Chang-Seok Ki

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

531 papers	8,601 citations	42 h-index	64 g-index
555 ext. papers	9,893 ext. citations	3.8 avg, IF	5.88 L-index

#	Paper	IF	Citations
531	Prevalence of granular corneal dystrophy type 2-related p.R124H variant in a South Korean population. *Molecular Vision*, **2021**, 27, 283-287	2.3	
530	Development of Crohn's Disease in a Child With -related Congenital Chloride Diarrhea: Report of the First Case in East Asia and a Novel Missense Variant. *Annals of Laboratory Medicine*, **2021**, 41, 255-257	3.1	0
529	Carrier frequency and incidence estimation of Smith-Lemli-Opitz syndrome in East Asian populations by Genome Aggregation Database (gnomAD) based analysis. *Orphanet Journal of Rare Diseases*, **2021**, 16, 166	4.2	1
528	Metaphyseal Dysplasia Without Hypotrichosis Caused by RNA Component of Mitochondrial RNA-Processing Endoribonuclease () Gene Variants: The First Case in Korea. *Annals of Laboratory Medicine*, **2021**, 41, 346-349	3.1	
527	Case report of juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome: first report in Korea with a novel mutation in the gene. *Translational Pediatrics*, **2021**, 10, 1369-1376	4.2	1
526	A Novel Heterozygous Missense Variant Cluster in c.[5954C>G;6314C>T;6334C>T;6843G>C] causes a Coffin-Siris Syndrome. *Annals of Laboratory Medicine*, **2021**, 41, 350-353	3.1	3
525	Proposal of a New Prognostic Model for Differentiated Thyroid Cancer with Promoter Mutations. *Cancers*, **2021**, 13,	6.6	2
524	Genetic Analysis of Korean Adult Patients with Nontuberculous Mycobacteria Suspected of Primary Ciliary Dyskinesia Using Whole Exome Sequencing. *Yonsei Medical Journal*, **2021**, 62, 224-230	3	
523	Recent Advances in the Clinical Application of Next-Generation Sequencing. *Pediatric Gastroenterology, Hepatology and Nutrition*, **2021**, 24, 1-6	2.3	3
522	Comparison of Longitudinal Changes of Cerebral Small Vessel Disease Markers and Cognitive Function Between Subcortical Vascular Mild Cognitive Impairment With and Without Variant: A 5-Year Follow-Up Study. *Frontiers in Neurology*, **2021**, 12, 586366	4.1	
521	Promoter Mutations and the 8th Edition TNM Classification in Predicting the Survival of Thyroid Cancer Patients. *Cancers*, **2021**, 13,	6.6	7
520	Molecular classification of follicular thyroid carcinoma based on TERT promoter mutations. *Modern Pathology*, **2021**,	9.8	2
519	A nonsense variant in NME5 causes human primary ciliary dyskinesia with radial spoke defects. *Clinical Genetics*, **2020**, 98, 64-68	4	10
518	Determining the best candidates for next-generation sequencing-based gene panel for evaluation of early-onset epilepsy. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1376	2.3	7
517	Flow Cytometry for the Diagnosis of Primary Immunodeficiency Diseases: A Single Center Experience. *Allergy, Asthma and Immunology Research*, **2020**, 12, 292-305	5.3	5
516	PSEN1 variants in Korean patients with clinically suspicious early-onset familial Alzheimer's disease. *Scientific Reports*, **2020**, 10, 3480	4.9	3
515	Carrier Frequency of Spinal Muscular Atrophy in a Large-scale Korean Population. *Annals of Laboratory Medicine*, **2020**, 40, 326-330	3.1	1

514	De Novo SCN8A Pathogenic Variant (c.5630A>G; p.Asn1877Ser) Presenting with a Relatively Mild Phenotype. *Annals of Child Neurology*, **2020**, 28, 118-121	0.2	
513	The First Korean Case of De Novo Proximal 4p Deletion Syndrome in a Child With Developmental Delay. *Annals of Laboratory Medicine*, **2020**, 40, 435-437	3.1	
512	The First Korean Case of De Novo Proximal 4p Deletion Syndrome in a Child With Developmental Delay. *Annals of Laboratory Medicine*, **2020**, 40, 435-437	3.1	1
511	Identification of MECP2 Duplication Using Low-Depth Whole-Genome Sequencing-Based Copy Number Variation Analysis. *Laboratory Medicine Online*, **2020**, 10, 165	0.2	
510	Moyamoya Disease and Spectrums of RNF213 Vasculopathy. *Translational Stroke Research*, **2020**, 11, 580-589	7.8	28
509	Spontaneous Pisa syndrome in a patient with early-onset Alzheimer's disease. *Neurological Sciences*, **2020**, 41, 1297-1299	3.5	2
508	A Novel Heterozygous Missense Variant (c.667G>T;p.Gly223Cys) in That Interferes With Cadherin-Related 23 and Harmonin Interaction Causes Autosomal Dominant Nonsyndromic Hearing Loss. *Annals of Laboratory Medicine*, **2020**, 40, 224-231	3.1	3
507	The first familial case of inherited intellectual developmental disorder with dysmorphic facies and behavioral abnormalities (IDDFBA) with a novel FBXO11 variant. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2788-2792	2.5	1
506	mutations in ALS cause dysregulation of calcium homeostasis and stress granule dynamics. *Science Translational Medicine*, **2020**, 12,	17.5	12
505	Distribution and clinical impact of apolipoprotein E4 in subjective memory impairment and early mild cognitive impairment. *Scientific Reports*, **2020**, 10, 13365	4.9	3
504	Highly Sensitive and Specific Molecular Test for Mutations in the Diagnosis of Thyroid Nodules: A Prospective Study of -Prevalent Population. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	4
503	High prevalence of increased sitosterol levels in hypercholesterolemic children suggest underestimation of sitosterolemia incidence. *PLoS ONE*, **2020**, 15, e0238079	3.7	5
502	Analysis of dementia-related gene variants in APOE ε4 noncarrying Korean patients with early-onset Alzheimer's disease. *Neurobiology of Aging*, **2020**, 85, 155.e5-155.e8	5.6	6
501	Characterization of a novel LQT3 variant with a selective efficacy of mexiletine treatment. *Scientific Reports*, **2019**, 9, 12997	4.9	3
500	Multimodal imaging analyses in patients with genetic and sporadic forms of small vessel disease. *Scientific Reports*, **2019**, 9, 787	4.9	3
499	Is Cross-reactivity with Nontuberculous Mycobacteria a Systematic Problem in the Xpert MTB/RIF Assay?. *Tuberculosis and Respiratory Diseases*, **2019**, 82, 88-89	3.2	5
498	GenoType NTM-DR Performance Evaluation for Identification of Mycobacterium avium Complex and Mycobacterium abscessus and Determination of Clarithromycin and Amikacin Resistance. *Journal of Clinical Microbiology*, **2019**, 57,	9.7	19
497	Prevalence of fragile X-associated tremor/ataxia syndrome: A survey of essential tremor patients with cerebellar signs or extrapyramidal signs. *Brain and Behavior*, **2019**, 9, e01337	3.4	4

496	Challenges and Considerations in Sequence Variant Interpretation for Mendelian Disorders. *Annals of Laboratory Medicine*, **2019**, 39, 421-429	3.1	15
495	A novel SMAD6 variant in a patient with severely calcified bicuspid aortic valve and thoracic aortic aneurysm. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e620	2.3	6
494	Long-term natural history of non-cavitary nodular bronchiectatic nontuberculous mycobacterial pulmonary disease. *Respiratory Medicine*, **2019**, 151, 1-7	4.6	17
493	Ring Finger Protein 213 Variant and Plaque Characteristics, Vascular Remodeling, and Hemodynamics in Patients With Intracranial Atherosclerotic Stroke: A High-Resolution Magnetic Resonance Imaging and Hemodynamic Study. *Journal of the American Heart Association*, **2019**, 8, e011996	6	16
492	Phenotypic association of presence of a somatic GNAQ mutation with port-wine stain distribution in capillary malformation. *Head and Neck*, **2019**, 41, 4143-4150	4.2	2
491	De novo mutations in COL4A5 identified by whole exome sequencing in 2 girls with Alport syndrome in Korea. *Korean Journal of Pediatrics*, **2019**, 62, 193-197	2.4	4
490	A case ofFLNAgene mutation with respiratory insufficiency and periventricular heterotopia. *Allergy Asthma & Respiratory Disease*, **2019**, 7, 158	0.3	0
489	Mycobacterium shimoidei Pulmonary Disease: The First Case in Korea. *Laboratory Medicine Online*, **2019**, 9, 166	0.2	
488	A Case of Urinary Tract Infection Caused by the Emerging Uropathogen Actinotignum schaalii. *Laboratory Medicine Online*, **2019**, 9, 94	0.2	
487	Drug susceptibility patterns of Mycobacterium abscessus and Mycobacterium massiliense isolated from respiratory specimens. *Diagnostic Microbiology and Infectious Disease*, **2019**, 93, 107-111	2.9	16
486	Genetic variants of PARK genes in Korean patients with early-onset Parkinson's disease. *Neurobiology of Aging*, **2019**, 75, 224.e9-224.e15	5.6	15
485	Comparison between DiaPlexQ™STI6 and GeneFinder™STD I/STD II multiplex Real-time PCR Kits in the detection of six sexually transmitted disease pathogens. *Journal of Clinical Laboratory Analysis*, **2019**, 33, e22703	3	4
484	Comparative Evaluation Between the RealStar PCR Kit and the AmpliSens ()-FRT PCR Kit for Detecting in Non-HIV Immunocompromised Patients. *Annals of Laboratory Medicine*, **2019**, 39, 176-182	3.1	9
483	Compound Heterozygous Pathogenic Variants of the 15-Hydroxyprostaglandin Dehydrogenase Gene in a Patient With Hypertrophic Osteoarthropathy: First Case in Korea. *Annals of Laboratory Medicine*, **2019**, 39, 105-108	3.1	
482	Analysis of ATXN2 trinucleotide repeats in Korean patients with amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2018**, 67, 201.e5-201.e8	5.6	4
481	Comparación del efecto del aliskireno frente a controles negativos en la rigidez aórtica de los pacientes con síndrome de Marfan tratados con atenolol. *Revista Espanola De Cardiologia*, **2018**, 71, 743-749	1.5	2
480	Prospective monitoring of adenovirus infection and type analysis after allogeneic hematopoietic cell transplantation: A single-center study in Korea. *Transplant Infectious Disease*, **2018**, 20, e12885	2.7	6
479	Significance of circulating Epstein-Barr virus DNA monitoring after remission in patients with extranodal natural killer T cell lymphoma. *Annals of Hematology*, **2018**, 97, 1427-1436	3	5

478	The Impact of APOE e4 in Alzheimer's Disease Differs According to Age. *Journal of Alzheimer's Disease*, **2018**, 61, 1377-1385	4.3	7
477	Treatment outcomes of macrolide-susceptible Mycobacterium abscessus lung disease. *Diagnostic Microbiology and Infectious Disease*, **2018**, 90, 293-295	2.9	14
476	Frequency of hereditary neuropathy with liability to pressure palsies (HNPP) due to 17p11.2 deletion in a Korean newborn population. *Orphanet Journal of Rare Diseases*, **2018**, 13, 40	4.2	5
475	Nonsyndromic Peripheral Pulmonary Artery Stenosis Is Associated With Homozygosity of RNF213 p.Arg4810Lys Regardless of Co-occurrence of Moyamoya Disease. *Chest*, **2018**, 153, 404-413	5.3	27
474	Molecular genotyping of the non-invasive encapsulated follicular variant of papillary thyroid carcinoma. *Histopathology*, **2018**, 72, 648-661	7.3	42
473	Mutations in and in Moxifloxacin-Resistant Mycobacterium avium Complex and Mycobacterium abscessus Complex Clinical Isolates. *Antimicrobial Agents and Chemotherapy*, **2018**, 62,	5.9	9
472	First Molecular Diagnosis of a Patient with Unverricht-Lundborg Disease in Korea. *Yonsei Medical Journal*, **2018**, 59, 798-800	3	3
471	Repeated Intrathecal Mesenchymal Stem Cells for Amyotrophic Lateral Sclerosis. *Annals of Neurology*, **2018**, 84, 361-373	9.4	51
470	The Etiologies of Chronic Progressive Cerebellar Ataxia in a Korean Population. *Journal of Clinical Neurology (Korea*, **2018**, 14, 374-380	1.7	11
469	A Case of Cruoricaptor ignavus Isolated From the Blood of a Patient With Ewing Sarcoma. *Annals of Laboratory Medicine*, **2018**, 38, 613-615	3.1	
468	Nontuberculous Mycobacterial Lung Diseases Caused by Mixed Infection with Mycobacterium avium Complex and Mycobacterium abscessus Complex. *Antimicrobial Agents and Chemotherapy*, **2018**, 62,	5.9	9
467	Changing Epidemiology of Nontuberculous Mycobacterial Lung Diseases in a Tertiary Referral Hospital in Korea between 2001 and 2015. *Journal of Korean Medical Science*, **2018**, 33, e65	4.7	40
466	Development of Macrolide Resistance and Reinfection in Refractory Mycobacterium avium Complex Lung Disease. *American Journal of Respiratory and Critical Care Medicine*, **2018**, 198, 1322-1330	10.2	33
465	Malignant intercostal psammomatous melanotic schwannoma in a patient with Carney complex. *Korean Journal of Internal Medicine*, **2018**, 33, 1256-1257	2.5	0
464	Differences in drug susceptibility pattern between Mycobacterium avium and Mycobacterium intracellulare isolated in respiratory specimens. *Journal of Infection and Chemotherapy*, **2018**, 24, 315-318	2.2	22
463	Comparison of the Effect of Aliskiren Versus Negative Controls on Aortic Stiffness in Patients With Marfan Syndrome Under Treatment With Atenolol. *Revista Espanola De Cardiologia (English Ed)*, **2018**, 71, 743-749	0.7	1
462	Intermittent Antibiotic Therapy for Recurrent Nodular Bronchiectatic Mycobacterium avium Complex Lung Disease. *Antimicrobial Agents and Chemotherapy*, **2018**, 62,	5.9	10
461	Identification of Compound Heterozygous EYS Variants in a Korean Patient with Retinitis Pigmentosa. *Laboratory Medicine Online*, **2018**, 8, 66	0.2	1

460	A Variant Identified in a Sporadic Amyotrophic Lateral Sclerosis Patient Impairs Microtubule Stability and Axonal Mitochondria Distribution. *Experimental Neurobiology*, **2018**, 27, 550-563	4	7
459	Cav-1 (Caveolin-1) and Arterial Remodeling in Adult Moyamoya Disease. *Stroke*, **2018**, 49, 2597-2604	6.7	22
458	PITX2-related Axenfeld-Rieger Syndrome with a Novel Pathogenic Variant (c.475_476delCT). *Annals of Laboratory Medicine*, **2018**, 38, 283-286	3.1	
457	Bronchiectasis and Recurrent Respiratory Infections with a Gain-of-Function Variant: First Case in Korea. *Yonsei Medical Journal*, **2018**, 59, 1004-1007	3	6
456	Identification of Homozygous Likely Pathogenic Variant of ALDH3A2 in a Korean Boy with Sjgren-Larsson Syndrome. *Annals of Laboratory Medicine*, **2018**, 38, 80-82	3.1	2
455	Analysis of frontotemporal dementia, amyotrophic lateral sclerosis, and other dementia-related genes in 107 Korean patients with frontotemporal dementia. *Neurobiology of Aging*, **2018**, 72, 186.e1-186.e7	5.6	20
454	Identification of pathogenic variants in genes related to channelopathy and cardiomyopathy in Korean sudden cardiac arrest survivors. *Journal of Human Genetics*, **2017**, 62, 615-620	4.3	4
453	The relationship of 19 functional polymorphisms in iodothyronine deiodinase and psychological well-being in hypothyroid patients. *Endocrine*, **2017**, 57, 115-124	4	7
452	A novel likely pathogenic variant in the RAB28 gene in a Korean patient with cone-rod dystrophy. *Ophthalmic Genetics*, **2017**, 38, 587-589	1.2	8
451	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	9.7	17
450	Mycobacteriological characteristics and treatment outcomes in extrapulmonary Mycobacterium abscessus complex infections. *International Journal of Infectious Diseases*, **2017**, 60, 49-56	10.5	34
449	Ultrasonographic prediction of highly aggressive telomerase reverse transcriptase (TERT) promoter-mutated papillary thyroid cancer. *Endocrine*, **2017**, 57, 234-240	4	8
448	Liquid culture enhances diagnosis of patients with milder forms of non-tuberculous mycobacterial lung disease. *International Journal of Tuberculosis and Lung Disease*, **2017**, 21, 345-350	2.1	1
447	Atypical presentations of MERS-CoV infection in immunocompromised hosts. *Journal of Infection and Chemotherapy*, **2017**, 23, 769-773	2.2	30
446	Identification of a novel PML-RARG fusion in acute promyelocytic leukemia. *Leukemia*, **2017**, 31, 1992-1995	10.7	30
445	Distribution and clinical significance of Mycobacterium avium complex species isolated from respiratory specimens. *Diagnostic Microbiology and Infectious Disease*, **2017**, 88, 125-137	2.9	29
444	Performance evaluation of the Cobas TaqMan MTB assay on respiratory specimens according to clinical application. *International Journal of Infectious Diseases*, **2017**, 64, 42-46	10.5	2
443	Outcomes of complex lung disease based on clinical phenotype. *European Respiratory Journal*, **2017**, 50,	13.6	98

442	Reassessing the significance of the PAH c.158G>A (p.Arg53His) variant in patients with hyperphenylalaninemia. *Journal of Pediatric Endocrinology and Metabolism*, **2017**, 30, 1211-1218	1.6	7
441	Identification of Pathogenic Variants in the CHM Gene in Two Korean Patients With Choroideremia. *Annals of Laboratory Medicine*, **2017**, 37, 438-442	3.1	3
440	Clinical characteristics of pregnancies complicated by congenital myotonic dystrophy. *Obstetrics and Gynecology Science*, **2017**, 60, 323-328	1.9	7
439	Acromicric Dysplasia Caused by a Novel Heterozygous Mutation of FBN1 and Effects of Growth Hormone Treatment. *Annals of Laboratory Medicine*, **2017**, 37, 92-94	3.1	6
438	Identification of the PROM1 Mutation p.R373C in a Korean Patient With Autosomal Dominant Stargardt-like Macular Dystrophy. *Annals of Laboratory Medicine*, **2017**, 37, 536-539	3.1	7
437	HDR syndrome with a novel mutation in GATA3 mimicking a congenital X-linked stapes gusher: a case report. *BMC Medical Genetics*, **2017**, 18, 121	2.1	3
436	The Rap activator Gef26 regulates synaptic growth and neuronal survival via inhibition of BMP signaling. *Molecular Brain*, **2017**, 10, 62	4.5	5
435	A Korean Patient with Early Juvenile Form of Metachromatic Leukodystrophy: Biochemical and Molecular Genetic Investigation. *Laboratory Medicine Online*, **2017**, 7, 41	0.2	
434	Report on the Project for Establishment of the Standardized Korean Laboratory Terminology Database, 2015. *Journal of Korean Medical Science*, **2017**, 32, 695-699	4.7	5
433	PenicilliumSpecies Other ThanTalaromyces marneffeiProducing Red Pigment from Clinical Specimens: Isolation ofTalaromyces albobiverticillius. *Laboratory Medicine Online*, **2017**, 7, 211	0.2	2
432	Diverse Phenotypic Expression of Cardiomyopathies in a Family with TNNI3 p.Arg145Trp Mutation. *Korean Circulation Journal*, **2017**, 47, 270-277	2.2	4
431	First Report of Familial Dysalbuminemic Hyperthyroxinemia With an ALB Variant. *Annals of Laboratory Medicine*, **2017**, 37, 63-65	3.1	10
430	A Novel De Novo Pathogenic Variant in FOXF1 in a Newborn with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. *Yonsei Medical Journal*, **2017**, 58, 672-675	3	8
429	Novel Pathogenic Variant (c.580C>T) in the CPS1 Gene in a Newborn With Carbamoyl Phosphate Synthetase 1 Deficiency Identified by Whole Exome Sequencing. *Annals of Laboratory Medicine*, **2017**, 37, 58-62	3.1	7
428	Performance Evaluation of the PowerChek MERS (upE & ORF1a) Real-Time PCR Kit for the Detection of Middle East Respiratory Syndrome Coronavirus RNA. *Annals of Laboratory Medicine*, **2017**, 37, 494-498	3.1	10
427	Importance of Specimen Type and Quality in Diagnosing Middle East Respiratory Syndrome. *Annals of Laboratory Medicine*, **2017**, 37, 81-83	3.1	8
426	Laboratory Identification of Leptotrichia Species Isolated From Bacteremia Patients at a Single Institution. *Annals of Laboratory Medicine*, **2017**, 37, 272-276	3.1	5
425	Novel SLC37A4 Mutations in Korean Patients With Glycogen Storage Disease Ib. *Annals of Laboratory Medicine*, **2017**, 37, 261-266	3.1	14

424	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	3.1	4
423	Development of a One-Step Multiplex PCR Assay for Differential Detection of Major Mycobacterium Species. *Journal of Clinical Microbiology*, **2017**, 55, 2736-2751	9.7	21
422	Clinical, biochemical and molecular characterization of Korean patients with mucolipidosis II/III and successful prenatal diagnosis. *Orphanet Journal of Rare Diseases*, **2017**, 12, 11	4.2	13
421	Mycobacterial Characteristics and Treatment Outcomes in Mycobacterium abscessus Lung Disease. *Clinical Infectious Diseases*, **2017**, 64, 309-316	11.6	133
420	Clinical Characteristics and Treatment Outcomes of Patients with Macrolide-Resistant Mycobacterium massiliense Lung Disease. *Antimicrobial Agents and Chemotherapy*, **2017**, 61,	5.9	19
419	Genetic and functional analysis of TBK1 variants in Korean patients with sporadic amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2017**, 50, 170.e1-170.e6	5.6	16
418	Refining Dynamic Risk Stratification and Prognostic Groups for Differentiated Thyroid Cancer With TERT Promoter Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 1757-1764	5.6	28
417	Novel 4-bp Intronic Deletion (c.1560+5_1560+8del) [corrected] in LEMD3 in a Korean Patient With Osteopoikilosis. *Annals of Laboratory Medicine*, **2017**, 37, 540-543	3.1	
416	A Case of Antley-Bixler Syndrome With a Novel Likely Pathogenic Variant (c.529G>C) in the POR Gene. *Annals of Laboratory Medicine*, **2017**, 37, 559-562	3.1	5
415	Clinical Characteristics and Treatment Outcomes of Patients with Acquired Macrolide-Resistant Mycobacterium abscessus Lung Disease. *Antimicrobial Agents and Chemotherapy*, **2017**, 61,	5.9	27
414	Frequency and significance of rare RNF213 variants in patients with adult moyamoya disease. *PLoS ONE*, **2017**, 12, e0179689	3.7	8
413	Ultrasound and clinicopathological features of papillary thyroid carcinomas with BRAF and TERT promoter mutations. *Oncotarget*, **2017**, 8, 108946-108957	3.3	12
412	Resistance mechanisms of linezolid-nonsusceptible enterococci in Korea: low rate of 23S rRNA mutations in Enterococcus faecium. *Journal of Medical Microbiology*, **2017**, 66, 1730-1735	3.2	18
411	Caveolin-1, Ring finger protein 213, and endothelial function in Moyamoya disease. *International Journal of Stroke*, **2016**, 11, 999-1008	6.3	27
410	Clinical Characteristics, Treatment Outcomes, and Resistance Mutations Associated with Macrolide-Resistant Mycobacterium avium Complex Lung Disease. *Antimicrobial Agents and Chemotherapy*, **2016**, 60, 6758-6765	5.9	63
409	TERT promoter mutations and long-term survival in patients with thyroid cancer. *Endocrine-Related Cancer*, **2016**, 23, 813-23	5.7	53
408	Analytical and clinical evaluation of the Abbott RealTime hepatitis B sequencing assay. *Journal of Clinical Virology*, **2016**, 85, 27-30	14.5	1
407	Response to Biesecker. *Genetics in Medicine*, **2016**, 18, 417	8.1	

406	Congenital insensitivity to pain and anhydrosis due to a rare mutation and that is complicated by inflammatory bowel disease and amyloidosis: a case report. *Clinical Case Reports (discontinued)*, **2016**, 4, 997-1000	0.7	3
405	Clinical significance of smear positivity for acid-fast bacilli after 6 months of treatment in patients with drug-susceptible pulmonary tuberculosis. *Medicine (United States)*, **2016**, 95, e4540	1.8	6
404	Directly converted patient-specific induced neurons mirror the neuropathology of FUS with disrupted nuclear localization in amyotrophic lateral sclerosis. *Molecular Neurodegeneration*, **2016**, 11, 8	19	23
403	PHKA2 mutation spectrum in Korean patients with glycogen storage disease type IX: prevalence of deletion mutations. *BMC Medical Genetics*, **2016**, 17, 33	2.1	11
402	Evaluation of three real-time PCR assays for differential identification of Mycobacterium tuberculosis complex and nontuberculous mycobacteria species in liquid culture media. *Diagnostic Microbiology and Infectious Disease*, **2016**, 85, 186-91	2.9	13
401	Triage of patients with AUS/FLUS on thyroid cytopathology: effectiveness of the multimodal diagnostic techniques. *Cancer Medicine*, **2016**, 5, 769-77	4.8	21
400	Highly Concordant Key Genetic Alterations in Primary Tumors and Matched Distant Metastases in Differentiated Thyroid Cancer. *Thyroid*, **2016**, 26, 672-82	6.2	33
399	Epstein-Barr virus reactivation in extranodal natural killer/T-cell lymphoma patients: a previously unrecognized serious adverse event in a pilot study with romidepsin. *Annals of Oncology*, **2016**, 27, 508-13	10.3	41
398	Patient fibroblasts-derived induced neurons demonstrate autonomous neuronal defects in adult-onset Krabbe disease. *Oncotarget*, **2016**, 7, 74496-74509	3.3	16
397	CLEC4C p.K210del variant causes impaired cell surface transport in plasmacytoid dendritic cells of amyotrophic lateral sclerosis. *Oncotarget*, **2016**, 7, 24942-9	3.3	3
396	Comparative evaluation of the AdvanSure Mycobacteria GenoBlot assay and the GenoType Mycobacterium CM/AS assay for the identification of non-tuberculous mycobacteria. *Journal of Medical Microbiology*, **2016**, 65, 1422-1428	3.2	12
395	[Secondary publication] Sudden Aortic Rupture in Ehlers-Danlos Syndrome Type IV. *Korean Journal of Legal Medicine*, **2016**, 40, 61	0.2	
394	Delayed Diagnosis of Atypical Mowat-Wilson Syndrome. *Laboratory Medicine Online*, **2016**, 6, 57	0.2	
393	Survey of Clinical Laboratory Practices for 2015 Middle East Respiratory Syndrome Coronavirus Outbreak in the Republic of Korea. *Annals of Laboratory Medicine*, **2016**, 36, 154-61	3.1	7
392	Possible Transfusion-Related Acute Lung Injury Following Convalescent Plasma Transfusion in a Patient With Middle East Respiratory Syndrome. *Annals of Laboratory Medicine*, **2016**, 36, 393-5	3.1	35
391	Identification of mucorales from clinical specimens: a 4-year experience in a single institution. *Annals of Laboratory Medicine*, **2016**, 36, 60-3	3.1	16
390	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, 603-6	3.1	5
389	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, 203-8	3.1	9

388	Clinical Characteristics of Marfan Syndrome in Korea. *Korean Circulation Journal*, **2016**, 46, 841-845	2.2	9
387	Germline TP53 Mutation and Clinical Characteristics of Korean Patients With Li-Fraumeni Syndrome. *Annals of Laboratory Medicine*, **2016**, 36, 463-8	3.1	15
386	Guidelines for the Laboratory Diagnosis of Middle East Respiratory Syndrome Coronavirus in Korea. *Infection and Chemotherapy*, **2016**, 48, 61-9	3.9	8
385	Two Likely Pathogenic Variants of COL2A1 in Unrelated Korean Patients With Ocular-Only Variants of Stickler Syndrome: The First Molecular Diagnosis in Korea. *Annals of Laboratory Medicine*, **2016**, 36, 166-9	3.1	2
384	Analysis of intrapatient heterogeneity uncovers the microevolution of Middle East respiratory syndrome coronavirus. *Journal of Physical Education and Sports Management*, **2016**, 2, a001214	2.8	39
383	Clinical and endocrine characteristics and genetic analysis of Korean children with McCune-Albright syndrome: a retrospective cohort study. *Orphanet Journal of Rare Diseases*, **2016**, 11, 113	4.2	11
382	A Polymorphism in RNF213 Is a Susceptibility Gene for Intracranial Atherosclerosis. *PLoS ONE*, **2016**, 11, e0156607	3.7	40
381	Comparative Evaluation of Three Homogenization Methods for Isolating Middle East Respiratory Syndrome Coronavirus Nucleic Acids From Sputum Samples for Real-Time Reverse Transcription PCR. *Annals of Laboratory Medicine*, **2016**, 36, 457-62	3.1	19
380	Diffuse sclerosing variant of papillary thyroid carcinoma: major genetic alterations and prognostic implications. *Histopathology*, **2016**, 69, 45-53	7.3	32
379	MERS-CoV outbreak following a single patient exposure in an emergency room in South Korea: an epidemiological outbreak study. *Lancet, The*, **2016**, 388, 994-1001	40	205
378	Control of an Outbreak of Middle East Respiratory Syndrome in a Tertiary Hospital in Korea. *Annals of Internal Medicine*, **2016**, 165, 87-93	8	42
377	Identification of mutations in Korean patients with amyotrophic lateral sclerosis using multigene panel testing. *Neurobiology of Aging*, **2016**, 37, 209.e9-209.e16	5.6	21
376	Determinants of Basal Collaterals in Moyamoya Disease: Clinical and Genetic Factors. *European Neurology*, **2016**, 75, 178-85	2.1	7
375	Identification and characterization of NF1 splicing mutations in Korean patients with neurofibromatosis type 1. *Journal of Human Genetics*, **2016**, 61, 705-9	4.3	7
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