Claude Ferec

List of Publications by Year
in descending order

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398 papers	16,869 citations	18887 64 h-index	26792 111 g-index
421 all docs	421 docs citations	421 times ranked	17164 citing authors

#	Article	IF	Citations
1	NGS mismapping confounds the clinical interpretation of the PRSS1 p.Ala16Val (c.47C>T) variant in chronic pancreatitis. Gut, 2022, 71, 841-842.	6.1	8
2	Mutations in the most divergent Î±â€tubulin isotype, Î±8â€tubulin, cause defective platelet biogenesis. Journal of Thrombosis and Haemostasis, 2022, 20, 461-469.	1.9	4
3	The novel c.634+<scp>4A</scp>>G splicing variant in<scp>RHCE</scp>results in weak C and e antigen expression in a pregnant woman originated from Japan. Transfusion, 2022, 62, 758-763.	0.8	0
4	Trypsinogen (PRSS1 and PRSS2) gene dosage correlates with pancreatitis risk across genetic and transgenic studies: a systematic review and re-analysis. Human Genetics, 2022, 141, 1327-1338.	1.8	8
5	The CEL-HYB1 Hybrid Allele Promotes Digestive Enzyme Misfolding and Pancreatitis in Mice. Cellular and Molecular Gastroenterology and Hepatology, 2022, 14, 55-74.	2.3	8
6	Functionally deficient TRPV6 variants contribute to hereditary and familial chronic pancreatitis. Human Mutation, 2022, 43, 228-239.	1.1	7
7	Variants in the pancreatic CUB and zona pellucida-like domains 1 (CUZD1) gene in early-onset chronic pancreatitis - A possible new susceptibility gene. Pancreatology, 2022, 22, 564-571.	0.5	4
8	3D Chromatin Organization Involving MEIS1 Factor in the cis-Regulatory Landscape of GJB2. International Journal of Molecular Sciences, 2022, 23, 6964.	1.8	0
9	Evaluation of aminopyrrolidine amide to improve chloride transport in CFTR-defective cells. Bioorganic and Medicinal Chemistry Letters, 2022, 72, 128866.	1.0	2
10	Clinical interpretation of PRSS1 variants in patients with pancreatitis. Clinics and Research in Hepatology and Gastroenterology, 2021, 45, 101497.	0.7	9
11	Splicing analysis of SLC40A1 missense variations and contribution to hemochromatosis type 4 phenotypes. Blood Cells, Molecules, and Diseases, 2021, 87, 102527.	0.6	5
12	The reversion variant (p.Arg90Leu) at the evolutionarily adaptive p.Arg90 site in CELA3B predisposes to chronic pancreatitis. Human Mutation, 2021, 42, 385-391.	1.1	6
13	The p.E152K-STIM1 mutation deregulates Ca2+ signaling contributing to chronic pancreatitis. Journal of Cell Science, 2021, 134, .	1.2	4
14	Scale and Scope of Gene-Alcohol Interactions in Chronic Pancreatitis: A Systematic Review. Genes, 2021, 12, 471.	1.0	9
15	CFTR Cooperative Cis-Regulatory Elements in Intestinal Cells. International Journal of Molecular Sciences, 2021, 22, 2599.	1.8	3
16	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
17	A <scp>Peruvian</scp> patient carrying the novel <scp>RHCE</scp>*<scp>cE</scp>(<scp>c.382Gâ€‰>â€‰C</scp>) missense allele in the <scp>RH</scp> blood group system. Transfusion, 2021, 61, E41-E43.	0.8	1
18	Digenic Inheritance and Gene-Environment Interaction in a Patient With Hypertriglyceridemia and Acute Pancreatitis. Frontiers in Genetics, 2021, 12, 640859.	1.1	7

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19	Missense RHD single nucleotide variants induce weakened D antigen expression by altering splicing and/or protein expression. Transfusion, 2021, 61, 2468-2476.	0.8	1
20	Heterozygous <scp>HMGB1</scp> lossâ€ofâ€function variants are associated with developmental delay and microcephaly. Clinical Genetics, 2021, 100, 386-395.	1.0	3
21	The three common polymorphisms p.A986S, p.R990G and p.Q1011E in the calcium sensing receptor (CASR) are not associated with chronic pancreatitis. Pancreatology, 2021, 21, 1299-1304.	0.5	3
22	Whole-Genome Sequencing Improves the Diagnosis of DFNB1 Monoallelic Patients. Genes, 2021, 12, 1267.	1.0	4
23	Splicing Outcomes of 5â€² Splice Site GT>GC Variants That Generate Wild-Type Transcripts Differ Significantly Between Full-Length and Minigene Splicing Assays. Frontiers in Genetics, 2021, 12, 701652.	1.1	9
24	No Convincing Evidence to Support a Bimodal Age of Onset in Idiopathic Chronic Pancreatitis. Clinical Gastroenterology and Hepatology, 2021, , .	2.4	1
25	Chronic Pancreatitis: The True Pathogenic Culprit within the SPINK1 N34S-Containing Haplotype Is No Longer at Large. Genes, 2021, 12, 1683.	1.0	5
26	The corrected breakpoint sequence of the homozygous SPINK1 deletion causing severe infantile isolated exocrine pancreatic insufficiency. Human Mutation, 2021, 42, 216-217.	1.1	0
27	Nationâ€wide investigation of RHD variants in Thai blood donors: Impact for molecular diagnostics. Transfusion, 2021, 61, 931-938.	0.8	4
28	The Interplay between the Unfolded Protein Response, Inflammation and Infection in Cystic Fibrosis. Cells, 2021, 10, 2980.	1.8	12
29	Most unambiguous loss-of-function CPA1 mutations are unlikely to predispose to chronic pancreatitis. Gut, 2020, 69, 785-786.	6.1	6
30	Variants That Affect Function of Calcium Channel TRPV6 Are Associated With Early-Onset Chronic Pancreatitis. Gastroenterology, 2020, 158, 1626-1641.e8.	0.6	77
31	Defining Blood Group Gene Reference Alleles by Long-Read Sequencing: Proof of Concept in the <i>ACKR1</i> Gene Encoding the Duffy Antigens. Transfusion Medicine and Hemotherapy, 2020, 47, 23-32.	0.7	10
32	Comprehensive Molecular Analysis of Serologically D-Negative and Weak/Partial D Phenotype in Thai Blood Donors. Transfusion Medicine and Hemotherapy, 2020, 47, 54-60.	0.7	12
33	Analysis of GPRC6A variants in different pancreatitis etiologies. Pancreatology, 2020, 20, 1262-1267.	0.5	1
34	Clinical interpretation of SPINK1 and CTRC variants in pancreatitis. Pancreatology, 2020, 20, 1354-1367.	0.5	5
35	Role of the Common PRSS1-PRSS2 Haplotype in Alcoholic and Non-Alcoholic Chronic Pancreatitis: Meta- and Re-Analyses. Genes, 2020, 11, 1349.	1.0	14
36	Pathogenic and likely pathogenic variants in at least five genes account for approximately 3% of mild isolated nonsyndromic thrombocytopenia. Transfusion, 2020, 60, 2419-2431.	0.8	6

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37	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.	1.1	4
38	5â€² splice site GC>GT and GT>GC variants differ markedly in terms of their functionality and pathogenicity. Human Mutation, 2020, 41, 1358-1364.	1.1	7
39	The Changing Epidemiology of Cystic Fibrosis: Incidence, Survival and Impact of the CFTR Gene Discovery. Genes, 2020, 11, 589.	1.0	151
40	Identification and functional characterization of a novel heterozygous missense variant in the LPL associated with recurrent hypertriglyceridemiaâ€induced acute pancreatitis in pregnancy. Molecular Genetics & Genomic Medicine, 2020, 8, e1048.	0.6	11
41	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.	0.5	5
42	The Experimentally Obtained Functional Impact Assessments of 5' Splice Site GT>GC Variants Differ Markedly from Those Predicted. Current Genomics, 2020, 21, 56-66.	0.7	16
43	Molecular characterization of rare D--/D-- variants in individuals of Indian origin. Blood Transfusion, 2020, , .	0.3	3
44	Blood transcriptomic biomarker as a surrogate of ischemic brain gene expression. Annals of Clinical and Translational Neurology, 2019, 6, 1681-1695.	1.7	17
45	<scp>CFTR</scp> gene variants: a predisposition factor to aquagenic palmoplantar keratoderma. British Journal of Dermatology, 2019, 181, 1097-1099.	1.4	14
46	Pitfalls in the interpretation of CFTR variants in the context of incidental findings. Human Mutation, 2019, 40, 2239-2246.	1.1	9
47	Common variants in glyoxalase I do not increase chronic pancreatitis risk. PLoS ONE, 2019, 14, e0222927.	1.1	0
48	Molecular model of the ferroportin intracellular gate and implications for the human iron transport cycle and hemochromatosis type 4A. FASEB Journal, 2019, 33, 14625-14635.	0.2	11
49	Overexpression of certain transient receptor potential and Orai channels in prostate cancer is associated with decreased risk of systemic recurrence after radical prostatectomy. Prostate, 2019, 79, 1793-1804.	1.2	15
50	Natural history of SPINK1 germline mutation related-pancreatitis. EBioMedicine, 2019, 48, 581-591.	2.7	37
51	Compound Heterozygosity for Novel Truncating Variants in the LMOD3 Gene as the Cause of Polyhydramnios in Two Successive Fetuses. Frontiers in Genetics, 2019, 10, 835.	1.1	5
52	Characterization of GJB2 cis-regulatory elements in the DFNB1 locus. Human Genetics, 2019, 138, 1275-1286.	1.8	7
53	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	1.1	41
54	First estimate of the scale of canonical 5â€² splice site GT>GC variants capable of generating wildâ€type transcripts. Human Mutation, 2019, 40, 1856-1873.	1.1	25

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55	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. Brain, 2019, 142, 1573-1586.	3.7	49
56	Functional analysis of novel RHD variants: splicing disruption is likely to be a common mechanism of variant D phenotype. Transfusion, 2019, 59, 1367-1375.	0.8	12
57	Porphyromonas, a potential predictive biomarker of Pseudomonas aeruginosa pulmonary infection in cystic fibrosis. BMJ Open Respiratory Research, 2019, 6, e000374.	1.2	12
58	A novel JK null allele in a Brazilian patient with sickle cell disease (SCD). Transfusion, 2019, 59, 2459-2460.	0.8	1
59	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
60	Toward a clinical diagnostic pipeline for SPINK1 intronic variants. Human Genomics, 2019, 13, 8.	1.4	8
61	Protease-Sensitive Pancreatic Lipase Variants Are Associated With Early Onset Chronic Pancreatitis. American Journal of Gastroenterology, 2019, 114, 974-983.	0.2	48
62	Performance of semiconductor sequencing platform for nonâ€invasive prenatal genetic screening for fetal aneuploidy: results from a multicenter prospective cohort study in a clinical setting. Ultrasound in Obstetrics and Gynecology, 2019, 54, 246-254.	0.9	9
63	Functional characterization and phenotypic spectrum of three recurrent disease-causing deep intronic variants of the CFTR gene. Journal of Cystic Fibrosis, 2019, 18, 468-475.	0.3	36
64	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	3.7	44
65	Screening for Regulatory Variants in 460 kb Encompassing the CFTR Locus in Cystic Fibrosis Patients. Journal of Molecular Diagnostics, 2019, 21, 70-80.	1.2	7
66	Weak D type 1, 2 and 3 subtype alleles are rare in the Western French population. Transfusion Medicine, 2019, 29, 209-210.	0.5	6
67	An essential role for Î±4A-tubulin in platelet biogenesis. Life Science Alliance, 2019, 2, e201900309.	1.3	34
68	Comprehensive phenotypic and molecular investigation of RhD and RhCE variants in Moroccan blood donors. Blood Transfusion, 2019, 17, 151-156.	0.3	6
69	Molecular basis of weak D expression in the Indian population and report of a novel, predominant variant RHD allele. Transfusion, 2018, 58, 1540-1549.	0.8	24
70	RHD-Positive Alleles among D- C/E+ Individuals from India. Transfusion Medicine and Hemotherapy, 2018, 45, 173-177.	0.7	9
71	Do pregnancies reduce iron overload in HFE hemochromatosis women? results from an observational prospective study. BMC Pregnancy and Childbirth, 2018, 18, 53.	0.9	2
72	Monoallelic Mutations to DNAJB11 Cause Atypical Autosomal-Dominant Polycystic Kidney Disease. American Journal of Human Genetics, 2018, 102, 832-844.	2.6	208

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73	PRSS1 copy number variants and promoter polymorphisms in pancreatitis: common pathogenetic mechanism, different genetic effects. Gut, 2018, 67, 592-593.	6.1	12
74	Genome-wide association study identifies inversion in the CTRB1-CTRB2 locus to modify risk for alcoholic and non-alcoholic chronic pancreatitis. Gut, 2018, 67, 1855-1863.	6.1	97
75	The CTRB1-CTRB2 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. Gut, 2018, 67, 1368-1369.	6.1	12
76	Adhesion, proliferation and osteogenic differentiation of human MSCs cultured under perfusion with a marine oxygen carrier on an allogenic bone substitute. Artificial Cells, Nanomedicine and Biotechnology, 2018, 46, 95-107.	1.9	18
77	SPINK1 , PRSS1 , CTRC , and CFTR Genotypes Influence Disease Onset and Clinical Outcomes in Chronic Pancreatitis. Clinical and Translational Gastroenterology, 2018, 9, e204.	1.3	76
78	Novel long-range regulatory mechanisms controlling PKD2 gene expression. BMC Genomics, 2018, 19, 515.	1.2	3
79	Primary sclerosing cholangitis is associated with abnormalities in CFTR. Journal of Cystic Fibrosis, 2018, 17, 666-671.	0.3	11
80	The SLC40A1 R178Q mutation is a recurrent cause of hemochromatosis and is associated with a novel pathogenic mechanism. Haematologica, 2018, 103, 1796-1805.	1.7	19
81	Identification of compound heterozygous variants in the noncoding RNU4ATAC gene in a Chinese family with two successive foetuses with severe microcephaly. Human Genomics, 2018, 12, 3.	1.4	12
82	Novel diagnostic tool for prediction of variant spliceogenicity derived from a set of 395 combined in silico/in vitro studies: an international collaborative effort. Nucleic Acids Research, 2018, 46, 7913-7923.	6.5	71
83	Estimating the age of p.(Phe508del) with family studies of geographically distinct European populations and the early spread of cystic fibrosis. European Journal of Human Genetics, 2018, 26, 1832-1839.	1.4	45
84	Large phenotypic spectrum associated with two new deep intronic variants on the CFTR gene. , 2018, , .		0
85	Calumenin contributes to ER-Ca2+ homeostasis in bronchial epithelial cells expressing WT and F508del mutated CFTR and to F508del-CFTR retention. Cell Calcium, 2017, 62, 47-59.	1.1	11
86	No significant enrichment of rare functionally defective CPA1 variants in a large Chinese idiopathic chronic pancreatitis cohort. Human Mutation, 2017, 38, 959-963.	1.1	19
87	First report of Rh_{null} individuals in the Indian population and characterization of the underlying molecular mechanisms. Transfusion, 2017, 57, 1944-1948.	0.8	5
88	Evaluation of quantitative PCR for early diagnosis of Pseudomonas aeruginosa infection in cystic fibrosis: a prospective cohort study. Clinical Microbiology and Infection, 2017, 23, 203-207.	2.8	27
89	Sex chromosome aneuploidies and copy-number variants: a further explanation for neurodevelopmental prognosis variability?. European Journal of Human Genetics, 2017, 25, 930-934.	1.4	19
90	Identification of a functional enhancer variant within the chronic pancreatitisâ€associatedSPINK1c.101A>G (p.Asn34Ser)â€containing haplotype. Human Mutation, 2017, 38, 1014-1024.	1.1	18

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91	CFTR-France, a national relational patient database for sharing genetic and phenotypic data associated with rareCFTRvariants. Human Mutation, 2017, 38, 1297-1315.	1.1	62
92	Intragenic deletion of the WDR45 gene in a male with encephalopathy, severe psychomotor disability, and epilepsy. American Journal of Medical Genetics, Part A, 2017, 173, 1444-1446.	0.7	10
93	HEMOXCell, a New Oxygen Carrier Usable as an Additive for Mesenchymal Stem Cell Culture in Platelet Lysate-Supplemented Media. Artificial Organs, 2017, 41, 359-371.	1.0	24
94	PKD2 -Related Autosomal Dominant Polycystic Kidney Disease: Prevalence, Clinical Presentation, Mutation Spectrum, andÂPrognosis. American Journal of Kidney Diseases, 2017, 70, 476-485.	2.1	50
95	In vitro and in silico evidence against a significant effect of theSPINK1c.194G>A variant on pre-mRNA splicing. Gut, 2017, 66, 2195-2196.	6.1	12
96	Sequential analysis of 18 genes in polycythemia vera and essential thrombocythemia reveals an association between mutational status and clinical outcome. Genes Chromosomes and Cancer, 2017, 56, 354-362.	1.5	12
97	A genetic overview of Atlantic coastal populations from Europe and North-West Africa based on a 17 X-STR panel. Forensic Science International: Genetics, 2017, 27, 167-171.	1.6	9
98	Severe infantile isolated exocrine pancreatic insufficiency caused by the complete functional loss of theSPINK1gene. Human Mutation, 2017, 38, 1660-1665.	1.1	24
99	Deciphering the molecular basis of ferroportin resistance to hepcidin: Structure/function analysis of rare SLC40A1 missense mutations found in suspected hemochromatosis type 4 patients. Transfusion Clinique Et Biologique, 2017, 24, 462-467.	0.2	4
100	GJB2 mutations: Genotypic and phenotypic correlation in a cohort of 690 hearing-impaired patients, toward a new mutation?. International Journal of Pediatric Otorhinolaryngology, 2017, 102, 80-85.	0.4	10
101	Analysis of the R1b-DF27 haplogroup shows that a large fraction of Iberian Y-chromosome lineages originated recently in situ. Scientific Reports, 2017, 7, 7341.	1.6	27
102	Diagnostic value of targeted nextâ€generation sequencing in suspected hemochromatosis patients with a single copy of the HFE p.Cys282Tyr causative allele. American Journal of Hematology, 2017, 92, E664-E666.	2.0	4
103	NGS and blood group systems: State of the art and perspectives. Transfusion Clinique Et Biologique, 2017, 24, 240-244.	0.2	9
104	From genetic variability to phenotypic expression of blood group systems. Transfusion Clinique Et Biologique, 2017, 24, 472-475.	0.2	8
105	In silico prioritization and further functional characterization of SPINK1 intronic variants. Human Genomics, 2017, 11, 7.	1.4	10
106	GNPAT polymorphism rs11558492 is not associated with increased severity in a large cohort of HFE p.Cys282Tyr homozygous patients. Hepatology, 2017, 65, 1069-1071.	3.6	4
107	Analysis of the Impact of Known SPINK1 Missense Variants on Pre-mRNA Splicing and/or mRNA Stability in a Full-Length Gene Assay. Genes, 2017, 8, 263.	1.0	10
108	In silico search for modifier genes associated with pancreatic and liver disease in Cystic Fibrosis. PLoS ONE, 2017, 12, e0173822.	1.1	14

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109	Buserelin alleviates chloride transport defect in human cystic fibrosis nasal epithelial cells. PLoS ONE, 2017, 12, e0187774.	1.1	1
110	Pathogenetics of Chronic Pancreatitis. , 2017, , 63-77.		0
111	Next generation sequencing with a semi-conductor technology (Ion Torrent PGMâ„¢) for HLA typing: overall workflow performance and debate. Annales De Biologie Clinique, 2016, 74, 449-456.	0.2	3
112	No Association Between CELâ€“HYB Hybrid Allele and Chronic Pancreatitis in Asian Populations. Gastroenterology, 2016, 150, 1558-1560.e5.	0.6	59
113	The experience of extended blood group genotyping by nextâ€generation sequencing (NGS): investigation of patients with sickleâ€cell disease. Vox Sanguinis, 2016, 111, 418-424.	0.7	36
114	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 695-703.	2.6	87
115	The p.Leu96Pro Missense Mutation in the BMP6 Gene Is Repeatedly Associated With Hyperferritinemia in Patients ofÂFrench Origin. Gastroenterology, 2016, 151, 769-770.	0.6	11
116	Discovery and Functional Annotation ofPRSS1Promoter Variants in Chronic Pancreatitis. Human Mutation, 2016, 37, 1149-1152.	1.1	5
117	Mutations in GANAB , Encoding the Glucosidase IIÎ± Subunit, Cause Autosomal-Dominant Polycystic Kidney and Liver Disease. American Journal of Human Genetics, 2016, 98, 1193-1207.	2.6	345
118	Mutational status of synchronous and metachronous tumor samples in patients with metastatic non-small-cell lung cancer. BMC Cancer, 2016, 16, 210.	1.1	26
119	Identification of novel variant A alleles within the ABO gene. Transfusion, 2016, 56, 1244-1246.	0.8	1
120	Analysis of long-range interactions in primary human cells identifies cooperativeCFTRregulatory elements. Nucleic Acids Research, 2016, 44, 2564-2576.	6.5	19
121	Highlighting the impact of cascade carrier testing in cystic fibrosis families. Journal of Cystic Fibrosis, 2016, 15, 452-459.	0.3	8
122	Digging deeper into the intronic sequences of theSPINK1gene: TableÂ1. Gut, 2016, 65, 1055-1056.	6.1	10
123	Function and regulation of TRPM7, as well as intracellular magnesium content, are altered in cells expressing Î”F508-CFTR and G551D-CFTR. Cellular and Molecular Life Sciences, 2016, 73, 3351-3373.	2.4	8
124	Clarifying the clinical relevance ofSPINK1intronic variants in chronic pancreatitis. Gut, 2016, 65, 884-886.	6.1	32
125	New clues to the evolutionary history of the main European paternal lineage M269: dissection of the Y-SNP S116 in Atlantic Europe and Iberia. European Journal of Human Genetics, 2016, 24, 437-441.	1.4	26
126	Comprehensive PKD1 and PKD2 Mutation Analysis in Prenatal Autosomal Dominant Polycystic Kidney Disease. Journal of the American Society of Nephrology: JASN, 2016, 27, 722-729.	3.0	68

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127	The PROPKD Score. Journal of the American Society of Nephrology: JASN, 2016, 27, 942-951.	3.0	245
128	Overrepresentation of Rare CASR Coding Variants in a Sample of Young French Patients With Idiopathic Chronic Pancreatitis. Pancreas, 2015, 44, 996-998.	0.5	15
129	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.	1.1	6
130	G551Dâ€CFTR needs more bound actin than wildâ€type CFTR to maintain its presence in plasma membranes. Cell Biology International, 2015, 39, 978-985.	1.4	7
131	Genetic and Electrophysiological Characteristics of Recurrent Acute Pancreatitis. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, 675-679.	0.9	22
132	Complex Multiple-Nucleotide Substitution Mutations Causing Human Inherited Disease Reveal Novel Insights into the Action of Translesion Synthesis DNA Polymerases. Human Mutation, 2015, 36, 1034-1038.	1.1	12
133	Report of 2 CTRC Intronic Mutations Associated With Acute or Chronic Pancreatitis and Delineation of Their Pathogenic Molecular Mechanisms. Pancreas, 2015, 44, 999-1001.	0.5	2
134	Insights into RHCE Molecular Analysis in Samples with Partial D Variants: the Experience of Western France. Transfusion Medicine and Hemotherapy, 2015, 42, 372-377.	0.7	2
135	Impact of the CFTR-Potentiator Ivacaftor on Airway Microbiota in Cystic Fibrosis Patients Carrying A G551D Mutation. PLoS ONE, 2015, 10, e0124124.	1.1	67
136	Rapid detection of the mature form of cystic fibrosis transmembrane regulator by surface plasmon resonance. Analytical Methods, 2015, 7, 226-236.	1.3	6
137	Concurrent Nucleotide Substitution Mutations in the Human Genome Are Characterized by a Significantly Decreased Transition/Transversion Ratio. Human Mutation, 2015, 36, 333-341.	1.1	9
138	Dual NRASQ61R and BRAFV600E mutation-specific immunohistochemistry completes molecular screening in melanoma samples in a routine practice. Human Pathology, 2015, 46, 1582-1591.	1.1	27
139	Genome-wide association study identifies TF as a significant modifier gene of iron metabolism in HFE hemochromatosis. Journal of Hepatology, 2015, 62, 664-672.	1.8	62
140	Small-scale high-throughput sequencingâ€"based identification of new therapeutic tools in cystic fibrosis. Genetics in Medicine, 2015, 17, 796-806.	1.1	31
141	Advancement in recombinant protein production using a marine oxygen carrier to enhance oxygen transfer in a CHO-S cell line. Artificial Cells, Nanomedicine and Biotechnology, 2015, 43, 186-195.	1.9	13
142	HFE hemochromatosis: influence of dietary iron intake on the iron overload of C282Y homozygous patients. Annals of Hematology, 2015, 94, 1225-1227.	0.8	2
143	A recombined allele of the lipase gene CEL and its pseudogene CELP confers susceptibility to chronic pancreatitis. Nature Genetics, 2015, 47, 518-522.	9.4	157
144	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. Human Genetics, 2015, 134, 589-603.	1.8	25

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145	Extensive functional analyses of RHD splice site variants: Insights into the potential role of splicing in the physiology of Rh. Transfusion, 2015, 55, 1432-1443.	0.8	25
146	Characterization of the second <scp>HFE</scp> gross deletion highlights the potential importance of Aluâ€mediated recombination in haemochromatosis. British Journal of Haematology, 2015, 168, 759-762.	1.2	0
147	Identification of a functionalPRSS1promoter variant in linkage disequilibrium with the chronic pancreatitis-protecting rs10273639. Gut, 2015, 64, 1837-1838.	6.1	35
148	Molecular RHD-RHCE Analysis by Multiplex PCR of Short Fluorescent Fragments. Methods in Molecular Biology, 2015, 1310, 97-104.	0.4	1
149	Polymorphisms atPRSS1â€“PRSS2andCLDN2â€“MORC4loci associate with alcoholic and non-alcoholic chronic pancreatitis in a European replication study. Gut, 2015, 64, 1426-1433.	6.1	105
150	Comprehensive CFTR gene analysis of the French cystic fibrosis screened newborn cohort: implications for diagnosis, genetic counseling, and mutation-specific therapy. Genetics in Medicine, 2015, 17, 108-116.	1.1	37
151	Sequential Analysis By Next Generation Sequencing of 18 Genes in Polycythemia Vera and Essential Thrombocythemia Reveals a Association Between Mutational Status and Clinical Outcome after 3-Year Follow-up. Blood, 2015, 126, 2808-2808.	0.6	0
152	Biophysical Characterisation of Calumenin as a Charged F508del-CFTR Folding Modulator. PLoS ONE, 2014, 9, e104970.	1.1	9
153	Genetics and Pathogenesis of Autosomal Dominant Polycystic Kidney Disease: 20 Years On. Human Mutation, 2014, 35, 1393-1406.	1.1	74
154	Cystic Fibrosis Transmembrane Conductance Regulator Protein (CFTR) Expression in the Developing Human Brain. Journal of Histochemistry and Cytochemistry, 2014, 62, 791-801.	1.3	34
155	Comprehensive functional annotation of 18 missense mutations found in suspected hemochromatosis type 4 patients. Human Molecular Genetics, 2014, 23, 4479-4490.	1.4	46
156	Nextâ€generation sequencing is a credible strategy for blood group genotyping. British Journal of Haematology, 2014, 167, 554-562.	1.2	57
157	A New and More Accurate Estimate of the Rate of Concurrent Tandem-Base Substitution Mutations in the Human Germline: â^¼0.4% of the Single-Nucleotide Substitution Mutation Rate. Human Mutation, 2014, 35, 392-394.	1.1	15
158	Effects of a Novel Archaeal Tetraether-Based Colipid on the In Vivo Gene Transfer Activity of Two Cationic Amphiphiles. Molecular Pharmaceutics, 2014, 11, 2973-2988.	2.3	19
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