Sung Wook Park

List of Publications by Year
in descending order

188 papers	3,903 citations	172457 29 h-index	175258 52 g-index
196 all docs	196 docs citations	196 times ranked	7909 citing authors

#	Article	IF	Citations
1	Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a caseâ€“control study. Lancet Neurology, The, 2011, 10, 898-908.	10.2	294
2	A Single Recurrent Mutation in the 5â€²-UTR of IFITM5 Causes Osteogenesis Imperfecta Type V. American Journal of Human Genetics, 2012, 91, 343-348.	6.2	216
3	Viral Load Kinetics of MERS Coronavirus Infection. New England Journal of Medicine, 2016, 375, 1303-1305.	27.0	186
4	Association of DRD3 and GRIN2B with impulse control and related behaviors in Parkinson's disease. Movement Disorders, 2009, 24, 1803-1810.	3.9	148
5	Large-scale replication and heterogeneity in Parkinson disease genetic loci. Neurology, 2012, 79, 659-667.	1.1	119
6	Importance of Low-Range CAG Expansion and CAA Interruption in SCA2 Parkinsonism. Archives of Neurology, 2007, 64, 1510.	4.5	112
7	Sequential Analysis of Viral Load in a Neonate and Her Mother Infected With Severe Acute Respiratory Syndrome Coronavirus 2. Clinical Infectious Diseases, 2020, 71, 2236-2239.	5.8	111
8	A multi-centre clinico-genetic analysis of the VPS35 gene in Parkinson disease indicates reduced penetrance for disease-associated variants. Journal of Medical Genetics, 2012, 49, 721-726.	3.2	94
9	The Alu-Rich Genomic Architecture of SPAST Predisposes to Diverse and Functionally Distinct Disease-Associated CNV Alleles. American Journal of Human Genetics, 2014, 95, 143-161.	6.2	87
10	Molecular characterization of D- Korean persons: development of a diagnostic strategy. Transfusion, 2005, 45, 345-352.	1.6	85
11	Evidence of Severe Acute Respiratory Syndrome Coronavirus 2 Reinfection After Recovery from Mild Coronavirus Disease 2019. Clinical Infectious Diseases, 2021, 73, e3002-e3008.	5.8	68
12	Enteroviral meningitis without pleocytosis in children. Archives of Disease in Childhood, 2012, 97, 874-878.	1.9	62
13	Mitochondrial DNA C4171A/ND1 is a novel primary causative mutation of Leber's hereditary optic neuropathy with a good prognosis. Annals of Neurology, 2002, 51, 630-634.	5.3	56
14	The LRRK2 G2385R variant is a risk factor for sporadic Parkinson's disease in the Korean population. Parkinsonism and Related Disorders, 2010, 16, 85-88.	2.2	56
15	Interleukin-10 receptor mutations in children with neonatal-onset Crohnâ€™s disease and intractable ulcerating enterocolitis. European Journal of Gastroenterology and Hepatology, 2013, 25, 1.	1.6	50
16	Molecular Characterization ofFZD4,LRP5, andTSPAN12in Familial Exudative Vitreoretinopathy. , 2015, 56, 5143.		46
17	Genetic Mutation Profiles in Korean Patients with Inherited Retinal Diseases. Journal of Korean Medical Science, 2019, 34, e161.	2.5	44
18	Usefulness of a Rapid Real-time PCR Assay in Prenatal Screening for Group B Streptococcus Colonization. Annals of Laboratory Medicine, 2013, 33, 39-44.	2.5	43

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19	Viral RNA in Blood as Indicator of Severe Outcome in Middle East Respiratory Syndrome Coronavirus Infection. Emerging Infectious Diseases, 2016, 22, 1813-1816.	4.3	41
20	COQ6 Mutations in Children With Steroid-Resistant Focal Segmental Glomerulosclerosis and Sensorineural Hearing Loss. American Journal of Kidney Diseases, 2017, 70, 139-144.	1.9	40
21	The wide clinical spectrum and nigrostriatal dopaminergic damage in spinocerebellar ataxia type 6. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 529-532.	1.9	39
22	Clinical and Genetic Characteristics of Korean Occult Macular Dystrophy Patients. , 2013, 54, 4856.		37
23	The protective effect of LRRK2 p.R1398H on risk of Parkinson's disease is independent of MAPT and SNCA variants. Neurobiology of Aging, 2014, 35, 266.e5-266.e14.	3.1	36
24	Analysis of the Vaginal Microbiome by Next-Generation Sequencing and Evaluation of its Performance as a Clinical Diagnostic Tool in Vaginitis. Annals of Laboratory Medicine, 2016, 36, 441-449.	2.5	35
25	Consecutive analysis of mutation spectrum in the dystrophin gene of 507 Korean boys with Duchenne/Becker muscular dystrophy in a single center. Muscle and Nerve, 2017, 55, 727-734.	2.2	35
26	Clinical course of non-severe aplastic anemia in adults. International Journal of Hematology, 2010, 91, 770-775.	1.6	33
27	Microevolution of Outbreak-Associated Middle East Respiratory Syndrome Coronavirus, South Korea, 2015. Emerging Infectious Diseases, 2016, 22, 327-30.	4.3	33
28	Nightâ€time gastric acid suppression by tegoprazan compared to vonoprazan or esomeprazole. British Journal of Clinical Pharmacology, 2022, 88, 3288-3296.	2.4	33
29	Antituberculosis medication as a possible epigenetic factor of Leber's hereditary optic neuropathy. Clinical and Experimental Ophthalmology, 2010, 38, 363-366.	2.6	32
30	A variant of Desbuquois dysplasia characterized by advanced carpal bone age, short metacarpals, and elongated phalanges: Report of seven cases. American Journal of Medical Genetics, Part A, 2010, 152A, 875-885.	1.2	32
31	Respiratory Viral Infections after Hematopoietic Stem Cell Transplantation in Children. Journal of Korean Medical Science, 2013, 28, 36.	2.5	31
32	Chimerism Monitoring after Allogeneic Hematopoietic Stem Cell Transplantation Using Quantitative Real-Time PCR of Biallelic Insertion/Deletion Polymorphisms. Journal of Molecular Diagnostics, 2014, 16, 679-688.	2.8	31
33	VSX1 Gene and Keratoconus. Cornea, 2012, 31, 746-750.	1.7	30
34	Populationâ€specific frequencies for LRRK2 susceptibility variants in the genetic epidemiology of Parkinson's disease (GEOâ€PD) consortium. Movement Disorders, 2013, 28, 1740-1744.	3.9	30
35	Phenotype analysis in patients with early onset Parkinsonâ€™s disease with and without parkin mutations. Journal of Neurology, 2011, 258, 2260-2267.	3.6	29
36	Proteomic Profiling of Serum from Patients with Tuberculosis. Annals of Laboratory Medicine, 2014, 34, 345-353.	2.5	29

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37	Mitochondrial DNA Variant Discovery in Normal-Tension Glaucoma Patients by Next-Generation Sequencing. , 2014, 55, 986.		29
38	Efficient Molecular Genetic Diagnosis of Enlarged Vestibular Aqueducts in East Asians. Genetic Testing and Molecular Biomarkers, 2009, 13, 679-687.	0.7	28
39	Glutathione S-transferase A1 polymorphisms and acute graft-vs.-host disease in HLA-matched sibling allogeneic hematopoietic stem cell transplantation. Clinical Transplantation, 2007, 21, 207-213.	1.6	27
40	Spectrum of the mitochondrial DNA mutations of Leber's hereditary optic neuropathy in Koreans. Journal of Neurology, 2003, 250, 278-281.	3.6	26
41	Congenital subependymal giant cell astrocytoma: clinical considerations and expression of radial glial cell markers in giant cells. Child's Nervous System, 2008, 24, 1499-1503.	1.1	26
42	Infantile Vitreous Hemorrhage as the Initial Presentation of X-linked Juvenile Retinoschisis. Korean Journal of Ophthalmology: KJO, 2009, 23, 118.	1.1	26
43	A multi-institutional study of the prevalence of BRCA1 and BRCA2 large genomic rearrangements in familial breast cancer patients. BMC Cancer, 2014, 14, 645.	2.6	26
44	Monochorionic dizygotic twins with discordant sex and confined blood chimerism. European Journal of Pediatrics, 2014, 173, 1249-1252.	2.7	26
45	Longitudinal proteomic profiling provides insights into host response and proteome dynamics in COVIDâ€19 progression. Proteomics, 2021, 21, e2000278.	2.2	26
46	Reclassification of BRCA1 and BRCA2 variants of uncertain significance: a multifactorial analysis of multicentre prospective cohort. Journal of Medical Genetics, 2018, 55, 794-802.	3.2	25
47	Two Parkinson's disease patients with Î±-synuclein gene duplication and rapid cognitive decline. Movement Disorders, 2010, 25, 957-959.	3.9	23
48	Pitfalls of Multiple Ligation-Dependent Probe Amplifications in Detecting DMD Exon Deletions or Duplications. Journal of Molecular Diagnostics, 2016, 18, 253-259.	2.8	23
49	Hereditary hemolytic anemia in Korea from 2007 to 2011: A study by the Korean Hereditary Hemolytic Anemia Working Party of the Korean Society of Hematology. Blood Research, 2013, 48, 211.	1.3	22
50	Association between moyamoya syndrome and the RNF213 c.14576G>A variant in patients with neurofibromatosis Type 1. Journal of Neurosurgery: Pediatrics, 2016, 17, 717-722.	1.3	22
51	Low contribution of BRCA1/2 genomic rearrangement to high-risk breast cancer in the Korean population. Familial Cancer, 2009, 8, 505-508.	1.9	20
52	Relative contribution of SCA2, SCA3 and SCA17 in Korean patients with parkinsonism and ataxia. Parkinsonism and Related Disorders, 2011, 17, 338-342.	2.2	20
53	Revisit of multiple epiphyseal dysplasia: Ethnic difference in genotypes and comparison of radiographic features linked to the COMP and MATN3 genes. American Journal of Medical Genetics, Part A, 2011, 155, 2669-2680.	1.2	20
54	A case report and literature review of Fanconi Anemia (FA) diagnosed by genetic testing. Italian Journal of Pediatrics, 2015, 41, 38.	2.6	20

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55	Molecular basis and diagnosis of thalassemia. Blood Research, 2021, 56, S39-S43.	1.3	20
56	QT Prolongation and Life Threatening Ventricular Tachycardia in a Patient Injected With Intravenous Meperidine (DemerolÂ®). Korean Circulation Journal, 2011, 41, 342.	1.9	19
57	Clinical and mutational spectrum in Korean patients with Rubinsteinâ€“Taybi syndrome: The spectrum of brain MRI abnormalities. Brain and Development, 2015, 37, 402-408.	1.1	19
58	The Relation Between Endothelial Nitric Oxide Synthase Polymorphisms and Normal Tension Glaucoma. Journal of Glaucoma, 2017, 26, 1030-1035.	1.6	19
59	Long QT Syndrome and Dilated Cardiomyopathy with SCN5A p.R1193Q Polymorphism: Cardioverterâ€Defibrillator Implantation at 27 Months. PACE - Pacing and Clinical Electrophysiology, 2012, 35, e243-6.	1.2	18
60	Mitochondrial Dysfunction of Immortalized Human Adipose Tissue-Derived Mesenchymal Stromal Cells from Patients with Parkinsonâ€™s Disease. Experimental Neurobiology, 2013, 22, 283-300.	1.6	18
61	Parkin mutation and deep brain stimulation outcome. Journal of Clinical Neuroscience, 2014, 21, 107-110.	1.5	18
62	Genotype-phenotype analysis of von Hippel-Lindau syndrome in Korean families: HIF-Î± binding site missense mutations elevate age-specific risk for CNS hemangioblastoma. BMC Medical Genetics, 2016, 17, 48.	2.1	18
63	NovelCOL2A1Variant (c.619G>A, p.Gly207Arg) Manifesting as a Phenotype Similar to Progressive Pseudorheumatoid Dysplasia and Spondyloepiphyseal Dysplasia, Stanescu Type. Human Mutation, 2015, 36, 1004-1008.	2.5	17
64	Mutational spectrum of the SPAST and ATL1 genes in Korean patients with hereditary spastic paraplegia. Journal of the Neurological Sciences, 2015, 357, 167-172.	0.6	17
65	Diagnostic Application of an Extensive Gene Panel for Leber Congenital Amaurosis with Severe Genetic Heterogeneity. Journal of Molecular Diagnostics, 2015, 17, 100-105.	2.8	17
66	Clinical andABCB11profiles in Korean infants with progressive familial intrahepatic cholestasis. World Journal of Gastroenterology, 2016, 22, 4901.	3.3	17
67	Leber's hereditary optic neuropathy mutations in ethambutol-induced optic neuropathy. Journal of Neurology, 2003, 250, 87-89.	3.6	16
68	A novel double mutation in cis in MFN2 causes Charcotâ€“Marieâ€“Tooth neuropathy type 2A. Neurogenetics, 2012, 13, 275-280.	1.4	16
69	Sudden Cardiac Arrest during Anesthesia in a 30-Month-Old Boy with Syndactyly: A Case of Genetically Proven Timothy Syndrome. Journal of Korean Medical Science, 2013, 28, 788.	2.5	16
70	SCA2 family presenting as typical Parkinson's disease: 34 year follow up. Parkinsonism and Related Disorders, 2017, 40, 69-72.	2.2	16
71	Polymorphisms of the methylenetetrahydrofolate reductase gene and clinical outcomes in HLA-matched sibling allogeneic hematopoietic stem cell transplantation. Annals of Hematology, 2006, 86, 41-48.	1.8	15
72	Impact of vitamin D receptor gene polymorphisms on clinical outcomes of HLAâ€matched sibling hematopoietic stem cell transplantation. Clinical Transplantation, 2012, 26, 476-483.	1.6	15

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73	A mutation analysis of the AGL gene in Korean patients with glycogen storage disease type III. Journal of Human Genetics, 2014, 59, 42-45.	2.3	15
74	Hb variants in Korea: effect on HbA1c using five routine methods. Clinical Chemistry and Laboratory Medicine, 2017, 55, 1234-1242.	2.3	15
75	False Homozygous Deletions of SMN1 Exon 7 Using Dra I PCR-RFLP Caused by a Novel Mutation in Spinal Muscular Atrophy. Genetic Testing and Molecular Biomarkers, 2009, 13, 511-513.	0.7	14
76	SNPs in axon guidance pathway genes and susceptibility for Parkinsonâ€™s disease in the Korean population. Journal of Human Genetics, 2011, 56, 125-129.	2.3	14
77	Impact of cytokine gene polymorphisms on risk and treatment outcomes of aplastic anemia. Annals of Hematology, 2011, 90, 515-521.	1.8	14
78	The Korean undiagnosed diseases program: lessons from a one-year pilot project. Orphanet Journal of Rare Diseases, 2019, 14, 68.	2.7	14
79	Detailed analysis of phenotypes and genotypes in megalencephaly-capillary malformation-polymicrogyria syndrome caused by somatic mosaicism of PIK3CA mutations. Orphanet Journal of Rare Diseases, 2020, 15, 205.	2.7	14
80	Î²-THALASSEMIA IN THE KOREAN POPULATION. Hemoglobin, 2002, 26, 135-145.	0.8	13
81	Molecular and Clinical Characteristics of Myotonic Dystrophy Type 1 in Koreans. Annals of Laboratory Medicine, 2008, 28, 483-492.	2.5	13
82	Evaluation of Two Hepatitis C Virus Genotyping Assays Based on the 5â€² Untranslated Region (UTR): the Limitations of 5â€² UTR-Based Assays and the Need for a Supplementary Sequencing-Based Approach. Journal of Clinical Microbiology, 2012, 50, 3741-3743.	3.9	13
83	A multi-institutional study on the association between BRCA1/BRCA2 mutational status and triple-negative breast cancer in familial breast cancer patients. Breast Cancer Research and Treatment, 2014, 146, 63-69.	2.5	13
84	Screening for MAPT and PGRN mutations in Korean patients with PSP/CBS/FTD. Parkinsonism and Related Disorders, 2010, 16, 305-306.	2.2	12
85	Choline Acetyltransferase 2384G>A Polymorphism and the Risk of Alzheimer Disease. Alzheimer Disease and Associated Disorders, 2012, 26, 81-87.	1.3	12
86	First Case of Mycobacterium longobardum Infection. Annals of Laboratory Medicine, 2013, 33, 356-359.	2.5	12
87	Comparison of orthopaedic manifestations of multiple epiphyseal dysplasia caused by MATN3 versus COMP mutations: a case control study. BMC Musculoskeletal Disorders, 2014, 15, 84.	1.9	12
88	Case of mild Schmid-type metaphyseal chondrodysplasia with novel sequence variation involving an unusual mutational site of the COL10A1 gene. European Journal of Medical Genetics, 2015, 58, 175-179.	1.3	12
89	Effect of Next-Generation Exome Sequencing Depth for Discovery of Diagnostic Variants. Genomics and Informatics, 2015, 13, 31.	0.8	12
90	First Korean Case of Mycobacterium arupense Tenosynovitis. Annals of Laboratory Medicine, 2014, 34, 321-324.	2.5	11

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91	Large Deletions of TSPAN12 Cause Familial Exudative Vitreoretinopathy (FEVR). , 2016, 57, 6902.		11
92	Hereditary Fructose Intolerance Diagnosed in Adulthood. Gut and Liver, 2021, 15, 142-145.	2.9	11
93	A Case of Brain Abscess Caused by Propionibacterium acnes 13 Months after Neurosurgery and Confirmed by 16S rRNA Gene Sequencing. Annals of Laboratory Medicine, 2011, 31, 122-126.	2.5	10
94	Polymorphisms in Genes That Regulate Cyclosporine Metabolism Affect Cyclosporine Blood Levels and Clinical Outcomes in Patients Who Receive Allogeneic Hematopoietic Stem Cell Transplantation. Biology of Blood and Marrow Transplantation, 2012, 18, 37-43.	2.0	10
95	External Quality Assessment of MERS-CoV Molecular Diagnostics During the 2015 Korean Outbreak. Annals of Laboratory Medicine, 2016, 36, 230-234.	2.5	10
96	Spinocerebellar ataxia type 2 in seven Korean families: CAG trinucleotide expansion and clinical characteristics. Journal of Korean Medical Science, 1999, 14, 659.	2.5	9
97	The First Korean Case of Lysinuric Protein Intolerance: Presented with Short Stature and Increased Somnolence. Journal of Korean Medical Science, 2012, 27, 961.	2.5	9
98	A Case Report of Fanconi Anemia Diagnosed by Genetic Testing Followed by Prenatal Diagnosis. Annals of Laboratory Medicine, 2012, 32, 380-384.	2.5	9
99	Identification of Two Novel NPM1 Mutations in Patients with Acute Myeloid Leukemia. Annals of Laboratory Medicine, 2013, 33, 60-64.	2.5	9
100	Clinical Characteristics of Pediatric Thalassemia in Korea: A Single Institute Experience. Journal of Korean Medical Science, 2013, 28, 1645.	2.5	9
101	The Impact of Methylenetetrahydrofolate Reductase C677T Polymorphism on Patients Undergoing Allogeneic Hematopoietic Stem Cell Transplantation with Methotrexate Prophylaxis. PLoS ONE, 2016, 11, e0163998.	2.5	9
102	Evaluation of a Real-Time Reverse Transcription-PCR (RT-PCR) Assay for Detection of Middle East Respiratory Syndrome Coronavirus (MERS-CoV) in Clinical Samples from an Outbreak in South Korea in 2015. Journal of Clinical Microbiology, 2017, 55, 2554-2555.	3.9	9
103	Establishment of Pediatric Reference Intervals for Routine Laboratory Tests in Korean Population: A Retrospective Multicenter Analysis. Annals of Laboratory Medicine, 2021, 41, 155-170.	2.5	9
104	Comparison of the analytical and clinical performances of Abbott RealTime High Risk HPV, Hybrid Capture 2, and DNA Chip assays in gynecology patients. Diagnostic Microbiology and Infectious Disease, 2013, 76, 432-436.	1.8	8
105	Mutational analysis of paediatric patients with tuberous sclerosis complex in Korea: genotype and epilepsy. Epileptic Disorders, 2014, 16, 449-455.	1.3	8
106	Genetic Polymorphisms in Autophagyâ€Associated Genes in Korean Children With Earlyâ€Onset Crohn Disease. Journal of Pediatric Gastroenterology and Nutrition, 2015, 61, 285-291.	1.8	8
107	Whole Exome Sequencing Identifies Novel Genetic Alterations in Patients with Pheochromocytoma/Paraganglioma. Endocrinology and Metabolism, 2020, 35, 909-917.	3.0	8
108	A KOREAN FAMILY WITH A DOMINANTLY INHERITED Î²-THALASSEMIA DUE TO Hb DURHAM-N.C./BRESCIA [Î²114(G16)Leuâ†'Pro]. Hemoglobin, 2001, 25, 79-89.	0.8	7

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109	LCA5, a Rare Genetic Cause of Leber Congenital Amaurosis in Koreans. Ophthalmic Genetics, 2009, 30, 54-55.	1.2	7
110	Molecular Characterization of the NF2 Gene in Korean Patients with Neurofibromatosis Type 2: A Report of Four Novel Mutations. Annals of Laboratory Medicine, 2010, 30, 190-194.	2.5	7
111	Comparison of Modified Multiple-locus Variable-number Tandem-repeat Fingerprinting with Pulsed-field Gel Electrophoresis for Typing Clinical Isolates of Staphylococcus aureus. Annals of Laboratory Medicine, 2012, 32, 50-56.	2.5	7
112	Characteristics of hereditary nonpolyposis colorectal cancer patients with double primary cancers in endometrium and colorectum. Obstetrics and Gynecology Science, 2015, 58, 112.	1.6	7
113	Genotyping Influenza Virus by Next-Generation Deep Sequencing in Clinical Specimens. Annals of Laboratory Medicine, 2016, 36, 255-258.	2.5	7
114	Diverse Phenotypic Expression of Cardiomyopathies in a Family with TNNI3 p.Arg145Trp Mutation. Korean Circulation Journal, 2017, 47, 270.	1.9	7
115	Spectrum ofMNX1Pathogenic Variants and Associated Clinical Features in Korean Patients with Currarino Syndrome. Annals of Laboratory Medicine, 2018, 38, 242-248.	2.5	7
116	Comparative genomics of Mycoplasma pneumoniae isolated from children with pneumonia: South Korea, 2010â€“2016. BMC Genomics, 2019, 20, 910.	2.8	7
117	Rates of Coinfection Between SARS-CoV-2 and Other Respiratory Viruses in Korea. Annals of Laboratory Medicine, 2022, 42, 110-112.	2.5	7
118	Genotypic profile and phenotype correlations of -associated retinopathy in Koreans. Molecular Vision, 2019, 25, 679-690.	1.1	7
119	Predicted pathogenic missense mutation of PGRN found in a normal control. Annals of Neurology, 2010, 67, 415-416.	5.3	6
120	Childhood Brugada Syndrome in Two Korean Families. Korean Circulation Journal, 2010, 40, 143.	1.9	6
121	False Homozygosity Results in HLA Genotyping due to Loss of Chromosome 6 in a Patient with Acute Lymphoblastic Leukemia. Annals of Laboratory Medicine, 2011, 31, 302-306.	2.5	6
122	A Case Report of a Male Patient With Hb Hammersmith [Î²42(CD1)Pheâ†'Ser, TTT>TCT]. Hemoglobin, 2012, 36, 161-165.	0.8	6
123	A Case of Near Total Aortic Replacement in an Adolescent With Loeys-Dietz Syndrome. Korean Circulation Journal, 2012, 42, 288.	1.9	6
124	RANTES Polymorphisms and the Risk of Graft-versus-Host Disease in Human Leukocyte Antigen-Matched Sibling Allogeneic Hematopoietic Stem Cell Transplantation. Acta Haematologica, 2013, 129, 137-145.	1.4	6
125	Absence of CHN1 in two patients with a bilateral absence of cranial nerves IV and VI. Graefe's Archive for Clinical and Experimental Ophthalmology, 2015, 253, 491-492.	1.9	6
126	A Unique Mutational Spectrum of MLC1 in Korean Patients With Megalencephalic Leukoencephalopathy With Subcortical Cysts: p.Ala275Asp Founder Mutation and Maternal Uniparental Disomy of Chromosome 22. Annals of Laboratory Medicine, 2017, 37, 516-521.	2.5	6

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127	Ophthalmoplegia Diagnosis. Ophthalmology, 2009, 116, 813-814.e2.	5.2	5
128	Jervell and Lange-Nielsen Syndrome: Novel Compound Heterozygous Mutations in theKCNQ1in a Korean Family. Journal of Korean Medical Science, 2010, 25, 1522.	2.5	5
129	Glutaric Aciduria Type 1 in Korea: Report of Two Novel Mutations. Journal of Korean Medical Science, 2010, 25, 957.	2.5	5
130	Molecular identification of the novel Gî³-î² hybrid hemoglobin: Hb Gî³-î² Ulsan (Gî³ through 13; î² from 19). Blood Cells, Molecules, and Diseases, 2010, 45, 276-279.	1.4	5
131	A Novel Mutation of the TAZ Gene in Barth Syndrome: Acute Exacerbation after Contrast-Dye Injection. Journal of Korean Medical Science, 2013, 28, 784.	2.5	5
132	Novel mutation in the ATL1 with autosomal dominant hereditary spastic paraplegia presented as dysautonomia. Autonomic Neuroscience: Basic and Clinical, 2014, 185, 141-143.	2.8	5
133	Noninvasive prenatal test of single-gene disorders by linked-read direct haplotyping: application in various diseases. European Journal of Human Genetics, 2021, 29, 463-470.	2.8	5
134	Leukemic stem cell phenotype is associated with mutational profile in acute myeloid leukemia. Korean Journal of Internal Medicine, 2021, 36, 401-412.	1.7	5
135	Young-Onset Parkinsonâ€™s Disease with Impulse Control Disorder Due to Novel Variants of F-Box Only Protein 7. Journal of Movement Disorders, 2020, 13, 225-228.	1.3	5
136	Consistent count regionâ€“copy number variation (CCR-CNV): an expandable and robust tool for clinical diagnosis of copy number variation at the exon level using next-generation sequencing data. Genetics in Medicine, 2022, 24, 663-672.	2.4	5
137	Identification of a GDF5 Mutation in a Korean Patient with Brachydactyly Type C without Foot Involvement. Annals of Laboratory Medicine, 2013, 33, 150-152.	2.5	4
138	Biochemical and Genetic Analysis of Seven Korean Individuals With Suspected Metachromatic Leukodystrophy. Annals of Laboratory Medicine, 2015, 35, 458-462.	2.5	4
139	Menkes disease in Korea: ATP7A mutation and epilepsy phenotype. Brain and Development, 2015, 37, 223-229.	1.1	4
140	Identification of a Novel De Novo Mutation of the TAZ Gene in a Korean Patient with Barth Syndrome. Journal of Cardiovascular Imaging, 2016, 24, 153.	0.8	4
141	Bietti Crystalline Retinopathy Confirmed by Mutation of CYP4V2 Gene in a Korean Patient. Korean Journal of Ophthalmology: KJO, 2016, 30, 81.	1.1	4
142	Novel MT-ND5 gene mutation identified in Leber's hereditary optic neuropathy patient using mitochondrial genome sequencing. Journal of the Neurological Sciences, 2017, 375, 301-303.	0.6	4
143	Application of Multigene Panel Sequencing in Patients with Prolonged Rate-corrected QT Interval and No Pathogenic Variants Detected in KCNQ1, KCNH2, and SCN5A. Annals of Laboratory Medicine, 2018, 38, 54-58.	2.5	4
144	Clinical and genetic profiling of nevoid basal cell carcinoma syndrome in Korean patients by whole-exome sequencing. Scientific Reports, 2021, 11, 1163.	3.3	4

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145	Patient HSP70â€hom TG haplotype is associated with decreased transplantâ€related mortality and improved survival after sibling HLAâ€matched hematopoietic stem cell transplantation. Clinical Transplantation, 2010, 24, 459-466.	1.6	3
146	Brain Imaging Studies in Leber's Congenital Amaurosis: New Radiologic Findings Associated with the Complex Trait. Korean Journal of Ophthalmology: KJO, 2010, 24, 360.	1.1	3
147	Detection of very large off-ladder alleles at the PentaE locus in a 15 locus autosomal STR database of 199 Korean individuals. Forensic Science International: Genetics, 2012, 6, e189-e191.	3.1	3
148	Successful birth with preimplantation genetic diagnosis using single-cell allele-specific PCR and sequencing in a woman with hypochondroplasia due toFGFR3mutation (c.1620C>A, p.N540K). Clinical and Experimental Reproductive Medicine, 2013, 40, 42.	1.5	3
149	MtDNA m.3472T > C could be classified as a primary mutation of Leber's hereditary optic neuropathy. Journal of the Neurological Sciences, 2017, 380, 174-176.	0.6	3
150	Patients diagnosed with long QT syndrome after repair of congenital heart disease. PACE - Pacing and Clinical Electrophysiology, 2018, 41, 1435-1440.	1.2	3
151	Evaluation of the new Abbott Real-Time EBV assay: fully automated quantification of EBV in whole blood by targeting BLLF1. Diagnostic Microbiology and Infectious Disease, 2019, 94, 135-139.	1.8	3
152	Accuracy and Performance Evaluation of Triplet Repeat Primed PCR as an Alternative to Conventional Diagnostic Methods for Fragile X Syndrome. Annals of Laboratory Medicine, 2021, 41, 394-400.	2.5	3
153	Determination of Clinical Characteristics of Mycobacterium kansasii-Derived Species by Reanalysis of Isolates Formerly Reported as M. kansasii. Annals of Laboratory Medicine, 2021, 41, 463-468.	2.5	3
154	Congenital Stationary Night Blindness due to Novel TRPM1 Gene Mutations in a Korean Patient. Korean Journal of Ophthalmology: KJO, 2020, 34, 170.	1.1	3
155	Retinitis Pigmentosa Associated with Bardet-Biedl Syndrome with BBS9 Gene Mutation in a Korean Patient. Korean Journal of Ophthalmology: KJO, 2020, 34, 94.	1.1	3
156	SnackNTM: An Open-Source Software for Sanger Sequencing-based Identification of Nontuberculous Mycobacterial Species. Annals of Laboratory Medicine, 2022, 42, 213-248.	2.5	3
157	Inherited protein S deficiency due to a novel nonsense mutation in the PROS1 gene in the patient with recurrent vascular access thrombosis: A case report. Kidney Research and Clinical Practice, 2012, 31, 72-75.	2.2	2
158	The First Study on Nucleotide-level Identification of Hb Koriyama in a Patient with Severe Hemolytic Anemia. Annals of Laboratory Medicine, 2012, 32, 99-101.	2.5	2
159	Hereditary Spastic Paraplegia with a Novel SPAST Mutation Misdiagnosed with Subacute Combined Degeneration. Experimental Neurobiology, 2013, 22, 128-131.	1.6	2
160	Direct Haplotyping-Based Noninvasive Prenatal Test for Myotonic Dystrophy Type 1 with Large CTG Expansion. Clinical Chemistry, 2020, 66, 614-615.	3.2	2
161	Evaluation of the AccuPowerÂ® RV1 Real-Time RT-PCR Kit and the AccuPowerÂ® RV1 Multiplex Kit for SARS-CoV-2 and Influenza Virus Detection. Laboratory Medicine Online, 2021, 11, 290-296.	0.2	2
162	Clinical Utility of Methylation-Specific Multiplex Ligation-Dependent Probe Amplification for the Diagnosis of Praderâ€“Willi Syndrome and Angelman Syndrome. Annals of Laboratory Medicine, 2022, 42, 79-88.	2.5	2

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