Seung-Tae Lee

List of Publications by Citations

157 papers	1,769 citations	22 h-index	36 g-index
174 ext. papers	2,292 ext. citations	3.9 avg, IF	4.7 L-index

#	Paper	IF	Citations
157	Whole-genome fingerprint of the DNA methylome during human B cell differentiation. *Nature Genetics*, **2015**, 47, 746-56	36.3	209
156	Guidelines for Laboratory Diagnosis of Coronavirus Disease 2019 (COVID-19) in Korea. *Annals of Laboratory Medicine*, **2020**, 40, 351-360	3.1	199
155	A global DNA methylation and gene expression analysis of early human B-cell development reveals a demethylation signature and transcription factor network. *Nucleic Acids Research*, **2012**, 40, 11339-51	20.1	79
154	Clinical implication of highly sensitive detection of the BRAF V600E mutation in fine-needle aspirations of thyroid nodules: a comparative analysis of three molecular assays in 4585 consecutive cases in a BRAF V600E mutation-prevalent area. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, 2299-306	5.6	78
153	Targeted gene panel and genotype-phenotype correlation in children with developmental and epileptic encephalopathy. *Epilepsy Research*, **2018**, 141, 48-55	3	49
152	Association study of 27 annotated genes for clozapine pharmacogenetics: validation of preexisting studies and identification of a new candidate gene, ABCB1, for treatment response. *Journal of Clinical Psychopharmacology*, **2012**, 32, 441-8	1.7	48
151	Genome-wide CpG island methylation and intergenic demethylation propensities vary among different tumor sites. *Nucleic Acids Research*, **2016**, 44, 1105-17	20.1	34
150	Distinct frequencies and mutation spectrums of genetic thrombophilia in Korea in comparison with other Asian countries both in patients with thromboembolism and in the general population. *Haematologica*, **2014**, 99, 561-9	6.6	33
149	Periconceptional folate consumption is associated with neonatal DNA methylation modifications in neural crest regulatory and cancer development genes. *Epigenetics*, **2015**, 10, 1166-76	5.7	30
148	Efficient strategy for the molecular diagnosis of intractable early-onset epilepsy using targeted gene sequencing. *BMC Medical Genomics*, **2018**, 11, 6	3.7	29
147	Mutant enrichment with 3'-modified oligonucleotides a practical PCR method for detecting trace mutant DNAs. *Journal of Molecular Diagnostics*, **2011**, 13, 657-68	5.1	29
146	Association of a single nucleotide polymorphism near the interleukin-28B gene with response to hepatitis C therapy in Asian patients. *Journal of Gastroenterology and Hepatology (Australia)*, **2011**, 26, 1374-9	4	28
145	Effects of 7 hemoglobin variants on the measurement of glycohemoglobin by 14 analytical methods. *Clinical Chemistry*, **2007**, 53, 2202-5	5.5	28
144	The Efficacy of Ketogenic Diet for Specific Genetic Mutation in Developmental and Epileptic Encephalopathy. *Frontiers in Neurology*, **2018**, 9, 530	4.1	27
143	Accuracy of Next-Generation Sequencing for Molecular Diagnosis in Patients With Infantile Nystagmus Syndrome. *JAMA Ophthalmology*, **2017**, 135, 1376-1385	3.9	27
142	Molecular screening of the TSH receptor (TSHR) and thyroid peroxidase (TPO) genes in Korean patients with nonsyndromic congenital hypothyroidism. *Clinical Endocrinology*, **2011**, 75, 715-21	3.4	27
141	Validation and optimization of the Ion Torrent S5 XL sequencer and Oncomine workflow for BRCA1 and BRCA2 genetic testing. *Oncotarget*, **2017**, 8, 34858-34866	3.3	27

140	Epigenetic remodeling in B-cell acute lymphoblastic leukemia occurs in two tracks and employs embryonic stem cell-like signatures. *Nucleic Acids Research*, **2015**, 43, 2590-602	20.1	26
139	Genetic contribution to variation in DNA methylation at maternal smoking-sensitive loci in exposed neonates. *Epigenetics*, **2016**, 11, 664-673	5.7	23
138	Clinical Evaluation of Massively Parallel RNA Sequencing for Detecting Recurrent Gene Fusions in Hematologic Malignancies. *Journal of Molecular Diagnostics*, **2019**, 21, 163-170	5.1	23
137	Determination of pharmacokinetic properties of clozapine and norclozapine in Korean schizophrenia patients. *International Clinical Psychopharmacology*, **2009**, 24, 139-44	2.2	22
136	Detection of Germline Mutations in Patients with Epithelial Ovarian Cancer Using Multi-gene Panels: Beyond BRCA1/2. *Cancer Research and Treatment*, **2018**, 50, 917-925	5.2	22
135	Mutation analysis of PAH gene and characterization of a recurrent deletion mutation in Korean patients with phenylketonuria. *Experimental and Molecular Medicine*, **2008**, 40, 533-40	12.8	21
134	Clinical and genetic analysis of Korean patients with congenital insensitivity to pain with anhidrosis. *Muscle and Nerve*, **2009**, 40, 855-9	3.4	20
133	Identification of a Novel BRCA1 Pathogenic Mutation in Korean Patients Following Reclassification of BRCA1 and BRCA2 Variants According to the ACMG Standards and Guidelines Using Relevant Ethnic Controls. *Cancer Research and Treatment*, **2017**, 49, 1012-1021	5.2	20
132	Exploring the Prevalence of Clozapine Phenotypic Poor Metabolizers in 4 Asian Samples: They Ranged Between 2% and 13. *Journal of Clinical Psychopharmacology*, **2019**, 39, 644-648	1.7	18
131	Mutation profiling of 19 candidate genes in acute myeloid leukemia suggests significance of DNMT3A mutations. *Oncotarget*, **2016**, 7, 54825-54837	3.3	17
130	Variants of cancer susceptibility genes in Korean BRCA1/2 mutation-negative patients with high risk for hereditary breast cancer. *BMC Cancer*, **2018**, 18, 83	4.8	16
129	The effect of trauma and PTSD on telomere length: An exploratory study in people exposed to combat trauma. *Scientific Reports*, **2017**, 7, 4375	4.9	16
128	Multiplex ligation-dependent probe amplification screening of isolated increased HbF levels revealed three cases of novel rearrangements/deletions in the beta-globin gene cluster. *British Journal of Haematology*, **2010**, 148, 154-60	4.5	16
127	Identification of an Arg35X mutation in the PDCD10 gene in a patient with cerebral and multiple spinal cavernous malformations. *Journal of the Neurological Sciences*, **2008**, 267, 177-81	3.2	16
126	Effect of mesenchymal stem cell transplantation on the engraftment of human hematopoietic stem cells and leukemic cells in mice model. *International Journal of Hematology*, **2008**, 87, 327-37	2.3	16
125	Comparison of Clinical Outcomes of Pathologic Mutation, Variants of Unknown Significance, or Wild Type Epithelial Ovarian Cancer Patients. *Cancer Research and Treatment*, **2017**, 49, 408-415	5.2	16
124	Efficacy of Stiripentol in Dravet Syndrome with or without SCN1A Mutations. *Journal of Clinical Neurology (Korea*, **2018**, 14, 22-28	1.7	14
123	Targeted gene panel sequencing in early infantile onset developmental and epileptic encephalopathy. *Brain and Development*, **2020**, 42, 438-448	2.2	12

122	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	3.7	12
121	Bone marrow flow cytometry in staging of patients with B-cell non-Hodgkin lymphoma. *Annals of Laboratory Medicine*, **2015**, 35, 187-93	3.1	12
120	Diagnostic application of clinical exome sequencing in Leber congenital amaurosis. *Molecular Vision* , **2017**, 23, 649-659	2.3	12
119	Interaction of allergy history and antibodies to specific varicella-zoster virus proteins on glioma risk. *International Journal of Cancer*, **2014**, 134, 2199-210	7.5	11
118	Sequential array comparative genomic hybridization analysis identifies copy number changes during blastic transformation of chronic myeloid leukemia. *Leukemia Research*, **2012**, 36, 418-21	2.7	11
117	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	3.1	11
116	PTPRG inhibition by DNA methylation and cooperation with RAS gene activation in childhood acute lymphoblastic leukemia. *International Journal of Cancer*, **2014**, 135, 1101-9	7.5	11
115	Identification of a rare 3 bp BRAF gene deletion in a thyroid nodule by mutant enrichment with 3'-modified oligonucleotides polymerase chain reaction. *Annals of Laboratory Medicine*, **2012**, 32, 238-41	3.1	11
114	The proliferative history shapes the DNA methylome of B-cell tumors and predicts clinical outcome. *Nature Cancer*, **2020**, 1, 1066-1081	15.4	11
113	BRCA1 and BRCA2 mutation predictions using the BRCAPRO and Myriad models in Korean ovarian cancer patients. *Gynecologic Oncology*, **2017**, 145, 137-141	4.9	10
112	Telomere length in alcohol dependence: A role for impulsive choice and childhood maltreatment. *Psychoneuroendocrinology*, **2017**, 83, 72-78	5	10
111	SLC38A8 mutations result in arrested retinal development with loss of cone photoreceptor specialization. *Human Molecular Genetics*, **2020**, 29, 2989-3002	5.6	10
110	Systematic evaluation of gene variants linked to hearing loss based on allele frequency threshold and filtering allele frequency. *Scientific Reports*, **2019**, 9, 4583	4.9	9
109	SNP-based next-generation sequencing reveals low-level mixed chimerism after allogeneic hematopoietic stem cell transplantation. *Annals of Hematology*, **2018**, 97, 1731-1734	3	9
108	Prevalence and clinical implications of germline predisposition gene mutations in patients with acute myeloid leukemia. *Scientific Reports*, **2020**, 10, 14297	4.9	9
107	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	3.1	9
106	Targeted next generation sequencing can serve as an alternative to conventional tests in myeloid neoplasms. *PLoS ONE*, **2019**, 14, e0212228	3.7	8
105	Whole exome sequencing identifies mutational signatures of vitreoretinal lymphoma. *Haematologica*, **2020**, 105, e458-460	6.6	8

104	Genetic and clinical features of SCN8A developmental and epileptic encephalopathy. *Epilepsy Research*, **2019**, 158, 106222	3	8
103	Two cases of acute lymphoblastic leukemia with an e1a3 BCR-ABL1 fusion transcript. *Annals of Laboratory Medicine*, **2015**, 35, 159-61	3.1	8
102	Clinical features and mutations in the ENG, ACVRL1, and SMAD4 genes in Korean patients with hereditary hemorrhagic telangiectasia. *Journal of Korean Medical Science*, **2009**, 24, 69-76	4.7	8
101	Copy number variations and multiallelic variants in Korean patients with Leber congenital amaurosis. *Molecular Vision*, **2020**, 26, 26-35	2.3	8
100	Detection of recurrent, rare, and novel gene fusions in patients with acute leukemia using next-generation sequencing approaches. *Hematological Oncology*, **2020**, 38, 82-88	1.3	8
99	Detection of MYD88 L265P in patients with lymphoplasmacytic lymphoma/Waldenstrom macroglobulinemia and other B-cell non-Hodgkin lymphomas. *Blood Research*, **2016**, 51, 181-186	1.4	8
98	Molecular characterization and clinical impact of t(11;15)(q23;q14-15) MLL-CASC5 rearrangement. *Haematologica*, **2014**, 99, e11-3	6.6	7
97	A novel COL3A1 gene mutation in patient with aortic dissected aneurysm and cervical artery dissections. *Heart and Vessels*, **2008**, 23, 144-8	2.1	7
96	Chronic Myeloid Leukemia With Rare Variant b2a3 (e13a3) BCR-ABL1 Fusion. *Annals of Laboratory Medicine*, **2016**, 36, 287-9	3.1	7
95	t(12;17)(p13;q12)/TAF15-ZNF384 Rearrangement in Acute Lymphoblastic Leukemia. *Annals of Laboratory Medicine*, **2016**, 36, 396-8	3.1	7
94	Clinical characteristics of KCNQ2 encephalopathy. *Brain and Development*, **2021**, 43, 244-250	2.2	7
93	Targeted panel sequencing identifies a novel mutations in a patient with Bosch-Boonstra-Schaaf optic atrophy syndrome. *Ophthalmic Genetics*, **2019**, 40, 359-361	1.2	6
92	A cryptic ETV6/ABL1 rearrangement represents a unique fluorescence in situ hybridization signal pattern in a patient with B acute lymphoblastic leukemia. *Annals of Laboratory Medicine*, **2014**, 34, 475-7	3.1	6
91	Thalidomide, cyclophosphamide and dexamethasone induction therapy: feasibility for myeloma patients destined for autologous stem cell transplantation. *Acta Haematologica*, **2014**, 132, 226-32	2.7	6
90	Analytical validation of the droplet digital PCR assay for diagnosis of spinal muscular atrophy. *Clinica Chimica Acta*, **2020**, 510, 787-789	6.2	6
89	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	3.7	5
88	Proband-Only Clinical Exome Sequencing for Neurodevelopmental Disabilities. *Pediatric Neurology*, **2019**, 99, 47-54	2.9	5
87	Antigen Expression Patterns of Plasma Cell Myeloma: An Association of Cytogenetic Abnormality and International Staging System (ISS) for Myeloma. *Journal of Clinical Laboratory Analysis*, **2015**, 29, 505-10	3	5

86	Genetic diagnosis and clinical characteristics by etiological classification in early-onset epileptic encephalopathy with burst suppression pattern. *Epilepsy Research*, **2020**, 163, 106323	3	5
85	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	3.1	5
84	Molecular characterization and clinical course of MLL-ACTN4 rearrangement in therapy-related hematologic malignancies. *Haematologica*, **2014**, 99, e49-51	6.6	5
83	BRAF V600E and MAP2K1 mutations in hairy cell leukemia and splenic marginal zone lymphoma cases. *Annals of Laboratory Medicine*, **2015**, 35, 257-9	3.1	5
82	Acute lymphoblastic leukemia with mature B-cell phenotype and t(9;11;11)(p22;q23;p11.2): a case study and literature review. *Annals of Laboratory Medicine*, **2014**, 34, 166-9	3.1	5
81	Serial Detection of L265P Mutation in the Aqueous Humor of a Patient with Vitreoretinal Lymphoma for Disease Monitoring. *Ocular Immunology and Inflammation*, **2021**, 29, 485-489	2.8	5
80	Fusobacterium nucleatum in biopsied tissues from colorectal cancer patients and alcohol consumption in Korea. *Scientific Reports*, **2020**, 10, 19915	4.9	5
79	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	3.1	5
78	Clinical utility of targeted NGS panel with comprehensive bioinformatics analysis for patients with acute lymphoblastic leukemia. *Leukemia and Lymphoma*, **2019**, 60, 3138-3145	1.9	4
77	Genetic Analysis and Clinical Characteristics of Hereditary Pheochromocytoma and Paraganglioma Syndrome in Korean Population. *Endocrinology and Metabolism*, **2020**, 35, 858-872	3.5	4
76	Usefulness of Flow Cytometric Analysis for Detecting Leptomeningeal Diseases in Non-Hodgkin Lymphoma. *Annals of Laboratory Medicine*, **2016**, 36, 209-14	3.1	4
75	Frequency and Clinical Characteristics of Intrachromosomal Amplification of Chromosome 21 in Korean Childhood B-lineage Acute Lymphoblastic Leukemia. *Annals of Laboratory Medicine*, **2016**, 36, 475-80	3.1	4
74	Evaluation of an amplicon-based next-generation sequencing panel for detection of BRCA1 and BRCA2 genetic variants. *Breast Cancer Research and Treatment*, **2016**, 158, 433-40	4.4	4
73	A subset of CD45+/CD19 - cells in bone marrow may be associated with clinical outcomes of patients with mantle cell lymphoma. *Leukemia and Lymphoma*, **2015**, 56, 3052-7	1.9	3
72	Next-Generation Sequencing in Korean Children With Autism Spectrum Disorder and Comorbid Epilepsy. *Frontiers in Pharmacology*, **2020**, 11, 585	5.6	3
71	Difference in Risk of Breast and Ovarian Cancer According to Putative Functional Domain Regions in Korean BRCA1/2 Mutation Carriers. *Clinical Breast Cancer*, **2018**, 18, 362-373.e1	3	3
70	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, 421-4	3.8	3
69	First report on familial hemophagocytic lymphohistiocytosis with an abnormal immunophenotype and T cell monoclonality in Korea. *Annals of Laboratory Medicine*, **2015**, 35, 155-8	3.1	3

68	Identification of Mixed Lineage Leukemia Gene (MLL)/MLLT10 Fusion Transcripts by Reverse Transcription-PCR and Sequencing in a Case of AML With a FISH-Negative Cryptic MLL Rearrangement. *Annals of Laboratory Medicine*, **2015**, 35, 469-71	3.1	3
67	Discrepancy in genotyping of apolipoprotein E between allele-specific PCR and fluorescence resonance energy transfer or sequencing. *Annals of Laboratory Medicine*, **2010**, 30, 325-8	3.1	3
66	Measurement of tyrosine hydroxylase transcripts in bone marrow using biopsied tissue instead of aspirates for neuroblastoma. *Pediatric Blood and Cancer*, **2010**, 55, 273-8	3	3
65	Clinical Implications of Genetic Testing for Hereditary Breast and Ovarian Cancer Syndrome in the Era of Genomic Medicine: Clinicians' Perspectives. *Journal of Breast Disease*, **2016**, 4, 1-9	0.3	3
64	The phenotype and treatment of SCN2A-related developmental and epileptic encephalopathy. *Epileptic Disorders*, **2020**, 22, 563-570	1.9	3
63	Trends in contralateral prophylactic mastectomy rate according to clinicopathologic and socioeconomic status. *Annals of Surgical Treatment and Research*, **2019**, 97, 113-118	2	3
62	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, 194-6	3.1	3
61	Distinct Clinical Courses of Epithelial Ovarian Cancer with Mutations in 5' and 3' Exons. *Anticancer Research*, **2018**, 38, 6947-6953	2.3	3
60	Two cases of in-frame deletion mutation in juxtamembrane domain of the FLT3. *Leukemia and Lymphoma*, **2016**, 57, 486-488	1.9	2
59	Missed Heterozygous Deletion in Study of Next-Generation Sequencing for Molecular Diagnosis in Patients With Infantile Nystagmus Syndrome. *JAMA Ophthalmology*, **2019**, 137, 1465-1466	3.9	2
58	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	2
57	A Somatic p.Phe29del Mutation of Connexin 26 (GJB2) Manifesting as Acantholytic Dyskeratotic Epidermal Nevus. *JAMA Dermatology*, **2019**, 155, 633-635	5.1	2
56	Mutant Enrichment with 3'-Modified Oligonucleotides (MEMO)-Quantitative PCR for Detection of NPM1 Mutations in Acute Myeloid Leukemia. *Journal of Clinical Laboratory Analysis*, **2015**, 29, 361-5	3	2
55	Reanalysis of Genomic Sequencing Results in a Clinical Laboratory: Advantages and Limitations. *Frontiers in Neurology*, **2020**, 11, 612	4.1	2
54	Mowat-Wilson syndrome presenting with fever-associated seizures. *Epileptic Disorders*, **2017**, 19, 481-485	1.9	2
53	Molecular characterization of near-complete trisomy 17p syndrome from inverted duplication in association with cryptic deletion of 17pter. *Gene*, **2014**, 537, 343-7	3.8	2
52	Clinical application of catalytically cleavable fluorescence probe technology for multiplexing quantification of BCR-ABL1 fusion transcripts. *Clinica Chimica Acta*, **2014**, 428, 72-76	6.2	2
51	Analysis of acute myeloid leukemia in Korean patients with sole trisomy 6. *Annals of Laboratory Medicine*, **2014**, 34, 402-4	3.1	2

50	Changes in DNA methylation after 6-week exercise training in colorectal cancer survivors: A preliminary study. *Asia-Pacific Journal of Clinical Oncology*, **2020**,	1.9	2
49	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	4.9	2
48	Analysis of BRCA1/2 variants of unknown significance in the prospective Korean Hereditary Breast Cancer study. *Scientific Reports*, **2021**, 11, 8485	4.9	2
47	A Child With Lymphangioma Due to Somatic Mutation in PIK3CA Successfully Treated With Everolimus. *Pediatric Neurology*, **2019**, 91, 65-67	2.9	2
46	Newborn hereditary elliptocytosis confirmed by familial genetic testing. *International Journal of Laboratory Hematology*, **2020**, 42, e20-e22	2.5	2
45	Differential genetic diagnoses of adult post-lingual hearing loss according to the audiogram pattern and novel candidate gene evaluation. *Human Genetics*, **2021**, 1	6.3	2
44	Associations of single nucleotide polymorphisms related to insulin resistance with polycystic ovary syndrome. *Annals of Clinical and Laboratory Science*, **2014**, 44, 277-82	0.9	2
43	Deletion of 20p13 and Duplication of 20p13p12.3 in a Patient with Delayed Speech and Development. *Annals of Laboratory Medicine*, **2018**, 38, 77-79	3.1	1
42	Simultaneous occurrence of angioimmunoblastic T-cell lymphoma and plasma cell leukemia. *Annals of Laboratory Medicine*, **2015**, 35, 149-51	3.1	1
41	Molecular detection of tyrosine hydroxylase in the peripheral blood of patients with neuroblastoma: useful at diagnosis but not predictive of subsequent relapse during off-therapy follow-up. *Pediatric Hematology and Oncology*, **2011**, 28, 16-23	1.7	1
40	The Role of Ion Channel-Related Genes in Autism Spectrum Disorder: A Study Using Next-Generation Sequencing. *Frontiers in Genetics*, **2021**, 12, 595934	4.5	1
39	M323V Syndrome Presents with Infantile Nystagmus. *Genes*, **2021**, 12,	4.2	1
38	Disparate treatment outcomes according to presence of pathogenic mutations in West syndrome. *Epilepsia*, **2021**, 62, 1656-1664	6.4	1
37	Real-world data on the survival outcome of patients with newly diagnosed Waldenstrm macroglobulinemia. *Korean Journal of Internal Medicine*, **2021**, 36, 668-678	2.5	1
36	Prevalence of cancer susceptibility variants in patients with multiple Lynch syndrome related cancers. *Scientific Reports*, **2021**, 11, 14807	4.9	1
35	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, 191-3	3.1	1
34	Standards and Guidelines for Reporting Diagnostic Test Results in Acute Leukemia Patients: Bone Marrow Examination, Flow Cytometry, and Cytogenetic/Molecular Genetics Tests. *Laboratory Medicine Online*, **2021**, 11, 1-10	0.2	1
33	Eif2b3 mutants recapitulate phenotypes of vanishing white matter disease and validate novel disease alleles in zebrafish. *Human Molecular Genetics*, **2021**, 30, 331-342	5.6	1

32	DYNC2H1 variants cause Leber congenital amaurosis without syndromic features. *Clinical Genetics*, **2021**, 100, 111-113	4	1
31	Status of BRCA1/2 Genetic Testing Practices in Korea (2014). *Laboratory Medicine Online*, **2018**, 8, 107	0.2	1
30	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	1.6	1
29	NOTCH2 missplicing can occur in relation to apoptosis. *Blood*, **2015**, 126, 1731-2	2.2	0
28	A comparative study of next-generation sequencing and fragment analysis for the detection and allelic ratio determination of FLT3 internal tandem duplication.. *Diagnostic Pathology*, **2022**, 17, 14	3	0
27	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	1.6	0
26	Targeted next generation sequencing of circulating tumor DNA provides prognostic information for management in breast cancer patients.. *Annals of Translational Medicine*, **2022**, 10, 28	3.2	0
25	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*, **2021**, 59, 102629	1.6	0
24	Recurrent somatic mutations and low germline predisposition mutations in Korean ALL patients. *Scientific Reports*, **2021**, 11, 8893	4.9	0
23	Clinical Implementation of Targeted Gene Sequencing for Malformation of Cortical Development. *Pediatric Neurology*, **2020**, 103, 27-34	2.9	0
22	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	5.5	0
21	Application of CRISPR/Cas9-based mutant enrichment technique to improve the clinical sensitivity of plasma EGFR testing in patients with non-small cell lung cancer.. *Cancer Cell International*, **2022**, 22, 82	6.4	0
20	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	0.2	0
19	Cytogenetic testing by fluorescence in situ hybridization is improved by plasma cell sorting in multiple myeloma.. *Scientific Reports*, **2022**, 12, 8287	4.9	0
18	Clinical Significance of Revised Banff Criteria in the Diagnosis of Antibody-Mediated Rejection. *Transplantation Proceedings*, **2019**, 51, 1488-1490	1.1	
17	A case of CD5-positive mature B-cell neoplasm with t(10;14)(q24;q11.2) and trisomy 12. *Leukemia and Lymphoma*, **2015**, 56, 1550-1	1.9	
16	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	3	
15	Mutant Thr95Ile Transthyretin-Related Cardiac Amyloidosis With Polyneuropathy. *Circulation Journal*, **2019**, 83, 2328	2.9	

14	The first Korean case of childhood acute myeloid leukemia with inv(11)(p15q22)/NUP98-DDX10 rearrangement: a rare but recurrent genetic abnormality. *Annals of Laboratory Medicine*, **2014**, 34, 478-80	2.1
13	Secondary Germline CDKN2A Mutation Identified using Liquid Biopsy in a Patient with Esophageal Cancer. *Laboratory Medicine Online*, **2022**, 12, 63-67	0.2
12	Comprehensive Sequencing Identifies High Frequency of Copy Number Changes in Korean Patients with Acute Lymphoblastic Leukemia. *Blood*, **2018**, 132, 5166-5166	2.2
11	Diagnostic Challenge: Primary Bone Marrow Diffuse Large B-cell Lymphoma Mimicking Systemic Autoimmune Diseases. *Laboratory Medicine Online*, **2019**, 9, 242	0.2
10	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	3.1
9	Clinical Relevance of CD45+/CD19- Stem-like Tumor Cells in Patients with Mantle Cell Lymphoma: A Single Center Experience. *Blood*, **2014**, 124, 1622-1622	2.2
8	Characterization of the DNA Methylome during Human B-Cell Differentiation. *Blood*, **2014**, 124, 4346-4346	2.2
7	Report of the Korean Association of External Quality Assessment Service on Next-Generation Sequencing Analysis for Somatic Variants (2018–2020). *Journal of Laboratory Medicine and Quality Assurance*, **2021**, 43, 65-71	0.6
6	In Silico identification of a common mobile element insertion in exon 4 of RP1. *Scientific Reports*, **2021**, 11, 13381	4.9
5	A Case of Spontaneous Regression and Recurrence of Primary Vitreoretinal Lymphoma. *Ocular Immunology and Inflammation*, **2021**, 1-4	2.8
4	Genetic Characteristics of Polycythemia Vera and Essential Thrombocythemia in Korean Patients. *Journal of Clinical Laboratory Analysis*, **2016**, 30, 1061-1070	3
3	Low CtBP2 expression is associated with a stem cell-like signature and adverse clinical outcome in childhood B-cell lymphoblastic leukemia. *Leukemia*, **2021**, 35, 2684-2687	10.7
2	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	3.1
1	Status of Next-Generation Sequencing-Based Genetic Diagnosis in Hematologic Malignancies in Korea (2017-2018). *Laboratory Medicine Online*, **2021**, 11, 25-31	0.2