

Jong Rak Choi

List of Publications by Year in descending order

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111
papers

1,172
citations

516215

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500791

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114
all docs

114
docs citations

114
times ranked

2444
citing authors

#	ARTICLE	IF	CITATIONS
1	Association of BRAF ^{V600E} Mutation with Poor Clinical Prognostic Factors and US Features in Korean Patients with Papillary Thyroid Microcarcinoma. <i>Radiology</i> , 2009, 253, 854-860.	3.6	117
2	Targeted gene panel and genotype-phenotype correlation in children with developmental and epileptic encephalopathy. <i>Epilepsy Research</i> , 2018, 141, 48-55.	0.8	72
3	Efficient strategy for the molecular diagnosis of intractable early-onset epilepsy using targeted gene sequencing. <i>BMC Medical Genomics</i> , 2018, 11, 6.	0.7	55
4	Dual priming oligonucleotide-based multiplex PCR analysis for detection of BRAF ^{V600E} mutation in FNAB samples of thyroid nodules in BRAF ^{V600E} mutation-prevalent area. <i>Head and Neck</i> , 2010, 32, 490-498.	0.9	53
5	Accuracy of Next-Generation Sequencing for Molecular Diagnosis in Patients With Infantile Nystagmus Syndrome. <i>JAMA Ophthalmology</i> , 2017, 135, 1376.	1.4	43
6	Clinical Evaluation of Massively Parallel RNA Sequencing for Detecting Recurrent Gene Fusions in Hematologic Malignancies. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 163-170.	1.2	33
7	Application of <i>BRAF</i> , <i>NRAS</i> , <i>KRAS</i> mutations as markers for the detection of papillary thyroid cancer from FNAB specimens by pyrosequencing analysis. <i>Clinical Chemistry and Laboratory Medicine</i> , 2013, 51, 1673-1680.	1.4	30
8	Beneficial Chromosomal Integration of the Genes for CTX-M Extended-Spectrum β -Lactamase in <i>Klebsiella pneumoniae</i> for Stable Propagation. <i>MSystems</i> , 2020, 5, .	1.7	30
9	Validation and optimization of the Ion Torrent S5 XL sequencer and OncoPrint workflow for <i>BRCA1</i> and <i>BRCA2</i> genetic testing. <i>Oncotarget</i> , 2017, 8, 34858-34866.	0.8	29
10	Study of peripheral BRAFV600E mutation as a possible novel marker for papillary thyroid carcinomas. <i>Head and Neck</i> , 2013, 35, 1630-1633.	0.9	26
11	Proper Indication of BRAFV600E Mutation Testing in Fine-Needle Aspirates of Thyroid Nodules. <i>PLoS ONE</i> , 2013, 8, e64505.	1.1	23
12	Diagnostic Value of BRAFV600E Mutation Analysis of Thyroid Nodules According to Ultrasonographic Features and the Time of Aspiration. <i>Annals of Surgical Oncology</i> , 2011, 18, 792-799.	0.7	22
13	Whole exome sequencing identifies mutational signatures of vitreoretinal lymphoma. <i>Haematologica</i> , 2020, 105, e458-460.	1.7	21
14	SNP-based next-generation sequencing reveals low-level mixed chimerism after allogeneic hematopoietic stem cell transplantation. <i>Annals of Hematology</i> , 2018, 97, 1731-1734.	0.8	20
15	Next-generation sequencing with comprehensive bioinformatics analysis facilitates somatic mosaic APC gene mutation detection in patients with familial adenomatous polyposis. <i>BMC Medical Genomics</i> , 2019, 12, 103.	0.7	20
16	The effect of trauma and PTSD on telomere length: An exploratory study in people exposed to combat trauma. <i>Scientific Reports</i> , 2017, 7, 4375.	1.6	19
17	Proband-Only Clinical Exome Sequencing for Neurodevelopmental Disabilities. <i>Pediatric Neurology</i> , 2019, 99, 47-54.	1.0	19
18	Clinical characteristics of KCNQ2 encephalopathy. <i>Brain and Development</i> , 2021, 43, 244-250.	0.6	18

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19	Detection of Immunoglobulin Heavy Chain Gene Clonality by Next-Generation Sequencing for Minimal Residual Disease Monitoring in B-Lymphoblastic Leukemia. <i>Annals of Laboratory Medicine</i> , 2017, 37, 331-335.	1.2	17
20	Routine Chromosomal Microarray Analysis is Necessary in Korean Patients With Unexplained Developmental Delay/Mental Retardation/Autism Spectrum Disorder. <i>Annals of Laboratory Medicine</i> , 2015, 35, 510-518.	1.2	16
21	Phenotypic and Genotypic Characterization of <i>Acinetobacter</i> spp. Panel Strains: A Cornerstone to Facilitate Antimicrobial Development. <i>Frontiers in Microbiology</i> , 2019, 10, 559.	1.5	15
22	Detection of recurrent, rare, and novel gene fusions in patients with acute leukemia using next-generation sequencing approaches. <i>Hematological Oncology</i> , 2020, 38, 82-88.	0.8	15
23	Telomere length in alcohol dependence: A role for impulsive choice and childhood maltreatment. <i>Psychoneuroendocrinology</i> , 2017, 83, 72-78.	1.3	14
24	Development and Comparison of Warfarin Dosing Algorithms in Stroke Patients. <i>Yonsei Medical Journal</i> , 2016, 57, 635.	0.9	13
25	A multicenter phase II study of sorafenib in combination with erlotinib in patients with advanced non-small cell lung cancer (KCSG-0806). <i>Lung Cancer</i> , 2016, 93, 1-8.	0.9	13
26	Genetic and clinical features of SCN8A developmental and epileptic encephalopathy. <i>Epilepsy Research</i> , 2019, 158, 106222.	0.8	13
27	Systematic evaluation of gene variants linked to hearing loss based on allele frequency threshold and filtering allele frequency. <i>Scientific Reports</i> , 2019, 9, 4583.	1.6	13
28	Analytical validation of the droplet digital PCR assay for diagnosis of spinal muscular atrophy. <i>Clinica Chimica Acta</i> , 2020, 510, 787-789.	0.5	13
29	Diagnostic application of clinical exome sequencing in Leber congenital amaurosis. <i>Molecular Vision</i> , 2017, 23, 649-659.	1.1	13
30	A Case of del(13)(q22) with Multiple Major Congenital Anomalies, Imperforate Anus and Penoscrotal Transposition. <i>Yonsei Medical Journal</i> , 2001, 42, 558.	0.9	12
31	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. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 392-399.	0.6	12
32	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. <i>Yonsei Medical Journal</i> , 2005, 46, 347.	0.9	11
33	Therapy-related myelodysplastic syndrome with der(17)t(12;17)(q13;p13) as a new recurrent cytogenetic abnormality after treatment for chronic lymphocytic leukemia. <i>Leukemia Research</i> , 2009, 33, 1001-1004.	0.4	11
34	Targeted next generation sequencing can serve as an alternative to conventional tests in myeloid neoplasms. <i>PLoS ONE</i> , 2019, 14, e0212228.	1.1	11
35	Precision Medicine through Next-Generation Sequencing in Inherited Eye Diseases in a Korean Cohort. <i>Genes</i> , 2022, 13, 27.	1.0	11
36	Breakpoint mapping by whole genome sequencing identifies PTH2R gene disruption in a patient with midline craniosynostosis and a de novo balanced chromosomal rearrangement. <i>Journal of Medical Genetics</i> , 2015, 52, 706-709.	1.5	10

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37	<i>FLT3</i> Internal Tandem Duplication in Patients With Acute Myeloid Leukemia Is Readily Detectable in a Single Next-Generation Sequencing Assay Using the Pindel Algorithm. <i>Annals of Laboratory Medicine</i> , 2019, 39, 327-329.	1.2	10
38	<i>Fusobacterium nucleatum</i> in biopsied tissues from colorectal cancer patients and alcohol consumption in Korea. <i>Scientific Reports</i> , 2020, 10, 19915.	1.6	10
39	Reanalysis of Genomic Sequencing Results in a Clinical Laboratory: Advantages and Limitations. <i>Frontiers in Neurology</i> , 2020, 11, 612.	1.1	10
40	Genetic diagnosis and clinical characteristics by etiological classification in early-onset epileptic encephalopathy with burst suppression pattern. <i>Epilepsy Research</i> , 2020, 163, 106323.	0.8	10
41	The phenotype and treatment of <i>SCN2A</i> -related developmental and epileptic encephalopathy. <i>Epileptic Disorders</i> , 2020, 22, 563-570.	0.7	10
42	Copy number variations and multiallelic variants in Korean patients with Leber congenital amaurosis. <i>Molecular Vision</i> , 2020, 26, 26-35.	1.1	10
43	Acute erythroleukemia with der(1;7)(q10;p10) as a sole acquired abnormality after treatment with azathioprine. <i>Cancer Genetics and Cytogenetics</i> , 2008, 186, 58-60.	1.0	9
44	Korean Society for Laboratory Medicine Practice Guidelines for the Molecular Diagnosis of Middle East Respiratory Syndrome During an Outbreak in Korea in 2015. <i>Annals of Laboratory Medicine</i> , 2016, 36, 203-208.	1.2	9
45	Ig Gene Clonality Analysis Using Next-Generation Sequencing for Improved Minimal Residual Disease Detection with Significant Prognostic Value in Multiple Myeloma Patients. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 48-56.	1.2	9
46	t(12;17)(p13;q12)/ <i>TAF15-ZNF384</i> Rearrangement in Acute Lymphoblastic Leukemia. <i>Annals of Laboratory Medicine</i> , 2016, 36, 396-398.	1.2	8
47	Eif2b3 mutants recapitulate phenotypes of vanishing white matter disease and validate novel disease alleles in zebrafish. <i>Human Molecular Genetics</i> , 2021, 30, 331-342.	1.4	8
48	Novel indel mutation in the N gene of SARS-CoV-2 clinical samples that were diagnosed positive in a commercial RT-PCR assay. <i>Virus Research</i> , 2021, 297, 198398.	1.1	8
49	A case of Klinefelter syndrome with retroperitoneal teratoma. <i>Yonsei Medical Journal</i> , 2000, 41, 136.	0.9	7
50	Chronic Myeloid Leukemia With Rare Variant b2a3 (e13a3) <i>BCR-ABL1</i> Fusion. <i>Annals of Laboratory Medicine</i> , 2016, 36, 287-289.	1.2	7
51	Application of Multiplex Ligation-Dependent Probe Amplification Assay for Genotyping Major Blood Group Systems Including DEL Variants in the D-Negative Korean Population. <i>Annals of Laboratory Medicine</i> , 2018, 38, 32-38.	1.2	7
52	Somatic mosaic truncating mutations of PPM1D in blood can result from expansion of a mutant clone under selective pressure of chemotherapy. <i>PLoS ONE</i> , 2019, 14, e0217521.	1.1	7
53	Identification of cell morphology parameters from automatic hematology analyzers to predict the peripheral blood CD34-positive cell count after mobilization. <i>PLoS ONE</i> , 2017, 12, e0174286.	1.1	7
54	Bone Marrow Chimerism Detection Using Next Generation Sequencing Based on Single Nucleotide Polymorphisms Following Liver Transplantation: Comparison With Short Tandem Repeat-PCR. <i>Annals of Laboratory Medicine</i> , 2016, 36, 82-84.	1.2	6

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55	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
56	A Novel KPC Variant KPC-55 in Klebsiella pneumoniae ST307 of Reinforced Meropenem-Hydrolyzing Activity. Frontiers in Microbiology, 2020, 11, 561317.	1.5	6
57	Next-Generation Sequencing in Korean Children With Autism Spectrum Disorder and Comorbid Epilepsy. Frontiers in Pharmacology, 2020, 11, 585.	1.6	6
58	Trajectory of genetic alterations associated with colistin resistance in <i>Acinetobacter baumannii</i> during an in-hospital outbreak of infection. Journal of Antimicrobial Chemotherapy, 2021, 77, 69-73.	1.3	6
59	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
60	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
61	A novel association between relaxin receptor polymorphism and hematopoietic stem cell yield after mobilization. PLoS ONE, 2017, 12, e0179986.	1.1	5
62	Concomitant <i>AID</i> Expression and <i>BCL7A</i> Loss Associates With Accelerated Phase Progression and Imatinib Resistance in Chronic Myeloid Leukemia. Annals of Laboratory Medicine, 2017, 37, 177-179.	1.2	5
63	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
64	Clinical Implementation of Targeted Gene Sequencing for Malformation of Cortical Development. Pediatric Neurology, 2020, 103, 27-34.	1.0	5
65	Disparate treatment outcomes according to presence of pathogenic mutations in West syndrome. Epilepsia, 2021, 62, 1656-1664.	2.6	5
66	Amplification of the Chromosomal <i>bla</i> _{CTX-M-14} Gene in Escherichia coli Expanding the Spectrum of Resistance under Antimicrobial Pressure. Microbiology Spectrum, 2022, 10, e0031922.	1.2	5
67	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
68	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
69	Recurrent somatic mutations and low germline predisposition mutations in Korean ALL patients. Scientific Reports, 2021, 11, 8893.	1.6	4
70	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
71	Non-age related Y chromosome loss in an elderly patient with acute promyelocytic leukemia. Leukemia Research, 2009, 33, e114-e115.	0.4	3
72	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

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73	An 18.3-Mb Duplication on Chromosome 14q With Multiple Cardiac Anomalies and Clubfoot Was Identified by Microarray Analysis. <i>Annals of Laboratory Medicine</i> , 2016, 36, 194-196.	1.2	3
74	Minor <i>BCR-ABL1</i> -Positive Acute Myeloid Leukemia Associated With the <i>NPM1</i> Mutation and <i>FLT3</i> Internal Tandem Duplication. <i>Annals of Laboratory Medicine</i> , 2016, 36, 263-265.	1.2	3
75	Effects of Neutralization by Soluble ABH Antigens Produced by Transplanted Kidneys From ABO-Incompatible Secretor Donors. <i>Annals of Laboratory Medicine</i> , 2017, 37, 254-260.	1.2	3
76	Newborn hereditary elliptocytosis confirmed by familial genetic testing. <i>International Journal of Laboratory Hematology</i> , 2020, 42, e20-e22.	0.7	3
77	Chimerism Assay Using Single Nucleotide Polymorphisms Adjacent and in Linkage-Disequilibrium Enables Sensitive Disease Relapse Monitoring after Hematopoietic Stem-Cell Transplantation. <i>Clinical Chemistry</i> , 2021, 67, 781-787.	1.5	3
78	Tissue plasminogen activator and plasminogen activator inhibitor-1 in human choledochal bile. <i>Yonsei Medical Journal</i> , 2000, 41, 119.	0.9	2
79	A Novel Silent Substitution (C8516T) in Exon 9 of the Human PROC Gene. <i>Yonsei Medical Journal</i> , 2001, 42, 364.	0.9	2
80	NOTCH2 missplicing can occur in relation to apoptosis. <i>Blood</i> , 2015, 126, 1731-1732.	0.6	2
81	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. <i>Annals of Laboratory Medicine</i> , 2016, 36, 191-193.	1.2	2
82	Mowat-Wilson syndrome presenting with fever-associated seizures. <i>Epileptic Disorders</i> , 2017, 19, 481-485.	0.7	2
83	A Case of Therapy-Related Acute Leukemia With Mixed Phenotype With <i>BCR-ABL1</i> After Treatment of Diffuse Large B-Cell Lymphoma. <i>Annals of Laboratory Medicine</i> , 2017, 37, 166-168.	1.2	2
84	In Silico identification of a common mobile element insertion in exon 4 of RP1. <i>Scientific Reports</i> , 2021, 11, 13381.	1.6	2
85	Establishment of a novel human iPSC line (YCMi003-A) from a patient with dilated cardiomyopathy carrying genetic variant LMNA p.Asp364His. <i>Stem Cell Research</i> , 2021, 56, 102508.	0.3	2
86	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. <i>Stem Cell Research</i> , 2022, 59, 102629.	0.3	2
87	<i>De novo</i> <i>HCN1</i> Mutation Identified by Next-Generation Sequencing in a Patient with Early Infantile Epileptic Encephalopathy: Case Report. <i>Laboratory Medicine Online</i> , 2022, 12, 134-137.	0.0	2
88	A Single-Center Experience on HLA Typing with 11 Loci Next Generation Sequencing in Korean Patients with Hematologic Disease. <i>Diagnostics</i> , 2022, 12, 1074.	1.3	2
89	Real-world data on prognostic value of measurable residual disease assessment by fragment analysis or next-generation sequencing in multiple myeloma. <i>British Journal of Haematology</i> , 2022, , .	1.2	2
90	Cytogenetic testing by fluorescence in situ hybridization is improved by plasma cell sorting in multiple myeloma. <i>Scientific Reports</i> , 2022, 12, 8287.	1.6	2

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91	Evaluation of an Automated Coagulation Analyzer Coapresta 2000. <i>Laboratory Medicine Online</i> , 2011, 1, 94.	0.0	1
92	Cys482Trp Missense Mutation in the Coagulation Factor XI Gene (F11) in a Korean Patient with Factor XI Deficiency. <i>Annals of Laboratory Medicine</i> , 2014, 34, 332-335.	1.2	1
93	Bone marrow hypoplasia, isochromosome 8q and deletion of chromosome 6q preceding B-cell lymphoma. <i>Blood Research</i> , 2014, 49, 200.	0.5	1
94	Birt-Hogg-Dub� syndrome incidentally diagnosed during asthma management. <i>Allergy Asthma & Respiratory Disease</i> , 2015, 3, 232.	0.3	1
95	Isochromosome 1q in Childhood Burkitt Lymphoma: The First Reported Case in Korea. <i>Annals of Laboratory Medicine</i> , 2015, 35, 663-665.	1.2	1
96	A patient with B-cell acute lymphoblastic leukemia with PAX5-ETV6 rearrangement with dic(9;12)(p13;p13) identified by chromosomal microarray. <i>Annals of Hematology</i> , 2018, 97, 1505-1507.	0.8	1
97	Deletion of 20p13 and Duplication of 20p13p12.3 in a Patient with Delayed Speech and Development. <i>Annals of Laboratory Medicine</i> , 2018, 38, 77-79.	1.2	1
98	Derivation of YCMi005-A, a human-induced pluripotent stem cell line, from a patient with dilated cardiomyopathy carrying missense variant in TPM1 (p. Glu192Lys). <i>Stem Cell Research</i> , 2022, 60, 102707.	0.3	1
99	Noncanonical Splice Site and Deep Intronic <i>FRMD7</i> Variants Activate Cryptic Exons in X-linked Infantile Nystagmus. <i>Translational Vision Science and Technology</i> , 2022, 11, 25.	1.1	1
100	von Willebrand disease with G4022A mutation (vWd Sungnam): a case report. <i>Journal of Korean Medical Science</i> , 1999, 14, 93.	1.1	0
101	A novel t(1;12)(q21;q24) in a patient with myelodysplastic syndrome. <i>Annals of Hematology</i> , 2010, 89, 513-516.	0.8	0
102	Isodicentric Chromosome 15 Syndrome in a Korean Patient With Caf�-au-lait Spots. <i>Annals of Laboratory Medicine</i> , 2015, 35, 474-476.	1.2	0
103	Report of the Korean Association of External Quality Assessment Service on Next-Generation Sequencing Analysis for Somatic Variants (2018�2020). <i>Journal of Laboratory Medicine and Quality Assurance</i> , 2021, 43, 65-71.	0.1	0
104	Expanding the Non-Invasive Diagnosis of Acute Rejection in Kidney Transplants Through Detection of Donor-Derived DNA in Urine: Proof-of-Concept Study. <i>Annals of Laboratory Medicine</i> , 2021, 41, 469-478.	1.2	0
105	Clinical Meaning of INNO-LiPA Test in the Diagnosis of Rifampin Resistant Tuberculosis. <i>Tuberculosis and Respiratory Diseases</i> , 2003, 55, 344.	0.2	0
106	Difference in serum nephrin expression between normal and preeclamptic pregnancies: A preliminary study. <i>Korean Journal of Obstetrics & Gynecology</i> , 2012, 55, 546.	0.1	0
107	Comprehensive Sequencing Identifies High Frequency of Copy Number Changes in Korean Patients with Acute Lymphoblastic Leukemia. <i>Blood</i> , 2018, 132, 5166-5166.	0.6	0
108	MON-195 Genetic Analysis and Clinical Characteristics of Hereditary Paraganglioma and Pheochromocytoma Syndrome in Korean Population. <i>Journal of the Endocrine Society</i> , 2020, 4, .	0.1	0

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109	Concomitant Diagnosis of Primary Bone Marrow B-Cell Non-Hodgkin Lymphoma and Essential Thrombocythemia: A Case Report. <i>Annals of Laboratory Medicine</i> , 2022, 42, 282-285.	1.2	0
110	Secondary Germline CDKN2A Mutation Identified using Liquid Biopsy in a Patient with Esophageal Cancer. <i>Laboratory Medicine Online</i> , 2022, 12, 63-67.	0.0	0
111	Abstract LB547: Ultra-deep targeted sequencing of circulating tumor DNA with colorectal cancer patients for prediction of treatment response and post-surgical follow-up. <i>Cancer Research</i> , 2022, 82, LB547-LB547.	0.4	0