Jong Rak Choi

List of Publications by Year
in descending order

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111	1,172	516215	500791
papers	citations	16 h-index	28 g-index

114	114	114	2444
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Ig Gene Clonality Analysis Using Next-Generation Sequencing for Improved Minimal Residual Disease Detection with Significant Prognostic Value in Multiple Myeloma Patients. Journal of Molecular Diagnostics, 2022, 24, 48-56.	1.2	9
2	Concomitant Diagnosis of Primary Bone Marrow B-Cell Non-Hodgkin Lymphoma and Essential Thrombocythemia: A Case Report. Annals of Laboratory Medicine, 2022, 42, 282-285.	1.2	0
3	Generation of a human induced pluripotent stem cell line YCMi004-A from a patient with dilated cardiomyopathy carrying a protein-truncating mutation of the Titin gene and its differentiation towards cardiomyocytes. Stem Cell Research, 2022, 59, 102629.	0.3	2
4	Secondary Germline CDKN2A Mutation Identified using Liquid Biopsy in a Patient with Esophageal Cancer. Laboratory Medicine Online, 2022, 12, 63-67.	0.0	0
5	Derivation of YCMi005-A, a human-induced pluripotent stem cell line, from a patient with dilated cardiomyopathy carrying missense variant in TPM1 (p. Glu192Lys). Stem Cell Research, 2022, 60, 102707.	0.3	1
6	De novo HCN1 Mutation Identified by Next-Generation Sequencing in a Patient with Early Infantile Epileptic Encephalopathy: Case Report. Laboratory Medicine Online, 2022, 12, 134-137.	0.0	2
7	Precision Medicine through Next-Generation Sequencing in Inherited Eye Diseases in a Korean Cohort. Genes, 2022, 13, 27.	1.0	11
8	Amplification of the Chromosomal bla _{CTX-M-14} Gene in Escherichia coli Expanding the Spectrum of Resistance under Antimicrobial Pressure. Microbiology Spectrum, 2022, 10, e0031922.	1.2	5
9	A Single-Center Experience on HLA Typing with 11 Loci Next Generation Sequencing in Korean Patients with Hematologic Disease. Diagnostics, 2022, 12, 1074.	1.3	2
10	Realâ€world data on prognostic value of measurable residual disease assessment by fragment analysis or nextâ€generation sequencing in multiple myeloma. British Journal of Haematology, 2022, , .	1.2	2
11	Cytogenetic testing by fluorescence in situ hybridization is improved by plasma cell sorting in multiple myeloma. Scientific Reports, 2022, 12, 8287.	1.6	2
12	Abstract LB547: Ultra-deep targeted sequencing of circulating tumor DNA with colorectal cancer patients for prediction of treatment response and post-surgical follow-up. Cancer Research, 2022, 82, LB547-LB547.	0.4	0
13	Noncanonical Splice Site and Deep Intronic FRMD7 Variants Activate Cryptic Exons in X-linked Infantile Nystagmus. Translational Vision Science and Technology, 2022, 11, 25.	1.1	1
14	Clinical characteristics of KCNQ2 encephalopathy. Brain and Development, 2021, 43, 244-250.	0.6	18
15	Eif2b3 mutants recapitulate phenotypes of vanishing white matter disease and validate novel disease alleles in zebrafish. Human Molecular Genetics, 2021, 30, 331-342.	1.4	8
16	Chimerism Assay Using Single Nucleotide Polymorphisms Adjacent and in Linkage-Disequilibrium Enables Sensitive Disease Relapse Monitoring after Hematopoietic Stem-Cell Transplantation. Clinical Chemistry, 2021, 67, 781-787.	1.5	3
17	Recurrent somatic mutations and low germline predisposition mutations in Korean ALL patients. Scientific Reports, 2021, 11, 8893.	1.6	4
18	Novel indel mutation in the N gene of SARS-CoV-2 clinical samples that were diagnosed positive in a commercial RT-PCR assay. Virus Research, 2021, 297, 198398.	1.1	8

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19	Disparate treatment outcomes according to presence of pathogenic mutations in West syndrome. Epilepsia, 2021, 62, 1656-1664.	2.6	5
20	Report of the Korean Association of External Quality Assessment Service on Next-Generation Sequencing Analysis for Somatic Variants (2018â€“2020). Journal of Laboratory Medicine and Quality Assurance, 2021, 43, 65-71.	0.1	0
21	In Silico identification of a common mobile element insertion in exon 4 of RP1. Scientific Reports, 2021, 11, 13381.	1.6	2
22	Expanding the Non-Invasive Diagnosis of Acute Rejection in Kidney Transplants Through Detection of Donor-Derived DNA in Urine: Proof-of-Concept Study. Annals of Laboratory Medicine, 2021, 41, 469-478.	1.2	0
23	Establishment of a novel human iPSC line (YCMi003-A) from a patient with dilated cardiomyopathy carrying genetic variant LMNA p.Asp364His. Stem Cell Research, 2021, 56, 102508.	0.3	2
24	Trajectory of genetic alterations associated with colistin resistance in Acinetobacter baumannii during an in-hospital outbreak of infection. Journal of Antimicrobial Chemotherapy, 2021, 77, 69-73.	1.3	6
25	The Role of Ion Channel-Related Genes in Autism Spectrum Disorder: A Study Using Next-Generation Sequencing. Frontiers in Genetics, 2021, 12, 595934.	1.1	4
26	Clinical Implementation of Targeted Gene Sequencing for Malformation of Cortical Development. Pediatric Neurology, 2020, 103, 27-34.	1.0	5
27	Newborn hereditary elliptocytosis confirmed by familial genetic testing. International Journal of Laboratory Hematology, 2020, 42, e20-e22.	0.7	3
28	Detection of recurrent, rare, and novel gene fusions in patients with acute leukemia using nextâ€generation sequencing approaches. Hematological Oncology, 2020, 38, 82-88.	0.8	15
29	Analytical validation of the droplet digital PCR assay for diagnosis of spinal muscular atrophy. Clinica Chimica Acta, 2020, 510, 787-789.	0.5	13
30	Genetic heterogeneity and prognostic impact of recurrent ANK2 and TP53 mutations in mantle cell lymphoma: a multi-centre cohort study. Scientific Reports, 2020, 10, 13359.	1.6	4
31	A Novel KPC Variant KPC-55 in Klebsiella pneumoniae ST307 of Reinforced Meropenem-Hydrolyzing Activity. Frontiers in Microbiology, 2020, 11, 561317.	1.5	6
32	Fusobacterium nucleatum in biopsied tissues from colorectal cancer patients and alcohol consumption in Korea. Scientific Reports, 2020, 10, 19915.	1.6	10
33	Next-Generation Sequencing in Korean Children With Autism Spectrum Disorder and Comorbid Epilepsy. Frontiers in Pharmacology, 2020, 11, 585.	1.6	6
34	Reanalysis of Genomic Sequencing Results in a Clinical Laboratory: Advantages and Limitations. Frontiers in Neurology, 2020, 11, 612.	1.1	10
35	Genetic diagnosis and clinical characteristics by etiological classification in early-onset epileptic encephalopathy with burst suppression pattern. Epilepsy Research, 2020, 163, 106323.	0.8	10
36	Whole exome sequencing identifies mutational signatures of vitreoretinal lymphoma. Haematologica, 2020, 105, e458-460.	1.7	21

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37	Beneficial Chromosomal Integration of the Genes for CTX-M Extended-Spectrum Î²-Lactamase in Klebsiella pneumoniae for Stable Propagation. MSystems, 2020, 5, .	1.7	30
38	The phenotype and treatment of SCN2Aâ€related developmental and epileptic encephalopathy. Epileptic Disorders, 2020, 22, 563-570.	0.7	10
39	MON-195 Genetic Analysis and Clinical Characteristics of Hereditary Paraganglioma and Pheochromocytoma Syndrome in Korean Population. Journal of the Endocrine Society, 2020, 4, .	0.1	0
40	Copy number variations and multiallelic variants in Korean patients with Leber congenital amaurosis. Molecular Vision, 2020, 26, 26-35.	1.1	10
41	Next-generation sequencing with comprehensive bioinformatics analysis facilitates somatic mosaic APC gene mutation detection in patients with familial adenomatous polyposis. BMC Medical Genomics, 2019, 12, 103.	0.7	20
42	Genetic and clinical features of SCN8A developmental and epileptic encephalopathy. Epilepsy Research, 2019, 158, 106222.	0.8	13
43	Clinical utility of targeted NGS panel with comprehensive bioinformatics analysis for patients with acute lymphoblastic leukemia. Leukemia and Lymphoma, 2019, 60, 3138-3145.	0.6	5
44	Somatic mosaic truncating mutations of PPM1D in blood can result from expansion of a mutant clone under selective pressure of chemotherapy. PLoS ONE, 2019, 14, e0217521.	1.1	7
45	Phenotypic and Genotypic Characterization of Acinetobacter spp. Panel Strains: A Cornerstone to Facilitate Antimicrobial Development. Frontiers in Microbiology, 2019, 10, 559.	1.5	15
46	The TECTA mutation R1890C is identified as one of the causes of genetic hearing loss: a case report. BMC Medical Genetics, 2019, 20, 57.	2.1	6
47	Systematic evaluation of gene variants linked to hearing loss based on allele frequency threshold and filtering allele frequency. Scientific Reports, 2019, 9, 4583.	1.6	13
48	Targeted next generation sequencing can serve as an alternative to conventional tests in myeloid neoplasms. PLoS ONE, 2019, 14, e0212228.	1.1	11
49	Proband-Only Clinical Exome Sequencing for Neurodevelopmental Disabilities. Pediatric Neurology, 2019, 99, 47-54.	1.0	19
50	FLT3 Internal Tandem Duplication in Patients With Acute Myeloid Leukemia Is Readily Detectable in a Single Next-Generation Sequencing Assay Using the Pindel Algorithm. Annals of Laboratory Medicine, 2019, 39, 327-329.	1.2	10
51	Clinical Evaluation of Massively Parallel RNA Sequencing for Detecting Recurrent Gene Fusions in Hematologic Malignancies. Journal of Molecular Diagnostics, 2019, 21, 163-170.	1.2	33
52	SNP-based next-generation sequencing reveals low-level mixed chimerism after allogeneic hematopoietic stem cell transplantation. Annals of Hematology, 2018, 97, 1731-1734.	0.8	20
53	Targeted gene panel and genotype-phenotype correlation in children with developmental and epileptic encephalopathy. Epilepsy Research, 2018, 141, 48-55.	0.8	72
54	Efficient strategy for the molecular diagnosis of intractable early-onset epilepsy using targeted gene sequencing. BMC Medical Genomics, 2018, 11, 6.	0.7	55

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55	A patient with B-cell acute lymphoblastic leukemia with PAX5-ETV6 rearrangement with dic(9;12)(p13;p13) identified by chromosomal microarray. Annals of Hematology, 2018, 97, 1505-1507.	0.8	1
56	Application of Multiplex Ligation-Dependent Probe Amplification Assay for Genotyping Major Blood Group Systems Including DEL Variants in the D-Negative Korean Population. Annals of Laboratory Medicine, 2018, 38, 32-38.	1.2	7
57	Deletion of 20p13 and Duplication of 20p13p12.3 in a Patient with Delayed Speech and Development. Annals of Laboratory Medicine, 2018, 38, 77-79.	1.2	1
58	Comprehensive Sequencing Identifies High Frequency of Copy Number Changes in Korean Patients with Acute Lymphoblastic Leukemia. Blood, 2018, 132, 5166-5166.	0.6	0
59	Telomere length in alcohol dependence: A role for impulsive choice and childhood maltreatment. Psychoneuroendocrinology, 2017, 83, 72-78.	1.3	14
60	Accuracy of Next-Generation Sequencing for Molecular Diagnosis in Patients With Infantile Nystagmus Syndrome. JAMA Ophthalmology, 2017, 135, 1376.	1.4	43
61	The effect of trauma and PTSD on telomere length: An exploratory study in people exposed to combat trauma. Scientific Reports, 2017, 7, 4375.	1.6	19
62	Detection of Immunoglobulin Heavy Chain Gene Clonality by Next-Generation Sequencing for Minimal Residual Disease Monitoring in B-Lymphoblastic Leukemia. Annals of Laboratory Medicine, 2017, 37, 331-335.	1.2	17
63	A novel association between relaxin receptor polymorphism and hematopoietic stem cell yield after mobilization. PLoS ONE, 2017, 12, e0179986.	1.1	5
64	Mowat-Wilson syndrome presenting with fever-associated seizures. Epileptic Disorders, 2017, 19, 481-485.	0.7	2
65	Concomitant AID Expression and BCL7A Loss Associates With Accelerated Phase Progression and Imatinib Resistance in Chronic Myeloid Leukemia. Annals of Laboratory Medicine, 2017, 37, 177-179.	1.2	5
66	A Case of Therapy-Related Acute Leukemia With Mixed Phenotype With BCR-ABL1 After Treatment of Diffuse Large B-Cell Lymphoma. Annals of Laboratory Medicine, 2017, 37, 166-168.	1.2	2
67	Effects of Neutralization by Soluble ABH Antigens Produced by Transplanted Kidneys From ABO-Incompatible Secretor Donors. Annals of Laboratory Medicine, 2017, 37, 254-260.	1.2	3
68	Identification of cell morphology parameters from automatic hematology analyzers to predict the peripheral blood CD34-positive cell count after mobilization. PLoS ONE, 2017, 12, e0174286.	1.1	7
69	Validation and optimization of the Ion Torrent S5 XL sequencer and Oncomine workflow for BRCA1 and BRCA2 genetic testing. Oncotarget, 2017, 8, 34858-34866.	0.8	29
70	Diagnostic application of clinical exome sequencing in Leber congenital amaurosis. Molecular Vision, 2017, 23, 649-659.	1.1	13
71	Isolated 9p Duplication With der(Y)t(Y;9)(q12;p13.2) in a Male Patient With Cardiac Defect and Mental Retardation Confirmed by Chromosomal Microarray. Annals of Laboratory Medicine, 2016, 36, 191-193.	1.2	2
72	An 18.3-Mb Duplication on Chromosome 14q With Multiple Cardiac Anomalies and Clubfoot Was Identified by Microarray Analysis. Annals of Laboratory Medicine, 2016, 36, 194-196.	1.2	3

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73	Frequency and Clinical Characteristics of Intrachromosomal Amplification of Chromosome 21 in Korean Childhood B-lineage Acute Lymphoblastic Leukemia. Annals of Laboratory Medicine, 2016, 36, 475-480.	1.2	5
74	Chronic Myeloid Leukemia With Rare Variant b2a3 (e13a3) BCR-ABL1 Fusion. Annals of Laboratory Medicine, 2016, 36, 287-289.	1.2	7
75	Korean Society for Laboratory Medicine Practice Guidelines for the Molecular Diagnosis of Middle East Respiratory Syndrome During an Outbreak in Korea in 2015. Annals of Laboratory Medicine, 2016, 36, 203-208.	1.2	9
76	Development and Comparison of Warfarin Dosing Algorithms in Stroke Patients. Yonsei Medical Journal, 2016, 57, 635.	0.9	13
77	Minor BCR-ABL1-Positive Acute Myeloid Leukemia Associated With the NPM1 Mutation and FLT3 Internal Tandem Duplication. Annals of Laboratory Medicine, 2016, 36, 263-265.	1.2	3
78	Bone Marrow Chimerism Detection Using Next Generation Sequencing Based on Single Nucleotide Polymorphisms Following Liver Transplantation: Comparison With Short Tandem Repeat-PCR. Annals of Laboratory Medicine, 2016, 36, 82-84.	1.2	6
79	t(12;17)(p13;q12)/TAF15-ZNF384 Rearrangement in Acute Lymphoblastic Leukemia. Annals of Laboratory Medicine, 2016, 36, 396-398.	1.2	8
80	Evaluation of an amplicon-based next-generation sequencing panel for detection of BRCA1 and BRCA2 genetic variants. Breast Cancer Research and Treatment, 2016, 158, 433-440.	1.1	4
81	A multicenter phase II study of sorafenib in combination with erlotinib in patients with advanced non-small cell lung cancer (KCSG-0806). Lung Cancer, 2016, 93, 1-8.	0.9	13
82	NOTCH2 missplicing can occur in relation to apoptosis. Blood, 2015, 126, 1731-1732.	0.6	2
83	Birt-Hogg-DubÃ© syndrome incidentally diagnosed during asthma management. Allergy Asthma & Respiratory Disease, 2015, 3, 232.	0.3	1
84	Isodicentric Chromosome 15 Syndrome in a Korean Patient With CafÃ©-au-lait Spots. Annals of Laboratory Medicine, 2015, 35, 474-476.	1.2	0
85	Isochromosome 1q in Childhood Burkitt Lymphoma: The First Reported Case in Korea. Annals of Laboratory Medicine, 2015, 35, 663-665.	1.2	1
86	Routine Chromosomal Microarray Analysis is Necessary in Korean Patients With Unexplained Developmental Delay/Mental Retardation/Autism Spectrum Disorder. Annals of Laboratory Medicine, 2015, 35, 510-518.	1.2	16
87	Breakpoint mapping by whole genome sequencing identifiesPTH2Rgene disruption in a patient with midline craniosynostosis and a de novo balanced chromosomal rearrangement. Journal of Medical Genetics, 2015, 52, 706-709.	1.5	10
88	Clinical Implication of Highly Sensitive Detection of the BRAFV600E Mutation in Fine-Needle Aspirations According to the Thyroid Bethesda System in Patients With Conventional Papillary Thyroid Carcinoma. Annals of Otology, Rhinology and Laryngology, 2015, 124, 392-399.	0.6	12
89	Cys482Trp Missense Mutation in the Coagulation Factor XI Gene (F11) in a Korean Patient with Factor XI Deficiency. Annals of Laboratory Medicine, 2014, 34, 332-335.	1.2	1
90	Bone marrow hypoplasia, isochromosome 8q and deletion of chromosome 6q preceding B-cell lymphoma. Blood Research, 2014, 49, 200.	0.5	1

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91	Study of peripheral BRAFV600Emutation as a possible novel marker for papillary thyroid carcinomas. Head and Neck, 2013, 35, 1630-1633.	0.9	26
92	Application of BRAF, NRAS, KRAS mutations as markers for the detection of papillary thyroid cancer from FNAB specimens by pyrosequencing analysis. Clinical Chemistry and Laboratory Medicine, 2013, 51, 1673-1680.	1.4	30
93	Proper Indication of BRAFV600E Mutation Testing in Fine-Needle Aspirates of Thyroid Nodules. PLoS ONE, 2013, 8, e64505.	1.1	23
94	Difference in serum nephrin expression between normal and preeclamptic pregnancies: A preliminary study. Korean Journal of Obstetrics & Gynecology, 2012, 55, 546.	0.1	0
95	Evaluation of an Automated Coagulation Analyzer Coapresta 2000. Laboratory Medicine Online, 2011, 1, 94.	0.0	1
96	Diagnostic Value of BRAFV600E Mutation Analysis of Thyroid Nodules According to Ultrasonographic Features and the Time of Aspiration. Annals of Surgical Oncology, 2011, 18, 792-799.	0.7	22
97	Dual priming oligonucleotideâ€“based multiplex PCR analysis for detection of BRAF^{V600E} mutation in FNAB samples of thyroid nodules in BRAF^{V600E} mutationâ€“prevalent area. Head and Neck, 2010, 32, 490-498.	0.9	53
98	A novel t(1;12)(q21;q24) in a patient with myelodysplastic syndrome. Annals of Hematology, 2010, 89, 513-516.	0.8	0
99	Association of BRAF^{V600E}Mutation with Poor Clinical Prognostic Factors and US Features in Korean Patients with Papillary Thyroid Microcarcinoma. Radiology, 2009, 253, 854-860.	3.6	117
100	Therapy-related myelodysplastic syndrome with der(17)t(12;17)(q13;p13) as a new recurrent cytogenetic abnormality after treatment for chronic lymphocytic leukemia. Leukemia Research, 2009, 33, 1001-1004.	0.4	11
101	Non-age related Y chromosome loss in an elderly patient with acute promyelocytic leukemia. Leukemia Research, 2009, 33, e114-e115.	0.4	3
102	der(1)t(1;19)(p13;p13.1) in two elderly patients with myeloid neoplasms: New case reports and review of the literature. Leukemia Research, 2009, 33, e128-e131.	0.4	3
103	Acute erythroleukemia with der(1;7)(q10;p10) as a sole acquired abnormality after treatment with azathioprine. Cancer Genetics and Cytogenetics, 2008, 186, 58-60.	1.0	9
104	Effectiveness of Real-Time Quantitative PCR Compare to Repeat PCR for the Diagnosis of Charcot-Marie-Tooth Type 1A and Hereditary Neuropathy with Liability to Pressure Palsies. Yonsei Medical Journal, 2005, 46, 347.	0.9	11
105	Clinical Meaning of INNO-LiPA Test in the Diagnosis of Rifampin Resistant Tuberculosis. Tuberculosis and Respiratory Diseases, 2003, 55, 344.	0.2	0
106	A Case of del(13)(q22) with Multiple Major Congenital Anomalies, Imperforate Anus and Penoscrotal Transposition. Yonsei Medical Journal, 2001, 42, 558.	0.9	12
107	A Novel Silent Substitution (C8516T) in Exon 9 of the Human PROC Gene. Yonsei Medical Journal, 2001, 42, 364.	0.9	2
108	Tissue plasminogen activator and plasminogen activator inhibitor-1 in human choledochal bile. Yonsei Medical Journal, 2000, 41, 119.	0.9	2

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109	A case of Klinefelter syndrome with retroperitoneal teratoma. Yonsei Medical Journal, 2000, 41, 136.	0.9	7
110	von Willebrand disease with G4022A mutation (vWd Sungnam): a case report. Journal of Korean Medical Science, 1999, 14, 93.	1.1	0
111	Diagnostic Efficacy of Plasma Urokinase-type Plasminogen Activator and Plasminogen Activator Inhibitor-2 in Differentiation of Hepatocellular Carcinoma from Cirrhosis. Thrombosis and Haemostasis, 1995, 74, 864-867.	1.8	6