Mi-Ae Jang

List of Publications by Year
in descending order

57	642	687363 13	642732 23
papers	citations	h-index	g-index

59	59	59	1561
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in DDX58, which Encodes RIG-I, Cause Atypical Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 266-274.	6.2	169
2	Distribution of Nontuberculous Mycobacteria by Multigene Sequence-Based Typing and Clinical Significance of Isolated Strains. Journal of Clinical Microbiology, 2014, 52, 1207-1212.	3.9	46
3	Frequency and spectrum of actionable pathogenic secondary findings in 196 Korean exomes. Genetics in Medicine, 2015, 17, 1007-1011.	2.4	41
4	Challenges and Considerations in Sequence Variant Interpretation for Mendelian Disorders. Annals of Laboratory Medicine, 2019, 39, 421-429.	2.5	31
5	Recent insights regarding the molecular basis of myeloproliferative neoplasms. Korean Journal of Internal Medicine, 2020, 35, 1-11.	1.7	25
6	Association of CFTR gene variants with nontuberculous mycobacterial lung disease in a Korean population with a low prevalence of cystic fibrosis. Journal of Human Genetics, 2013, 58, 298-303.	2.3	23
7	Chronic lymphocytic leukemia in Korean patients: frequent atypical immunophenotype and relatively aggressive clinical behavior. International Journal of Hematology, 2013, 97, 403-408.	1.6	19
8	Frequency of the moyamoya-related RNF213 p.Arg4810Lys variant in 1,516 Korean individuals. BMC Medical Genetics, 2015, 16, 109.	2.1	19
9	The t(11;14)(q13;q32) Translocation as a Poor Prognostic Parameter for Autologous Stem Cell Transplantation in Myeloma Patients With Extramedullary Plasmacytoma. Clinical Lymphoma, Myeloma and Leukemia, 2015, 15, 227-235.	0.4	19
10	Effect of Accreditation on Accuracy of Diagnostic Tests in Medical Laboratories. Annals of Laboratory Medicine, 2017, 37, 213-222.	2.5	16
11	Frequency and significance of rare RNF213 variants in patients with adult moyamoya disease. PLoS ONE, 2017, 12, e0179689.	2.5	15
12	Identification of TSC1 and TSC2 Mutations in Korean Patients With Tuberous Sclerosis Complex. Pediatric Neurology, 2012, 46, 222-224.	2.1	14
13	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, 238-241.	2.5	13
14	Identification of a Novel De Novo Variant in the PAX3 Gene in Waardenburg Syndrome by Diagnostic Exome Sequencing: The First Molecular Diagnosis in Korea. Annals of Laboratory Medicine, 2015, 35, 362-365.	2.5	10
15	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
16	A NovelDe NovoPathogenic Variant inFOXF1in a Newborn with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. Yonsei Medical Journal, 2017, 58, 672.	2.2	10
17	Clinical Utility of a Diagnostic Approach to Detect Genetic Abnormalities in Multiple Myeloma: A Single Institution Experience. Annals of Laboratory Medicine, 2018, 38, 196-203.	2.5	10
18	Identification of DCX Gene Mutation in Lissencephaly Spectrum With Subcortical Band Heterotopia Using Whole Exome Sequencing. Pediatric Neurology, 2013, 48, 411-414.	2.1	9

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19	Dose Estimation Curves Following In Vitro X-ray Irradiation Using Blood From Four Healthy Korean Individuals. Annals of Laboratory Medicine, 2019, 39, 91-95.	2.5	9
20	A Novel de novo Mutation in the G6PD Gene in a Korean Boy with Glucose-6-phosphate Dehydrogenase Deficiency: Case Report. Annals of Clinical and Laboratory Science, 2015, 45, 446-8.	0.2	9
21	Isolation and Identification of Geosmithia argillacea from a Fungal Ball in the Lung of a Tuberculosis Patient. Annals of Laboratory Medicine, 2013, 33, 136-140.	2.5	8
22	Gardnerella vaginalis in Recurrent Urinary Tract Infection Is Associated with Dysbiosis of the Bladder Microbiome. Journal of Clinical Medicine, 2022, 11, 2295.	2.4	8
23	Novel Mutations in CEBPA in Korean Patients with Acute Myeloid Leukemia with a Normal Karyotype. Annals of Laboratory Medicine, 2012, 32, 153-157.	2.5	7
24	Asp58Ala is the Predominant Mutation of the TTR Gene in Korean Patients with Hereditary Transthyretinâ€Related Amyloidosis. Annals of Human Genetics, 2015, 79, 99-107.	0.8	7
25	Diverse Phenotypic Expression of Cardiomyopathies in a Family with TNNI3 p.Arg145Trp Mutation. Korean Circulation Journal, 2017, 47, 270.	1.9	7
26	Novel Pathogenic Variant (c.3178G>A) in the SMC1A Gene in a Family With Cornelia de Lange Syndrome Identified by Exome Sequencing. Annals of Laboratory Medicine, 2015, 35, 639-642.	2.5	6
27	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
28	Clinical impact of CD5 expression in Korean patients with diffuse large B-cell lymphoma. Blood Research, 2017, 52, 193.	1.3	6
29	A Rare Case of Essential Thrombocythemia with Coexisting JAK2 and MPL Driver Mutations. Journal of Korean Medical Science, 2020, 35, e168.	2.5	6
30	Sequence variation data ofF8andF9genes in functionally validated control individuals: implications on the molecular diagnosis of hemophilia. Blood Research, 2013, 48, 206.	1.3	5
31	Evaluation of Performance of the Real-Q NTM-ID Kit for Rapid Identification of Eight Nontuberculous Mycobacterial Species. Journal of Clinical Microbiology, 2014, 52, 4053-4055.	3.9	5
32	Reporting Quality of Diagnostic Accuracy Studies in Laboratory Medicine: Adherence to Standards for Reporting of Diagnostic Accuracy Studies (STARD) 2015. Annals of Laboratory Medicine, 2020, 40, 245-252.	2.5	5
33	Rare Cases of PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome in a Korean Family Identified by Next Generation Sequencing. Journal of Korean Medical Science, 2020, 35, e96.	2.5	5
34	A novel splice-site mutation c.42-2A>T (IVS1-2A>T) of SERPINC1 in a Korean family with inherited antithrombin deficiency. Blood Coagulation and Fibrinolysis, 2011, 22, 742-745.	1.0	4
35	A Novel Mutation (c.200T>C) in the NAGLU Gene of a Korean Patient with copolysaccharidosis IIIB. Annals of Laboratory Medicine, 2013, 33, 221-224.	2.5	4
36	Novel and recurrent mutations in the F13A1 gene in unrelated Korean patients with congenital factor XIII deficiency. Blood Coagulation and Fibrinolysis, 2015, 26, 46-49.	1.0	4

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37	A novelBTKgene mutation, c.82delC (p.Arg28 Alafs[*]5), in a Korean family with X-linked agammaglobulinemia. Korean Journal of Pediatrics, 2016, 59, S49.	1.9	4
38	Extra X Chromosome in Mosaic Klinefelter Syndrome Is Associated with a Hematologic Malignancy. Annals of Laboratory Medicine, 2013, 33, 297-299.	2.5	3
39	First Report on Familial Hemophagocytic Lymphohistiocytosis with an Abnormal Immunophenotype and T Cell Monoclonality in Korea. Annals of Laboratory Medicine, 2015, 35, 155-158.	2.5	3
40	Identification of p.Glu131Lys Mutation in the IHH Gene in a Korean Patient With Brachydactyly Type A1. Annals of Laboratory Medicine, 2015, 35, 387-389.	2.5	3
41	A novel nonsense mutation Tyr301* of PROS1 causing protein S deficiency. Blood Coagulation and Fibrinolysis, 2015, 26, 223-224.	1.0	3
42	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, 166-169.	2.5	3
43	WNT16 elevation induced cell senescence of osteoblasts in ankylosing spondylitis. Arthritis Research and Therapy, 2021, 23, 301.	3.5	3
44	Effect of the Standardization of Diagnostic Tests on the Prevalence of Diabetes Mellitus and Impaired Fasting Glucose. Journal of Korean Medical Science, 2018, 33, e81.	2.5	2
45	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.	2.5	2
46	Clinical Manifestations and Genetic Analysis of 5 Korean Choroideremia Patients Initially Diagnosed With Retinitis Pigmentosa. Journal of Korean Medical Science, 2022, 37, e5.	2.5	2
47	Identification of PRODH mutations in Korean neonates with type I hyperprolinemia. Annals of Clinical and Laboratory Science, 2013, 43, 31-6.	0.2	2
48	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, 478-480.	2.5	1
49	Simultaneous Occurrence of Angioimmunoblastic T-cell Lymphoma and Plasma Cell Leukemia. Annals of Laboratory Medicine, 2015, 35, 149-151.	2.5	1
50	A unique case of dendritic cell neoplasm from monocyte-derived myeloid origin with distinct immunophenotype and cytomorphology. Annals of Hematology, 2014, 93, 877-878.	1.8	0
51	Response to Biesecker. Genetics in Medicine, 2016, 18, 417.	2.4	0
52	Correspondence: Response to â€œEvaluating the Cumulative Impact of Ionizing Radiation Exposure With Diagnostic Geneticsâ€. Annals of Laboratory Medicine, 2019, 39, 419-420.	2.5	0
53	Distinctive Severe Ocular Abnormalities and Epilepsy Accompanied by a Novel ZEB2 Mutation in a Child with Mowat-Wilson Syndrome. Annals of Child Neurology, 0, , .	0.1	0
54	Clinical utility of comprehensive approach to detect genetic abnormalities in multiple myeloma.. Journal of Clinical Oncology, 2016, 34, 8034-8034.	1.6	0

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55	A Novel Pathogenic Variant (c.592_599del) in PCDH19 in a Korean Family with Epilepsy. Annals of Child Neurology, 2019, 27, 152-154.	0.1	0
56	Identification of MECP2 Duplication Using Low-Depth Whole-Genome Sequencing-Based Copy Number Variation Analysis. Laboratory Medicine Online, 2020, 10, 165.	0.2	0
57	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.	2.5	0