Jong-Won Kim

List of Publications by Year
in descending order

118 papers	2,126 citations	331670 21 h-index	289244 40 g-index
124 all docs	124 docs citations	124 times ranked	5280 citing authors

#	Article	IF	Citations
1	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases. JAMA Oncology, 2017, 3, 636.	7.1	376
2	Mutations in DDX58, which Encodes RIG-I, Cause Atypical Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 266-274.	6.2	169
3	SORL1 Is Genetically Associated with Late-Onset Alzheimerâ€™s Disease in Japanese, Koreans and Caucasians. PLoS ONE, 2013, 8, e58618.	2.5	149
4	Video laryngoscopy vs. direct laryngoscopy: Which should be chosen for endotracheal intubation during cardiopulmonary resuscitation? A prospective randomized controlled study of experienced intubators. Resuscitation, 2016, 105, 196-202.	3.0	62
5	DUOX2 Mutations Are Frequently Associated With Congenital Hypothyroidism in the Korean Population. Annals of Laboratory Medicine, 2016, 36, 145-153.	2.5	60
6	Video laryngoscopy improves the first-attempt success in endotracheal intubation during cardiopulmonary resuscitation among novice physicians. Resuscitation, 2015, 89, 188-194.	3.0	57
7	Analysis of intrapatient heterogeneity uncovers the microevolution of Middle East respiratory syndrome coronavirus. Journal of Physical Education and Sports Management, 2016, 2, a001214.	1.2	48
8	Lack of association of the interleukin-1? gene polymorphism with Alzheimer's disease in a Korean population. Annals of Neurology, 2001, 49, 817-818.	5.3	42
9	NRAMP1gene polymorphisms in patients with rheumatoid arthritis in Koreans. Journal of Korean Medical Science, 2000, 15, 83.	2.5	39
10	Large-scale clinical validation of biomarkers for pancreatic cancer using a mass spectrometry-based proteomics approach. Oncotarget, 2017, 8, 42761-42771.	1.8	34
11	Mutation analysis of the GNE gene in Korean patients with distal myopathy with rimmed vacuoles. Journal of Human Genetics, 2006, 51, 137-140.	2.3	33
12	How much experience do rescuers require to achieve successful tracheal intubation during cardiopulmonary resuscitation?. Resuscitation, 2018, 133, 187-192.	3.0	33
13	Mutation analysis of PAH gene and characterization of a recurrent deletion mutation in Korean patients with phenylketonuria. Experimental and Molecular Medicine, 2008, 40, 533.	7.7	32
14	No association between the genes for butyrylcholinesterase K variant and apolipoprotein E4 in late-onset Alzheimer's disease. , 1999, 88, 113-115.		29
15	A genome-wide association study identifies novel loci associated with susceptibility to chronic myeloid leukemia. Blood, 2011, 117, 6906-6911.	1.4	28
16	SNP Linkage Analysis and Whole Exome Sequencing Identify a Novel POU4F3 Mutation in Autosomal Dominant Late-Onset Nonsyndromic Hearing Loss (DFNA15). PLoS ONE, 2013, 8, e79063.	2.5	28
17	Preliminary Report of Multisession Gamma Knife Radiosurgery for Benign Perioptic Lesions: Visual Outcome in 22 Patients. Journal of Korean Neurosurgical Society, 2008, 44, 67.	1.2	28
18	Linkage and association of schizophrenia with genetic variations in the locus of neuregulin 1 in Korean population. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 281-286.	1.7	26

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19	Dried Blood Spot Testing for Seven Steroids Using Liquid Chromatography-Tandem Mass Spectrometry With Reference Interval Determination in the Korean Population. Annals of Laboratory Medicine, 2015, 35, 578-585.	2.5	26
20	Principles of Genetic Counseling in the Era of Next-Generation Sequencing. Annals of Laboratory Medicine, 2018, 38, 291-295.	2.5	25
21	Performance of Confirmatory Interferon-Î³ Release Assays in School TB Outbreaks. Chest, 2012, 141, 983-988.	0.8	23
22	Triage of patients with AUS / FLUS on thyroid cytopathology: effectiveness of the multimodal diagnostic techniques. Cancer Medicine, 2016, 5, 769-777.	2.8	22
23	Urinary neutrophil gelatinase-associated lipocalin as a biomarker of acute kidney injury in sepsis patients in the emergency department. Clinica Chimica Acta, 2019, 495, 552-555.	1.1	22
24	Performance Evaluation of SpliceAI for the Prediction of Splicing of NF1 Variants. Genes, 2021, 12, 1308.	2.4	22
25	Mutation profiling of 19 candidate genes in acute myeloid leukemia suggests significance of DNMT3A mutations. Oncotarget, 2016, 7, 54825-54837.	1.8	22
26	Improved Detection of Germline Mutations in Korean VHL Patients by Multiple Ligation-dependent Probe Amplification Analysis. Journal of Korean Medical Science, 2009, 24, 77.	2.5	21
27	Performance Evaluation of Allplex Respiratory Panels 1, 2, and 3 for Detection of Respiratory Viruses and Influenza A Virus Subtypes. Journal of Clinical Microbiology, 2017, 55, 479-484.	3.9	21
28	Clinical and genetic analysis of HLXB9 gene in Korean patients with Currarino syndrome. Journal of Human Genetics, 2007, 52, 698-701.	2.3	20
29	EnsemPro: An ensemble approach to predicting transcription start sites in human genomic DNA sequences. Genomics, 2008, 91, 259-266.	2.9	20
30	NOTCH3 variants in patients with subcortical vascular cognitive impairment: a comparison with typical CADASIL patients. Neurobiology of Aging, 2015, 36, 2443.e1-2443.e7.	3.1	20
31	Nonsyndromic cleft lip with or without cleft palate and cancer: Evaluation of a possible common genetic background through the analysis of GWAS data. Genomics Data, 2016, 10, 22-29.	1.3	19
32	Menopausal hormone therapy and mild cognitive impairment: a randomized, placebo-controlled trial. Menopause, 2018, 25, 870-876.	2.0	18
33	Genetic Prediction of Antidepressant Drug Response and Nonresponse in Korean Patients. PLoS ONE, 2014, 9, e107098.	2.5	17
34	The First Korean Family With Hereditary Gelsolin Amyloidosis Caused by p.D214Y Mutation in the GSN Gene. Annals of Laboratory Medicine, 2016, 36, 259-262.	2.5	16
35	Comparative analysis of BRCA1 and BRCA2 variants of uncertain significance in patients with breast cancer: a multifactorial probability-based model versus ACMG standards and guidelines for interpreting sequence variants. Genetics in Medicine, 2016, 18, 1250-1257.	2.4	16
36	PHKA2 mutation spectrum in Korean patients with glycogen storage disease type IX: prevalence of deletion mutations. BMC Medical Genetics, 2016, 17, 33.	2.1	16

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37	Suggestion of BRCA1 c.5339T>C (p.L1780P) variant confer from â€˜unknown significanceâ€™ to â€˜Likely pathogenicâ€™ based on clinical evidence in Korea. Breast, 2017, 33, 109-116.	2.2	16
38	Identification of novel PKD1 and PKD2 mutations in Korean patients with autosomal dominant polycystic kidney disease. BMC Medical Genetics, 2014, 15, 129.	2.1	15
39	Prevalence and oncologic outcomes of BRCA 1/2 mutations in unselected triple-negative breast cancer patients in Korea. Breast Cancer Research and Treatment, 2019, 173, 385-395.	2.5	15
40	Clinical Practice Guidelines for Pre-Analytical Procedures of Plasma Epidermal Growth Factor Receptor Variant Testing. Annals of Laboratory Medicine, 2022, 42, 141-149.	2.5	15
41	Clinical, biochemical and molecular characterization of Korean patients with mucolipidosis II/III and successful prenatal diagnosis. Orphanet Journal of Rare Diseases, 2017, 12, 11.	2.7	14
42	HMGCLL1 is a predictive biomarker for deep molecular response to imatinib therapy in chronic myeloid leukemia. Leukemia, 2019, 33, 1439-1450.	7.2	14
43	Presymptomatic Identification of CDH1 Germline Mutation in a Healthy Korean Individual with Family History of Gastric Cancer. Annals of Laboratory Medicine, 2014, 34, 386-389.	2.5	13
44	Application of whole exome sequencing to a rare inherited metabolic disease with neurological and gastrointestinal manifestations: A congenital disorder of glycosylation mimicking glycogen storage disease. Clinica Chimica Acta, 2015, 444, 50-53.	1.1	13
45	Improvement in Traineesâ€™ Attitude and Resuscitation Quality With Repeated Cardiopulmonary Resuscitation Training. Simulation in Healthcare, 2016, 11, 250-256.	1.2	13
46	Novel Alzheimerâ€™s disease risk variants identified based on whole-genome sequencing of APOE Îµ4 carriers. Translational Psychiatry, 2021, 11, 296.	4.8	13
47	Genomeâ€widely significant evidence of linkage of schizophrenia to chromosomes 2p24.3 and 6q27 in an SNPâ€Based analysis of Korean families. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 647-652.	1.7	12
48	Comparison of the AdvanSureâ„¢ real-time RT-PCR and SeeplexÂ® RV12 ACE assay for the detection of respiratory viruses. Journal of Virological Methods, 2015, 224, 42-46.	2.1	12
49	A novel SMAD6 variant in a patient with severely calcified bicuspid aortic valve and thoracic aortic aneurysm. Molecular Genetics & Genomic Medicine, 2019, 7, e620.	1.2	12
50	Cataloging Coding Sequence Variations in Human Genome Databases. PLoS ONE, 2008, 3, e3575.	2.5	12
51	Comparison of the AnyplexTM II RV16 and SeeplexÂ® RV12 ACE assays for the detection of respiratory viruses. Diagnostic Microbiology and Infectious Disease, 2014, 79, 419-421.	1.8	11
52	Evaluation of the Real-Q BRAF V600E Detection Assay in Fine-Needle Aspiration Samples of Thyroid Nodules. Journal of Molecular Diagnostics, 2015, 17, 431-437.	2.8	11
53	Detection of MYD88 L265P in patients with lymphoplasmacytic lymphoma/Waldenstrom macroglobulinemia and other B-cell non-Hodgkin lymphomas. Blood Research, 2016, 51, 181.	1.3	11
54	DeviCNV: detection and visualization of exon-level copy number variants in targeted next-generation sequencing data. BMC Bioinformatics, 2018, 19, 381.	2.6	11

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55	Genotype-Related Clinical Characteristics and Myocardial Fibrosis and Their Association with Prognosis in Hypertrophic Cardiomyopathy. Journal of Clinical Medicine, 2020, 9, 1671.	2.4	11
56	Comparison of the PowerChek SARS-CoV-2, Influenza A&B, RSV Multiplex Real-time PCR Kit and BioFire Respiratory Panel 2.1 for simultaneous detection of SARS-CoV-2, influenza A and B, and respiratory syncytial virus. Journal of Virological Methods, 2021, 298, 114304.	2.1	11
57	Identification and characterization of NF1 splicing mutations in Korean patients with neurofibromatosis type 1. Journal of Human Genetics, 2016, 61, 705-709.	2.3	10
58	Multifocality in a Patient with Cribriformâ€“Morular Variant of Papillary Thyroid Carcinoma Is an Important Clue for the Diagnosis of Familial Adenomatous Polyposis. Thyroid, 2019, 29, 1606-1614.	4.5	10
59	Red cell distribution width as a novel marker for predicting high-risk from upper gastro-intestinal bleeding patients. PLoS ONE, 2017, 12, e0187158.	2.5	9
60	A New Integrated Newborn Screening Workflow Can Provide a Shortcut to Differential Diagnosis and Confirmation of Inherited Metabolic Diseases. Yonsei Medical Journal, 2018, 59, 652.	2.2	9
61	Heritability estimates of individual psychological distress symptoms from genetic variation. Journal of Affective Disorders, 2019, 252, 413-420.	4.1	9
62	Statins Enhance the Molecular Response in Chronic Myeloid Leukemia when Combined with Tyrosine Kinase Inhibitors. Cancers, 2021, 13, 5543.	3.7	9
63	High frequency ofJAK2exon 12 mutations in Korean patients with polycythaemia vera: novel mutations and clinical significance. Journal of Clinical Pathology, 2016, 69, 737-741.	2.0	8
64	Serum Lactate Could Predict Mortality in Patients With Spontaneous Subarachnoid Hemorrhage in the Emergency Department. Frontiers in Neurology, 2020, 11, 975.	2.4	7
65	Distribution and clinical impact of apolipoprotein E4 in subjective memory impairment andÂearly mild cognitive impairment. Scientific Reports, 2020, 10, 13365.	3.3	7
66	Functional and Structural Changes in the Membrane-Bound O-Acyltransferase Family Member 7 (MBOAT7) Protein: The Pathomechanism of a Novel MBOAT7 Variant in Patients With Intellectual Disability. Frontiers in Neurology, 2022, 13, 836954.	2.4	7
67	BRAFV600E andMAP2K1Mutations in Hairy Cell Leukemia and Splenic Marginal Zone Lymphoma Cases. Annals of Laboratory Medicine, 2015, 35, 257-259.	2.5	6
68	Evaluation of the iNtRON VRE vanA/vanB Real-Time PCR Assay for Detection of Vancomycin-Resistant Enterococci. Annals of Laboratory Medicine, 2015, 35, 76-81.	2.5	6
69	Performance Evaluation of the Real-Q Cytomegalovirus (CMV) Quantification Kit Using Two Real-Time PCR Systems for Quantifying CMV DNA in Whole Blood. Annals of Laboratory Medicine, 2016, 36, 603-606.	2.5	6
70	Korean Society for Genetic Diagnostics Guidelines for Validation of Next-Generation Sequencing-Based Somatic Variant Detection in Hematologic Malignancies. Annals of Laboratory Medicine, 2019, 39, 515-523.	2.5	6
71	Analytical Validation of a Pan-Cancer Panel for Cell-Free Assay for the Detection of EGFR Mutations. Diagnostics, 2021, 11, 1022.	2.6	6
72	Current Status of Clinical Diagnosis and Genetic Analysis of Hereditary Hemorrhagic Telangiectasia in South Korea: Multicenter Case Series and a Systematic Review. Neurointervention, 2019, 14, 91-98.	0.8	6

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73	Prevalence and clinical characterization of BRCA1 and BRCA2 mutations in Korean patients with epithelial ovarian cancer. Cancer Science, 2021, 112, 5055-5067.	3.9	6
74	A Simple and Rapid Method Based on Liquid Chromatographyâ€“Tandem Mass Spectrometry for the Measurement of Î±-L-Iduronidase Activity in Dried Blood Spots: An Application to Mucopolysaccharidosis I (Hurler) Screening. Annals of Laboratory Medicine, 2015, 35, 41-49.	2.5	5
75	Clinical Characteristics and Genotype-Phenotype Correlation of Korean Patients with Spinal and Bulbar Muscular Atrophy. Yonsei Medical Journal, 2015, 56, 993.	2.2	5
76	Performance of the Real-Q EBV Quantification Kit for Epstein-Barr Virus DNA Quantification in Whole Blood. Annals of Laboratory Medicine, 2017, 37, 147-150.	2.5	5
77	Nextâ€generation sequencing reveals unique combination of mutations in cis of CSF3R in atypical chronic myeloid leukemia. Journal of Clinical Laboratory Analysis, 2020, 34, e23064.	2.1	5
78	Performance Evaluation of the PowerChek SARS-CoV-2, Influenza A & B Multiplex Real-Time PCR Kit in Comparison with the BioFire Respiratory Panel. Annals of Laboratory Medicine, 2022, 42, 473-477.	2.5	5
79	Challenges in assessing pathogenicity based on frequency of variants in mismatch repair genes: an extreme case of a MSH2 variant and a meta-analysis. Gene, 2014, 546, 421-424.	2.2	4
80	Safe azathioprine treatment in a pediatric ulcerative colitis patient with TPMT*16 by thiopurine metabolite monitoring. Clinica Chimica Acta, 2014, 437, 101-102.	1.1	4
81	CDH1 mutations in gastric cancers are not associated with family history. Pathology Research and Practice, 2020, 216, 152941.	2.3	4
82	A Population-Based Analysis of BRCA1/2 Genes and Associated Breast and Ovarian Cancer Risk in Korean Patients: A Multicenter Cohort Study. Cancers, 2021, 13, 2192.	3.7	4
83	Direct-to-consumer genetic testing. Genomics and Informatics, 2019, 17, e34.	0.8	4
84	Genetic Counseling and Long-Term Surveillance Using a Multidisciplinary Approach in von Hippelâ€“Lindau Disease. Annals of Laboratory Medicine, 2022, 42, 352-357.	2.5	4
85	Two novel FAH gene mutations in a patient with hereditary tyrosinemia type I. Annals of Clinical and Laboratory Science, 2014, 44, 317-23.	0.2	4
86	Discrepancy in Genotyping of Apolipoprotein E between Allele-Specific PCR and Fluorescence Resonance Energy Transfer or Sequencing. Annals of Laboratory Medicine, 2010, 30, 325-328.	2.5	3
87	One case of endometrial cancer occurrence: Over 10 years after colon cancer in Lynch family. Obstetrics and Gynecology Science, 2013, 56, 408.	1.6	3
88	Genetic Counseling Status and Perspectives Based on a 2018 Professional Survey in Korea. Annals of Laboratory Medicine, 2020, 40, 232-237.	2.5	3
89	Quantitative and Qualitative QC of Next-Generation Sequencing for Detecting Somatic Variants: An Example of Detecting Clonal Hematopoiesis of Indeterminate Potential. Clinical Chemistry, 2020, 66, 832-841.	3.2	3
90	Whole-genome sequencing reveals KRTAP1-1 as a novel genetic variant associated with antidepressant treatment outcomes. Scientific Reports, 2021, 11, 4552.	3.3	3

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91	Linkage and Association Analyses of Schizophrenia with Genetic Variations on Chromosome 22q11 in Koreans. Psychiatry Investigation, 2016, 13, 630.	1.6	3
92	Clinical Utility of Plasma Cell-Free DNA EGFR Mutation Analysis in Treatment-NaÃ¯ve Stage IV Non-Small Cell Lung Cancer Patients. Journal of Clinical Medicine, 2022, 11, 1144.	2.4	3
93	Clinical application of catalytically cleavable fluorescence probe technology for multiplexing quantification of BCRâ€“ABL1 fusion transcripts. Clinica Chimica Acta, 2014, 428, 72-76.	1.1	2
94	Mutant Enrichment with 3â€²â€Modified Oligonucleotides (MEMO)â€Quantitative PCR for Detection of NPM1 Mutations in Acute Myeloid Leukemia. Journal of Clinical Laboratory Analysis, 2015, 29, 361-365.	2.1	2
95	Clinical severity of viral myocarditis is not associated with a mutation of dystrophin gene cleavage sites. International Journal of Cardiology, 2015, 194, 21-22.	1.7	2
96	Complete STK11 Deletion and Atypical Symptoms in Peutz-Jeghers Syndrome. Annals of Laboratory Medicine, 2017, 37, 462-464.	2.5	2
97	Status of BRCA1/2 Genetic Testing Practices in Korea (2014). Laboratory Medicine Online, 2018, 8, 107.	0.2	2
98	Local Laboratory Testing of Germline BRCA Mutations vs. Myriad: A Single-Institution Experience in Korea. Diagnostics, 2021, 11, 370.	2.6	2
99	Clinicopathological Characterization of Double Heterozygosity for BRCA1 and BRCA2 Variants in Korean Breast Cancer Patients. Cancer Research and Treatment, 2022, 54, 827-833.	3.0	2
100	Detection Methods and Status of CAT Interruption of ATXN1 in Korean Patients With Spinocerebellar Ataxia Type 1. Annals of Laboratory Medicine, 2022, 42, 274-277.	2.5	2
101	Necessity of Multiplex Ligation Probe Amplification in Genetic Tests: Germline variant analysis of the APC gene in familial adenomatous polyposis patients. Cancer Genetics, 2022, 262-263, 95-101.	0.4	2
102	The role of pleural fluid MAGE RTâ€nested PCR in the diagnosis of malignant pleural effusion. Thoracic Cancer, 2012, 3, 320-325.	1.9	1
103	Analytical and clinical evaluation of the Abbott RealTime hepatitis B sequencing assay. Journal of Clinical Virology, 2016, 85, 27-30.	3.1	1
104	Evidence that 6q25.1 variant rs6931104 confers susceptibility to chronic myeloid leukemia through RMND1 regulation. PLoS ONE, 2019, 14, e0218968.	2.5	1
105	Psychological effects and risk perception after genetic counseling. Journal of Genetic Medicine, 2021, 18, 38-43.	0.2	1
106	The First Korean Case of SLC12A3 Aberrant Skipping of Two Exons Detected by RNA Splicing Analysis. Case Reports in Nephrology and Dialysis, 2021, 11, 210-213.	0.6	1
107	The First Korean Case of Gorlinâ€“Goltz Syndrome Caused by a PTCH2 Pathogenic Variant Identified via Whole Exome Sequencing. Laboratory Medicine Online, 2020, 10, 175.	0.2	1
108	Regulated sandbox and disease-related genetic tests as a direct-to-consumer test. Journal of the Korean Medical Association, 2019, 62, 240.	0.3	1

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109	Intravascular Stent Graft with Polyurethane and Metallic Stent: Experimental Study. Journal of the Korean Radiological Society, 1997, 36, 955.	0.0	1
110	A Case of Next-generation Sequencing Gene Testing: Points to be Considered in Testing and Reporting. Annals of Laboratory Medicine, 2022, 42, 296-297.	2.5	1
111	Variation spectrum of MECP2 in Korean patients with Rett and Rett-like syndrome: a literature review and reevaluation of variants based on the ClinGen guideline. Journal of Human Genetics, 0, , .	2.3	1
112	P3-100: The distribution and clinical impact of apolipoprotein e4 among patients with subjective memory impairment and early mild cognitive impairment. , 2015, 11, P658-P658.		0
113	Genetic Characteristics of Polycythemia Vera and Essential Thrombocythemia in Korean Patients. Journal of Clinical Laboratory Analysis, 2016, 30, 1061-1070.	2.1	0
114	Reply to Letter: Comparing direct and video laryngoscopy for urgent intubation during chest compression. Resuscitation, 2017, 111, e3.	3.0	0
115	A novel type of +2-base pair frameshift CALR mutation in a patient with myeloproliferative neoplasm. European Journal of Haematology, 2018, 101, 216-219.	2.2	0
116	Diagnostic Classification and Genomic Analyses of Cancer. Laboratory Medicine Online, 2021, 11, 223-229.	0.2	0
117	Genome-Wide Single-Nucleotide Polymorphism-Array Based Karyotyping Detects Clonal Aberrations, and Predicts the Risk of Imatinib Failure In Chronic Myeloid Leukemia.. Blood, 2010, 116, 3387-3387.	1.4	0
118	Importance of family segregation in the American College of Medical Genetics and Genomics and Association of Molecular Pathology guidelines: Case of a Korean family with autosomal dominant polycystic disease. Journal of Genetic Medicine, 2020, 17, 51-54.	0.2	0