Structure and Underlying Mutational Mechanism of a Novel Type of Duplication CNV Responsible for Hereditary Multiple Osteochondromas. Human Mutation, 2015, 36, 758-763.	2.5	6
132	Most unambiguous loss-of-function <i>CPA1</i> mutations are unlikely to predispose to chronic pancreatitis. Gut, 2020, 69, 785-786.	12.1	6
133	Pathogenic and likely pathogenic variants in at least five genes account for approximately 3% of mild isolated nonsyndromic thrombocytopenia. Transfusion, 2020, 60, 2419-2431.	1.6	6
134	The reversion variant (p.Arg90Leu) at the evolutionarily adaptive p.Arg90 site in CELA3B predisposes to chronic pancreatitis. Human Mutation, 2021, 42, 385-391.	2.5	6
135	The effect of laparoscopic excisional and ablative surgery on ovarian reserve in patients with endometriomas. Medicine (United States), 2021, 100, e24362.	1.0	6
136	Role of nonâ€ DNA conformations in initiating the nonallelic homologous recombinationâ€ derived <i>Se<sup>fus</sup></i> allele and the interlocus gene conversionâ€ derived <i>Sec1â€FUT2â€Sec1</i> hybrid allele. Transfusion, 2008, 48, 1522-1523.	1.6	5
137	Discovery and Functional Annotation of<i>PRSS1</i> Promoter Variants in Chronic Pancreatitis. Human Mutation, 2016, 37, 1149-1152.	2.5	5
138	Characterization of CEL-DUP2: Complete duplication of the carboxyl ester lipase gene is unlikely to influence risk of chronic pancreatitis. Pancreatology, 2020, 20, 377-384.	1.1	5
139	Splicing analysis of SLC40A1 missense variations and contribution to hemochromatosis type 4 phenotypes. Blood Cells, Molecules, and Diseases, 2021, 87, 102527.	1.4	5
140	Chronic Pancreatitis: The True Pathogenic Culprit within the SPINK1 N34S-Containing Haplotype Is No Longer at Large. Genes, 2021, 12, 1683.	2.4	5
141	TPX2 silencing mediated by joint action of microvesicles and ultrasonic radiation inhibits the migration and invasion of SKOV3 cells. Molecular Medicine Reports, 2018, 17, 7627-7635.	2.4	4
142	EXT1 and EXT2 Variants in 22 Chinese Families With Multiple Osteochondromas: Seven New Variants and Potentiation of Preimplantation Genetic Testing and Prenatal Diagnosis. Frontiers in Genetics, 2020, 11, 607838.	2.3	4
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