

Mi-Ae Jang

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5534278/publications.pdf>

Version: 2024-02-01

57
papers

642
citations

687363

13
h-index

642732

23
g-index

59
all docs

59
docs citations

59
times ranked

1561
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical Manifestations and Genetic Analysis of 5 Korean Choroideremia Patients Initially Diagnosed With Retinitis Pigmentosa. <i>Journal of Korean Medical Science</i> , 2022, 37, e5.	2.5	2
2	<i>Gardnerella vaginalis</i> in Recurrent Urinary Tract Infection Is Associated with Dysbiosis of the Bladder Microbiome. <i>Journal of Clinical Medicine</i> , 2022, 11, 2295.	2.4	8
3	WNT16 elevation induced cell senescence of osteoblasts in ankylosing spondylitis. <i>Arthritis Research and Therapy</i> , 2021, 23, 301.	3.5	3
4	Reporting Quality of Diagnostic Accuracy Studies in Laboratory Medicine: Adherence to Standards for Reporting of Diagnostic Accuracy Studies (STARD) 2015. <i>Annals of Laboratory Medicine</i> , 2020, 40, 245-252.	2.5	5
5	A Rare Case of Essential Thrombocythemia with Coexisting <i>JAK2</i> and <i>MPL</i> Driver Mutations. <i>Journal of Korean Medical Science</i> , 2020, 35, e168.	2.5	6
6	Rare Cases of <i>PLOD1</i> -Related Kyphoscoliotic Ehlers-Danlos Syndrome in a Korean Family Identified by Next Generation Sequencing. <i>Journal of Korean Medical Science</i> , 2020, 35, e96.	2.5	5
7	Recent insights regarding the molecular basis of myeloproliferative neoplasms. <i>Korean Journal of Internal Medicine</i> , 2020, 35, 1-11.	1.7	25
8	Identification of <i>MECP2</i> Duplication Using Low-Depth Whole-Genome Sequencing-Based Copy Number Variation Analysis. <i>Laboratory Medicine Online</i> , 2020, 10, 165.	0.2	0
9	The First Korean Case of De Novo Proximal 4p Deletion Syndrome in a Child With Developmental Delay. <i>Annals of Laboratory Medicine</i> , 2020, 40, 435-437.	2.5	0
10	The First Korean Case of De Novo Proximal 4p Deletion Syndrome in a Child With Developmental Delay. <i>Annals of Laboratory Medicine</i> , 2020, 40, 435-437.	2.5	2
11	Challenges and Considerations in Sequence Variant Interpretation for Mendelian Disorders. <i>Annals of Laboratory Medicine</i> , 2019, 39, 421-429.	2.5	31
12	Correspondence: Response to "Evaluating the Cumulative Impact of Ionizing Radiation Exposure With Diagnostic Genetics". <i>Annals of Laboratory Medicine</i> , 2019, 39, 419-420.	2.5	0
13	Dose Estimation Curves Following In Vitro X-ray Irradiation Using Blood From Four Healthy Korean Individuals. <i>Annals of Laboratory Medicine</i> , 2019, 39, 91-95.	2.5	9
14	A Novel Pathogenic Variant (c.592_599del) in <i>PCDH19</i> in a Korean Family with Epilepsy. <i>Annals of Child Neurology</i> , 2019, 27, 152-154.	0.1	0
15	Clinical Utility of a Diagnostic Approach to Detect Genetic Abnormalities in Multiple Myeloma: A Single Institution Experience. <i>Annals of Laboratory Medicine</i> , 2018, 38, 196-203.	2.5	10
16	Effect of the Standardization of Diagnostic Tests on the Prevalence of Diabetes Mellitus and Impaired Fasting Glucose. <i>Journal of Korean Medical Science</i> , 2018, 33, e81.	2.5	2
17	Effect of Accreditation on Accuracy of Diagnostic Tests in Medical Laboratories. <i>Annals of Laboratory Medicine</i> , 2017, 37, 213-222.	2.5	16
18	Clinical impact of CD5 expression in Korean patients with diffuse large B-cell lymphoma. <i>Blood Research</i> , 2017, 52, 193.	1.3	6

#	ARTICLE	IF	CITATIONS
19	Diverse Phenotypic Expression of Cardiomyopathies in a Family with TNNI3 p.Arg145Trp Mutation. Korean Circulation Journal, 2017, 47, 270.	1.9	7
20	A Novel <i>De Novo</i> Pathogenic Variant in <i>FOXF1</i> in a Newborn with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. Yonsei Medical Journal, 2017, 58, 672.	2.2	10
21	Frequency and significance of rare RNF213 variants in patients with adult moyamoya disease. PLoS ONE, 2017, 12, e0179689.	2.5	15
22	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
23	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
24	Response to Biesecker. Genetics in Medicine, 2016, 18, 417.	2.4	0
25	A novel <i>BTK</i> gene mutation, c.82delC (p.Arg28 Alafs [*]), in a Korean family with X-linked agammaglobulinemia. Korean Journal of Pediatrics, 2016, 59, S49.	1.9	4
26	Clinical utility of comprehensive approach to detect genetic abnormalities in multiple myeloma.. Journal of Clinical Oncology, 2016, 34, 8034-8034.	1.6	0
27	Frequency of the moyamoya-related RNF213 p.Arg4810Lys variant in 1,516 Korean individuals. BMC Medical Genetics, 2015, 16, 109.	2.1	19
28	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
29	Novel Pathogenic Variant (c.3178G>A) in the <i>SMC1A</i> Gene in a Family With Cornelia de Lange Syndrome Identified by Exome Sequencing. Annals of Laboratory Medicine, 2015, 35, 639-642.	2.5	6
30	Simultaneous Occurrence of Angioimmunoblastic T-cell Lymphoma and Plasma Cell Leukemia. Annals of Laboratory Medicine, 2015, 35, 149-151.	2.5	1
31	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
32	Identification of a Novel <i>De Novo</i> Variant in the <i>PAX3</i> 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
33	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
34	Asp58Ala is the Predominant Mutation of the <i>TTR</i> Gene in Korean Patients with Hereditary Transthyretin-Related Amyloidosis. Annals of Human Genetics, 2015, 79, 99-107.	0.8	7
35	Mutations in DDX58, which Encodes RIG-I, Cause Atypical Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 266-274.	6.2	169
36	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

#	ARTICLE	IF	CITATIONS
37	A novel nonsense mutation Tyr301* of PROS1 causing protein S deficiency. Blood Coagulation and Fibrinolysis, 2015, 26, 223-224.	1.0	3
38	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
39	Frequency and spectrum of actionable pathogenic secondary findings in 196 Korean exomes. Genetics in Medicine, 2015, 17, 1007-1011.	2.4	41
40	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
41	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
42	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
43	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
44	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
45	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
46	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
47	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
48	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
49	Extra X Chromosome in Mosaic Klinefelter Syndrome Is Associated with a Hematologic Malignancy. Annals of Laboratory Medicine, 2013, 33, 297-299.	2.5	3
50	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
51	Sequence variation data of F8 and F9 genes in functionally validated control individuals: implications on the molecular diagnosis of hemophilia. Blood Research, 2013, 48, 206.	1.3	5
52	Identification of PRODH mutations in Korean neonates with type I hyperprolinemia. Annals of Clinical and Laboratory Science, 2013, 43, 31-6.	0.2	2
53	Identification of TSC1 and TSC2 Mutations in Korean Patients With Tuberous Sclerosis Complex. Pediatric Neurology, 2012, 46, 222-224.	2.1	14
54	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

#	ARTICLE	IF	CITATIONS
55	Identification of a Rare 3 bp BRAF Gene Deletion in a Thyroid Nodule by Mutant Enrichment with 3'-Modified Oligonucleotides Polymerase Chain Reaction. <i>Annals of Laboratory Medicine</i> , 2012, 32, 238-241.	2.5	13
56	A novel splice-site mutation c.42-2A>T (IVS1-2A>T) of SERPINC1 in a Korean family with inherited antithrombin deficiency. <i>Blood Coagulation and Fibrinolysis</i> , 2011, 22, 742-745.	1.0	4
57	Distinctive Severe Ocular Abnormalities and Epilepsy Accompanied by a Novel <i>ZEB2</i> Mutation in a Child with Mowat-Wilson Syndrome. <i>Annals of Child Neurology</i> , 0, , .	0.1	0